

**Living with Leber Hereditary Optic
Neuropathy: Exploring Experiences and
Perceptions of a Disruptive Mitochondrial
Condition**

Lydia Harper

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School of Social Sciences, Cardiff University

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Abstract

This thesis explores the experiences and perceptions of people living with Leber hereditary optic neuropathy (LHON) and the healthcare professionals charged with diagnosing and treating the condition. LHON is the first disease linked to a mitochondrial mutation, characteristically resulting in bilateral sight loss over a period of 6–12 weeks from the initial onset and predominantly (but not exclusively) affecting young men in their teens and early twenties. As with other mitochondrial conditions, there is currently no cure for LHON, and treatment options to slow the progress of the condition are limited. Qualitative research exploring the effect of LHON following a sudden and dramatic loss of sight has been absent from the literature. Addressing this gap, my study involves: (1) semi-structured interviews (N=41) with affected men and women, mothers who carry the condition, and genetic ophthalmologists; (2) participant observation over a period of nine months in two genetic ophthalmology clinics located in UK hospitals. Drawing upon key theoretical and empirical contributions from medical sociology and beyond, such as the work of Bury (1982) and Charmaz (1983), I explore the past, present and future lives of people with LHON by describing their chronic illness trajectory. Drawing predominantly on the interview data, I document their experience of the initial symptoms of sight loss, the challenges of receiving a formal diagnosis (as an uncertain, contested and often misdiagnosed condition), the aftermath of receiving a genetic diagnosis for participants and their wider family, and the disruption to everyday, mundane moments in people’s daily lives. Moreover, I unpack how people restore their former self-images (Charmaz 1987, 1991), gain control over their lives, and regain some sense of ‘normality’ (Davis 1995), whilst also reflecting on future aspirations with respect to treatment options and reproductive imaginaries. To conclude, I acknowledge how my thesis contributes to knowledge by uncovering the multi-faceted experience of people living with sudden bilateral sight loss—a group who have, thus far, been invisible in the sociological literature.

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List of Abbreviations

CRN	Clinical Research Network
CT	Computerised tomography
CVI	Certificate of Visual Impairment
CVS	Chorionic villus sampling
EMA	European Medicines Agency
ERG	Electroretinography
LHON	Leber hereditary optic neuropathy
MST	Maternal spindle transfer
MRI	Magnetic resonance imaging
NHS	National Health Service
NICE	National Institute for Health and Care Excellence
NIHR	National Institute of Healthcare Research
OCT	Optical coherence tomography
PNT	Pro-nuclear transfer
REC	Research Ethics Committee
RGCs	Retinal ganglion cells
RNIB	Royal National Institute of Blind People
SpR	Specialist Registrar
VFT	Visual Fields Test
VI	Visual impairment

Chapter One: Introduction

This thesis provides an account of the experiences and perceptions of a group of people living with Leber hereditary optic neuropathy (LHON) and the healthcare professionals charged with diagnosing and treating the condition. LHON is a neurodegenerative disease characteristically resulting in bilateral sight loss over a period of 6–12 weeks from the initial onset, and predominantly (but not exclusively), affecting young males in their teens and early twenties. It is also one of the most common inherited optic neuropathies encountered in clinical practice (Abu-Amero 2011; Yu-Wai-Man *et al.* 2014).¹

LHON was originally suspected of being an X-linked² disease until Erickson (1972) suggested that the inheritance of the condition was compatible with maternal inheritance, indicating that LHON was an underlying mutation of mitochondrial DNA (mtDNA) (Yen *et al.* 2006). In 1988, Wallace *et al.* confirmed for the first time the link between mtDNA mutations and human disease, with LHON caused by a point mutation—namely m.11778G>A—within the mitochondrial genome (Wallace 1988). This was followed by the discovery in 1991 of the m.3460G>A mutation (Howell *et al.* 1991; Huoponen *et al.* 1991) and in 1992 the m.14484T>C mutation (Johns *et al.* 1992; Mackey and Howell 1992). Over 95 per cent of LHON cases have been identified as attributable to one of these pathogenic mutations and classed as a rare mitochondrial disease (i.e. affecting less than 5 in 10,000 of the general population Rare Disease UK 2016), with on average approximately 30 new cases a year across the UK (Moore and Burton 2008). However, this figure may be underestimated as the condition is often misdiagnosed in clinical practice (Moore

¹ In England and Wales, inherited optic neuropathies affect at least 1 in 10,000 of the population (Newman and Biousse 2004) and are a significant cause of certifiable blindness (Yu-Wai-Man *et al.* 2014,) accounting for approximately 20.2 of the Certificates of Visual Impairment (CVI) issued to adults (aged 16–64) (Liew *et al.* 2014).

² Females have two copies of the X chromosome, one of which will almost certainly be normal and, thus, compensate for the faulty chromosome. Males have one X chromosome and one Y chromosome, and so do not have the 'normal' X chromosome to compensate. As such men can develop X-linked conditions, such as, Duchenne muscular dystrophy, haemophilia and fragile X syndrome.

and Burton 2008). As with other mitochondrial conditions, there is currently no cure and treatment options to slow the progress of LHON are limited.

LHON displays a marked gender bias, and is characterised by incomplete penetrance, which means that not all carriers will lose their sight. Approximately 50 per cent of males and approximately 10 per cent of females who carry the mutation developing sight loss, typically between the ages of 15 and 35 (Newman and Biousse 2004; Yu-Wai-Man *et al.* 2014). Approximately 90 per cent of carriers will become symptomatic before the age of 50 (Spruit *et al.* 2006; Yu-Wai-Man and Chinnery 2011). However, there are reports of the condition occurring in children as young as two years old and in adults in their 70s and 80s (Chinnery *et al.* 2001; Newman and Biousse 2004; Yu-Wai-Man *et al.* 2011).

LHON is a disease that targets the optic disc (also referred to as the optic nerve head—which connects the eye to the brain) and the retina (the light-sensitive tissue lining the back of the eye), both of which are contained within the posterior segment of the eye (Appendix 1). The disease is characterised by the selective loss of retinal ganglion cells (which transmit visual information from the retina to the brain), leading to optic atrophy (death of cells) and blindness. In the pre-symptomatic phase, people may become aware of problems with colour vision (dyschromatopsia) along the red–green axis (Yu-Wai-Man *et al.* 2011). In the acute phase, people experience painless blurring of the central vision in one eye over a period of 6–12 weeks (Man *et al.* 2002; Hudson *et al.* 2007), with the fellow eye becoming affected, on average, within one year (Yu-Wai-Man *et al.* 2014). It is unusual for people with LHON to experience loss of vision in only one eye (Yu-Wai-Man and Chinnery 2011). The visual acuity (clarity of vision) at the point of maximum sight loss can range from only experiencing light perception, or only being able to see hand movements. In the majority of cases the sight loss is permanent, and people are registered as severely sight impaired (previously referred to as blind).

The genes that cause LHON can be found in the DNA of cellular structures, mitochondria which are the minute structures present in the cytoplasm (the gel-like substance enclosed within the cell membrane) of the cells in the human body and are responsible for converting enzymes (sugars and fats) into the energy required

for the cells to function efficiently. They have been described as the ‘powerhouse’ or ‘batteries’ of the cell (Nuffield Council on Bioethics 2012: 18). Mitochondrial disease is the umbrella term given to a diverse group of progressive and multisystem diseases which occur at any age (Haas 2007; Munnich *et al.* 1996). Women pass on mitochondrial DNA to their children via the mitochondria in their egg. Although men can inherit a mitochondrial disease, they cannot pass it onto their children. Mitochondrial diseases vary in severity, have the potential to cause chronic morbidity, and can prove fatal. Unpredictable in nature, the symptoms of mitochondrial diseases may vary between individuals, ranging from no apparent symptoms to life-threatening conditions.

This thesis

Only limited scholarship has considered the lived experience of people living with LHON, with the exception of three previous research studies, which adopted a psychological perspective, utilising quantitative (Kirkman *et al.* 2009b; Dator 2014) and qualitative research methods (Ferguson and de Abreu 2016). Kirkman *et al.*'s (2009b) research used questionnaires to assess the quality of life of LHON carriers from the UK, Netherlands and Germany. Dator's (2014) PhD thesis used an on-line survey to assess psychological distress experienced by 65 unaffected mothers and 52 unaffected siblings following the diagnosis of an affected relative. The third study, conducted by Ferguson and de Abreu (2016), used in-depth interviews to explore the lived experience of seven men following a LHON diagnosis. There are no sociological studies that have considered the impact of LHON. My thesis plugs this gap.

The aim of this thesis is to adopt qualitative research methods to gather rich, informative, textured data to explore the impact of LHON on individuals and how relationships within the family are re-negotiated following a LHON diagnosis. Using participant observation undertaken over a period of nine months in two genetic ophthalmology clinics located in UK hospitals, and in-depth interviews (N=41), I unpack the experience of children and young people living with sight loss as a result of LHON; adults who have experienced sight loss later in life; mothers who carry the LHON mutation and who have given birth to a child who subsequently loses their sight; and women who are considering their reproductive options knowing that, if

they conceive ‘naturally’, they will pass LHON on to their children. I also uncover the experiences of visually impaired mothers bringing up sighted children.

This thesis is located within a long tradition of sociological scholarship including key theoretical and empirical contributions from medical sociology and beyond, such as the work of Bury (1982), Charmaz (1983, 1987) and Goffman (1959, 1968), which has explored individuals’ subjective experience of living with chronic illness. Grounded in key contributions of how chronic illness is an assault on the integrity of the body and the self (Frank 1995; Charmaz 1983, 1987), I explore participants’ narratives to highlight the impact of sudden sight loss on their sense of self and how this amounts to a ‘biographical disruption’, that is, a ‘disruption of taken-for-granted assumptions and behaviours’ followed by a ‘re-thinking of a person’s biography and self-concept’ (Bury 1982: 168). Adopting Bury’s conceptual framework I seek to understand the stories of individuals—as ‘wounded storytellers’ in body and voice (Frank 1996: xi) — and how they account to others for their sight loss. During in-depth interviews I facilitate participants in uncovering their past, explicating their present and imagining their future. Illness narratives, identified by Frank (1995), resonate with participants in this study: (1) restitution (returning to the former self); (2) chaos (trying to make sense of the illness and not envisaging returning to normal health), and; (3) quest (striving to achieve a new self).

Similar to Frank’s observations, participants in this study move through a number of stages commencing with the pre-diagnosis—where symptoms are first experienced, and the quest for a diagnosis begins. Often, the diagnosis is not forthcoming. LHON is frequently misdiagnosed by ophthalmologists who have not previously encountered the condition in clinical practice, relegating participants to the status of what Jeffrey (1979: 90) refers to as ‘normal rubbish’. Patients are judged by healthcare professionals, in such instances, to be time-wasters or attention seekers who fabricate their symptoms. Failure to attach a label or give a name to their symptom’s often means that they become ‘medical orphans’ (Aronowitz 2001: 803) and are often denied entry to the Parsonian (1951) ‘sick role’, thus being deprived of the opportunity to access welfare benefits, employment and educational support.

In the months which follow the diagnosis, participants strive to achieve a sense of equilibrium. Experiencing biographical disruption, they seek to make sense of their altered position, including questioning why this has happened to them. It is in this stage that they are no longer able to undertake taken-for-granted mundane tasks, such as making a cup of tea, reading a newspaper or using their mobile phone. However, with the passing of time, some participants move into the process of narrative reconstruction (Williams 1984) and, through biographical repair work, they commence the process of 'reconstructing the self'. By adopting coping strategies to restore their former self-images (Charmaz 1987: 296), and become what Frank (1995: 62) refers to as 'successfully ill', participants see themselves arriving at the point of moving on with their lives.

In the final stage of participants' sight loss, the majority of them look to the future. Irrespective of age or gender, they claim that they have the same aspirations as others —such as having a successful career, being financially independent, buying their first home, getting married, and having children. However, not all participants reach this point. Some continue their quest to find answers for their sight loss and, by adopting narratives of hope, they invest emotionally and, in some cases, financially in finding new treatment options to cure their sight loss. I am not suggesting that participants move through the stages in a linear fashion; some move between the stages mirroring the wider sight loss literature. Indeed, over time they moved back and forth, experiencing what Charmaz (1991: 51) characterises as 'good days' and 'bad days'.

Based on the aims of this research, and a review of the sight loss and chronic illness literature, this thesis addresses the following broad research question:

What are the subjective experiences and perceptions of people affected by LHON, and how are relationships within the family renegotiated following the diagnosis?

The following subsidiary questions are addressed in the thesis:

1. What are the challenges presented to genetic ophthalmologists in the diagnosis and treatment of LHON?

2. To what extent does LHON disrupt an individual's biography and what are the implications for their identity and sense of self?
3. How do people with LHON imagine their future?
4. Are women influenced by their experiential knowledge of LHON when making reproductive decisions?

Thesis structure

This thesis comprises eight chapters. Having introduced my research in this opening chapter, in Chapter Two, I explore the previous sight loss literature which is primarily located within psychology and disability studies. LHON is a chronic condition as, although not terminal, it currently has no cure. Given the paucity of sociological scholarship exploring the experiences of living with sudden sight loss, I draw on the chronic illness literature, identifying key themes including uncertainty, biographical disruption (Bury 1982), loss of self (Charmaz 1983, 1987), and stigma (Goffman 1968), in order to ground my arguments.

Chapter Three outlines the methodological approach I adopted in addressing the research questions. I outline the process of obtaining NHS ethical and R&D approval to undertake data collection in two UK hospitals. I detail the practical difficulties I encountered in accessing an unco-operative site, and the impact that this had on my original intention to undertake an ethnographic study.

Chapter Four marks the beginning of the empirical chapters. I begin the chapter by describing how some participants initially ignored their symptoms. Medical attention was often sought only when participants were no longer able to undertake everyday mundane tasks. Drawing upon interview data and some participant observations, I unpack how a LHON diagnosis is accomplished in the eye clinic. Here, I recognise how LHON is the subject of diagnostic uncertainty and contestation as it is frequently misdiagnosed by ophthalmologists who have not yet encountered the condition in clinical practice.

Chapter Five considers the initial reactions of participants to sudden sight loss. Here I draw upon Bury's (1982, 1987, 1991) concept of biographical disruption to explore how following their diagnosis, participants cope with the emotional impact of experiencing sudden sight loss. A genetic diagnosis has repercussions for both the

immediate and extended family. I explore the complex moral dilemmas that arise when decisions are made to tell or withhold information from relatives who may also be at risk of developing LHON. Finally, I consider notions of guilt and self-blame (Arribas-Ayllon *et al.* 2008) experienced by mothers who were unaware of their carrier status when their children were conceived.

Chapter Six explores the longer-term implications of living with LHON. Once again, I draw upon Bury's conceptual framework of biographical disruption. Bury explicitly referred to biographical disruption as an event occurring at the *onset* of chronic illness, however, I suggest that his concept is relevant when exploring the longer-term consequences of living with LHON as participants reported experiencing repeated incidents of disruption *throughout* the trajectory of the condition, irrespective of the age of onset.

Chapter Seven looks to the future by considering participants' narratives of hope associated with the development of new treatment options, which are currently the subject of clinical trials, to restore sight. Drawing predominantly on interview data and some observational data, I locate my discussion within the sociology of hope (Petersen and Wilkinson 2014; Brown *et al.* 2015; Petersen 2015). This chapter is presented in two parts. In the first, I introduce the concept of hope and explore how participants invest both emotionally and, in some cases, financially in a new drug designed to restore sight. In the second part, I explore narratives of hope associated with the use of mitochondrial replacement techniques (MRTs) to avoid the birth of a child with LHON.

In Chapter Eight I conclude the thesis by further unpacking the findings of the empirical chapters. I reflect on the research questions posed in the introductory chapter and discuss to what extent I have answered them. I will discuss how my research addresses the gaps identified in previous LHON and wider sight loss studies. However, I will also discuss how my findings have highlighted the inadequacies of ophthalmology training, particularly in the diagnosis of LHON, which potentially have repercussions for the effectiveness of new treatment options. I will also refer to the variability in the provision of genetic counselling and

referrals to Local Authority Sensory Impairment Services in England and Wales.
Finally, I briefly outline suggestions for future research.

Chapter Two: Literature Review

Introduction

In the previous chapter, I explained that the aim of this thesis is to use qualitative research to explore the subjective experiences and perceptions of people living with Leber hereditary optic neuropathy (LHON). In this chapter, I consider the previous sight loss literature to provide context for the four empirical chapters that follow. Recently, the scientific literature reviewing the pathogenesis of LHON has increased considerably. This renewed interest is, in part, attributable to the promise of new treatments currently the subject of clinical trials in the UK, Europe and North America. While I discuss the small number of psychological studies exploring individual experiences of LHON, there have been, as yet, no research looking at LHON from a sociological perspective. Therefore, this review will draw on the wider sight loss literature located within medical sociology, psychology and disability studies. By bringing together contributions from the three disciplines, I unpack the depth and breadth of the individual experience of sight loss.

This chapter will be presented in three sections. In the first section, I consider the key findings from three research studies (located within psychology studies). The studies have adopted both quantitative (Kirkman *et al.* 2009b; Dator 2014) and qualitative (Ferguson and de Abreu 2016) research methods to explore the experiences of people affected by, or who are carriers of, the LHON mutation. In section two, I review the wider sight loss literature. LHON is characterised by sudden (bilateral) sight loss, presenting unique challenges to individuals. As such I therefore focus on research undertaken with people with *acquired* (as opposed to *congenital*) sight loss. Given the paucity of the current LHON sight loss literature, I include in this literature review, research exploring sudden traumatic sight loss experienced by ex-service personnel. In addition, I have taken the decision to include research that considers *progressive* eye conditions. My rationale for adopting this approach is that although the manner of the sight loss may vary, I would suggest that both groups ultimately face the same challenges in coping with their sight loss. Finally, drawing on Goffman's (1968) work on stigma together with

other contributions, I explore how stigmatising attitudes towards the visually impaired shape their decisions to disclose or conceal their disability.

It is evident from reviewing the sight loss literature, that the primary focus of this work has been on the 'medical model of disability', which depicts disability as 'an individual failing and a personal tragedy' (Barnes and Mercer 2010: 1). In focusing on the medical model, such research highlights symptoms, diagnosis and treatment, with scant regard given to the social model of disability which acknowledges that, by adopting disabling practices, it is society that marginalises those who are living with a disability (Thomas 2004, 2007; Scambler 2009; Barnes and Mercer 2010). In his research with visually impaired people living in New York, Ainlay (1989—cited in Green *et al.* 2002: 258) laments the 'sighted world that has systematically avoided the world of blindness and segregated those who experience it from mainstream life'.

In opposition to the dominance of the medical model, there is an evolving body of sociological scholarship which considers the challenges encountered by the visually impaired in their everyday interactions with sighted persons (Hetherington 1999, 2000; Schillmeier 2006, 2007a, 2007b, 2008; Måseide and Grøttland 2015). Måseide and Grøttland (2015: 594) note that, for the visually impaired, 'sharing physical and social space with the sighted may imply special challenges for blind persons with regard to interaction order and social identities'. Similarly, in his ethnographic research with the visually impaired, Schillmeier (2007a: 195) observes how 'ordinary acts of everyday life make up the complex and contingent scenarios of disabilities and create enabling and disabling (dis/abling) practices'.

This literature is situated alongside the chronic illness scholarship that explores the experience of individuals living with illness, disease and/or disability. The narratives of the participants in this thesis regarding the diagnosis and prognosis of LHON parallel key themes in the chronic illness scholarship including biographical disruption (Bury 1982), loss of self (Charmaz 1983), uncertainty (Fox 1957; Atkinson 1984, 1995), and stigmatisation (Goffman 1968). Over the next three sections I will draw upon these key theoretical and empirical contributions from sociology and the wider sight loss literature to explore the lived experience of individuals and their families following a LHON diagnosis.

Previous LHON research

There is limited qualitative research in sociology which explores the lived experience of LHON. In this section, I refer to three studies, that explore the experiences of people who are affected by or are carriers of the LHON mutation. Kirkman and colleagues (2009b) undertook a large scale quantitative study to evaluate the quality of life of LHON carriers (196 affected with sight loss and 206 unaffected carriers) from the UK, Netherlands and Germany. Telephone interviews were conducted using the visual function index (VF-14) questionnaire,³ with the findings concluding that LHON had a strong detrimental impact on the majority of the activities associated with daily living and quality of life. When compared with research undertaken with other inherited and acquired ophthalmic conditions, LHON was associated with the worst mean VF-14 score, attributable to the fact that the majority of the LHON participants had been issued with a certificate of visual impairment (CVI).⁴ The study also concluded that the VF-14 score did not worsen over time. The authors also refer to previous clinical research undertaken by Van Senu (1963) where he noted that of 352 participants diagnosed with LHON from 27 large Dutch families, nearly half of his participants' social and economic status dropped (resulting from loss of employment) following the onset of LHON.

The second study—a PhD thesis using quantitative research methods (Dator 2014)—examined the relationship between psychological well-being, social support and spirituality in unaffected carriers of LHON who have experience of living with an affected family member. The participants (65 mothers and 52 siblings) were invited to complete a demographic questionnaire and four validated assessments (Outcome Questionnaire-45,⁵ via an internet survey. Dator's findings suggest that both mothers and siblings experienced higher levels of psychological

³ The VF-14 Questionnaire measures the ability of individuals to perform 14 vision-dependent activities of daily living that are rated in terms of degree of difficulty, with a possible score of 5 for each question: 0 (unable to do), 1 (great deal of difficulty), 2 (moderate difficulty), 3 (little difficulty), and 4 (no difficulty) (Kirkman *et al.* 2009: 3113).

⁴ A Certificate of Vision Impairment (CVI) is (usually) completed by a consultant ophthalmologist and certifies that the patient has a visual impairment. The CVI also acts as a referral to Social Services who instigate a care assessment (RNIB 2017).

⁵The Outcome Questionnaire-45 (OQ-45) was developed by Lambert *et al.* (1996). The 45-item self-report instrument requires patients to rate their functioning on a 5-point Likert scale. The OQ-45 was designed to assess common symptoms across a wide range of adult mental disorders and syndromes, including stress-related illness.

stress than a normative sample, and siblings experienced significantly higher levels of interpersonal problems and social role dysfunction than a normative sample. The findings also indicated that mothers of affected children displayed significantly lower levels of psychological stress when perceiving that the levels of support from other family members had increased. Similarly, unaffected siblings with perceived higher levels of social support from other family members displayed significantly lower levels of psychological stress, interpersonal problems, and social role dysfunction.

The two studies potentially allow the findings to be generalisable to the wider LHON population. However, the use of standard instruments to assess quality of life and psychological functioning using pre-determined responses is of limited value when the aim of the research—as in my thesis—is to unpack the complex emotional responses experienced by individuals affected by LHON and their family members who are also at risk of losing their sight.

In contrast, a third (small) study on LHON (Ferguson and de Abreu 2016) used in-depth semi-structured interviews with seven affected men with an age range of 21–62 years. Focusing on their lived experience of LHON, the authors identified six key themes across the data, including the theme of ‘psycho-social losses’ (2016: 113), where the loss of vision is said to affect the individual’s ability to navigate in the world, resulting in irritation and decreased capabilities. Participants reported the loss of social/communication skills as they were unable to detect non-verbal communications, which led to feelings of frustration. The loss of independence and freedom featured strongly in the data, with participants again expressing their frustration when, for example, they boarded the wrong bus or train, and all of the participants expressing feelings of stress and frustration at their perceived loss of skill, ability, and independence.

The second theme, ‘attitudes and coping strategies’ (2016: 114), documents how participants adopt a pragmatic approach to visual impairment by developing various coping strategies, such as telephoning in advance of meeting to ensure that the participant arrived at the right place. Theme three explores the ‘development of practical skills’ (2016: 115), with participants explaining how they acquired new skills which enabled them to use the latest assistive technologies or learnt to touch

type on a computer. The fourth theme explored 'identity' (2016: 115), where all participants registered as visually impaired (registration is voluntary)—acknowledging their blind identity, and where participants who retain a good level of residual vision explained how this brought them into conflict with the public's perception that blind people live in total darkness. The authors suggest that this perception results in LHON participants' skills and abilities being overlooked when interacting with the sighted.

The fifth theme that the authors discussed was 'regaining independence' (2016: 116); participants adopted strategies dependent on the level of their residual vision to enable them to live and work independently. This included using large print on files to make them easier to locate at work, undertaking white cane training, learning new walking routes, and learning to use new assistive technologies. The final theme identified was that of 'recurrent loss' (2016: 117). The participants acknowledged that there are always moments which evoke a feeling of loss for their sight. Whilst individuals adopted practical and emotional coping mechanisms to enable them to lead a fulfilling life, they also recognised that they have fewer options in life as a result of their sight loss, which at times leads to a feeling of loss and frustration reminiscent of their early period of adjustment. Ferguson and de Abreu (2016) suggest that one of the fundamental findings from the research is that the process of adjustment has no conclusion; from time to time, the challenges and restrictions of visual impairment overwhelmed the coping mechanisms their participants had developed. Despite the insight offered into the experience of living with LHON, Ferguson and de Abreu's (2016: 120) study has a number of limitations. The authors acknowledge that their participants were active members of the LHON online community, the implication being that those who volunteered to be interviewed 'represent a sample of the population who have highly developed coping mechanisms allowing them to live a fulfilling life'. The study did not include affected women. I would suggest that including women in the research provides the opportunity to explore issues surrounding the inheritable nature of LHON and the factors that influence women's reproductive choices.

Sight loss

The majority of the current sight loss research is located in psychology and as such is mostly quantitative in nature, primarily using standard instruments to assess mood and social functioning (Boulton *et al.* 2006; Thurston 2010) and tends to focus on conditions that cause progressive sight loss, such as retinitis pigmentosa (Nemschick *et al.* 1986; Hayeems *et al.* 2005; Jangra *et al.* 2007; Bittner *et al.* 2010). More recently, some researchers acknowledging the inadequacy of quantitative research methods to explore the individual experience of visual impairment, have supplemented their data with in-depth semi-structured interviews (Stanford *et al.* 2009; Thurston *et al.* 2013).

Given the limited qualitative research exploring LHON, I unpack the literature exploring sudden traumatic sight loss experienced by ex-service personnel deployed in Iraq and Afghanistan (Stevelink *et al.* 2015a, 2015b; Stevelink and Fear 2016) and research exploring progressive sight loss including glaucoma (Green *et al.* 2002), diabetic retinopathy (Devenney and O'Neill 2011) and retinitis pigmentosa (Hayeems *et al.* 2005; Fourie 2007), to identify commonalities across acquired sight loss conditions. According to this work (Hewson 1997; Thurston *et al.* 2010), when an individual loses their sight, they experience a number of responses, including: *emotional responses* (e.g. shock particularly when the loss is sudden and unexpected, sadness, anger and guilt); *physical responses* (e.g. emptiness, headaches and exhaustion); *behavioural responses* (e.g. isolation, insomnia and crying), and; *cognitive responses* (e.g. denial and confusion).

As explained in Chapter One, LHON is a condition that predominantly affects young people at a particularly vulnerable time in their lives, as they move from childhood through adolescence, ultimately arriving at adulthood. Evidence suggests that childhood visual impairment has the potential to adversely affect educational attainment (Corn *et al.* 2002) and the ability to participate in activities that involve an element of physicality, such as playing ball games (Khadka *et al.* 2012), leading to social disadvantages, including having fewer friends (Cochrane *et al.* 2008) and opportunities to socialise and develop interpersonal skills (Huurre and Aro 2000). Adolescents with sight loss are said to be particularly susceptible to feelings of worry, and to experiencing problems in forming romantic relationships, close

friendships and being accepted by peer groups (Pinquart and Pfeiffer 2014). Indeed, the process of adjusting to sight loss is said to negatively impact a child's self-esteem. Research undertaken by Bowen (2010) with 60 children with visual impairment found that self-esteem plays a key role in determining life chances, academic success and mental and emotional health. Drawing upon the work of Maslow (1968), Bowen (2010: 47) claims that nurturing of self-esteem is one of the top five needs in the hierarchy of human requirements:

'[...] the commodity termed self-esteem can be described as the procedure by which individuals set a value on themselves, appreciate their own worth and recognise their attributes as a person, so they can achieve a quiet sense of self respect in the process.'

Research undertaken with visually impaired adults also suggests that acquired sight loss results in a loss of self-esteem (Lyons and Sullivan 1998) and independence (Horowitz 2003; Senra *et al.* 2011), affecting people's capacity to fully participate in everyday life (Lamoureux *et al.* 2004; Vu *et al.* 2005; Senra *et al.* 2011). The visually impaired are said to feel as if they are 'incompetent' (Dodds 1989: 11), 'useless' (Devenney and O'Neill (2011: 712) or to have reverted to 'child mode' (Southwell 2012: 111) when they attempt to undertake previously taken-for-granted tasks (for example, reading instructions, cooking a meal, applying makeup). This is because the tasks are impossible to accomplish without help from family, friends and the use of low vision aids (such as a white cane and magnifiers).

Experiencing feelings of helplessness and incompetence have also been linked to an inability to accept sight loss (Dodds 1989). The loss of independent living skills, resulting in social isolation and lower self-confidence, have previously been reported in the Network 1000 study (Douglas *et al.* 2006) and is a recurring theme in the literature (Burmedi *et al.* 2002; Percival *et al.* 2005). In their research with 900 adults in the UK with low vision, Bruce *et al.* (2007) explored perceptions of inclusion by family and friends in the context of social isolation. They concluded that the visually impaired are more likely than the population in general to feel a sense of exclusion as a result of a severe lack of social support. Participants in the study were more likely to be living alone, receive fewer visitors, have no hobbies, and go shopping less than once a month and on their own. Sight loss is said to negatively impact on emotional well-being (Thurston *et al.* 2013) and quality of life (Hassell *et*

al. 2006), resulting in increased emotional distress (Scott *et al.* 2001) anxiety, worry, suicidal thoughts (De Leo *et al.* 1999), and uncertainty about the future (Thurston *et al.* 2010; RNIB 2017a).

There is an established link between sight loss and depression (Horowitz and Reinhardt 2000; Burmedi *et al.* 2002; Desrosiers *et al.* 2009; Renauld *et al.* 2010), loneliness and mental health problems (Leamon *et al.* 2014). Individuals with sight loss are said to experience consequential comorbidities, including a higher risk of falls (Leamon *et al.* 2014) and an increased risk of mortality (Vu *et al.* 2005). A recurring theme within the sight loss literature is that of experiencing loss not only of sight but also of future plans (including working abroad), independence (which impacts on self-esteem) and hobbies and pastimes (Baus 1999; Thurston *et al.* 2010; RNIB 2017). Charmaz (1983: 168) has also considered what she terms a 'loss of self' following the onset of chronic illness. Charmaz highlights the multifaceted experience of loss for her participants, as they experienced social isolation following the loss of friendships and the loss of independence when they were no longer able to drive. The themes identified by Charmaz are present in the sight loss literature but also have salience for participants in this thesis (discussed in Chapter Six).

When people lose their sight, they are also said to experience feelings of grief as they 'mourn the loss of the sighted self' (De Leo *et al.* 1999: 339) resulting, in some cases, in attempted suicide. However, it is not only people with sight loss who experiences feelings of grief. Research suggests that the diagnosis of visual impairment can have an immediate and lasting impact on family members (Tuttle 1986; Bambara *et al.* 2009). Grief is experienced by parents who mourn the loss of their child's ability (Solinit and Stark 1961; Blacher 1984; Fortier and Wanlass 1984; Anderegg *et al.* 1992; Hewson 1997) and the loss of the 'idealised child' (Hewson 1997: 1131). In his research with mothers of children diagnosed with Down's syndrome, Thomas (2014: 287) explains how mothers spoke of their grief when learning of their child's diagnosis, framing the diagnosis in terms of loss, 'the child having disrupted preconstructed expectations of their child and the mother role'. Mirroring Thomas's findings, mothers participating in this thesis also expressed feelings of grief following their child's diagnosis (discussed in Chapter Five).

The feeling of grief experienced by individuals when they lose their sight is said to be similar to that experienced when a close family member dies (Kübler-Ross 1969; Hewson 1997; Baus 1999; De Leo *et al.* 1999; Fourie 2007; Murray *et al.* 2010; Thurston *et al.* 2010; Stevelink *et al.* 2015a; Ferguson and de Abreu 2016). Kübler-Ross (1969) developed the five-stage theoretical grief model within the context of the emotional responses of terminally ill patients and their relatives to death and dying. The dying and the bereaved are thought to experience feelings of denial, anger, bargaining, depression and acceptance. Kübler-Ross's grief model has been applied in diverse contexts including sight loss (Adams and Pearlman 1970; Giarratana-Oehler 1976; Schainholz 2000), chronic illness (Telford *et al.* 2006), and marriage breakdown (Somary and Emery 1991). Within the sight loss literature, denial is defined as a refusal to acknowledge the deterioration in sight, which manifests in a number of ways, including a refusal to participate in rehabilitation services (Bergeron and Wanet-Defalque 2012). Here, acceptance refers to recognising sight loss and adopting positive coping strategies (Bergeron and Wanet-Defalque 2012; Stevelink *et al.* 2015a; Ferguson and de Abreu 2016).

Kübler-Ross's model has been critiqued as being unrealistic within the context of sight loss (Dodds 1989; Hewson 1997; Baus 1999; Murray *et al.* 2010; Stevelink *et al.* 2015a). Murray *et al.* (2010: 79) argue that the assumption that people arrive at acceptance after they have 'completed the linear and time-bound grief process' is ill-founded, as acceptance of sight loss is far more complex and nuanced than Kübler-Ross's model suggests. As a consequence, alternative models of adjustment to sight loss have been developed. Dodds (1989: 11) explores feelings of loss within the context of the development of rehabilitation services. He argues that the traditional grief model has had a 'pervasive influence' since it suggests that the process of adjustment to sight loss and rehabilitation are separate entities. He argues that the loss model tacitly suggests that rehabilitation cannot take place until grieving for the loss of sight is *completed*, with rehabilitation only beginning when the point of acceptance has been reached. Dodds outlines three grounds to support his contention: the grief model does not reflect what actually happens in practice; the model suggests that individuals experience strong emotions in a natural order; and the learning of new skills can only take place when the individual is ready.

Building on the work of Bandura (1977), Dodds (1989) developed the self-efficacy model of adjustment in which he proposes that it is possible for individuals to adjust positively to sight loss by having realistic expectations of their future competencies, and thus help them to regain their independence. In contrast to the grief model, the self-efficacy model advocates that early intervention is not only possible, but essential if the individual is not to descend into 'learned helplessness syndrome', which develops as a direct result of being unable to function effectively at the most basic level (1989: 15).

Providing another alternative to the Kübler-Ross model of grief, Baus (1999)—in her work examining the physiological aspects of visual impairment— develops a grief model by adopting a four-phase approach. In phase one, there is an unwillingness to face the truth following diagnosis of sight loss, which leads to a state of paralysis and denial of the visual impairment. In the second phase, the paralysis gives way to anger, rage and fear—individuals are no longer able to hide their sight loss, resulting in limits being set by the outside world. In phase three, individuals experience depression and despair with the recognition that life will never be the same again; there is conformation of what one has lost, and at the same time, a search for what is still possible, leading to a process of withdrawal and isolation. Phase four marks a change in relationships between the individual and the world, with a new willingness to participate in activities of daily life and to adjust to life without sight. The individual once again becomes receptive to stimuli from outside and actively seeks contact with others, such as self-help groups. Baus (1999) acknowledged that the model represents the ideal case scenario; it is not watertight in practice, and the phases are not mutually dependent. In addition, Baus refuses to refer to phase four as 'acceptance' or 'agreement', as she considers that the 'process of adjustment to disability will never quite be finished and we may at any time come into contact with our loss and our grief' (1999: 43).

Recognising the limitations of the traditional loss model, Thurston *et al.* (2010) posit a theoretical five-stage model, presenting the transition from sight to blindness as experienced by 18 blind and partially-sighted participants in their study. In stage one, participants are diagnosed with a serious eye condition, leading to blindness which evokes feelings of shock, panic and disbelief. In the second stage, participants

who display no outward signs of sight loss adopt strategies of concealment. Participants demonstrated an overwhelming desire to remain the same for as long as possible, evidenced by a resistance to participating in rehabilitation services. In the third stage, referred to as the point of impact, lifestyle becomes affected (for example, the withdrawal of a driving licence). In stage four, participants accepted rehabilitation services and adopted the use of low vision aids.

These findings lend support to Dodds' self-efficacy model in which it is appropriate for rehabilitation to take place before arriving at acceptance of sight loss. The authors further contend that in stage four, the outward manifestations of sight loss are said to be emotionally challenging, leading to changes in perceptions by others as well as perceptions of self. In her work as a sight loss counsellor, Southwell (2012: 109) observed that many of her clients, following their sight loss (and similarly to the bereaved), refer to friends unexpectedly vanishing, which she suggests is a result of a 'subconscious fear of contagion, embarrassment and helplessness'. In the fifth stage, participants arrive at the position of accepting their sight loss as they complete the process of 'reconceptualization of self or of the condition' (Thurston *et al.* 2010: 107). Again, Thurston *et al.* acknowledge the limitations of their theoretical model, in that it was developed from a limited sample and that individual lived experiences will differ.

The reality that individuals may never really come to accept their sight loss has been echoed in research by Murray *et al.* (2010) who, using semi-structured interviews, explored grief and the needs of 10 adult participants (aged 26–56 years old) who had experienced either sudden vision loss (retinal detachment; nerve damage) or progressive vision loss (macular degeneration and diabetic retinopathy). The participants were divided into two groups: those who had lost their vision for up to six years at the time the interviews were being conducted (short-term participants) and participants who had lost their sight more than six years ago (long-term participants). The study found that the long-term participants—who were acutely aware of the continuing daily challenges of living with sight loss and who experienced recurring grief even after a number of years—did not conform to the traditional grief model. Participants instead embraced what is referred to as chronic, recurrent but episodic grief. In this model, grief is considered to be recurrent and

cyclical. It is not, however, constant or unrelenting, but can exacerbate difficult times and situations throughout the lifetime of the visually impaired. In contrast, the short-term participants appeared to have accepted sight loss on an intellectual and emotional level, thus validating the assumption of a 'linear, sequential and time-bound resolvable grief process' (Murray *et al.* 2010: 86). However, the authors argue that the short-term participants had not yet reached a point where they fully appreciated the long-term impact of sight loss.

In Stevelink *et al.* (2015a) interviews with 30 ex-servicemen under 55 years old, 10 of the participants had sustained traumatic irrevocable sight loss whilst deployed in Iraq and Afghanistan. Asked about their views on different aspects of visual impairment, the overarching theme identified by the authors for further discussion was: '*coping with a visual impairment and impact on daily life*'. Participants reported experiencing feelings of loss, (which included losing their job as a result of medical discharge), the breakdown of relationships and the loss of independence. Denial featured strongly in the data, with the ex-servicemen's attempts at tasks they used to do resulting in feelings of frustration and irritation. In adjusting to sight loss, the participants either adopted positive (adaptive) coping strategies, such as participating in rehabilitation, or negative (maladaptive) coping strategies, such as the use of illicit drugs and social withdrawal. When positive coping strategies were adopted by the participants they were helped to adjust to sight loss and overcome future challenges. Coping was considered to be an 'ongoing and dynamic process' (2015a: 5) with participants experiencing good and bad days notwithstanding that several years had passed since losing their sight. Echoing Charmaz's (1991) research with the chronically ill (discussed in Chapter Six), participants in my thesis also talk of experiencing good and bad days.

Stevelink *et al.* (2015a: 5) concluded that in dealing with loss, their participants, in contrast to the Kübler-Ross model, experienced a far broader range of emotions and behaviours which were seen to be more 'dynamic, highly interactive and a unique journey for every individual'. In some cases, the emotional responses occurred simultaneously as opposed to the linear process described by Kübler-Ross. For example, Kübler-Ross described acceptance as the final stage on the sight loss journey, but acceptance of sight loss may occur temporarily but interacts with new

periods of adjustment, and grief is characterised as a 'chronic, recurrent but episodic process' (Stevelink *et al.* 2015a: 6).

Similarly, in line with Murray *et al.* (2010) and Stevelink *et al.* (2015a), Ferguson and de Abreu (2016), reject Kübler-Ross's grief model favouring instead the Episodic Stress Response Model developed by Hewson (1997) in the context of loss of ability. Hewson's model talks in terms of primary stress, (occurring after an unexpected diagnosis), and secondary stress, (associated with daily frustrations, and judged to be not as inherently challenging to everyday life or the future). Ferguson and de Abreu conclude that how the seven men participating in their study evaluate primary and secondary stress is dependent on the context and individual personality. Ultimately, the authors argue that using Hewson's model helps to explain why feelings of loss and frustration are chronic and continue to be episodic throughout life.

The overwhelming rejection of Kübler-Ross's grief model in work exploring sight loss suggests that the process of adjustment to sight loss never reaches a conclusion as the visually impaired experience grief at intervals throughout their lives.

Sight loss and stigma

Stigmatisation is a recurring theme within the sight loss literature and is present in the narratives of participants in this thesis, and was reflected in their decision-making, for example, not disclosing their disability on job application forms or refusing to eat in public for fear of spilling food on their clothing. In this section, I consider the stigma literature in terms of chronic illnesses, where stigma is frequently depicted as an inevitable consequence of chronic illness with the chronically ill reporting that the stigma associated with their condition is more difficult to cope with than the illness itself (Green and Sobo 2000; Green 2009). More recently, Green (2009: 1) has challenged traditional perceptions that those living with chronic illness experience stigma and are stigmatized, suggesting that:

'[T]he old order in which the sick and disabled are disempowered and marginalised is being replaced by a world characterised by their increasing confidence and a reassertion of their essential personhood.'

There has been much scholarship devoted to the topic of stigma, resulting in an 'ever-widening circle' (Prior *et al.* 2003: 2191) of research, particularly on chronic illness, including: HIV (Green 1995); epilepsy (Schneider and Conrad 1980; Scambler and Hopkins 1986; Scambler 1989); mental illness (Link and Phelan 2001; Tyler and Slater 2018), and; inflammatory bowel disease (Thompson 2013; Saunders 2014).

However, social scientists have been criticised for studying stigma without demonstrating an understanding, or explicitly articulating, its 'theoretical ingredients and boundaries' (Livingston and Boyd 2010: 2150). This has prompted authors to suggest that stigma is constantly 'under-defined and overused' (Manzo 2004: 401), with the plethora of studies that have added depth and breadth to the concept of stigma, also being criticised for lacking conceptual clarity (Livingston and Boyd 2010) and introducing ambiguities (Weiss *et al.* 2006). This prompted Prior *et al.* (2003) to suggest that stigma 'is creaking under the burden of explaining a series of disparate, complex and unrelated processes to such an extent that the use of the term is in danger of obscuring as much as it enlightens' (2003: 219). Link and Phelan (2001) suggest that this ambiguity is in part attributable to the fact that stigma has been described in a range of unique contexts which has encouraged multi-disciplinary researchers (sociologists, psychologists, and anthropologists) to conceptualise stigma in diverse ways, a point also made by Green (2009), who agrees that there is an erroneous view that there is now a defined and shared understanding about the meaning of stigma.

Conceding the point that there may not be a 'universally accepted' (Hersh 2013: 246) definition of stigma in the literature, this thesis will draw on Erving Goffman's (1968) seminal work, *Stigma Notes on the Management of Spoiled identity*. Goffman (1968:15) argues that society categorises individuals on the basis of normative expectations distinguishing the 'normals' from the 'deviants'. Importantly, the relevance of Goffman's conceptual framework for understanding health-related stigma in contemporary society has been the focus of intense debate (Link and Phelan 2001; Scambler 2004, 2006, 2009, 2018; Weiss *et al.* 2006; Green 2009; Livingston and Boyd 2010; Hersh 2013; Tyler and Slater 2018). Within the realm of health research and policy, Weiss and colleagues identified four significant failings

of Goffman's conceptual framework of stigma: the use of what is now considered to be 'antiquated' language and concepts; the application of stigma to a vast array of circumstances outside the area of health and, significantly, the adoption of a conceptual framework based on notions of normalcy and deviance that is inadequate and inappropriate in the context of cross-cultural research; and policy-making which is incompatible with multicultural or pluralistic societies with excessive generalisation and lack of attention to underlying structures.

Similarly, Scambler (2009: 442), whilst conceding that Goffman's work provides a 'paradigm and exemplar' and that Goffman's thoughts on stigma remain insightful, also suggests that we should now move beyond Goffman's original telling contribution to ask and answer the questions which the original work overlooked (for example, 'social structures and axes of power' 2004: 29). Notwithstanding the criticism of Goffman's work, I would argue that within the realm of sight loss, it is insightful and relevant to the current thesis. For one, in his typology of stigmatised groups, Goffman places the blind in the category of individuals who have what he refers to as an 'abomination of the body' (1968: 14). Drawing a distinction between 'virtual social identity'—the stereotyped imputations that occur in everyday life—and 'actual social stigma'—the attributes that an individual actually possesses, for Goffman stigma is synonymous with possessing an 'attribute that is deeply discrediting' that arises from an 'undesired differentness' (1968: 13-14) which spoils one's identity. Goffman argues that once a 'discreditable' difference becomes known, the person may be transformed from a 'whole and usual' person to a 'tainted discounted one' (1968: 12). Referring to the concept of possessing a spoiled identity, Scott (1969 118) suggests in his study of the visually impaired in America that:

'The social identity of a man [woman], indeed his [her] whole personality, is spoiled when he [she] is blinded. That he [she] is regarded as a different and lesser person than others is sharply brought home to him[her] whenever he [she] has dealings with the sighted. A major component in the experience of being a blind man[woman] is defending the self from imputations of moral psychological and social inferiority.'

In his work, Goffman distinguishes between being 'discreditable'—where the stigmatising characteristic is not 'immediately perceived' nor has it been disclosed—and being 'discredited'—where the differentness is known about

already or is evident 'on the spot' (1968: 14). For some participants in this thesis this is a distinction that is particularly salient since LHON is an eye condition which presents a paradox with individuals being registered as severely sight impaired (blind) but still retaining sufficient residual peripheral vision not to require the use of low vision aids. In short, their eyes look 'normal' and their disability is 'invisible' to others (*discreditable*). In contrast, those who rely on a white cane or a guide dog may be said to be *discredited* as their sight loss is immediately apparent. In addition, I would suggest that for some of the participants in this thesis, Goffman's dramaturgical work, *The Presentation of Self in Everyday Life* (1959), is of particular relevance. Goffman was interested in face-to-face interaction, focusing on the structure of interaction in everyday life and how the social order is maintained. Using terms borrowed from the theatre, Goffman suggests that individuals throughout their lives undertake a process of impression management. In effect, they play different roles (for example parent, spouse, and employee) in much the same way as actors on a stage. To quote Shakespeare:

'All the world's a stage, and all the men merely players. They have their exits and their entrances. And one man in his time plays many parts' (Shakespeare, *As You Like It*, Act 2, Scene 7).

Goffman wrote of the 'front' [stage] where people undertake a performance; where in public spaces, when they perceived they are the subject of scrutiny, they conceal their stigmatising characteristic. In private spaces—Goffman's 'backstage'—they are able to relax, in those backstage moments they are true to themselves.

At this point, it is worth remembering that Goffman's work on stigma is not the product of original empirical research, but an account drawing upon several literatures. Goffman draws heavily on the work of the writer Hector Chevigny, *My Eyes Have a Cold Nose* (1947), whose work Goffman extensively quotes throughout his book. Chevigny, who at the age of 37 experienced bilateral sight loss following retinal detachment, wrote about his experience of sight loss and his refusal to be viewed by society as an object of pity relying on charitable handouts:

'Towards the blind the world presents a face it turns to no other group on earth. Everyone else must struggle for his existence, must fight for survival. The blind, however, need not want. Society, profoundly convinced of the utter helplessness of a man[woman] who has lost his

sight, stands ever ready to help him [her], whether this be a small thing as crossing the street or the larger one of food and shelter for the rest of his[her] days' (1947: 81).

Drawing upon Goffman's work, Scott (1969) in his study also refers to societal attitudes toward visual impairment:

'Blindness is a stigma carrying with it a series of moral imputations about character and personality. The stereotypical beliefs [...] lead normal people to feel that the blind are different; the fact that blindness is a stigma leads them to regard blind men[women] as their physical, psychological, moral, and emotional inferiors. Blindness is therefore a trait that discredits a man[woman] by spoiling both his identity and respectability' (1969: 24).

Scott further notes that when the stigmatised associate with 'others', barriers are created between them. The barriers result in the 'normals' shunning the stigmatised by adopting avoidance strategies which Scott believes are grounded in fear that 'direct contact with a blind person may be contaminating, or that the stigmatised person will somehow inflict physical or psychic damage' (1969: 24). Davis (1961) comments on the presence of the embarrassment factor in face-to-face interaction between 'normal' people and the visibly impaired. He argues that normal people are apt to make 'faux pas, slips of the tongue, revealing gestures and inadvertent remarks' (1961: 121–122) that betray their discomfort in dealing with other people who have a disability. Whilst the visually impaired may try to lead a normal life—and indeed consider themselves to be normal—they are forced to face the reality that, in their everyday encounters, others do not see them in the same way and are unwilling to deal with them on an 'equal footing' (Scott 1969: 24).

Covering and passing

Those living with a visible disability adopt strategies of 'concealment of the impaired self' (Lingsom 2008: 2), allowing them to be perceived by others as normal. Goffman (1968) describes two such strategies: covering (a process of tension management) and passing (a process of information management). By covering a stigmatising attribute, an individual adjusts their behaviour to ensure that their disability is kept as inconspicuous as possible. By doing so, tension or embarrassment for others (who are aware of the disability) is avoided during social interaction. Goffman (1968: 126) illustrates the point by referring to the reluctance of the partially

sighted to read a book in public. Although the sight loss is known about, it would necessitate holding the book extremely close to the eyes, thus 'express too glaringly the qualities of blindness'. Individuals can go to great lengths to ensure that a stigmatising attribute remains hidden from the gaze of others.

Those who possess a discrediting characteristic seek to conceal their differentness by adopting strategies of passing and so appear to 'others' as 'normal'. One of the primary motivations for passing is a fear of 'devaluation, exclusion or marginalisation' (Lingsom 2008: 5). Fourie acknowledges this point by noting that one motivation for passing is that individuals feel threatened by the 'perceived loss of normality in a society that seems to highly value the "normal"' (2007: 224). This point is also alluded to by Southwell who argues that passing stems from a 'deep-rooted need to appear "normal"' (2012: 108). In the context of sight loss, passing entails a reluctance to associate with other visually impaired people (Southwell 2012). Similarly, Wright's (1960) psychological perspective of disability observes that concealing disability does not make it disappear; it still remains in the eyes of the disabled person as a barrier to acceptance by the non-disabled group. Wright also argues that by not accepting their disability, the disabled have to pay the consequences of being in the ambiguous position of the marginalised who do not belong fully to any group:

'like a man [woman] without a country he [she] will wander in the search for acceptance that cannot be until he [she] accepts himself [herself]' (1960: 40).

There is also refusal by the visually impaired to utilise assistive technologies, as Hersh (2015) found in her research with 93 individuals with adult onset visual impairment. Her participants expressed a number of barriers to using the white cane, including concerns about the reaction of others who had known them before sight loss. This prompted feelings of shame and embarrassment. Other factors discouraging the use of the white cane included negative self-image as a consequence of negative public opinion; retaining a self-perception as sighted; allowing relatives to be over protective, and beliefs that the white cane is only for blind people. This popular misconception that blindness means living in total

darkness, whereas in reality only a very small proportion of visually impaired people experience no light perception.

Hersh's (2015) and Stevelink *et al.*'s (2015a) participants both report experiences of being verbally and/or physically abused when using the white cane. Participants claimed people jumped over or kicked the cane away. Such behaviour, resulting in a reluctance to use the white cane and feelings of vulnerability and fear of being assaulted as others may perceive them as weak and helpless. Page (1984) discusses covering and passing in the context of blind acceptors—those who try to ensure that they behave in ways that would be regarded as normal by the sighted (this would include looking directly at people when engaging in conversation). Blind rejecters, Page argues, ignore the sensitivities of the sighted and will adopt behaviours that are most practical for any given situation, for example using their fingers to eat rather than using cutlery.

Goffman (1968: 95) posits that, given the advantages of appearing normal, 'almost all persons who are in a position to pass will do on some occasion by intent'. However, passing comes at a price as normative expectations of behaviour (for example, not bumping into inanimate objects or spilling food and drink) are required, forcing the visually impaired to participate in 'self-censorship' and 'self-surveillance' and in so doing silence the 'blind-self' (Lingsom 2008: 14).

The sociological literature on stigma has predominantly focused on those who, because of their perceived differentness, experience stigma. Goffman argues that stigmatisation is not confined to those with a stigmatising attribute, but also affects those who are closely associated with them; 'the problems faced by the stigmatised persons spread out in waves' (1968: 43). Goffman refers to this phenomenon as courtesy stigma. In his research with the parents of children diagnosed with high functioning autism, Gray (2002: 735) refers to the 'stigma of affiliation that applies to people who associate with stigmatised groups rather than through any quality of their own'. The accounts given by the parents referred to experiencing both felt and engaged stigma (discussed in detail later). However, as with participants in this thesis, experiences of felt and enacted stigma blended together. Visually impaired parents in this thesis, to avoid their children being stigmatised, avoided using

'stigma symbols' (Goffman 1968: 124), for example they would not use a white cane when taking their children to school or to the park.

Goffman suggests that stigmatised individuals reach a point where they no longer feel the need to indulge in passing and undertake a process of voluntary disclosure: 'after laboriously learning to conceal, the individual may go on to unlearn his concealment' (1963: 125). This is the stage that Kübler-Ross (1969) calls acceptance and also relates to the alternative grief models developed by Murray *et al.* (2010) and Thurston *et al.* (2010), in which the visually impaired finally come to terms with the blind self and take part in rehabilitation services. Hayeems *et al.* (2005: 618), in their research with 43 participants with retinitis pigmentosa, refer to this process of voluntary disclosure as the 'action stage' where the visually impaired, in undertaking mobility training (for example, using the white cane), 'out' themselves.

Enacted and felt stigma

A significant contribution to Goffman's conceptualisation of stigma is provided by Scambler and Hopkins (1986) in their study of people diagnosed with epilepsy. The authors (1986: 33) developed the 'hidden distress model' of epilepsy, drawing a distinction between what they refer to as 'enacted' and 'felt' stigma. The former refers to overt acts of discrimination, whilst the latter encompasses feelings of shame and fear of being confronted with enacted stigma (Scambler and Hopkins 1986; Scambler 1989, 2004, 2009). Scambler (2009: 445) explains that the hidden distress model may be articulated by three propositions. Firstly, when faced with a medical diagnosis (in this case epilepsy), 'state-sanctioned, culturally authoritative and carrying legal weight, individuals develop a "special view of the world"'. This view is characterised by a powerful feeling of felt stigma, which results in acts of concealment of the disability. Secondly, by successfully managing their condition (with medication), others do not become aware of the illness. Finally, felt stigma was found to cause the most anxiety and was more disruptive to an individual's life than enacted stigma. Scambler and Hopkins (1986: 33) also draw a distinction between the concepts of stigma and deviance. Scambler (2006: 293) suggests that individuals are 'imperfect beings' with characteristics often beyond their control and, thus, stigma denotes an 'ontological deficit'. In contrast, deviance denotes a 'moral deficit' that is associated with conditions such as HIV/AIDS.

Scambler (2004, 2009), critiques the hidden distress model, suggesting that there is a risk that the concept, (1) potentially reinforces medical authority, by accepting the biomedical perspective, (2) reinforces the perception that disease is a personal tragedy, and (3) can imply that the sick are victims, who passively accept their fate. Scambler has called for the reframing of stigma within medical sociology, advocating a more ambitious sociology of illness-related stigma, one that moves away from the medical model of disability which depicts disability as personal tragedy. For Scambler, influenced by the work of disability activists, stigma is a social concept that is reproduced and sustained through social inequalities. More recently, Scambler (2018: 768) has argued that what is missing from Goffman's original work is 'the causal role of social structures like class, command, gender, and ethnicity'. In a similar vein, Tyler and Slater (2018) extending the conceptual framework espoused by Parker and Aggleton (2003), critically examine the anti-stigma mental health campaign (*Heads Together*). Focusing on the limitations of existing conceptual understandings of stigma, the authors suggest that to fully grasp the role and function of stigma in contemporary society, it is necessary to develop a more nuanced understanding of stigma as a cultural and political economy.

Summary

This chapter has presented an overview of the literature which provides the foundation for the empirical chapters that follow. I have highlighted that LHON is receiving considerable attention within the scientific community as a consequence of the promise of new treatment options to cure the condition. Yet this clamour is not reflected in social scientific contributions regarding the everyday lived realities of people with LHON. The paucity of sociological literature exploring LHON and sight loss more generally underpins my decision to align my thesis with key themes developed within sociology, including biographical disruption (Bury 1982); loss of self (Charmaz 1983); and stigma (Goffman 1968; Scambler and Hopkins 1986; Scambler 1989, 2004, 2006, 2009, 2018). I have also discussed psychology models of adjustment to sight loss (Dodds 1989; Baus 1999; Murray *et al.* 2010) and disability studies that advocate adopting a social model of disability (Oliver 1990; Thomas 2004, 2007; Barnes and Mercer 2013).

Evident from the literature is that the visually impaired, similar to the bereaved, experience a number of emotional responses to their sight loss, including denial and anger. However, the notion that people arrive at acceptance having moved through the stages of grief in a linear way is contested. Similarly, Murray *et al.*'s (2010) research is a point of departure for participants in this thesis who, irrespective of how long they have experienced sight loss, claim that they will never accept it (discussed in Chapter Six).

The literature also suggests that for the visually impaired there is an overwhelming desire to be perceived as 'normal' and avoid the stigmatising gaze of 'others'. People living with visual impairment face the dilemma of whether to conceal their sight loss or disclose information. However, disclosure is not a straightforward process as decisions must be made as to when to disclose information, and how much information to disclose (discussed in Chapter Six).

In the next chapter, I describe the methods employed to achieve the aims outlined in this thesis and to answer the research questions. This will be followed by four empirical chapters which describe the key findings from the research.

Chapter Three: A Methodological Discussion

Introduction

In this chapter, I outline the methodology I adopted in undertaking my research in the ophthalmic genetic clinic—a site that has not previously been the focus of sociological interest. The clinic provides the opportunity, firstly, to explore the work of an elite group of professionals—the genetic ophthalmologists (comprising clinician scientists and neuro-ophthalmologists) instrumental in diagnosing, treating and developing new treatment options to cure Leber hereditary optic neuropathy (LHON). Secondly, it facilitates interviews with participants diagnosed with LHON to explore their experiences and perceptions of living with a rare mitochondrial condition.

I take this opportunity to reflect on the challenges I encountered when I made the decision to undertake qualitative research in two genetic ophthalmology clinics located in NHS hospitals in two UK countries. Starting from a perhaps too-naïve position, I thought that once I had secured ethical approval from the Research Ethics Committee (REC), obtaining R&D permissions to access the two NHS sites would be a relatively straightforward process. In reality, obtaining NHS ethical approval and R&D permissions was the start of a long and complex journey in which I became embroiled in the National Institute of Health Research (NIHR) Clinical Research Network (CRN) portfolio adoption requirements and NHS governance training, both of which were mandatory requirements to access my chosen research sites. I provide an account of the practical difficulties I encountered when accessing an unco-operative site. I had originally intended to undertake an ethnographic study of the genetic eye clinic, however, ultimately, I was not able to resolve the access issues I encountered. Therefore, this thesis utilises in-depth qualitative semi-structured interviews supplemented with some observational data. I will explain how I analysed my data and developed the themes that emerged across the data set. Finally, I reflect upon my role as a researcher investigating a sensitive topic, highlighting the ethical issues I encountered during the data collection process.

Adopting a qualitative approach

One of the aims of this thesis is to understand the lived experience of sudden sight loss—a highly individual experience which is not easily quantifiable. Adopting qualitative research methods enables the collection of rich and informative data, thus bringing a greater depth of understanding of the impact of sudden sight loss on individuals and their families. Undertaking semi-structured interviews not only enabled me to be sensitive to the particular needs of my research participants, but also facilitated my participants' 'deliberations without restricting their responses to a narrow set of predefined areas' (Dovey-Pearce *et al.* 2007: 78) and, thus allowed me to capture their individualised in-depth thoughts, emotions and reactions to their sudden sight loss.

Qualitative research is the 'method of choice' (Ebrahim and Sullivan 1995: 196) within healthcare research, and this is particularly evident when the primary aim is to gain an understanding of the 'processes, events and relationships in the context of social and cultural situations'. Notwithstanding the undoubted continued popularity of qualitative research, it is not without its critics (Anderson 2010). Mays and Pope (1995: 109) identify the main areas of concern associated with qualitative research. Firstly, it is subject to bias as findings are based on anecdotal evidence, personal impressions and idiosyncrasies of the researcher and, as such, research findings lack 'scientific rigour.' Secondly, qualitative research is criticised for lacking generalisability with qualitative methods generating voluminous amounts of detailed information about a small number of people.

My research design builds in multiple sources and types of data, which means that understandings and experiences can be compared across policy, clinical and patient populations (Latimer 2008). Each of these accounts provides a different representation of LHON. Whilst the accounts may be qualitatively different, it is possible to build rigour into the analysis in the form of checks and balances. For example, fieldnotes are checked against interview transcripts, thus providing the opportunity to uncover how reality is constructed via multiple voices and positions across time and space (Latimer 2008). All these accounts are laid alongside each other to make comparisons, capture patterns, highlight similarities and identify any

deviations (Silverman 1993) to upset original interpretations or provide further explanations (Latimer 2008).

Preparing the ground—pre-fieldwork

In this section, I will discuss the preparatory work undertaken before accessing my research sites. I also provide an overview of the process of applying for NHS ethical approval and R&D permissions.

Locating a gatekeeper

Before applying for the necessary NHS ethical approval and R&D permissions, I became aware that there was a very small group of experts in the UK with an interest in the diagnosis and treatment of LHON. One leading expert, Dr Morgan, is an Honorary Consultant Ophthalmologist at St Tristan's Hospital in the UK. I approached Dr Morgan to act as gatekeeper and facilitate my research within the NHS. Involving a clinician at an early stage ensured that my research protocol was accurate in terms of the information I had outlined (for example, the inheritance, diagnosis and treatment options for LHON). It also ensured that potential access to research participants was accurate. Pope (2005) also argues that securing a gatekeeper located within the host organisation can legitimise the research and smooth the way for the researcher to access the site and be accepted by clinical and administrative support staff. My thesis provides an example of where Pope's claims do not always hold true (discussed later). Dr Morgan agreed to my request and explained that there were approximately 20 LHON patients, with on average 1–2 new referrals a year. One of the aims of my research was to observe how a LHON diagnosis was accomplished in the clinic—and how clinicians communicate complex genetic information in respect of inheritance to their patients. I realised that the opportunities to observe appointments with patients who had not received their diagnoses were going to be limited, mainly owing to the small number of referrals per year.

Dr Morgan made two suggestions, the first of which was that I observe appointments both in her Retina and Optic Nerve Screening Clinic and in her Ophthalmic Genetics Clinic. The Ophthalmic Genetics Clinic receives not only new patients referred from

other consultants within St Tristan's Hospital, but also referrals from other hospitals in the area. Attending these clinics provided the opportunity to observe the work of the ophthalmic genetics clinic and to understand how a genetic diagnosis is constructed for diverse eye conditions.⁶ Although the pattern of inheritance may differ (autosomal dominant, autosomal recessive, and, X-linked), the diagnostic procedures are similar to those used in identifying LHON (visual field test; optical coherence tomography (OCT); electroretinogram (ERG); and blood test). Dr Morgan explained that patients of interest would be identified and invited to participate in this study.

In consultation with my supervisors at the time, I made the decision that I would follow Dr Morgan's suggestion and amend my research protocol to enable me to undertake participant observation in the two clinics with adult participants and children aged 13–15 years, who had been referred with a suspected genetic eye condition. I decided that I would not conduct interviews with this group of participants as their deterioration in visual acuity was likely to be a gradual process, whereas my interest is the participant's reaction to *sudden* sight loss. The second suggestion made by Dr Morgan was that I contact Dr Penvenen, one of the other leading experts in LHON, and an Honorary Consultant Neuro-ophthalmologist at The Royal Albion Hospital. Dr Morgan agreed to contact Dr Penvenen on my behalf, and Dr Penvenen agreed to participate and indicated that his Optic Nerve Genetics Clinic located at The Royal Albion Hospital took place one afternoon a month, receiving referrals from hospitals in the UK. A major objective of his work is taking research from the laboratory into clinical practice. Dr Penvenen is a clinician scientist utilising next-generation sequencing technologies to identify the underlying genetic causes of eye conditions for families who do not have a confirmed diagnosis. During 2013–14, in excess of 20 LHON patients had been referred to Dr Penvenen.

To further maximise data collection, I also contacted stakeholders in the third sector who confirmed they would support my research by helping me to recruit participants. To maximise recruitment to my study, I attended two Patient Days organised by Dr Morgan and Dr Penvenen. Prior to submitting my research protocol,

⁶ For example, Retinitis Pigmentosa, Stargardt's Disease, and Cone-Rod Dystrophy.

I attended the genetic eye clinic at St Tristan's Hospital in order to orient myself and gain an understanding of the everyday, mundane processes and practices of the clinic. Attending the eye clinic was a useful exercise as it not only helped me develop my research protocol, but also gave me confidence in answering questions when I appeared before the REC.

Applying for NHS ethical approval and R&D permissions

The process of obtaining NHS ethical approval has been described as 'lengthy and bureaucratic' (Pope 2005: 1182), the system originally having been developed for the governance of clinical and biomedical research (Murphy and Dingwall 2007). This became apparent to me when I was asked questions on the application forms that were clearly not relevant to conducting qualitative research. Other researchers have made similar observations (Bosk and De Vries 2004; Israel and Hay 2006; Boden *et al.* 2009). Indeed, some scholars have suggested that social science research should not be the subject of ethical review (Schrag 2011), with arguments in part founded on the belief that the REC has a propensity to adopt an overly paternalistic attitude towards research participants (Edwards *et al.* 2004) and that competent participants should be allowed to make their own judgments about possible harm. More generally, the requirement to apply for ethical approval to undertake social science research in the NHS has been the subject of much debate (Dingwall 2006; Reed 2007; Hedgecoe 2008b, 2012; Nicholls *et al.* 2012).

I found the process of obtaining the necessary approvals both time-consuming and frustrating. In drafting the mandatory forms, I avoided the use of overly technical language, but instead included information that I hoped would reassure the NHS REC that I was competent to conduct the research. Having completed the forms, I booked my appointment via the Central Booking System and submitted the application via the Integrated Research Application System (IRAS). I attended a meeting with the NHS REC in October 2015. I was asked to undertake minor amendments to my invitation letters and information and consent forms. Within two weeks of making the necessary amendments, I received ethical approval.

Notwithstanding the criticisms of ethics committees, my experience was extremely positive. This is in stark contrast to my experience of applying for NHS R&D

permissions to access the two sites in two different countries. The R&D offices at St Tristan's Hospital and The Royal Albion Hospital advised me that I would not be given access to the sites unless my study was adopted by the NIHR CRN Portfolio of Studies in the UK. This began a time-consuming process of finding two experts to review my research protocol. The peer reviewers were required to be independent (in that the reviewers were external to Cardiff University) and not involved in the study in any way. They had to be experts in their field and be able to demonstrate knowledge of the relevant discipline in order to consider the clinical and/or service-based aspects of the protocol. The NIHR CRN suggested that the peer reviewers should include a sociologist and an ophthalmologist with experience of ophthalmic genetics, and particularly LHON, to review the project. Following favourable peer reviews, my study was adopted on to the CRN. In January 2016, I was issued with Letter of Access to undertake data collection at St Tristan's Hospital. The Royal Albion Hospital declined access to the site until I had undertaken online governance training (designed for clinicians) and passed an online test. I was eventually issued with an Honorary Contract to commence data collection at The Royal Albion Hospital at the end of March 2016; this delay was in part due to a change of personnel in the Human Resources department which resulted in paperwork being mislaid. The process of portfolio adoption and the requirement to undertake governance training caused a substantial delay in accessing The Royal Albion Hospital which, in turn, had repercussions for my data collection as I only had a narrow window of opportunity to undertake participant observation in Dr Penvenen's clinic (discussed later).

The research sites

As outlined, my fieldwork was undertaken in two sites: St Tristan's Hospital and The Royal Albion Hospital. Having explained the challenges I faced in gaining access, I will now take this opportunity to introduce the research sites.

St Tristan's Hospital

St Tristan's Hospital is an NHS teaching hospital located in an urban setting in the UK. The eye clinic is one of the busiest outpatient departments in the hospital. On entering the eye clinic waiting area, patients are requested to take a numbered ticket from a machine located to the side of the reception desk, sit down and wait for their

number to be called. The irony was not lost on me that this information is given to patients via a notice on the wall; patients who are visually impaired and do not have a relative or friend with them invariably do not see the notice and stand by the reception desk waiting to be booked in. Depending on how busy the appointment clerk is will influence whether the patient is booked in or told to take a ticket and sit down.

Once patients are booked in, they wait in the large waiting area for a specialist ophthalmic nurse, or Health Care Support Worker (HCSW), to call them into the vision testing room, where they confirm their personal details and have their visual acuity tested. Once the patient's pupils are dilated, they are either directed to have retinal imaging of their eyes or they are sent to the appropriate waiting area for the clinician to see them. The clinical assessment rooms are coloured coded: Orange 1-3; Purple 1-3; Black 1-3, and so on. To assist patients in finding their waiting area, corresponding coloured tram lines are marked out on the floor. Again, this is only of use if the patient is accompanied or has a sufficient level of visual acuity to distinguish the colours.

There are approximately 10 consultants working in the eye clinic engaged in a number of sub-specialties including oculoplastics; glaucoma; medical retina and uveitis. The majority of the consultants are assisted by specialist registrars (SpR). Dr Morgan is unusual in not routinely having a SpR working within the clinic. Patients are called in for their appointment by the clinician. On entering the clinical assessment rooms, it is immediately apparent that they are not rooms in the conventional sense of the word. They are actually semi-open plan work spaces, with walls replaced by partitions which are solid up to approximately 4-5ft high, and then glass to the ceiling. At the back of this area is another solid partition, again 4-5ft high, with a shelf on the top where patients' files are placed by the HCSW to indicate to the clinician which patients are ready for their examination. At the back of the work space is a narrow walkway used by clinical staff and, occasionally, by patients entering and exiting the assessment spaces. When sitting in the assessment space, it is possible to see into the spaces on either side and across the walkway. It is also possible to hear the consultation that is being conducted in the other spaces.

All the spaces have the same standard equipment,⁷ and a light switch which makes it possible for the clinician to dim the lights during the ophthalmic examination.

The Royal Albion Hospital

The Royal Albion is a large NHS teaching hospital located in a large metropolitan city in the UK. The hospital was built in the mid-19th century and currently has over 70 ophthalmologists working at the site and across its sister hospitals. The building is no longer fit for purpose and extensive refurbishment is ongoing. The Royal Albion Hospital is celebrated as a world-class centre of excellence for ophthalmic education and research. The NHS-funded eye clinic receives patients with a range of complex eye conditions from both the surrounding area and across the UK.

The eye clinic where Dr Penvenen examined LHON participants is located on the lower ground floor to which access is gained by using the stairs or a lift. On entering the clinic, visitors are confronted with a maze of corridors, some with uneven floors that slope down into the waiting areas which can make it difficult for the visually impaired to navigate. The ceilings are lower than one would expect, giving the impression of having descended into the bowels of the building. The clinical assessment rooms are accessed via the multiple waiting areas. There is no natural light in the eye clinic and the layout of the clinical assessment areas is similar to the spaces in St Tristan's Hospital. However, in contrast to St Tristan's, the clinical assessment rooms were cramped and felt claustrophobic. This may be attributed to the sheer number of clinicians and patients moving in and out of the assessment areas.

Data collection

Participant demographics

Previous qualitative research undertaken with LHON participants in the UK has focused on the lived experience of male participants (Ferguson and de Abreu 2016). I was keen to recruit both male and female affected participants to my study, as I wanted to explore to what extent (if any) gender impacted on how participants

⁷ A desk slit lamp, large ophthalmology examination chair, sink, one chair for relatives which is fixed to the floor, an illuminated Snellen visual acuity test chart displayed overhead, a cabinet containing eye drops, prescription forms, consent forms and leaflets for various procedures including fluorescence angiogram.

adjust to sudden sight loss. I was also keen to recruit participants who had been diagnosed later in life to obtain diverse experiences of living with sudden sight loss. During my data collection phase, I observed 49 appointments in the eye clinics (seven with LHON patients). I complemented my participant observation with 41 semi-structured interviews which commenced in April 2016 and continued until July 2017. The reason for the extended period of interviewing was because I was continuing to receive emails from prospective participants who had become aware of my study. I also presented my early findings at a patient conference which resulted in further potential research participants contacting me. I interviewed 16 affected males and 11 affected females. I also interviewed six mothers who carried one of the three primary mutations and had given birth to child who had developed sight loss, as well as one female carrier who was a sibling of one of the affected males and, at the time of the interview, was considering her reproductive options. The youngest participant was 14 at the date of the interview and, the oldest 71. The most recent diagnosis was 18 months prior to the interview and, the longest was 27 years. Finally, I interviewed seven professionals, including clinician scientists, ophthalmologists with a sub-specialty in neuro-ophthalmology, an Eye Clinic Liaison Officer (ECLLO), and a Trustee from a sight loss charity. I also interviewed a representative of the pharmaceutical company manufacturing the drug Idebenone which is currently involved in the Santhera Pharmaceutical LEROS⁸ clinical trial evaluating its effectiveness. I adopted a purposive sampling strategy, to ensure that I was able to answer the research questions. I selected participants, both men and women, of varying ages who had been diagnosed (affected or carriers) with LHON. The participants were recruited in the two eye clinics and from third sector charities. Ophthalmologists were recruited through the UK Eye Genetics Group (UK EGGG).

Invitations to participate

At St Tristan's hospital, the LHON participants were identified by Dr Morgan from a list that had prepared for an annual NHS clinical audit. Invitation letters were sent

⁸ LEROS is an open-label interventional Phase IV study designed to further assess the efficacy and safety of Raxone® (Idebenone) in the long-term treatment of LHON patients. The study is being undertaken in centres across the UK and internationally. Recruitment for the study commenced in May 2006 and is scheduled to end in April 2020.

out to the adult participants (Appendix 2) and invitations were sent to the parents of the child participants (Appendix 3). I was given the dates when these participants would be attending the clinic and I ensured that I was in clinic on those days. In respect of participants listed in the clinic with other inherited eye conditions, Dr Morgan' secretary viewed the list of patients two weeks in advance of their appointments and sent out invitation letters.

Where possible, emails were sent to research participants. Electronic communication is now the preferred method of communication for the blind and visually impaired since the development of software packages for computers, iPhones and android devices. The purpose of the invitation letters was to ensure, firstly, that participants were fully informed about the purpose and nature of the research and, secondly, that participants (if they decided to participate) understood the reason for, and the process of, taking their informed consent.

In The Royal Albion Hospital, the Clinical Research Fellow working with Dr Penvenen contacted LHON participants in the first instance. In the early stages, this worked very well. However, when the Clinical Research Fellow had finished her work with Dr Penvenen, no one was willing to take on the responsibility of accessing the information from the database. Eventually, a clerical assistant was allocated the task. However, The Royal Albion Hospital has a high turnover of support staff and she left very soon afterwards: I was back to square one. I sent numerous emails to try and resolve the situation, unfortunately without success. Murphy and Dingwall (2007) observe that ethnographers are extremely dependent upon the willingness and continued co-operation of the host organisation—without such co-operation, research can be obstructed or terminated. My experience at The Royal Albion Hospital impacted on my ability to undertake participant observation at the site; this meant that I was no longer able to undertake an ethnographic study and effectively brought this method of data collection to a premature end.

Families that had been identified by RNIB as having one or more members with LHON were sent the invitation letter by the Children and Family Support Manager. Those who wished to participate in my study were then invited to contact me directly by telephone or email. The Trustees of the LHON Society circulated details of my research to their members via their Facebook page. Again, anyone interested

in participating in the research was invited to contact me. Having sent out the invitation letters, I started to receive telephone calls and emails from prospective participants. After explaining my research, the participants were given the choice of having the information and consent form sent to them by post/email or me providing these at the clinic. I could go through the documents with them on the telephone or could wait until they attended the clinic if they felt more comfortable with this. The majority of my participants indicated they would prefer to wait to receive the documents in the clinic. Those who did want the forms sent ahead of the appointment were given the opportunity to discuss the consent form over the telephone before they signed or, again, were invited to wait until the clinic for further explanation before they signed.

Informed consent/assent

I drafted information and consent forms for my adult participants (Appendix 4) with information which varied according to where the participant was recruited. In addition, for the child participants (aged 13–15), I drafted parental information and consent forms (Appendix 5). As children do not legally have capacity to consent, I also prepared an assent form (Appendix 6) for the child to sign which contained simplified information in respect of my study. All the forms were drafted with reference to guidance issued by the National Research Ethics Service (2011) which recommends having two separate documents for the information sheet and consent or assent form. However, I had decided to produce one document containing both parts. This decision was taken on the basis that it would be more convenient for visually impaired research participants to have all information in one document as opposed to dealing with two separate documents.

I ensured that I read through the information sheet with the participant, either in person or on the telephone, to confirm that they had a grasp of the research and what would be expected of them should they consent to participation. I encouraged participants to ask questions. Participants were strongly encouraged to contact me if they had further questions at a later date. The invitation letters and consent/assent forms were also drafted in accordance with the '*See it Right*' Clear

Print Guidelines (2006) produced by RNIB,⁹ to ensure the documents were accessible to the participants. Research participants who were severely visually impaired or blind were given the option of receiving the information in Braille, or alternatively, the relevant information could be recorded on a CD. In any event, none of my participants requested Braille or a CD.

Interviews

In undertaking interviews, I prepared interview schedules which were designed to elicit in-depth narratives of the research participants' initial experiences of sight loss, the challenges encountered in receiving a diagnosis, and the information they received at the time of diagnosis (for example, prognosis, genetic information). I also wanted to explore how participants adapted to sight loss in the longer-term. I wanted to avoid a 'question and answer' type scenario, instead aimed to achieve what Wolfson (1976: 189) refers to as a 'conversational narrative', which I hoped would encourage participants to feel at ease and more likely to discuss their feelings and experiences. To achieve this, participants were asked an opening narrative style question in which they were invited to talk about their experience of LHON, starting with when they first realised they had a problem with their sight. This worked very well for the majority of the participants, some of whom initially spoke for in excess of 40 minutes without interruption. Adopting this style of interviewing ensured that topics that were considered important to the participants were not overlooked. However, where necessary, I did use the interview schedule (Appendix 7) when participants ran out of steam or if participants were finding it difficult to talk. Developing the interview schedules was an iterative process informed by my observations in the ophthalmology clinic, reading the literature, and modifying the questions as a result of transcribing the early interviews. Not all questions were put to every participant; questions were tailored to the personal circumstances of the individual. During interviews, I made notes that enabled me to return to any points my participants had made on which I wished them to elaborate.

⁹ The RNIB suggest that all documentation should be drafted in large print (Arial bold, font size 14) with a maximum of 70 characters per line, double line spacing, and justified to the left margin (but not the right margin).

My interviews were conducted either face-to-face in the two clinics, or over the telephone (due to the geographical location of the participants). I acknowledge the point made by Barlow *et al.* (2007) that telephone interviews may potentially impact on developing a rapport with participants. Similarly, it has been suggested (Chapple 1999; Novick 2008) that the absence of visual or nonverbal cues in telephone interviews could possibly result in the loss of contextual and nonverbal data, and compromise the researcher's ability to effectively probe and interpret participant accounts. All interviews were audio-recorded using a digital voice recorder with the consent of my participants. In St Tristan's Hospital, I was provided with a room located at the far end of the eye clinic to undertake interviews. This worked very well, and I was not interrupted by the clinical staff. The interviews in St Tristan's Hospital varied in duration from 55 minutes to 80 minutes. The first interviews I conducted at the hospital were with two young males (aged 20 and 14) who, I discovered later, had agreed to be interviewed because their mothers thought it would be good for them to have the opportunity to talk about living with LHON.

The two interviews were not a great success. The older participant attempted to avoid answering my questions by drinking from a plastic bottle of water; every time I asked him to discuss his emotions, he would crunch the bottle, so I could not understand what he was saying. A couple of times I gently reminded him that the digital recorder would not be able to pick up what he was saying, at which point he apologised and put the bottle down. The second interview with the 14 year old was equally challenging and was of shorter duration. He mostly answered "yes", "no" or "don't know" to the questions. At the end of the interview, he explained: "*I don't like talking about stuff. I don't like talking about personal things even with family or friends*". My experience of interviewing males was in sharp contrast to my female participants (particularly mothers) whose interviews lasted from 1 hour 45 minutes to 2 hours 45 minutes. Many of the mothers indicated they had enjoyed the experience. This resonates with the research undertaken by Brannen (1993) who interviewed mothers returning to work after maternity leave. However, unlike Brannen where a number of her participants were critical of the length of time the interviews took (2 hours 30 minutes), none of my participants complained about this. Indeed, at times I struggled to bring the interviews to a conclusion. At the

conclusion of the interview, if the participants indicated that they had more to say, I arranged a follow-up interview to take place. During interviews, the mothers often claimed that it was the first time anyone had asked them how they were feeling following their child's diagnosis and that they found the interview process cathartic. This resonated with the research undertaken with parents of children newly diagnosed with diabetes (Lowes and Paul 2006).

My experience with the two males is not unique; there is a considerable body of literature that has investigated the issue of gender in undertaking qualitative research, albeit focusing on female researchers interviewing female participants (Oakley 1981; Finch 1984). Brown (2001), a female researcher, investigated the issue of gender in two research studies. In the first study, 17 men were interviewed in general terms about their health and health needs. In the second study, 24 men who had had a heart attack were interviewed. Brown observed that in the first study she struggled to get the men to talk; some of the interviewees gave abrupt answers and refused to expand on their answers or provide personal information. In contrast, those in the second group, having been asked to tell the story of their heart attack, were much more willing to open up and talk about their experiences. Brown (2001: 190) concludes that, generally, men are not used to talking about health as it is not part of their 'day to day discourse'. What she believed distinguished the response of her two groups was not the gender of the interviewer, but the nature of the subject matter. The men in the first study did not want to be perceived as vulnerable; however, those in the second group deemed it acceptable because they had been affected by a significant health event. Participants in my thesis have been exposed to a serious life event, albeit not life-threatening. I am not convinced that Brown's argument holds true for my participants. I suspect that my participants' reticence to talk is more complex and likely to be linked to a number of factors including their age, maturity, when they were diagnosed, if they were still in a process of adjustment and the fact they do not want to appear to be vulnerable or different.

The interviews with affected children and adults focused on a number of issues, including: their experience of being diagnosed with LHON, focusing on their feelings at the time of diagnosis; to what extent they understood the information they were

given in respect of their genetic status; and their thoughts on undertaking genetic testing. The participants were asked to reflect on the coping strategies they adopted to deal with the uncertainty surrounding the diagnosis and treatment of the condition. Participants were also asked about their life before their sight loss and how they perceived the impact of their visual impairment on aspects of their everyday life, including independence, self-esteem, education, social experiences, well-being and identity. They were also asked to indicate the level of support they had received following sight loss from their Local Authority Sensory Care Team and other agencies. As LHON is a condition that is passed on maternally, participants were asked to discuss how their diagnosis had affected their relationship with their mother. In addition, female participants were asked to consider (where appropriate) whether they were aware of mitochondrial replacement techniques (MRTs) and, if so, whether they would consider using new technologies to avoid passing the condition on to their children. Finally, participants were asked to discuss their aspirations for the future.

I considered it appropriate to interview mothers as they may be said to have a dual role in the clinic, firstly, by acting as ‘gatekeeper to the patient’s body’ (Dimond 2014: 4), and secondly, following the diagnosis of LHON, they are transformed into patients in their own right. There is a dearth of literature that explores the experience of sighted mothers who are raising visually impaired children (Kelly 2005), and the experience of visually impaired mothers (Conley-Jung and Olkin 2001; Molden 2014; Fredrick 2015, 2017a, 2017b) raising sighted or visually impaired children. Mothers who carry the LHON mutation may or may not display any symptoms, and so mothers were asked to what extent they were aware of any members of the wider family who have sight loss and whether they knew that LHON was in the family. Mothers were asked to discuss, *inter alia*, their feelings when their child was diagnosed, and to what extent (if any) the diagnosis had altered their relationship with that child; their thoughts in respect of communicating their genetic test results to their other children and the wider family; and also, whether their carrier status would affect their future reproductive decision-making.

I conducted interviews with genetic ophthalmologists located in hospitals across the UK. The interviews were conducted over the telephone (participants’ choice) and

were of 30–45 minutes' duration. The interviews were arranged towards the end of my data collection; my rationale for adopting this approach was that I wanted to use my fieldnotes to construct an interview schedule and identify salient issues. Interviewing this group later in the study proved to be a good strategy. Firstly, I had a better understanding of the work of the genetic eye clinic. Secondly, my participants had identified a number of issues including the lengthy delay in being diagnosed: a number of participants were told they were making up their symptoms or had being misdiagnosed. Thirdly, participants gave the impression that LHON is a condition that can be difficult to identify for ophthalmologists who have not previously encountered the condition in clinical practice.

Genetic ophthalmologists were asked to comment on a range of issues, including (where relevant) the research they are currently undertaking in the laboratory, the potential effectiveness of new treatments, and the likely availability of new genetic reproductive technologies for mothers who carry the LHON mutation. They were also asked to consider whether there is a delay in referring patients to specialist genetic services and, if so, to what extent this affects patient outcomes; to what extent misdiagnosis by ophthalmologists who have no experience of the condition is prevalent; and how the referral process to specialist genetic ophthalmology clinics can be improved. They were also asked to recount the strategies they employed in explaining complex genetic information to children, young adults and their families, how they explain to patients the uncertainty associated with LHON, and whether they provided any genetic counselling.

Initially, it was not my intention to interview representatives from the third sector or the pharmaceutical industry. However, during the early stages of interviewing participants, there were a number of issues raised around the delay in diagnosing LHON which would potentially impact on the effectiveness of new treatment options. I interviewed a representative from sight loss charity and asked him to comment on a number of issues including why he thought delays in diagnosis occurred and how the charity was addressing this issue. I interviewed a representative from the pharmaceutical company manufacturing a drug that claims to stabilise sight loss, but also potentially restores sight.

Interviewing participants at The Royal Albion Hospital was more challenging as space in the CRU was at a premium. My participants were attending the hospital to take part in a clinical trial which required them to undertake six assessments during the day. The assessments were scheduled to take place either consecutively or with small gaps of approximately 20–30 minutes in between. The rooms I used were examination rooms with slit-lamps and other equipment used to undertake diagnostic testing, and it was not possible to book a room for the day. During my interviews, I was mindful of the time to ensure I did not over-run as I was anxious to ensure that my participants were not late for their next assessment.

One of the problems I encountered in the CRU was that my interviews were constantly interrupted by clinical research staff who were either looking for pieces of equipment or who were trying to eject me from the room because they had not booked a room. On one occasion, the Clinical Research Fellow interrupted the interview as she had forgotten to dilate my participant's pupils for the next test. We had to stop the interview until she had finished. Interviewing at the CRU was not ideal as I shuttled between participants and rooms. This resulted in the interviews, when transcribed, occasionally appearing disjointed as my participants lost their train of thought. Due to the time constraints at The Royal Albion Hospital, a number of the interviews were concluded over the telephone. Notwithstanding the problems I have highlighted, I still believe it was an appropriate idea to undertake the interviews at the CRU as it provided me with the opportunity to meet my participants, to establish a rapport, and to speak to them in an environment where they could be reminded of the matter at hand (i.e. their LHON diagnosis).

Participant observation

I commenced participant observation in St Tristan's Hospital in January 2016 and The Royal Albion Hospital in April 2016, with the intention of immersing myself in the clinic. I was interested in observing at first-hand the mundane and routine practices of the clinic (Latimer 2008), and how the medical staff interacted with each other and patients. As Silverman (2014) comments, there is no guarantee that what people *say* they do in a given situation is what they actually *do* in reality. I also wanted to observe how the genetic ophthalmologists accomplish a diagnosis using

their clinical judgment and advances in technology. All participants consented to me being present during their clinical consultations. My attendances at The Royal Albion Hospital were scheduled to coincide with the appointments arranged by Dr Penvenen's Clinical Research Fellow who was collecting data for their research study. I was able to observe Dr Penvenen's examination of two recently affected males who had been referred to the clinic from their local hospitals. However, I was not able to observe appointments conducted by the Clinical Research Fellow. During one of my early attendances at The Royal Albion Hospital, I asked the Fellow if I could observe the appointments of the participants who had signed a consent form. The Fellow indicated that she would rather I was not present in the assessment room whilst the tests were being undertaken. My experience resonates with Atkinson's (1995) experience of working with the haematologists. Whilst having secured access to the site and being sponsored by a senior clinician, Atkinson encountered initial hostility from junior doctors, who believed he was there to evaluate their competence. Atkinson resolved the problem by writing a personal note to each of the Haematologists in which he explained he was not there to criticise or evaluate their work. I was not able to resolve the issue with the Clinical Research Fellow as, shortly after I commenced data collection, the Fellow completed work on her project and left the Hospital.

Fieldnotes

Whilst observing appointments in the clinic, I used my digital voice recorder which was placed unobtrusively on the desk to capture the verbal exchanges between the clinician and the participants. I was initially concerned that the recorder might prove to be distraction for my participants. However, my impression was that participants forgot that the recorder was being used, as it became invisible sitting next to all the other equipment on the desk. I also supplemented the observations with fieldnotes which I wrote down in a series of notebooks. As I sat in the eye clinic, I made a conscientious effort to take contemporaneous fieldnotes. At times, the clinic moved at such a fast pace that it was impossible to do this. During the ophthalmic examinations the overhead lights are often dimmed, making it difficult to take notes as I was sat in semi-darkness. On those occasions, I endeavoured to ensure that the notes were written up after I left the clinic, relying on my memory

to accurately record what I had found unusual or interesting during the appointments. In the early stages of data collection, I tried to keep my writing legible as I knew I was going to be referring back to my fieldnotes, possibly 12 months later when I commenced writing up. However, as time went by and my handwriting deteriorated, I resorted to my own form of shorthand. Similar to the experience of Pope (2005: 1182), my fieldnotes are 'embarrassingly scruffy'. When I transcribed the digital recordings from the clinical appointments, I was able to read through my fieldnotes to add in the descriptive accounts of individuals, their dialogue, non-verbal communications and my reactions and reflections to events as they unfolded. I also included my detailed observations of the organisation of the eye clinics and the 'inanimate artefacts' (Latimer 2008: 9): the Snellen eye chart, slit-lamp, and other technologies used in accomplishing a diagnosis.

Data analysis

Early in my data analysis I made the decision that I would transcribe my participant observations and interview transcripts in full. I did not start coding my data until after the participant observations and interviews had been concluded. I had previously received training at Cardiff University in using computer assisted qualitative data analysis software (CAQDAS) which included both ATLAS.ti and NVivo 11. After I had transcribed my data, I started to use NVivo 11 (which I acknowledge is a good tool for organising data) but, of course, the decision on what parts of the transcripts to code and allocate to the themes or nodes rests with the researcher. I decided that, given the size of my study, manual coding would be more appropriate as it provided me with the opportunity not only to work in depth with the data to generate themes but, as Saldaña (2016: 22) has commented, it allowed me to demonstrate 'control and ownership' of the research. I also read through my fieldnotes. As Atkinson *et al.* (2001: 3) note, when fieldnotes are written, they do not become 'closed, completed final text: rather, they are indeterminate, subject to reading, rereading, coding, recording, interpreting, reinterpreting'. Reading the fieldnotes alongside the transcripts of the participant observations acted as an aide memoire.

I printed the transcripts and repeatedly read through them to identify themes which were coded using a variety of coloured highlighter pens. I analysed the data using

thematic analysis, primarily adopting the six-phase approach advocated by Braun and Clarke (2006).¹⁰ Adopting this approach enabled me to identify the main themes present in the data. In addition to identifying, analysing and reporting themes in my data, thematic analysis also facilitates ‘searching *across* a data set—be that a number of interviews or focus groups, or a range of texts—to find repeated patterns of meaning’ (Braun and Clarke 2006: 86, original emphasis). The popularity of thematic analysis may in part be attributable to the view that, as an analytical method, it is not tied to any particular discipline and is considered compatible with a number of diverse epistemological positions and research questions (Braun and Clarke 2006; Ritchie and Lewis 2014).

Thematic analysis has been criticised for the lack of guidance on how actually to identify a theme and, once identified, how the theme should be coded (Braun and Clarke 2006; Aronson 1995). Ryan and Bernard (2003: 780) account for this lack of clarity on the basis that themes are ‘abstract and often fuzzy constructs’. I chose thematic analysis for three reasons. Firstly, I had previous experience of using thematic analysis in earlier projects. Secondly, thematic analysis has been selected as it is equally suitable for use with both large datasets (De Brun *et al.* 2013) and with small data sets (Fielden *et al.* 2011), which is the case with my study. Finally, thematic analysis is considered to be particularly useful when investigating an under-researched area, such as LHON or where the research participants’ views are not known (Braun and Clark 2006). There is a dearth of literature exploring the impact of LHON, and therefore, I did not have any preconceived ideas before I commenced my analysis. Themes were developed inductively, with codes developed based on repeated reading of the raw data itself, ensuring that the codes and themes were strongly linked to the data (Braun and Clarke 2006). I also adopted an iterative approach, in that when I identified new points of interest, I was able to include this in my analysis.

¹⁰ Braun and Clarke (2006) refer to the six phase approach as: Phase 1: familiarising yourself with your data; Phase 2: generating initial codes; Phase 3: searching for themes; Phase 4: reviewing themes; Phase 5: defining and naming themes, and Phase 6: producing the report.

Ethical considerations and researcher reflexivity

Having outlined my methods for collecting data, I will now explore the ethical issues I encountered during the conduct of my research and reflect on my role as a researcher investigating a sensitive topic. In doing so, I pause to consider not only the emotional impact on my participants of taking part in my research, but also how I was affected by listening to my participants' narratives. During data collection, there were times when I called into question my role as a researcher as I became acutely aware of what Guillemin and Gillam (2004: 261) refer to as the dichotomy between 'procedural ethics'—obtaining ethical approval from the NHS REC, and 'ethics in practice'—the everyday ethical issues that arise during the conduct of the research study.

One of the first 'ethics in practice' problems I encountered was with the process of obtaining informed consent from my participants. It became apparent at a very early stage that the National Research Ethics Service (NRES) guidance for producing the consent forms are not designed for the visually impaired. The level of visual acuity of some of my participants meant that, even with font 14 and the use of computer software, they were unable to initial the boxes on the form or see where they needed to sign. I received a number of forms in the post or by email where the signature was in the wrong place, the date was missing, and/or the boxes had not been initialled. Some participants in the clinic asked me to initial the boxes for them. When it came to sign the consent forms, I used my finger to assist the participant in placing the pen at the right place on the form and guided them in placing their signature in the right place on the form. It has been suggested that the requirement for the visually impaired to sign the consent form should be updated to allow them, instead, to provide 'recorded audio consent' (Saleh 2004: 310).

Dr Morgan indicated that currently children (aged 13 and 14) with LHON were being treated in the clinic. The decision to involve children in this research project was not taken lightly. There has, in recent times, been a move away from adopting a paternalist attitude to children's involvement in research, with a growing recognition that children have the right to be involved in making decisions about their welfare (Piercy and Hargate 2004). Article 12 of the United Nations Convention on the Rights of the Child (1989) confirms that all children and young people who

are capable of forming their own views have a right to express those views freely in all matters affecting them. Previous research exploring children's expectations and experiences of attending the eye clinic is missing from the literature. I would suggest that involving children in this research adds another dimension to the research findings.

Children who are aged between 16–18 years old, are presumed in law to be competent to give consent. Notwithstanding this, it is considered to be good practice to encourage competent children to involve their families in the decision-making process (DH 2001). This was the approach I adopted. In respect of children under the age of 16, the position is more complex. The decision to assent or consent children was taken by me on a case-by-case basis and it would have been inappropriate for me to assume that children under 16 were not competent to consent. I obtained guidance on this subject from a number of sources. Medical research involving children is regulated by The Medicines for Human Use (Clinical Trials) Regulations 2004. Research that falls outside the regulations is subject to common law principles developed by the courts and to other statutory provisions including the Family Law Reform Act 1969. In addition, a number of research councils and professional bodies have provided guidance on when it is appropriate to consent or assent children in non-medical research.¹¹

Further guidance is provided by the judgment of Lord Fraser in the case of *Gillick v West Norfolk and Wisbech AHA* [1984] QB 581. Lord Fraser confirmed that in making the decision to obtain consent from children under 16; "It is not sufficient that the child understands the nature of the advice which is being given, the child must also have a sufficient maturity to understand what is involved". Children under 16 who are considered not competent to give consent should be asked to assent to the research after their parent or guardian has given consent. The National Children's

¹¹The Medical Research Council (2004) suggest that where children have sufficient understanding and intelligence to understand what is proposed, it is their consent and not that of the parents that is required by law. The Department of Health (DH) (2001) states that for a child's consent to be valid the child must: (1) be capable of taking that particular decision, in other words must demonstrate that they are competent; (2) be acting voluntarily and not coerced into giving consent; (3) be given sufficient information to enable them to make the decision.

Bureau (NCB) (2011) guidelines suggest that, ideally, informed consent should be obtained directly from the child by the researcher, thus enabling the researcher to satisfy themselves that informed consent has been given voluntarily. Given that I would not have had any previous contact with the child, the NCB confirms that it is appropriate for the parent/guardian or healthcare professional to provide the child with information in respect of the research and to make the decision whether the child is competent to consent. This was the approach I adopted, and the parent/guardian was asked to confirm whether their child was competent to consent.

Respecting the right to confidentiality of the research participants has also proved to be challenging as they are drawn from a small pool of individuals diagnosed with a rare condition. In anonymising data, the major concern has been to ensure that participants are not identified by family or friends. In my research, I have interviewed mothers, children and, on one occasion, siblings—and it is inevitable that participants with a familial connection may identify each other. Many of my participants have joined the LHON (national and international) on-line community, where they have developed friendships. They have placed in the public domain details of their condition and other personal information. A number of my participants attend lay and professional conferences. To overcome the potential for participants to be identified, I have allocated pseudonyms and changed the names of the two hospitals where I collected data. I have also changed some minor details in respect of the clinics and the participants. However, as a researcher it is not possible to legislate for every eventuality. Whilst I was presenting at a Patient Day, one of my participants 'outed' himself by disclosing to the audience that I had interviewed him: in that moment, any sense of anonymity and confidentiality disappeared.

Moreover, interviews involved asking participants to confront sensitive issues which they might find very emotionally challenging. For example, I interviewed a young male who became extremely angry and was shouting at me as he recounted how his parents were aware of his mother's carrier status but chose to have a child notwithstanding the risks. On occasions my participants, particularly mothers, became tearful when they talked about learning of their carrier status, and the

enormous guilt they felt when given their child's diagnosis. The affected participants explained the anxiety they felt that their visual acuity may deteriorate, or that they may never find a girlfriend/boyfriend, or one day have children of their own. Whenever I felt that participants were struggling with my questions, I offered them the opportunity to stop the interview and continue another time or to withdraw. None of the participants chose the latter option. Both Dr Morgan and Dr Penvenen agreed that participants could be referred to them for further advice. I also contacted a senior clinical geneticist at St Tristan's Hospital who confirmed that participants could be referred to his service for advice. In addition, I provided information in respect of sight loss charities that could give further support.

As qualitative researchers, we are anxious to provide a safe and comfortable space that empowers our participants to take part in the interviews and encourages them to open up and share with us their stories (Dickson-Swift *et al.* 2007). It is these disclosures that contribute to enhancing our knowledge and which help us to gain a deeper understanding of how individuals come to terms with living with chronic illness. At times, I was surprised at the level of information disclosed, given that I had never met the participant before. In some instances, participants informed me that having the opportunity to revisit their past and reflect on what had been a distressing and emotionally draining time for them was a cathartic experience. Birch and Miller (2000) comment that in the qualitative interview, the researcher becomes a catalyst for participants to reflect on and give voice to very private and, on occasions, deeply upsetting past events. The authors go on to suggest that this calls into question the judgment of the researcher in evaluating a good or successful interview. In my research, a number of my participants claimed that I had asked questions that no one else had asked before. Similarly, they said that they had disclosed to me information that they had not previously discussed with family or close friends because they wanted to 'protect' them. This is particularly the case with participants who had contemplated suicide or had been self-medicating on illicit drugs. Ultimately, the researcher is asking participants to relive traumatic events and has no way of knowing the long-term harm such disclosure will have on the participant.

The qualitative interview has the potential to provide a space in which the boundary between research and counselling, particularly when discussing sensitive topics, may become blurred (Dickson-Swift *et al.* 2006). This point is also made by Gale (1992) who claims that participants may view qualitative interviews as having more therapeutic value than a therapy interview. This is not surprising, considering that qualitative researchers utilise comparable skills to those adopted by therapists and counsellors when conducting therapeutic interviews, including the ability to listen to what is being said while also demonstrating empathy (Kvale 1996). However, if researchers are not mindful, they may inadvertently open the door to transforming the qualitative interview into an impromptu therapeutic session (Dickson-Swift *et al.* 2006)—one in which the qualitative researcher is ill-equipped to participate (Kvale 1996).

On occasions, interviews were considered by participants as an opportunity to canvass my view on recent advances in research on potential treatment options, particularly relating to mitochondrial replacement techniques, the use of Idebenone and gene therapy. In her research with the bereaved, Sque (2000: 27) claimed that participants seek information from the researcher which, if imparted, can itself have 'beneficial effects in eliminating latent misunderstanding and securing pertinent knowledge'. In qualitative research, Daly (1992: 5) advocates the concept of 'fair exchange' or reciprocity—where researcher and participant share information (Ackler *et al.* 1991). In sharing information, the researcher is seen to be giving something back to the participant and, as a result, this increases both the 'depth and quality' of the data (Dickson-Swift *et al.* 2007: 334). I found these moments difficult to negotiate, as I did not want to be perceived by my participants as someone who held themselves out as having expert knowledge. In response, suggested that the participants may like to discuss their questions with their ophthalmologist the next time they visited the clinic.

I also experienced the problem of the 'reluctant respondent' (Becker and Geer 1957; Adler and Adler 2003; Scott *et al.* 2012) where a participant, having agreed to take part in the interview, then fails to disclose information is well recognised. There may be a number of reasons for a participant's reluctance to speak: shyness, inability to articulate emotions, not wanting to answer questions, or not understanding the

questions (Becker and Geer 1957). Establishing a rapport enables the researcher to build a relationship with the participant and encourages them to impart their story (Dickson-Swift *et al.* 2007; Goodwin *et al.* 2003). Acknowledging the importance of developing a rapport with research participants, Scott *et al.* (2012) consider that this can be achieved by sharing information, finding common ground and on occasions using humour to break the ice. However, as Dickson-Swift and colleagues (2007) observe, the level of disclosure has been viewed as problematic by a number of feminist authors (Oakley 1981; Finch 1984), with concerns raised that participants may be 'seduced into disclosing information that they later regret' (Kvale 1996: 116). As Kvale explains, the interview is not produced as a result of a collaboration of two equal parties. There is a distinct asymmetry of power when the researcher decides on the topics to be discussed.

I acknowledge that I failed to build a rapport with the two young men discussed earlier. On reflection, I realise that my lack of confidence as a first-time researcher meant I over-compensated by trying to appear professional and this gave the impression of my being a little reserved. Having said that, I am not sure whether if I had been more experienced the males would have 'opened up' to me, it could have been that their level of maturity inhibited them, that they felt uncomfortable talking to me because I was a female or, irrespective of my gender or their age, that they just did not like talking about their emotions.

During the interviews, I heard some very positive and upbeat accounts of how participants on a daily basis overcome the challenges they face living in a sighted world. At times I marvelled at their resilience and fortitude in overcoming adversity. However, at other times I was privy to harrowing and disturbing accounts. One participant gave me a very detailed description of how he had been going to end his life; he had decided on the method and acquired the means to do so. He would have ended his life had it not been for the intervention of a school friend who happened to come across him and was able to talk him out of it. When undertaking research dealing with emotive topics, Lowes and Paul (2006) suggest that researchers should not under-estimate the potential impact such disclosures may have on their own emotional well-being. There has been little empirical research focusing directly on the experiences of researchers who may be affected by the stories they have been

told. Dickson-Swift *et al.* (2007) highlight the need for researchers investigating sensitive topics to be given appropriate training to deal with the emotional impact of their research and also to be provided with contact details for sources of professional advice and support. Similar to Atkinson (1997), I found my research was a source of mixed emotions. It gave me a great sense of personal satisfaction in highlighting an under-researched rare condition, but at the same time, there were occasions when I felt anxious and out of my depth.

Study limitations

LHON is a rare condition and this was reflected in the small sample (N=41). It would not be possible given the size of the sample to suggest that the findings are generalisable to the wider LHON community. Secondly, a number of the participants were recruited through a sight loss charity, and received regular newsletters updating them on recent developments in the treatment of LHON. Many of the participants also attend expert conferences and patient days, and It may be argued that this group are highly motivated and not representative of the wider LHON community. My intention to interview women who were considering their reproductive choice was limited as only two women were currently considering having children. As I explained in Chapter Three, I encountered a number of problems in gaining access to The Royal Albion Hospital which limited my opportunities to be present when participants attended the clinic to be receive their diagnosis. I have therefore relied on the retrospective account of participants. Whilst some participants had a vivid recollection of the events others were vague.

Summary

In this chapter, I have outlined the methodological challenges I encountered during my research. I have explained the problems I faced in gaining access to the two research sites, and how delays in securing the necessary R&D permissions in The Royal Albion Hospital had a significant impact on my data collection. Notwithstanding issues with accessing the two sites, I believe that the data I collected provides a valuable insight into the diagnosis and treatment of LHON. I have acknowledged the ethical issues that arise when discussing a sensitive topic and how I adopted strategies to minimise harm to participants. I have also referred

to the issues surrounding anonymity when interviewing participants drawn from a small pool of people who have been diagnosed with a rare condition.

I have explained my rationale for adopting qualitative research methods to explore the highly individual experience of sudden sight loss for people affected with LHON and their families. I have also described the processes of transcribing and analysing my data, justifying my decision to manually code my data.

Over the following four empirical chapters, I explore participants' chronic illness trajectory, highlighting the key findings from my research.

Chapter Four: “The Doctor Didn’t Believe Me”: Negotiating Diagnostic Uncertainty

Introduction

Over the following four empirical chapters, I outline a number of the key findings from my study. In this chapter, drawing upon interview and observational data, I explore participants’ accounts of receiving a diagnosis of Leber hereditary optic neuropathy (LHON). The unexpected loss of sight is the beginning of what is often, a tortuous process for participants, requiring multiple hospital appointments, undertaking numerous (often deleterious) diagnostic tests, and, on occasions, being subjected to the indignity of having their symptoms dismissed as attention-seeking behaviour. I commence the discussion by describing participants’ accounts of their initial reactions to experiencing problems with their eye sight and how symptoms were often ignored until they reached a critical point and medical attention was sought. Diagnosis is a key stage in participants’ illness trajectory and, as Jutel (2009: 278) has observed, ‘is integral to the system of medicine and the way it creates social order’. Diagnosis is also said to identify treatment options, predict outcomes and provide explanatory frameworks (Jutel 2009). I explore the boundaries of diagnosis and how assigning a label to symptoms legitimises participants’ claims to enter the ‘sick role’ (Parsons 1951). From here, I will unpack the narratives of participants regarding their experience of receiving the diagnosis of LHON. For many participants, receiving a diagnosis is the start of their quest for information with respect to the aetiology, prognosis, treatment and inheritance of LHON. In the final part of the chapter, I discuss the uncertainty surrounding LHON and why ophthalmologists fail to identify the condition in clinical practice. I will also describe their own interpretations of interacting with ophthalmologists who dismiss their symptoms as the product of an over-active imagination, thereby resulting in a contested diagnosis.

Experiencing symptoms of LHON

Reflecting the chronic illness literature (Bury 1982; Robinson 1988; Scambler 1989; Kelly 1992), Corbin and Strauss (1988: 22) suggest that when symptoms of illness appear, whilst some people in this thesis immediately sought medical attention,

others waited until the symptoms become 'undeniable, so visible or alarming that they can no longer be ignored'. I begin the discussion by unpacking the multiple interpretations participants attributed to experiencing initial symptoms of sight loss and how, through a process of deferral, some participants downgraded their symptoms until they reach a critical juncture, such as not able to see road signs when driving or read documents at work. It was at this point, when symptoms could be ignored no longer, that medical attention was sought. Mirroring research undertaken by Green and colleagues (2002) with people diagnosed with glaucoma, participants in this thesis, reflecting on their initial symptoms, referred to a range of problems with their eyes. However, only in retrospect (i.e. after their diagnosis) did they realise the significance of the symptoms. John started experiencing symptoms when he was 42 years old: *'I was looking at an email at work and the middle of the word was missing. I thought that's "strange", then I couldn't see the computer screen properly any more'*. At the time, John had been experiencing stress and drinking heavily, which is how he accounted for his symptoms. Sandra, who was diagnosed in 2015 aged 69, recalled an odd episode when her right pupil dilated: *'Looking back, it became apparent that something funny was happening with the right eye which was typified by text being broken up, by colours being distorted and by a lack of acuity'*. Sandra had recently been diagnosed with glaucoma and had undergone surgery to reduce the pressure in her eyes, which is how she accounted for the problem she was experiencing.

Problems with sight were often pushed to the margins of participants' lives and overlooked; this was particularly true of the younger participants as they constructed innocent explanations to account for their symptoms. David, who was diagnosed with LHON in 2011 aged 23, explained that he had been playing football prior to first noticing symptoms:

'It was quite a sunny day, so I thought I had damaged my eyes from the sun because I noticed, I think in my right eye, I kind of had that after image you get after looking at a bright light sometimes.'

Andy noticed that the road signs were blurred whilst he was on a long journey and thought his symptoms were due to tiredness. He was subsequently diagnosed when he was 20 in 2008: *'I was on a four-hour car journey, the [road] signs looked a bit*

blurry, I put it down to tiredness, but it was due to me having started to lose my sight, but I wasn't aware of that then'. Unaware of the seriousness of his symptoms, Andy went back-packing for six weeks, during which time his sight rapidly deteriorated.

Two women, Beth and Tina initially ignored their symptoms, notwithstanding that both shared the experience of living with a brother who had been diagnosed with LHON. Beth, who was diagnosed when she was aged 51 in 2015, explained that she had been wearing glasses for many years: *'I had noticed for a few weeks previously I was having difficulty reading. I couldn't really read, I just thought I needed to check my [glasses] prescription'*. Tina had for a number of years been experiencing recurring migraines and initially did not give any importance to her blurred vision: *'I had a massive migraine the day before. I was driving along, and I thought that the road sign isn't as crisp as it should be. I didn't initially think that there was a problem'*. Tina was subsequently diagnosed with LHON aged 30 in 2013.

Beth and Tina did not appreciate that women could be affected by LHON and, so, accounted for their symptoms as a consequence of ageing (Beth) or ongoing health issues (Tina). Irrespective of how they first experienced symptoms, all of the participants recounted feelings of devastation when they were given what was an unexpected diagnosis (discussed in Chapter Five). Having taken the decision to seek medical attention, participants in the first instance were referred to the eye casualty department. This heralded the start of what for many was a lengthy process to find a diagnosis.

Before dissecting the challenges which participants encountered when attending an eye clinic, I consider how a diagnosis has the power to legitimise illness by 'putting a name' to symptoms, providing entry to the sick role and providing an explanation for what would otherwise be inexplicable symptoms of sight loss (Jutel 2011: 1).

Defining the boundaries of diagnosis

The quest for a diagnosis has been recognised as one of the most pressing issues identified by patients suffering with symptoms of illness (Jutel 2011) and, unsurprisingly, this was evident from the interviews with participants in this thesis. In a world where uncertainty is 'omnipresent in life' (Corrigan 2012: 27), receiving

a diagnosis has the potential to deliver much sought after certainty for both the patient and the doctor, receiving a diagnosis is 'like being handed a road map in the middle of a forest' (Jutel 2011: 1). Without the road map, the doctor is left 'casting around in the dark' unable to articulate a plan of action (Pinder 1992: 8). Diagnosis is said to incorporate two distinct but interrelated characteristics, it is both a category (a label), specifying the pathological nature of the condition, and a process (an activity), utilised by the medical profession to assign the label (Blaxter 1978; Jutel 2009; Jutel and Nettleton 2011; Locock *et al.* 2016). As Blaxter (1978: 9) puts it, diagnosis as a process is dependent on diagnosis as a category, in that the process of describing is reliant on the descriptions 'acceptable or available in the relevant universe of knowledge'. In turn Jutel and Nettleton suggest that diagnosis has a third component—it is a consequence, arguing that the way in which diagnosis is organised, structured and delivered has consequences for patients as it has the potential to 'vindicate and blame; legitimise or stigmatise illness' (Jutel and Nettleton 2011: 797). Receiving a diagnosis gives individuals permission to be ill (Jutel and Nettleton 2011), with Nettleton (2013: 27) noting:

'a diagnosis: validates what counts as disease, offers explanations and coheres patients' symptoms, legitimises illness enabling patients to access the sick role, provides a means to access resources and facilitates their allocation, and forms the foundation of medical authority.'

In a more detailed sense, Parsons's (1951) conceptualisation of the sick role is founded on the premise that the social system is dependent on individuals being healthy and carrying out expected social roles (for example, being employed). Illness is a form of deviance and disrupts the ability to perform social roles, thus posing a threat to normative standards. People who are ill have temporary access to the sick role which is a form of 'sanctioned social deviance' (Barnes and Mercer 2010: 44). By entering the sick role, people obtain a 'claim for exemption' from undertaking 'normal social obligations' and responsibilities (Parsons 1951: 455). However, they must also adhere to the obligations placed upon them, such as acknowledging being ill, seeking appropriate medical intervention, and demonstrating the desire to get better.

Parsons's (1951) conceptualisation of the sick role has been criticised for providing an ideal-typical account of acute illness rather than taking account of chronic

illnesses, where recovery is unlikely. Those with a chronic illness, such as LHON, may be said to have a legitimate claim to the sick role and all its benefits. However, if individuals fail to follow medical advice to adjust their lifestyle to avoid further deterioration of their eye sight, one may wonder whether they are still entitled to the benefits that the sick role bestows upon them.

In the following extract from my observation at Dr Morgan's clinic in St Tristan's Hospital, John attends the clinic for his annual review, having experienced vision loss at the age of 42. At the time of his diagnosis, he was drinking and smoking heavily. Unaware of a previous family history of LHON, he was initially diagnosed with toxic amblyopia,¹² but was subsequently diagnosed with LHON. During the appointment, John raises concerns that his right eye is deteriorating, and it is evident that John has not been following medical advice regarding his smoking and alcohol consumption:

John: The only problem I have is in the past my right eye has always been the best eye. That's why I usually see the top letter [on the Snellen eye chart]. I couldn't see the top letter today. It might just be a temporary glitch, I don't know. My right eye seems to have gone a bit worse. My left eye is the same.

Dr: You are right, if you look back through the readings when you come (flicking through the file). In fact, looking at your notes you do not always see the top letter with the right eye. You obviously do feel that the right eye is the best eye. In general terms, the issue around protecting yourself is maintaining as healthy a lifestyle as you can.

John: I eat well, and I go to the gym, I occasionally drink too much at the rugby. I mean the problem I have is I try not to drink because if I drink, I smoke as well for some reason. I know I shouldn't.

Dr: Smoking is probably worse than the drinking. If there is any way that you can try to not smoke?

John: I know that. I am not a heavy smoker.

Dr: You are not really smoking regularly so the nicotine patches wouldn't help you. You don't need them daily.

¹²Toxic amblyopia presents as a painless bilateral loss of vision. Almost all patients complain of a blurring of vision and difficulty in reading small print. Patients may also complain of difficulty in differentiating red from green. The disease usually evolves over a period of several weeks to several months (Prakash *et al.* 2011).

John: No, that's true. Are the vapes [e-cigarettes] bad for you?

Dr: Nobody knows... nobody knows that they are good for you.

John: You can't really say?

Dr: Ingesting or taking in or breathing in any form of drug or chemical can't be neutral, it must have some effect. There are chemicals in them that are going into ... it's going to be absorbed. People will not know if it's dangerous and may not know for 5, 10, 15 or 20 years. I doubt if it's neutral, I doubt if it's got no effect. I don't necessarily think it is a good thing.

In his appointment, John articulates his concern that his eyesight may be deteriorating. When asked about his lifestyle, particularly his smoking, he becomes slightly irritated and is keen to present himself as adopting a healthy lifestyle, clarifying that he eats well and exercises on a regular basis. However, it is apparent that John's real concern is his level of smoking and drinking, and the impact this is having on his right eye. John is aware that alcohol consumption, and smoking are associated with an increased risk of sight loss for people with LHON (Yu-Wai-Man *et al.* 2014; Kirkman *et al.* 2009a). Dr Morgan subtly suggests that using e-cigarettes may not be a solution for him and that he should give up smoking altogether. I interviewed John after his appointment with Dr Morgan where he continued to be preoccupied with the perceived deterioration in his right eye and reflected on his lifestyle both before and since his sight loss:

How long had you been drinking heavily before you noticed the symptoms?

It was a couple of months. [...] I was living with a friend who died; he was an alcoholic. It's hard to say [how much], a couple of bottles of wine a day, maybe 2-3 bottles of wine a day at the time, I eventually went on to... sometimes it could be a bottle of vodka. [...] [drinking spirits] was only a temporary thing and then I stopped. Not on top of the wine. It would be wine or vodka or something. He was drinking as well, you see. He was a bad influence. I was living with him because I was going through a divorce. He died, and he was my best friend. It was a build-up of things. Then I got depressed about my eyesight. It wasn't a particularly good year for me (*laughs*). [...] I suppose with any eye condition ...even if you have good eyesight, it's a no, no because smoking can cause sight problems [...] I would like to stop drinking, but whether I will or not is another question. I will stop smoking. Friends keep trying to get me out on Friday and they pour me large vodkas and things, you know. I have told them I shouldn't be drinking. They are happy carrying on; they are not really thinking

about me. They say, "Have another drink". [...] I am a social creature, a party animal-type thing. I have tried to change as much as I can. I don't go out as frequently as I used to, drinking I mean. I don't drink in the week at all. At the weekends, if I go out, I drink more than I should (*laughs*).

(John, diagnosed aged 42 in 2009)

John is aware of the precarious position he is putting himself in by continuing to smoke and drink. However, he seeks to abrogate responsibility, firstly, by attributing the conduct to a friend who he was living with at the time of his sight loss and, more recently, his circle of friends, with John believing it was his drinking and smoking that triggered LHON. One of the other participants, David, also reflected on his lifestyle prior to his sight loss. David smoked and drank alcohol before his sight loss, suggesting that this may have triggered LHON:

'I was drinking quite a lot at the time (*laughs*). I was drinking after work, drinking to catch up with my friends and stuff, and then going into work hung-over quite a few times and things... and I would go to kind of... there were a couple of times when I went to all day parties and stuff, especially during the summer and things. And, yeah, like it was just a lot of binge drinking and stuff and drinking everything and anything.'

During his interview, David placed considerable emphasis on his previous drinking habits with respect to a diagnosis of LHON. Later in the interview, he told me that whilst he knows that smoking and drinking are not good for him and after initially giving up alcohol following his diagnosis, he has since started drinking again:

'I stopped drinking and smoking stuff for, like, six months or something and then it was after that I kind of thought to myself, you know, I quite enjoy drinking at least. If I am perfectly honest, (*laughs*) I do enjoy smoking cigarettes as well, but I know that's definitely bad for you; I would love to smoke if it had health benefits or wasn't bad for you (*laughs*). I enjoy drinking and my friends enjoy drinking and stuff and so I kind of felt I don't want LHON to stop me from living my life and enjoying the things I used to enjoy beforehand as well, and so, you know, that's why I am still drinking and stuff. I still do go a bit too wild sometimes, but before I lost my eyesight, there were times when, you know, I got through the stages when I thought I am drinking a bit too much, maybe I should quit for a few months and then start up again. So, you know, I would kind of stop and start.'

(David, diagnosed aged 23 in 2011)

John and David continue to undertake activities that they know may exacerbate their residual vision. Petersen (2006: 33), suggests that being unable to achieve or restore optimum health is often perceived as a 'weakness of the person and a failure to fulfil one's obligation as a citizen'. John and David, at times, are unable to follow medical advice—and thus continue to participate in 'deviant' (Parsons 1951: 453) behaviour, placing in jeopardy their legitimate claim to the sick role. There is of course no certainty that John's and David's sight loss can be attributed to their lifestyle choices. One of the abiding mysteries of LHON is that there are affected individuals who have never smoked, consumed alcohol or been exposed to toxins (Newman 2009). Ollie, who is a good example of this phenomenon, experienced sight loss when he was 18. He explained: *'I wasn't smoking; I led a very healthy life really. I had just done a charity cycle, I played football to a high level, I played cricket to a high level'*. As far as Ollie is aware, there is no family history of LHON and he remains puzzled as to why he was affected.

After their diagnosis other participants also scrutinised their previous behaviour. Gerry had recently retired from his job when his eyesight started to fail. He had a vague recollection of an aunt on his mother's side of the family who had an issue with her sight, but no one in her family was aware of what had caused her sight loss. Gerry pondered on the possible reasons for his sight loss, indicating that he had given up smoking many years ago. Gerry speculated that his previous employment may be the cause: *'I was working in a University research lab, I was working with chemicals, but I wasn't in day to day contact with them... whether there was anything there that triggered it.'* Declan, who experienced symptoms when he was 12 years old explained, that during a school rugby match, he was injured:

'I did play rugby for a local team, and it was when I was playing rugby for the school when my head was stamped on. They think that's what could have triggered it because it was around about the same week I can remember sort of noticing things.'

(Declan, diagnosed aged 12 in 2006)

Other people, when they received the diagnosis, researched their family tree to find relatives who had experienced sight loss. Tim was 22 years old when he experienced symptoms of LHON. He explained that after his diagnosis: *'We went back and looked*

at the family tree because going back to the early 1900s, you would know if anyone had been registered blind; there isn't anyone'. Tim's experience is fairly common: the majority of participants in this thesis were unaware of a family history of LHON.¹³

In the following section, I discuss the challenges which participants encountered in being diagnosed with LHON (with particular focus on the eye clinic), and how the diagnosis was the start of a quest to understand the aetiology and prognosis of their condition.

The clinical encounter

Medical sociologists have found the work of the clinic to be a fertile ground in which to undertake research—particularly in the field of genetic diseases. Studies have considered diverse topics including the use of pre-implantation diagnosis (Franklin and Roberts 2006), amniocentesis (Rapp 2000), genetic counselling (Bosk 1992), and dysmorphology (Shaw *et al.* 2003; Featherstone *et al.* 2005; Latimer *et al.* 2006; Latimer 2013). Previous research with families at risk of developing Huntington's disease (Konrad 2005) has highlighted the significant impact which knowledge gained from genetic testing can have on an individual's life and their relationships within the family. Konrad's study has resonance for this research in that it highlights the ethical dilemmas faced by women who are asymptomatic. In the case of LHON, this includes mothers who may not have experienced symptoms and who were unaware of their carrier status until their child was diagnosed. The identification of a genetic eye condition can have far-reaching consequences, not only for affected individuals, but also for other family members, including mothers and siblings (discussed in Chapter Five).

The clinic has been identified as an important site for the discovery of modern medical knowledge (Latimer *et al.* 2006). It is in the clinic that medical knowledge is produced and reproduced—where medical conditions are 'created' rather than simply being 'discovered' (Featherstone and Atkinson 2012: 25). It is through the 'spectacular display and representation of bodies, organs and pathologies'

¹³Evidence suggests that up to 40% of individuals do not have a family history of LHON (Harding *et al.* 1995). However, it is suspected that these families most likely represent cases where family history is difficult to trace given that de novo mutations are rare in LHON (Biousse *et al.* 1997; Man *et al.* 2003).

(Featherstone *et al.* 2005: 552) that disease is identified. Foucault (1973: 108) talks of the clinic as the place where the 'clinical gaze' is utilised to look into the patient's body and identify signs of disease. Similarly, Featherstone and Atkinson (2012: 17) refer to the clinic as a place for transforming patients and families into 'clinical entities'. In her ethnographic study of the dysmorphology clinic, Latimer (2013: 28) suggests that the clinic is also the place where new classifications begin to emerge for 'organising patients, medical knowledge and authority'. The process of categorisation is not undertaken by simply fitting individuals into existing diagnostic categories. As Featherstone and Atkinson (2012: 25) note, the categories are constantly evolving by a process of 'description, definition and classification'. Bowker and Star (1999: 86–87) expressed the view that classification systems may be said to 'provide a stabilising force between the natural and the social worlds...they hold in place sets of arrangements that allow us to read the natural as stable and objective and the social as tightly linked to it'. The act of naming symptoms is a 'prestige-enhancing' (Pinder 1992: 2) achievement for clinicians, setting them apart from the lay person and other medical professionals, confirming greater knowledge and status as well as medical authority (Jutel 2010: 1084; Friedson 1970). Until the medical profession acknowledges and classifies a condition as abnormal, it arguably does not officially exist (Blaxter 1978; Jutel 2009; Jutel and Nettleton 2011).

LHON is a condition traditionally diagnosed in the eye clinic by ophthalmologists who, utilising their clinical experience, identify the presence of classic symptoms (discussed in Chapter One). The ophthalmic genetics clinic is a domain that is increasing being reconfigured by the introduction of genetic technologies. The rapid expansion of next generation sequencing (NGS), and the advent of whole exome sequencing, has brought with it increased opportunities for molecular confirmation of a clinical diagnosis (McKibben *et al.* 2014; Ganne *et al.* 2015). Newman argues that NGS is becoming the 'new' standard for mitochondrial DNA (mtDNA) genome sequencing (Newman 2017: 56). Whilst testing for the three primary LHON mutations continues to be the norm, it is now possible to identify the rarer LHON mutations by undertaking complete mtDNA genome sequencing, which is no longer a 'difficult or exceptional process' (Newman 2017: 56). Featherstone and Atkinson (2012: 10) posit that advances in genetic technologies have not usurped the

practical and interpretive work of the clinician—who still retains the power to fix classifications. The authors suggest that ‘diagnosis is not wholly dependent on the use of laboratory technologies and the clinician’s perception is not reduced to laboratory findings’ Indeed. Hedgecoe (2008a) suggests that clinicians, relying on their acquired knowledge and experience, adopt genetic testing into their clinical practice only when they are persuaded of the clinical usefulness of such tests. The following extract illustrates that, in the identification of LHON it is the ophthalmologist’s judgement that is central to the diagnostic process. Sarah, whose son developed symptoms of LHON when he was 22 years old, attended the eye casualty department. During the day her son participated in numerous tests—which failed to identify the problem. As the day was drawing to a close, the eye casualty specialist registrar (SpR) referred Sarah’s son to a neuro-ophthalmologist who was working in the clinic on that day:

‘We went to A&E and they did a few basic tests and they were stumped by what was going on. After he had the basic initial examination, they said: “He hasn’t got a detached retina, he hasn’t got any bleeding in the eye, and he hasn’t got pressure behind the eye. We had better put him in to see Dr [neuro-ophthalmologist]”. He said straight away, “this is what I think it is”. He couldn’t guarantee what it was without doing the genetic test, but he knew straight away what it was. He said, “I hope it’s not, but this is what I think it is”.’

The neuro-ophthalmologist immediately identified the characteristic signs of LHON and arranged for a blood sample to be sent to the gene panel for confirmation. Sarah’s experience is an exception, as the majority of participants do not receive a diagnosis so (relatively) straightforwardly. Sally experienced symptoms in the mid-1970s prior to the discovery of the three LHON mutations:

‘It [symptoms] started when I was seven. I was passed from pillar to post. A lot of it is really clear because it was one of the most traumatic times in my life. I think they thought at some point that I had had a bang to the head. I seemed to be at the hospital every other day. It would either be to the eye bit or to the neurologist because I think they were looking for tumours and goodness knows what. I was admitted at one point for two weeks and I was on an elderly ward; again it was not very pleasant. I remember a lady dying. It was my first experience of death and it was really upsetting. They [doctors] even joked with me and said I would have my eye taken out and bounced up and down in the corridor to see what was wrong with it. It was ridiculous. For years, I never actually got a diagnosis’.

(Sally, experienced symptoms aged seven, diagnosed aged 29 in 1998)

The clinicians treating Sally trivialised her symptoms by joking that they would bounce her eye up and down the corridor and downgraded her care by discharging her from the eye clinic. Unaware that she had LHON, Sally gave birth to her sons. It was only in her late twenties after her youngest son also experienced sight loss that she was given her diagnosis. The lack of diagnosis has had a devastating impact on Sally and her family.

I asked Dr Morgan (consultant ophthalmologist and clinician scientist with a specialist interest in LHON) to explain the diagnostic process for patients attending the clinic with sudden unexplained sight loss:

‘Immediately you would do a dilated examination, you would do an OCT [Optical Coherence Tomography] and [retinal] imaging, you would request visual field tests—they would have to have that on their next visit anyway, you probably wouldn’t be able to do them that day. Then you would take the blood samples for just checking whether or not it could be any other inflammatory markers, anything else, causing it [sight loss]. Then you would consider doing electrophysiology and a brain scan. You would not do the gene test first visit—definitely not—but you would probably at least get them back once they have had their brain scan, which would be relatively urgent then do the genetic test. The genetic test would theoretically come at the end.’

(Dr Morgan, Ophthalmologist and Clinician Scientist)

I asked Dr Morgan why the ophthalmic diagnostic tests are not undertaken at the same time. Dr Morgan suggested: *‘We should be doing them at the same time. One of the problems with British medicine is that we do things in order. So, people come back for a year going from one test to another; that’s nonsense, you need to do all of these things at once’*. In the above extract, Dr Morgan describes the process of categorising patients, using both traditional diagnostic tests (such as slit-lamp examination, and OCT) and molecular testing. Dr Morgan, using clinical expertise, inspects the scans and photographs to detect subtle changes and abnormalities in the optic nerve that provide clues to the cause of the sight loss. Atkinson (1995: 196) describes accomplishing a diagnosis as akin to the work of a detective participating in ‘a kind of puzzle-solving activity’. Only after the eye has been subjected to a rigorous examination, and the clinician is highly suspicious that LHON may be responsible, is a blood sample sent to the laboratory. Drawing upon research in the dysmorphology

clinic Latimer *et al.* (2006) describe how patient categorisation is accomplished between the clinic and laboratory, and the decision to ask for a molecular test is a negotiated process, used alongside more traditional means of diagnosis. However, in the case of LHON, the blood test (on occasions) is inconclusive. Ginny was originally tested for LHON when she was 18. The test was negative, as she explains:

‘He [consultant] had a suspicion that it was LHON, but he didn’t do any more tests after it [blood test] came back negative. After that, he then said to my parents “I don’t know what’s wrong with her, she clearly can’t see. I am just going to register [certificate of severe visual impairment] her.”’

Ginny was referred twelve months later to another consultant who sent another blood sample to the gene panel for testing:

‘The first thing she said was she wanted to resend it [blood] because that was her... she was heavily suspicious that it was LHON and she was right. At the time of my first test, they had found the first two [mutations], then they found the third one.’

(Ginny, diagnosed aged 18 in 1999)

Tina recalls that when her blood sample was sent for genetic testing in 2013, the gene panel did not identify any of the three primary LHON mutations. Tina sent a second sample of blood for full DNA sequencing. On this occasion, Tina was diagnosed with one of the rarer LHON mutations.

For many participants, receiving a diagnosis was described as a pivotal moment and one that changed their lives forever. It was also described as the worst moment of their lives, as the manner in which the diagnosis was delivered to them was as traumatic as the diagnosis itself. The way in which diagnosis is communicated by healthcare professionals to patients has been identified as a key area of sociological interest (McLaughlin 2005). Previous studies have explored the trauma experienced by parents when told that their child has a life-limiting condition (Kerr *et al.* 1998; Williams *et al.* 2002). In her research with parents whose child has learning disabilities, Cunningham (1994), identifies three areas of particular concern to parents. Firstly, the diagnosis was delivered in an unsympathetic and insensitive manner. Secondly, inadequate information was given in respect of the diagnosis and

possible treatment options. Finally, there was a lack of signposting to access help and resources.

The views expressed by participants in this thesis mirror Cunningham's arguments. Some participants felt that the way they were given their diagnosis caused them unnecessary distress. This was particularly the case when an ophthalmologist told them that there were no treatment options or cure and they would be registered as severely sight impaired and discharged from the eye clinic. Many participants expressed the view that the prognosis could have been dealt with more sensitively. This was particularly the case when the inheritance of LHON was discussed. In the following extract, Marion, who attended the appointment with her affected son David, and her daughter, Naomi, a carrier, explained how the diagnosis was given:

'He (ophthalmologist) turned to us and he said, "there is no other way to break bad news", looking at me, "you are a carrier, you will pass it on to all your children" and then pointed to my daughter and said, "you will pass it on to all of your children; all of your children will be born carrying the disease". He said to my son "you will go blind". Bye, bye, thanks very much.'

Marion described the consultant's manner as abrupt; her daughter Naomi also alluded to this:

'I remember that moment of it [diagnosis] being a really horrible moment. He [consultant] was very blunt... said pointing at me "you carry it and all of your children will be affected". Doctors have always got a hard job delivering bad news, but it felt quite brutal. I remember that feeling of like hot tears that I couldn't control.'

Other participants gave similar accounts of the moment they received their diagnosis. Ginny, who was 18 when she was diagnosed, has a vivid recollection of the ophthalmologist giving her the LHON diagnosis: '*you are not going to die from it sort of thing and I thought he was an arrogant "what's it" (laughs)*'. Jake was diagnosed when he was 15 years old and remembers attending the appointment with his parents:

'The consultant basically said I am going to be certifying you blind now. I will never forget—there are certain things you will always remember. Bearing in mind my age at the time—I was looking forward to having a

moped and driving in the next year or so—he said, “you can get a third off your rail travel” and I just thought that’s not really great.’

(Jake, diagnosed aged 15 in 2005)

Participants also referred to the lack of information or specialist referrals to genetic counselling or sight loss agencies. Ollie (discussed earlier) sums up the experience of many of the participants:

‘The post-diagnosis care was, quite frankly, shambolic, in terms of the inability of some of the most sophisticated eyesight hospitals in the world to signpost you on to your local sensory team is quite frankly disgraceful. I had to basically *feel* my way to [name of local authority]. I turned up with a scrap of paper [Certificate of Visual Impairment], at which point they put me on to the sensory impairment team. If I hadn’t demonstrated that drive and determination to get there, I don’t think I would have been signposted to that place to this day. It is incredibly important for there to be a successful transition from a diagnosis on the medical side to a rehabilitative stage, which is really a partnership between the local council’s sensory impairment team and local charities.’

(Ollie, diagnosed aged 18 in 2009)

All participants raised concerns that when they received their diagnosis, they were given very little information before being discharged from the eye clinic. When participants were given the opportunity to see a genetic ophthalmologist, they ensured that they obtained as much information as possible. To illustrate this point, I now draw upon fieldnotes taken from an observation at in Dr Penvenen’s specialist LHON clinic in The Royal Albion Hospital. I arranged to be present in the clinic as Garyn was attending his first appointment with Dr Penvenen. Garyn experienced bilateral sight loss in 2015 when he was aged 21. Initially diagnosed with optic neuritis [inflammation of the optic nerve], the ophthalmologist in Garyn’s local hospital, suspicious that he may have LHON, sent a blood sample to the gene panel which confirmed the LHON m.14484T>C mutation.

Prior to the appointment, I spoke privately with Garyn, his mother, Lowri and his father. The account they gave of the delays in receiving a diagnosis are echoed in the narratives given by other participants. The family also expressed concerns that, following the diagnosis, they were given little information on the prognosis of LHON or risk to other family members of developing sight loss. In his study of the

interaction of clinicians and parents in a paediatric clinic, Strong (1979) suggests that the clinical appointment is conducted within a bureaucratic role format. Doctors and parents are subject to specific rules of engagement in which they adopt complementary roles to produce a ceremonial order of the clinic. Parents within the clinical hierarchy are relegated to being seen and only heard at the invitation of the clinician. It is clinicians who exercise their authority in determining what will be discussed.

The appointment between Dr Penvenen and Garyn began with a discussion of the family tree:

Dr: Looking at a very unusual family tree I have here [talking to Lowri], you are one of five siblings and you have an older brother who is 29 and the youngest is your sister, she is 20. There is a suggestion here of a distant cousin.

Lowri: A cousin of mine, she was diagnosed with MS going back about 30 years ago. She lost her vision. She is totally blind. They told her the MS had caused her to lose vision. She accepted that, but she saw a new consultant and he suggested that she had Leber's and they did the test and it came as the...

Dr: (turning to Garyn) when did you start having problems with your vision?

Garyn: Middle of July last year [2015]. It started in both eyes at the same time.

Lowri: (interrupting Garyn) we detected it in July on the night of his Graduation. We went out for a meal and I noticed he was squinting slightly at the menu. I suggested he went to have his eyes tested. It was just a bit of a squint and the following week everything had gone blurred.

In the early stages of the appointment, Lowri answers questions on Garyn's behalf, before he has the opportunity to answer the questions. At this point, Dr Penvenen examines Garyn's eyes and relays his observations:

Dr: There has been some visual recovery (reading the notes). Of the three mutations 14484 has the best visual prognosis and the best chance of spontaneous recovery. If you are going to recover any vision, and that normally happens within the first year, but it can happen up to five years.

Lowri: Excellent.

Dr Penvenen takes the opportunity to discuss the inheritance of LHON:

The important thing to say is that, in terms of the genetics, it goes down the maternal line because the mutation's spelling mistake is in your mitochondrial DNA. These mitochondria are like little batteries in your cells and they are the ones that produce enough energy. It is very straightforward—if the mitochondria in your cells are not working properly, you do not produce enough energy and, therefore, the cells at the back of the eye which form the optic nerve stop working properly. It is a very simple system: the eye is a camera, the brain is a computer, and in between the optic nerve is a high speed broadband cable. In Leber's optic neuropathy it is the cable which gets damaged. The eye is always fine, the brain we hope is okay (everyone laughs).

At this point, Lowri reaches into her handbag and produces a piece of paper on which she has written a list of questions and systematically reads through the list:

Lowri: Does age help in recovery? Because I have read up to the age of 20 or I heard it on YouTube.

Dr: Normally it is less than the age of 12. I think at the age of 21 you fit the adult criteria. I don't think that would apply in your case. Unfortunately, there are a lot of unanswered questions. I think in your case... because if it all goes well, and you recover some more vision... the important thing is to try and maintain a reasonably healthy lifestyle.

Once again checking her list, Lowri asks about the availability of new treatments:

Dr: There is the gene therapy. We are doing gene therapy at the moment but only for patients with the 11778 mutation and who have had the condition for less than one year. We are not doing gene therapy for your mutation, only the 11778 mutation.

Lowri: Is there an option to pay privately for the gene therapy?

Dr: No, there is none because this is entirely under research. The other thing that might happen as a result, if you are happy, I will keep you on my research database. There might be other clinical trials coming around the corner. Things are moving in the right direction, but things are pretty much limited at this point.

Lowri becomes tearful that Garyn is not eligible for the current gene therapy trial and continues to try and persuade Dr Penvenen to allow Garyn to take part in the trial. It is at this point that Dr Penvenen, reiterates that it would not be ethical for Garyn to participate in the gene therapy trial, nor is there the option to pay privately to be included. However, Dr Penvenen is able to placate Lowri by indicating he will

include Garyn's details on a database for future clinical trials. Lowri, now reassured that Garyn may be eligible to participate in future clinical trial, turns the focus of attention towards her concerns for her own eyesight. Here, it is worth remembering that when Garyn was diagnosed, Lowri also became a patient, as she is a carrier of the LHON mutation. Whilst Lowri is currently asymptomatic, she is at risk of developing problems with her eyesight. Lowri has entered the menopause and asks Dr Penvenen whether she should take hormone replacement therapy (HRT)¹⁴:

Dr: There are two mysteries with this condition. One is if you have the mutation, it does not mean you are going to lose vision. But purely from a statistical point of view, the risk is about 50 per cent if you are a man and 10 per cent if you are a woman. But there are known risk factors; if you smoke, then your risk is much higher. The second mystery is why does it tend to affect men in preference to women? We don't know why this is the case. There must be other genetic risk factors which influence whether or not you lose vision. What these other genetic risk factors are we don't know. As to whether a woman has an increased risk around the perimenopausal period is quite a contentious issue. What I normally say is that you should not be taking HRT just because of the Leber's problem. You should only be taking HRT if there are other reasons for taking it... so having very bad menopausal symptoms.

Throughout the appointment, Lowri was acting as a gatekeeper and transmitter of genetic knowledge by actively asking questions on behalf of the family. Arribas-Ayllon *et al.* (2008: 1522) suggest that the 'new genetics' still relies on the 'old structures of kinship', where responsibility for the family's health falls disproportionately on women. Garyn only spoke when asked a direct question, often deferring to his mother who repeatedly answered questions on his behalf. Garyn's father sat as a passive observer to the events that unfolded in front of him. Throughout the appointment, Lowri takes the lead and plays the role of the informed patient. Starke and Möller (2002: 245) argue that parents, in reacting to their child's diagnosis of a chronic illness, seek information and gain knowledge about the diagnosis as a way to 'restore order in a chaotic existence'. One of the first

¹⁴ Research (laboratory tests) have suggested that the female hormone oestrogen may protect cells from the effects of LHON (Giordano *et al.* 2010). However, there are also reports of cases where low oestrogen levels might have triggered LHON in females (Badura-Stronka *et al.* 2013).

places families seek information is from the internet, prompting Nettleton (2004) to suggest that formal medical knowledge has escaped (or 'e-scaped' to be more precise) and is no longer hidden within medical institutions but instead is available in books and through internet search engines. In a similar vein, Dew and Jutel (2014: 70) note that the availability of medical knowledge has reduced the authority of the doctor, creating the 'informed consumer rather than the acquiescent patient', with Petersen (2006) suggesting that patients and their families are in a unique position to provide valuable information to their doctor. Although to what extent it may be said that this acquired knowledge elevates them to the position of an expert is contentious. Whilst acknowledging that patients possess extensive knowledge of their own lives and their medical conditions, Prior (2003: 45) suggests this does not make them experts as they lack the necessary skills to gather medical facts and undertake the 'business of medicine'. Nonetheless, a number of participants in this thesis do consider that they know more about their condition than their ophthalmologist. Adrian, following his son's diagnosis, was instrumental in establishing a sight loss charity to support families with LHON and has acquired considerable expertise:

'I have become a mini expert in this [LHON]. The last few times that we saw the consultant ophthalmologist in [name of hospital], he actually said to my son "I don't know why you bother coming to see me, your father knows far more than I do about this". Without any question I do. On the other hand, I am not an ophthalmologist. [...] I can look at several photographs of the optic nerve and they all look the bloody same to me; it takes a trained eye. It takes the sort of years of knowledge and experience that I don't even begin to pretend to have. So, if I say I am an expert, I know just how much it is I don't know. I am trying to educate myself about the whole genetic side of things, as in the specific mutations et cetera, but I must admit I struggle; it doesn't come easy for me.'

Reinforcing the comments made by Prior, although considering himself an expert, Adrian acknowledges his limitations; he does not have the technical skill to look at the retinal imaging scans and identify LHON. Adrian has continued to acquire knowledge by attending both national and international patient conferences and is extremely proactive in working with other stakeholders to raise awareness of LHON (discussed in Chapter Seven).

Earlier in this chapter, I referred to Jutel's analogy of the roadmap used by the doctor to arrive at the right destination—in this case a diagnosis. However, ophthalmologists (when diagnosing LHON) frequently struggle to read the road map. In the following two sections, I consider the consequences of the failure by ophthalmologists to identify symptoms of LHON, resulting in diagnostic uncertainty and contested diagnoses.

Diagnostic uncertainty

It is apparent from the foregoing discussion that the art of diagnosis is a complex process. On occasions, it is also pervaded with uncertainty; this is particularly evident in the diagnosis of rare diseases (Rare Disease UK 2016). LHON is a condition that is shrouded in uncertainty in respect of its aetiology, treatment options and prognosis. The participants in this thesis present to the eye clinic with diverse symptoms; whilst some retain very poor residual vision, others maintain a good level of both central and peripheral vision. Conrad (1987) suggests that there are several types of uncertainty which, I would argue resonate with the experience of participants in this thesis. Firstly, the uncertainty felt by participants when they notice subtle changes in their eyes and seek medical attention. Secondly, medical uncertainty is experienced by clinicians who attempt to provide a diagnosis, often misdiagnosing LHON, and is also experienced by participants who are subjected to numerous tests including MRI scans, and imagine they have a life-threatening condition, such as a brain tumour. Thirdly, there is the uncertainty following the diagnosis when questions are asked such as 'will I be able to carry on working? Or will other people treat me differently?' Finally, there is what Conrad refers to as 'trajectory uncertainty' (1987: 8). LHON is a condition that is unpredictable, as some participants experience spontaneous recovery, whilst others become anxious that their sight will continue to deteriorate. There is also uncertainty as to whether other family members will become affected.

Atkinson (1995: 110) has argued that uncertainty is one of the most 'pervasive' and overworked themes in medical sociology, with an over-reliance on the concept in discussions around the boundaries of medical knowledge, medical education and the relationship between doctors and patients in the clinical consultation (Atkinson 1995). There is a considerable volume of literature within medical sociology that

has considered the concept of uncertainty within medical settings (Parsons 1951; Fox 1957, 1980; Davis 1960, 1963; Freidson 1970; Atkinson 1984, 1995). Much of the current scholarship has been developed with reference to the study 'Training for uncertainty' by Renée Fox (1957). In her work, Fox explored the education and socialisation of medical students and, in so doing, highlighted how in their early medical training, and later during their early professional development, they were able to identify the various dimensions of uncertainty in their own knowledge and the limits of medical knowledge, more generally. Fox argues that, in Western society, the doctor is regarded as an expert equipped with skills to cure all illnesses. However, this 'utopian view' (1957: 208) of the doctor is not consistent with reality. The knowledge and skills possessed by the doctor are not always adequate to identify disease or prescribe a cure, notwithstanding advances in medical science. The doctor's life, therefore, is full of uncertainty.

Fox (1957: 208) articulated two origins of uncertainty in diagnoses and medicine. Firstly, there are limitations of individual knowledge: 'no one can have at his [her] command all skills and all knowledge'. Secondly, the limitation of current medical knowledge means that it is not possible for medical students to have all the answers. As Groopman suggests (2007: 7), 'medicine is, at its core, an uncertain science'. Participants during interview with me, explained that when they attended eye casualty they were examined by a specialist registrar (SpR) who triages the patient and makes the decision to either refer into the eye clinic—to be examined by a consultant—or alternatively discharges them. For a number of participants, the SpR failed to include LHON in the differential diagnosis and they were subsequently discharged from the eye clinic and told to return if the symptoms persisted. This was the experience of Andy, who having been referred to eye clinic, was examined by a specialist registrar:

'He [registrar] did loads of different tests and things and said he thought that it was an eye virus and so, "People," he said "can get eye viruses. They are not totally uncommon but what it is with them is that the sight will come back in 10 weeks". He advised waiting 10 weeks to get my sight back. When I asked what might cause it, he said it could be stress.'

Andy was given a diagnosis of an eye virus. Other participants after undergoing numerous diagnostic tests were initially discharged from the eye clinic with a

diagnosis of optic neuritis. Timmermans and Buchbinder (2010: 410) suggest that prolonged diagnostic uncertainty is uncomfortable for patients and clinicians who, they suggest, 'will exert pressure to fold an incomplete characterization back into more conventional categories'. Andy's eyesight continued to deteriorate, and on returning to the hospital, he had further diagnostic tests before being diagnosed with the LHON m.11778G>A mutation:

'I went back to the hospital to see the ophthalmologist and they were quite concerned because they said it's affected the other eye so it's basically not the virus. I had various tests and I can't remember all of them, to be honest with you, brain scans, et cetera to see about MS [multiple sclerosis] and different blood tests and different things like that. They thought that there are three options. In effect, it's either LHON, or MS, or it's a brain tumour.'

Andy received his diagnosis five months after initially experiencing symptoms. One of the other male participants, Ollie, was 18 years old when he experienced symptoms of sight loss and, like Andy, also experienced delays in receiving his diagnosis:

'At the hospital, LHON is not a condition that they come across frequently at all. There was a huge amount of ambiguity and confusion into what I was suffering from. I was obviously suffering quite severe symptoms in terms of visual acuity and my visual field decreasing rapidly in my left eye. Within two months of suffering those problems, I think that problem had transferred to my right eye. I undertook a lumbar puncture, CT scan, MRI scans just to check whether there were any physiological issues, neuron-degradation in my head, to see whether it [could] be any other condition, including MS [multiple sclerosis]. But those tests came back negative. In terms of doctors spotting that, ironically, it wasn't anywhere which deals very specifically and strongly with VI. It was actually at [hospital] where a second year F2 medic, who was incredibly junior, spotted it and recognised it from his rare diseases module and said it could be Leber's. I had genetic tests which took another six weeks to turn around. It was seen that there was a point mutation on the 11778 gene on my mitochondrial DNA, which is effectively a positive diagnosis for LHON.'

Andy and Ollie's experience of being subjected to numerous diagnostic tests is typical of the experiences described by other participants. What is unusual in Ollie's case is that it was a medical student who had recently studied the rare disease module during his training. Given the delays which participants encountered in

obtaining their diagnosis, I asked Dr Penvenen to explain the challenges presented to ophthalmologists in diagnosing LHON:

‘This is a grey area, it can be from the no brainer where you have someone who is presenting with bi-lateral visual loss in a family known to have the disease. You may have someone, a young man who presents with a bi-lateral optic neuropathy and the original scans are normal with nothing but optic neuritis. Of course, LHON is going to be part of the differential diagnosis right from the start. So, if someone presenting [in the clinic], it can be very few things that are going to result in such a rapid bi-literal loss of vision. It is a medical emergency; as you can expect, everyone is freaking out by the time you have excluded inflammation, a tumour or something else in the brain. You have to think about LHON, that’s what I always drill into people’s brains.’

The experience of participants in this thesis suggest that LHON is not routinely considered. I asked Dr Penvenen if in reality LHON is always included in the differential diagnosis by the juniors:

‘I think probably, for the juniors, they would probably not think about this straightaway. In all fairness, if someone is presenting in eye casualty with bi-lateral visual loss, they should be really thinking about the more common causes because it [LHON] is still quite rare. I think it is a balance really. I think the juniors have this thing, looking at the common things— inflammation, optic neuritis, tumours and all the rest. I think by the time someone will present acutely and that they will be having initial investigations and get referred to a clinic for their re-assessment, I think in the bigger teaching hospitals, most of the patients will get referred to a neuro-ophthalmologist. I think the difficulty is when someone presents in a district general hospital or they move from eye casualty to a general clinic and then people still do not think about LHON because it is still quite rare, that’s where there are major delays.’

As highlighted in Chapter One, LHON has historically been characterised as a condition which predominantly affects young males in their late teens and early twenties (Yu-Wai-Man *et al.* 2014). However, a number of participants in this study (men and women) experienced symptoms in later life. The preoccupation with exclusively young males has contributed to misdiagnosis of the condition for a number of participants. Sandra was in her late sixties when she was eventually diagnosed with LHON. She had previously undergone surgery for glaucoma and realised that her sight was not improving. When asked if she was given any indication that they thought it might be LHON, she answered: *‘all that was said... the registrar had said to me at quite an early stage “oh it can’t be LHON, you are far too*

old". He said it in quite a jokey way (laughs)'. Gerry, who was diagnosed with LHON when he was 62, had a similar experience:

'I saw one of the junior members of the team and was talking to me about it and he was looking and thought he saw some form of cataract. I then saw a different consultant and she couldn't see any sign of a cataract but thought it was PVD [posterior vitreous detachment]. So it's the sack around the eye that holds the fluid in. When you get to a certain age, it starts to crack up and you get lots of floaty bits and distorted vision. She did a lot of prodding and poking; she was convinced that was it and told me not to worry, that it would be problematic at the moment, but it would eventually get better and my vision would return to normal. It [LHON] was missed because they automatically assumed that I was too old to have it because they saw it as a young person's problem; they dismissed the diagnosis as far as I was concerned because I was of the wrong age, it didn't fit his pre-determined concept. The three consultants at [hospital] missed it and were unaware of it.'

Whilst Gerry was initially examined by one of the juniors who failed to diagnose LHON, he was also examined by three consultants in his local hospital, who again failed to diagnose the condition. Sandra and Gerry's experience is consistent with Latimer's (1997, 1999, 2000) research in an acute medicine unit. Latimer (2000) observed that older patients' symptoms (breathlessness and chest pains) are reconfigured as social rather than medical problems. Latimer further suggests that 'older people have been targeted as inappropriate users, and even as "misusers" and "abusers" of acute health services' (Latimer 2000: 387).

The experiences of participants suggest that LHON is a condition that experienced ophthalmologists do not always consider when dealing with bi-lateral sight loss. When there is a known family history of the condition, LHON may still be overlooked in the differential diagnosis when a woman presents with vision loss. This is illustrated by the experience of Beth, whose brother had been diagnosed with LHON 25 years before. Beth first became aware of problems when she could no longer read a magazine. Having been referred to the eye clinic, several months passed before Beth's blood sample was sent to the gene panel:

'One week, I went five times [to hospital]. I had a VFT [visual fields test] and then I went for an MRI scan. All these tests they were doing, they were all coming back normal. First of all, we were hoping there would be a blocked blood vessel behind my eye which they could do something

about. I was even hoping it might be a tumour there. Anyway, he [consultant] eventually referred me. He said “There may be a chance you might have Leber’s”. He referred me to [consultant neuro-ophthalmologist]. He did various tests and all these tests were coming back normal. And I knew it was only going to point to Leber’s. So anyway, I decided to have the blood test. I paid for it myself.’

To speed up the diagnostic process, Beth paid privately for her blood sample to be sent to the gene panel. The delays in receiving genetic test results was another issue identified by participants, some of whom explained that they had waited up to eight weeks for confirmation of their LHON diagnosis. I asked Dr Penvenen whether it was usual for there to be delays in receiving the genetic test results:

‘I think the major issue is the fact that now, even in major centres, the turnaround time for tests for LHON is still eight weeks. It’s four months at [hospital] at the moment. There is also a problem with the delay in getting genetic results back.’

Dr Penvenen suggest that delays occur for a number of reasons. Firstly, hospitals do not always send samples to the gene panel straightaway. Financial constraints within the eye clinics result in samples being held back, particularly toward the end of the financial year. Secondly, in an age of austerity, cuts to the NHS budget has impacted on recruitment of specialist clinicians to undertake the testing.

In contrast to the older participants, very young participants were also considered to be unlikely to have LHON. Declan experienced symptoms when he was 11 years old and was subsequently diagnosed when he was 12 years old. His mother, Gwyneth, explained why LHON was initially dismissed:

‘He [consultant] didn’t think it was Leber’s because of him [son] being so young. He said it didn’t really affect boys until their late teens. That’s why it took such a long time for the diagnosis and making the connection because he was only 12.’

Four of the female participants in this thesis experienced an added dimension to the uncertainty surrounding their diagnosis as, in addition to being affected by sight loss, they also presented either simultaneously or at a later date with multiple

sclerosis-like (MS)¹⁵ symptoms (referred to as Harding's disease).¹⁶ Laura's account of being diagnosed is typical of the other three women in this study:

‘There were doubts about it [diagnosis] and we played a lot of ping pong because no one was singing on the same hymn sheet. It [diagnostic process] did go on for quite a long time; it took a long time for them all to say yes, it is Harding's disease. Then some consultants have said “what is Harding's disease?” [...] It's very confusing because some consultants say I have MS and I also have Leber's?’

(Laura, diagnosed aged 48 in 2012)

The terminology to describe the symptoms of sight loss with the MS-like symptoms is contested as I discovered when I attended Dr Morgan's clinic in St Tristan's Hospital. Two sisters attended the clinic who had been diagnosed with LHON, but also experienced the MS-like symptoms which they also referred to as Harding's disease. When I also used the terminology in conversation, Dr Morgan, quickly corrected me: *‘Don't call it that, if you walked around the eye clinic and asked any of the consultants what Harding's disease is, they wouldn't know what you were talking about’*. Here, it is clear that some general ophthalmologists are not familiar with LHON since it is so rare. It is only when an experienced genetic ophthalmologist examines the patient that the symptoms are observed. Reflecting on whether there is adequate training for ophthalmologists in identifying rare inherited eye conditions, Dr McDaid, a neuro-ophthalmologist with extensive experience of diagnosing and treating LHON patients, suggested that:

‘The problem is... of course, it is very easy to say they [juniors] should know more but the reality is that there are lots of things they should know more about and, you know, there is only a limited amount of time and there is only a limited amount of memory and brain power in any of us to cope with all the information.’

This view resonates with the work of Becker (1961) and his colleagues who observed that medical students are introduced to a vast body of knowledge which it

¹⁵ MS is a condition that can affect the brain and/or the spinal cord causing a wide range of symptoms including sight loss. MS is not directly inherited; however, research has suggested that people who are related to someone with MS are more likely to develop it (NHS Choices 2018).

¹⁶ LHON has also been associated with multiple sclerosis-like (MS) symptoms in females who carry the m.11778G>A mutation. Harding *et al.* (1992) first described the symptoms following their research with eight females who presented with bilateral, sequential sight loss. Six of the females later developed a neurological syndrome indistinguishable from MS.

is impossible to master. Fox also identifies a third source of uncertainty consisting of difficulty in distinguishing between 'personal ignorance or ineptitude and the limitations of present medical knowledge' (Fox 1957: 208). Later in the same interview, Dr McDaid suggests that:

'They [juniors] over-investigate because there are all sorts of ideas in their head and they don't have enough experience to be able to pin it down to just one or two and very quickly get to the answer with some clinical tests, instead they just order loads of tests. I have just done a ward round this morning at one of the hospitals I work at and, you know, the juniors there over the Christmas period have ordered all sorts of scans and blood tests which were total unnecessary and that does reflect that they are more junior. They are more anxious about missing something, they are less trusting of their own clinical skills to interpret the signs and so they do a lot of 'just in case' kind of investigations.'

Fox suggests that medical students are often expected to 'see before they know how to look or what to look for' (1957: 214). LHON is a rare condition that is not often encountered in the clinic. Dr Morgan was also asked whether there was adequate ophthalmology training for the rarer eye conditions:

'I think as trainee ophthalmologists, I know that all of them are completely aware of Leber's. I mean we were all aware of it even though we had never seen a patient. Everyone knows if you set them an exam question, they would all be able to write about it. But that's not the point. They need to be able to spot somebody that's suspicious. The point in a way is that it's not that they don't know about it, of course they know about it. You set any of them an exam question about it, and of course they will be able to parrot what it says in the text books. But there is a very big difference between knowing about the condition and being able to spot it.'

Ultimately, Fox concludes that training for uncertainty is an essential component of the medical curriculum as it prepares medical students to deal with the challenges they are confronted with in their medical training and clinical practice. Managing uncertainty is therefore considered to be an important aspect of professional socialisation. Fox's work has been critiqued, most notably by Atkinson (1984, 1995) who is critical of the treatment of uncertainty within the sociology of medicine. He delineates it as 'inadequate and incoherent' (1984: 949) and considers that it has lacked sufficient theoretical analysis. Atkinson proffers an alternative approach to

uncertainty by suggesting that whilst medical students are required to assimilate a considerable body of knowledge and grapple with over-burdened curricula, this does not necessarily result in uncertainty. On the contrary, he contends that medical students adopt a 'pragmatic approach to the accumulation and construction of a stock of knowledge' (1984: 952), which they call upon to navigate their way through medical training and clinical practice. For Atkinson, it is 'certainty' that reigns supreme. Atkinson goes on to argue that whilst 'medical knowledge and practice are inherently "uncertain" [...] the certainty of dogmatism and personal judgement are responses to that on the part of the clinician' (1984: 954).

Other sources of uncertainty within the clinical encounter have been considered by Olsen and Abeysinghe (2014), who focus on patients' presentation of information to the doctor. Patients may not disclose key symptoms because they fail to appreciate their significance, resulting in uncertainty in the mind of their doctor. Whilst acknowledging that patients do provide reliable accounts of their illness and family history, Gardner (2014: 152) suggests that, equally, patients can be emotional and allow irrational fears to hamper their ability to act as 'reliable observers of their condition'. In his study of paralytic poliomyelitis [polio], Davis (1960, 1963) identified several dimensions of uncertainty arising in the doctor-patient encounter. Davis (1963: 67) suggests that, on occasions, doctors "feign" uncertainty with the purpose of breaking bad news in a controlled way, allowing patients to come to terms with the diagnosis:

'Uncertainty is to some extent feigned by the doctor for the purpose of gradually—to use Goffman's [...] analogy— "cooling the mark out,"—i.e., getting the patient ultimately to accept and put up with a state-of -being that is intolerable to him.'

In his research with people diagnosed with epilepsy, Scambler (1989: 25) found that his participants often used real or feigned uncertainty as 'tools of negotiation' in an attempt to avoid being labelled as an epileptic. Davis (1960) also describes how doctors, when dealing with an uncertain diagnosis or prognosis, will either disclose their uncertainty (admission of uncertainty) or adopt a process of dissimulation in which they fail to communicate the uncertainty, but instead adopt delaying tactics or provide (possibly ineffective) treatment. Davis suggests that dissimulation is particularly prevalent when the doctor perceives that an admission would damage

their professional reputation. Dr McDaid was asked why he thought that general ophthalmologists were slow to refer cases onto specialists:

‘What is needed is for my colleagues who are not experts in this area, to have a lower threshold to refer onwards to specialist centres as quickly as possible. What I think should be improved is not the training of doctors but their willingness to say “I don’t know what this is and I better send it quickly on to a place where they see a lot of rare things and they can probably get to the bottom of it quicker than I can.” I think patients languish for a long time in non-specialist secondary care. Then it can be almost grudging. They are sort of “I don’t know, I am fed up with this patient coming every six months or every year and we still don’t know, let’s send it on”. Then you see the patient and, when you do get the diagnosis, you think “I wish I had met you two years ago”, we could have got to the whole thing a bit quicker. [...] I think what we need is for there to be a greater urgency and willingness for non-specialist ophthalmologists to say, “I don’t recognise this, I don’t know what it is” and rather than wasting a lot of time doing tests that are expensive to the NHS and which are on the wrong track, it would be better to send it to a specialist clinic’.

The majority of participants experienced uncertainty whilst waiting to be diagnosed with LHON and, as Dr McDaid notes, the process of receiving the diagnosis could be more effective if ophthalmologists were prepared to refer cases to specialist centres. However, receiving the diagnosis does not bring an end to experiences of uncertainty for some participants, who report experiencing further occurrences of uncertainty in relation to the prognosis of their condition and whether other family members will develop sight loss (discussed in Chapter Seven).

The medical merry-go-round—the challenge of unexplained symptoms

Building on the previous discussion of diagnostic categories and uncertainty prevalent in the diagnostic process, I now turn to consider the conflict that arises between the medical profession and patients when uncertainty in the diagnostic process results in medically unexplained symptoms (MUS) or contested illnesses—in Dumit’s terms, ‘illnesses you have to fight to get’ (2006: 578). Diagnoses are said to be contested when patients identify their symptoms with a specific clinical entity, where the very existence of the entity is disputed within mainstream medicine (Atkinson and Gregory 2008; Trundle *et al.* 2014). Contested illnesses are those for

which the medical profession has failed to find a pathological basis (Nettleton 2006). As Trundle *et al.* (2014: 165) note, conflict arises when ‘illnesses refuse to yield the level of proof that epidemiology, clinical medicine and toxicology require’. The authors go on to elaborate by suggesting that diagnosis is a contested process when a patient explains their symptoms to the doctor who, unable to observe any ‘organic disruption or pathology’ (2014: 166), makes a diagnosis based on the symptoms described by the patient.

The lack of a diagnosis relegates patients to the status of ‘medical orphans’ (Aronowitz 2001: 803). There are conditions that are extremely difficult to diagnose. For example, Lyme disease displays classic symptoms including (but not always) a distinctive circular rash [erythema migrans] which can appear with or without flu-like symptoms (Rebman *et al.* 2017). The disease is often diagnosed on the presence of the rash. However, if the diagnostic test is undertaken too soon or there is a delay in the rash developing, the disease can be dismissed as a virus (Olsen and Abeysinghe 2014). Similarly, LHON is a condition that has proved challenging to diagnose for ophthalmologists who have not encountered the condition in clinical practice. As with Lyme disease, the timing of the initial ophthalmic examination can be crucial, as Dr Morgan explains in the following extract:

‘They [patients] don’t necessarily always have a swollen disc when you see them... that might have subsided by the time you get to see them. The reasons why a general ophthalmologist might miss this is that if they are referred a patient by a GP, it’s not picked up as being urgent. By the time they [A&E] see the patient, they no longer have the classic signs. If you are sitting in casualty as a casualty doctor, you wouldn’t know that it’s Leber’s. It could be five or six or seven different things. You would be trying to exclude any cause of vision loss. The point is, if that person is sitting in front of you and they have virtually lost no vision yet, that can be very difficult. It’s completely dependent on at what stage you meet this person. The paleness of the optic nerve can take months to develop so the swelling can subside, and you will have a phase when maybe the disc is still slightly swollen, but it is a bit iffy. They are certainly not seeing normally, their colour vision is down, their field [of vision] is down, their acuity is down but they haven’t developed the optic atrophy yet. That can take months and, actually, what’s happening is that they are gradually losing more and more vision... it’s quite a difficult stage. Maybe that’s the kind of in-between stage that patients fall into.’

Dr Morgan identifies a number of issues associated with the timing of the ophthalmic examination. Firstly, delays in the referral process occur, either because patients delay acting upon symptoms, or secondly, patients make an appointment with their GP, who does not realise the significance of the symptoms and fail to make an urgent referral. Some participants indicated that when they presented in the eye clinic, the scans of their eyes appeared to be normal and, as such, they were thought to be making up or exaggerating their symptoms. Within the context of his research with patients experiencing symptoms of chronic fatigue syndrome (CFS) and multiple chemical sensitivity (MCS), Dumit (2006) identified a number of cultural, political and structural characteristics associated with contested diagnosis. Firstly, they frequently present as chronic conditions which fail to fit into mainstream understanding of disease. Secondly, they are of indeterminate aetiology and labelled 'biomental', reflecting the dispute as to whether they are primarily mental, psychiatric or biological. Thirdly, treatment strategies include both conventional and alternative medicine. Fourthly, they have fuzzy boundaries and are linked to comorbid conditions, other illnesses with similar symptoms, or are misdiagnosed. Finally, they are considered to be 'legally explosive,' in that patients who are denied disability status and welfare benefits resort to litigation to achieve recognition of their condition and challenge existing diagnostic categories (Dumit 2006: 578).

The lack of a diagnosis challenges medicine's 'modus operandi' which is to identify and treat symptoms. Trundle *et al.* (2014: 165) observe that when presented with a contested diagnosis, the medical profession is faced with managing two competing goals: 'the need to explain illness within the boundaries of scientific certainty and the desire to provide a successful outcome for the patient by returning them to good health'. Becker (1993), who together with colleagues described the socialisation of medical students in the USA, observed that patients who described multiple complaints, but no visible physical pathology, were considered to be 'crocks' (1993: 29). The patients were considered to no longer have the potential to add to the acquisition of medical knowledge. As the patients were not considered to be sick, it was impossible for the medical students to cure them. This, in turn, resulted in the patients becoming devalued in the eyes of the medical students (and discharged as a result) since they failed to provide the *stuff* that medical miracles are made of.

In his study of three Accident and Emergency (A&E) Departments in the UK, Jeffrey (1979) laid bare the process of labelling and categorisation of patients by the medical staff and, in doing so, illustrated how judgements are made about the moral worth of patients based not exclusively on their symptoms but also on their social identity. Patients who were perceived as ‘good’ were described by the medical staff in terms of their medical symptoms, such as head injuries or cardiac arrests—they were not considered to be the authors of their own misfortune. The ‘bad’ patients were identified as “normal rubbish” (for example, drunks, overdoses, tramps and timewasters) (1979: 94). Classification processes have been identified in more recent work too, where certain patients (for example the elderly and the chronically sick) are seen to belong—or not—in the clinical space (e.g. Latimer 1997, 1999, 2000; White *et al.* 2012; Hillman 2014). Gerry, discussed above, talked of his experience in the eye clinic, explaining that it had been a particularly difficult time for him as the consultant was unable to detect a problem with Gerry’s eyes, and failed to identify the symptoms as pathological:

‘He was absolutely clueless. He was looking at my retina and calling me back a couple of weeks later and doing the same thing. It was as though I was talking about the loss of sight and the fact that it was getting worse, but he couldn’t see anything physical that could be accounting for that and it was as though he wasn’t accepting what I was saying about the sight loss because he couldn’t see any physical manifestation to back up what I was saying. We got into an argument with him at one time because I asked isn’t there anything else he could do. I recounted this to my GP. I hadn’t made the connection... it was my GP who looked at me and said maybe he just thinks you are lying... the fact he can’t see anything and what you’re telling him isn’t matching up with what he is seeing.’

Other participants reported that ophthalmologists, who initially failed to identify symptoms of sight loss, suggested they were making up their symptoms or indulging in attention seeking behaviour and, as such, were wasting the ophthalmologists time. This was particularly evident when younger participants presented in the eye casualty department. Amy experienced symptoms in her left eye when she was 18. She was initially examined in eye casualty, but then referred into the eye clinic:

‘They kept referring me to each individual doctor in the hospital, like the next higher one. It was a nightmare. They were doing different tests on my eyes, but they kept finding nothing wrong with them. I saw a consultant and, again, nothing. And he just started scratching his head

and he was like “I don’t know what to do”. I saw a children’s specialist [paediatric ophthalmologist]. She [consultant] did loads of tests on my eyes and Mum was in the room with me. She [consultant] said “I am just going to have a word with your Mum. Do you want to walk out?” I was 18. Mum came out of the room and she looked really tearful and quite upset. I was like “What has she just said?” She [consultant] said I needed a psychiatric report because she thought I was going crazy. I could see some bits but not others. I could see bits because obviously my peripheral vision was still there. It was going but I had bits of sight. I just couldn’t believe it and at that time ...I am a very strong person and I don’t cry very often... but, that time, I broke down in tears because the whole time I was losing it [eyesight].’

(Amy, diagnosed aged 18 in 2011)

Amy’s experience is mirrored in research by Nettleton and colleagues (2014) who explored patient narratives within a neurology outpatients’ clinic in the UK. The research uncovered that a number of patients who were experiencing profound symptoms of illness failed to receive a clinical diagnosis. The authors highlighted recurring themes within the patient narratives which included talk about the illness being ‘imagined’ or ‘fake’ (2004: 47). Describing a similar experience to Amy, Jason, who at the age of 13 was unable to read the blackboard in his French class, spoke of his experience in the eye clinic:

‘I went to see Dr [name of consultant] and he did a series of different tests and looking at coloured dots [Ishihara test], looking in the eye, photographing the retina and the one where you put your chin on the thing [Slit-Lamp] and they look right up close, all sorts of pressure tests. He decided he knew what was wrong. He said to my Dad “Does he stress about things? Does he worry about things?” Dad said “No, not really”. “What about his school work, is he competitive, is he stressed about that?” My Dad gave quite an honest answer, said “No, because he doesn’t do very well at that. He fails at most things. He barely passes so why would he be stressed?” Because the guy was leading up to a diagnosis that it was psychosomatic. That was the diagnosis I walked away from him with. He said, “It will get better itself”. [That] was all I knew or understood.

Did your sight initially improve?

No. By then, I presumed that I had some kind of mental illness because of what I was told. It was me making it up or physically manifesting some kind of mental problem. In the midst of that, I think my Dad said “He is a lot of things but he is not nuts”. I then went through a series of other hospitals.

How many hospitals did you visit?

It was eight or nine, over probably 12–14 months. Fourteen months later I still believed that I am mentally ill.’

(Jason, diagnosed aged 13 in 1990)

Other participants reported that when the ophthalmologists identified out— of — the —ordinary symptoms and acknowledged that they were observing a condition they had not encountered before in clinical practice, participants were suddenly transformed into objects of medical interest (Becker 1993). Sally experienced sight loss when she was seven years old and was discharged from the eye clinic with a misdiagnosis of Neuromyelitis optica (extremely rare neurological condition). She has a vivid recollection of attending appointments with the consultant, who would then call in all the medical students in the eye clinic to examine her:

‘I remember there was always lots of [medical] students called in. I remember once there were lots of student’s visiting from Japan. I was laid in this chair like a dentist chair. I remember all these students looking at me; I just recall it as being really traumatic.’

The problem encountered by many participants is that their ophthalmologist did not recognise the characteristic symptoms of the condition, and several months passed before they received a diagnosis, or were discharged without a diagnosis. Whilst previously there were no treatment options for LHON, the landscape for the diagnosis and treatment of LHON is changing rapidly. New treatments are currently the subject of clinical trials to restore sight (discussed in Chapter Seven). However, delays in diagnosis have the potential to impact on the effectiveness of these new treatments. As Dr Penvenen outlined:

‘I think it [diagnosis] is really going to be a major issue. If, for example, the gene therapy trial is seen to work for patients with 0–6 months’ disease duration, but not from 6–12 months this will mean you have a very narrow critical window of opportunity. If and when there is a treatment that works, if the treatment only works in a very narrow window, then the pressure will be on [to diagnose LHON quickly].’

For patients with a rare condition, obtaining a diagnosis may be particularly troubling. In 2016, Rare Diseases UK published their report, *The Rare Reality*, in which they identified a number of key findings. Firstly, patients and families were frequently given limited information about their condition by health professionals. Secondly, patients face significant delays in securing a diagnosis, with the majority

receiving at least one incorrect diagnosis. Thirdly, patients experience difficulty in persuading health professionals to believe their symptoms and their condition and are initially written off as 'psychological' and, in some cases, 'neurotic'. Finally, not receiving a diagnosis is a significant barrier to accessing coordinated care and appropriate treatment. The lack of an effective treatment can exacerbate the challenges patients face on a daily basis. The themes identified in *The Rare Reality* report echo the narratives of participants in this research, who reflected back on their experience of receiving their diagnosis as one of the most traumatic times in their life, in part due to the fact that they were not believed by healthcare professionals, at the same time as they were losing their sight.

Summary

The quest to find a diagnosis is a thread which runs through this chapter. However, rather than a linear process, diagnosis, is on occasions pervaded with uncertainty and contestation. There are two main reasons for this. Firstly, LHON is a condition which relies upon the 'clinical gaze' (Foucault 1973: 108). There appears to be a lack of training for ophthalmologists in identifying rare conditions and, therefore, they fail to identify the characteristic symptoms of LHON in clinical practice. Secondly, participants who delay seeking medical advice may no longer display the textbook symptoms of LHON when they arrive in the clinic. As a result, LHON is overlooked in the differential diagnosis; participants report being discharged from the clinic with a diagnosis of optic neuritis, or they are discharged without a diagnosis as the ophthalmologist (failing to spot the symptoms) believes they are timewasters (Jeffrey 1979: 94) and are either inventing or exaggerating their sight loss.

My findings suggest that the discovery of the three primary LHON mutations, and the introduction of next-generation sequencing, has revolutionised the diagnosis of LHON. However, participants report experiencing significant delays in receiving their genetic test results (up to four months) in some hospitals. Whilst previously delays in diagnosis did not impact on the treatment of LHON, as there were no treatments available, LHON is now a condition that is located within a rapidly changing landscape, with new treatment options being developed. The clinical

evidence (Newman 2017) suggests that there is a narrow window of opportunity in which to start treatments to maximise their effectiveness.

Participants refer to the lack of information they receive at the time of their diagnosis. Referrals to genetic counselling services and sight loss are variable across the UK. Receiving a genetic diagnosis, whilst providing some certainty, also introduces an element of uncertainty for asymptomatic familial carriers as to whether they will also experience loss of vision in their lifetime or whether their children will experience sight loss (Yu-Wai-Man *et al.* 2014). The lack of information at the time of diagnosis provided the motivation for many participants to research LHON using the internet, and subsequently, they become lay experts in or at least extremely knowledgeable about, the condition.

In the next chapter, I will consider participants' initial reactions to their sight loss, and how they struggle to cope with bi-lateral sight loss in the weeks and months following their diagnosis, including dealing with notions of blame, shame, and responsibility.

Chapter Five: “I Don’t Blame My Mother”: Reactions to the Diagnosis

Introduction

In the previous chapter, I highlighted the contested nature and uncertainty surrounding the diagnosis and treatment of Leber’s hereditary optic neuropathy (LHON). In this chapter, I consider how people with LHON negotiate the aftermath (the early weeks and months) of their diagnosis. The point of diagnosis has been characterised as ‘biographically shattering, providing a symbolic marker in patients’ lives from which there is no return’ (Pinder 1992: 2). This is certainly true for many of the participants in my research who claimed that the diagnosis marked a pivotal point, with the realisation that life would never be the same again. They were confronted with an uncertain future, one over which an ‘ominous shadow’ was cast (Pinder 1992: 13). Although not terminal, LHON is a long-term condition that currently has no cure, and limited treatment options, and presents a ‘threat to self and identity’ (Conrad 1987: 11). Bury (1982: 171) has theorised that chronic illness is a major disruptive experience to everyday life—a ‘biographical disruption’ which alters an individual’s life course, forcing a ‘biographical shift from a perceived normal trajectory through relatively predictable chronological steps, to one fundamentally abnormal and inwardly damaging’. Focusing on the destructive effect on identity following the onset of chronic illness, Charmaz (1983: 168) refers to the ‘loss of self’ which she defines as ‘the crumbling away of former self-images without simultaneous development of equally valued new ones’. Participants in this thesis refer to their body letting them down, impacting on their self-esteem as they struggle to undertake simple tasks. Clarke and James (2003: 1387) suggest that the ‘*sine qua non* of the self is the body’, and when the body is no longer able to function as it once did, this has repercussions for the self.

In this chapter, I use the work of Bury (1982, 1987, 1991, 2001) as a foundation to explore the emotional impact of sudden sight loss on participants’ biographies. Bury’s work has been extremely influential in shaping the research into a wide range of chronic conditions, including osteoporosis (Sanders *et al.* 2002), multiple sclerosis (Green *et al.* 2007) and motor neurone disease (Locock *et al.* 2009).

However, there is limited research utilising this theory within the context of sight loss, although two notable exceptions are provided by Green and colleagues' (2002) research with people diagnosed with glaucoma and Larsson and Grassman's (2012) research with visually impaired people living with diverse chronic illnesses. Bury's work is particularly relevant to LHON as he developed his explanatory model from his research with relatively young participants—the majority were aged between 25 and 54—who had recently been diagnosed with rheumatoid arthritis, a condition traditionally associated with older people. Sight loss is often perceived in a similar way since it is expected to emerge in older age with the onset of conditions including Cataracts or Age Related Macular Degeneration (AMD). Yet, as highlighted in Chapter One, LHON affects individuals at any age, but predominantly young men in their teens and twenties.

Having considered the emotional effect of LHON, I will discuss the implications of a genetic diagnosis for the family. With some exceptions (notably Huoponen *et al.* 2002), little research has investigated how individuals diagnosed with LHON understand the inheritance of their condition, and to what extent their (lack of) knowledge influences their decision to share genetic information with family members. Disclosing genetic information raises complex social and ethical issues (Hallowell *et al.* 2003; Clarke *et al.* 2005; Keenan *et al.* 2005; Arribas-Ayllion *et al.* 2008). Drawing on data from my research, and previous research undertaken with diverse inherited conditions, I discuss how participants make the decision to disclose LHON to their family members. I also unpack discourses of guilt and self-blame expressed by mothers, and the extent to which their children blame them for their loss of sight.

Sight loss as biographical disruption

There has been considerable sociological interest in the subjective experiences of individuals diagnosed with chronic illness, which has produced a plethora of classic texts (Bury 1982, 1991; Charmaz 1983, 1991; Williams 1984; Conrad 1987; Corbin and Strauss 1987). Focusing on the onset of chronic illness and drawing on interview data with 30 people recently diagnosed with rheumatoid arthritis, Bury (1982: 167) highlights 'the problems of recognition and changes in life situation and relationships occasioned by the development of the illness'. Unusually for the

condition, the majority of Bury's participants were aged between 25 and 54. Bury observed that the distress experienced by his participants was, in part, underpinned by the cultural belief that arthritis is a condition that is the preserve of older people which, when experienced by the relatively young, suggests they are prematurely aging. George, who experienced symptoms of LHON when he was 16 years old, sums up the feelings of many younger participants:

'I don't want to sound selfish, but you expect people who are 80 and 90, their eyes to get worse as you get older, it's just life. But not when you are 16 and 17-years-old.'

Adopting Giddens's concept of 'critical situation' (1979), Bury posits that chronic illness may be viewed as a 'major kind of disruptive experience [...] where the structures of everyday life and forms of knowledge which underpin them are disrupted' (1982: 169). The onset of chronic illnesses, Bury suggests, is 'insidious' in nature as they 'creep-up' (1982: 170) on individuals. Bury's participants did not recognise their symptoms as the first signs of rheumatoid arthritis. Similarly, as discussed in Chapter Four, participants in this thesis initially dismissed symptoms of LHON as innocuous until their loss of sight reached a critical situation where they struggled to perform daily tasks.

When people try to make sense of the onset of their illness, Bury argues that they locate it within the wider context of their past lives. In this sense, chronic illness can be understood as biographical disruption in that it disturbs not only the physical body, but the trajectory of one's whole life. He further suggests that chronic illness 'involves a recognition of pain and suffering, possibly even of death, which are normally only seen as distant possibilities or the plight of others' (1982: 169), articulating three mutually-dependent aspects of disruption. Firstly, there is the 'disruption of taken-for-granted assumptions and behaviours: the breaching of common-sense boundaries' (1982: 169). In this initial stage, individuals ask 'what is going on here?' which Bury (1982: 169) suggests entails paying 'attention to bodily states not usually brought into consciousness', and decisions about seeking help. Secondly, there are disruptions in the explanatory frameworks normally adopted by individuals, necessitating a 'fundamental re-thinking of the person's biography and

self-concept' (1982: 169). Thirdly, individuals respond to the disruption by the 'mobilisation of resources, in facing an altered situation' (1982: 170).

The onset of LHON for participants was sudden, unexpected and characterised by some participants as a 'bolt from the blue'. They referred to varying levels of disruption being present in their lives in the weeks and months following the diagnosis. Some reflected on their past life which had been extremely active and contrasted this with the life they were now living. This echoes the work of Charmaz (1995: 662) who suggests that at the onset of chronic illness, 'people compare their present body with their past body; they assess the differences between then and now, and they measure the costs and risks of ordinary activities'. In the following extract, George reflects on his previous life, one in which his identity was defined by his sporting achievements:

'I was one of the popular kids in school... and then all of a sudden... I felt there was me who felt helpless in a way. I couldn't even bloody write my own notes down or read from my own book. It was quite hard, well, really hard—just mixed emotions. It was a horrific feeling. It's just like... I am not going to say I wanted to take my own life, but did I ever contemplate it, did it ever enter my mind? Yes, of course it did. Like I was always like top of the sports team, like I used to represent the school at athletics and things like that. I loved my sport, any sport. Obviously, I couldn't do it no more.'

(George, diagnosed aged 16 in 2004)

Similar comments are articulated by other participants, underlining the theme of biographical disruption which emerged strongly in the data irrespective of age or gender. For two of the affected women, the disruption was particularly acute. Although they were aware of the family history of the condition, they were given the impression by other family members that only men would lose their sight. This is illustrated in the following extracts, firstly by Kate whose two brothers had been diagnosed with LHON:

'I was always told by my relatives that it only affects the boys, but you know, I think there is a bit of... umm ...in the family, there is a bit of... they won't find any information out, they have got their heads stuck in the sand about it. I remember my Grandma, she lost her sight when she was 50. My relatives and the doctors, they didn't tell me that females could be affected. But, you know, my relatives could have [told me] because my

Grandma was affected. When I asked my aunts, they are like “oh, we are not quite sure”.’

(Kate, diagnosed aged 48 in 2012)

There are many reasons why Kate’s family may not have discussed the family history with her, including their own lack of knowledge, a desire to avoid causing her distress, or a wish not to allow LHON to influence the way she lived her life. The various factors influencing disclosure of a genetic diagnosis within the family will be discussed in greater detail later in this chapter. Similarly, Beth, whose younger brother was diagnosed when he was in his early 20s, explained that the family were fully aware of her carrier status, but had not recognised that women could be affected:

‘It never really occurred to me that it would be Leber’s because it’s normally... predominantly in the males. I have never worried for myself... never really worried about it.’

(Beth, diagnosed aged 51 in 2015)

In Pinder’s (1992) research with people diagnosed with Parkinson’s disease, she highlights that, after receiving the diagnosis, individuals experience a complexity of emotions and turmoil as one emotion is overtaken by another. This was mirrored in my interviews as participants expressed feelings of embarrassment, anger, frustration, denial and shock. For many of the participants, the diagnosis marked the start of a period of grieving for the loss of their sight. In the following extract, Ollie—in articulating his feelings—also sums up the thoughts expressed by other participants:

‘I was quite conscious of the fact that it was a grieving process. I probably did feel pretty sorry for myself, which I think is very important too. You have got to understand that you have reached a certain low, it would be really ignorant of someone to go through sight loss and not realise it is a huge blow to someone’s self-esteem and confidence. I had gone to University. All of a sudden, I didn’t have anything. I felt pretty sorry for myself.’

(Ollie, diagnosed aged 18 in 2009)

Ollie, after his diagnosis, talks of experiencing low self-esteem and a lack of self-confidence. Echoing Stevelink and colleagues’ (2015a) research with young ex-

service personnel who experienced sudden sight loss, Ollie's feelings are exacerbated because he was no longer able to continue with his education.

Participants also refer to experiencing a range of other emotions, including anger, an emotion that for a number of participants was, on occasions, inextricably linked to feelings of denial. Stevelink *et al.* (2015a) observed that their participants, who were in denial after sudden sight loss, continued to try and perform tasks they had previously undertaken with ease, resulting in feelings of frustration and irritation. In the following extract, Adam, who lost his sight when he was 14 years old in 2004, explains that he tried to carry on his life much as it had been before his sight loss:

'I was in denial by continuing to play football, continuing to try and be something that I wasn't in terms of "I am fully sighted". I still tried to continue to use a phone that didn't talk or didn't have any sort of magnification on it for a while. What was the point? I continued to try and do what I was doing as best I could. I was so angry. I was angry because this whole thing had disrupted my life. It's like in life in general you get yourself into a bit of a groove in life and that's where I had just got myself. The eyesight starts to deteriorate, it all kicks in and then you are out of that groove, you have to start again.'

In attempting to carry on with his life, Adam, is striving to retain a sense of normality (Davis 1995), discussed in more detail in Chapter Six. Jake was diagnosed with LHON when he was 16 years old and as with Adam, tried to hold on to his former life by continuing with his education and working part-time:

'I would say I was in denial and remember at the time I was in year 11. I was doing my GCSEs, I couldn't really stop. The school said I didn't have to do them if I didn't want to. I just thought I need normality, so I carried on going to school. I carried on trying to do my part-time job at the weekend for a long time. But I suppose when I talk about denial, I never tell new people [about sight loss] and I still struggle today. I am better now but I used to lie a lot or do anything to try and cover up. So, I would just pretend to look at my phone if I was lost. It sounds stupid.'

(Jake, diagnosed aged 16 in 2005)

Jake retains a good level of residual vision which gives him the opportunity to 'pass' (Goffman 1968: 92) as fully sighted and, so, avoids telling people that he is visually impaired. He also refers to 'covering' (Goffman 1968: 125) by pretending to look at his phone. Why Jake and other participants are motivated to avoid disclosing their

sight loss will be discussed in Chapter Six. Participants also referred to experiencing suicidal thoughts in the weeks and months following their diagnosis. In his interview, Ollie referred to having “*dark days*” when he experienced suicidal thoughts:

‘It is incredibly difficult to articulate what was going through my head, but I know that there were huge amounts of thoughts about suicide. I feel like it was a natural reflex of the brain to think about that. All you could think about was suicidal thoughts. I know that there are many people that have potentially committed suicide as a result [of LHON].’

Ollie’s comments are consistent with the sight loss literature (De Leo *et al.* 1999) and are typical of both the younger and older participants. At the heart of these emotions is the nature of sight loss as a ‘life changing experience’ (Horowitz 2004; Nyman *et al.* 2010; Pinquart and Pfeiffer 2011; Stevelink *et al.* 2015a), something observed by several of the participants, and highlighted by the following extract:

‘As soon as I knew this was permanent, and this would now be a way of life, suddenly everything became much more of a struggle. Literally two days after, I remember making a cup of tea and I was thinking “God, 22 years or however many years of making a cup of tea, you don’t even think about it, you get the milk out, you put the tea bag in, and you literally don’t even think about it”. Then all of a sudden, it’s a massive struggle. I just remember feeling really, really tired all the time because any menial little task took so much concentration. So yeah, everything was a struggle whatever the task was.’

(Tim, diagnosed aged 23 in 2013)

Ollie also articulates a range of emotions to explain his feelings at the time of the diagnosis:

‘I think the emotions at first are just utter disbelief, unhappiness, devastation. The way it affects people and the age it affects people. You are incredibly self-conscious about yourself, let alone having a disability. It’s a huge amount of shock, a huge amount of horror, and you initially think of “Why me?” You can’t really digest what’s just happened, let alone what’s going to happen in the future. The thought of having limited to no sight for the rest of your life is a pretty scary thought when you are 18. You just want to shut the world out and every time you wake up, you just want to believe it’s not happening.’

Ollie seeks an explanation for his sight loss—a ‘quest for meaning’ (Carricaburu and Pierret 1995; Pierret 2003)—and asks, ‘why me?’ Conrad (1987) suggests that when

given a diagnosis, people are faced with uncertainties which prompt them to question their identity—for example: who are they? How will they be perceived by others? How will the diagnosis impact on their life? What will the future hold for them? Conrad's comments are echoed by Stuart, who had left school at 16 without taking any exams. He was offered what he termed his “dream job” of working on the fishing boats. There was no possibility that he would be able to continue with his chosen occupation after his diagnosis. Stuart recounted his thoughts at the time of diagnosis and how his life would change:

‘I was a fisherman, I was quite an independent person and I did a lot of things on my own. I lived on my own... well, not on my own... I lived on the boat with a group of lads so realising that I was going to lose all of that terrified me. I was processing it all and thinking “What’s going to happen? What am I going to have to do? How is my life going to change?” I used to go out a lot with my friends socialising and I stopped that for a long time, about three months. I spent a lot of time at home, didn’t do the things I was doing before, didn’t play any sport, stopped going to the gym, yes, all sorts of things. But once I had processed it and realised the thing to do was to man up, find out what they [doctors] are going to do, what they can do, can’t do, what I need to do to move on.’

(Stuart, diagnosed aged 23 in 2013)

Stuart's level of residual vision gave him no choice but to give up a job he loved, and he struggles to understand why he has lost his sight. *‘I wondered what I have done to deserve this. All I have done ever since leaving school was I wanted to work hard and be happy and then this happens’*. Stuart's identity was embedded in his life as a fisherman. He lived on the boat with the other fishermen who were not only his work colleagues but also his friends. When he lost his sight, his life changed forever; he could not contemplate a future where he is not working as fisherman. Flurey *et al.* (2018: 115), in their research exploring the impact of rheumatoid arthritis on 22 men, argue that current scholarship has overlooked the impact of chronic illness on masculine identity. The authors suggest that ‘retaining hegemonic ideals of masculinity’ appeared to be important to many of the men, who tried to hold on to activities and roles valued by hegemonic masculinity (for example, sporting activities and paid employment) that they took part in before being diagnosed with the condition. The authors' findings resonate with the view expressed by Stuart, and other men in this thesis, who also struggled to adapt to their new identity. Other

participants also struggled to make sense of why they had lost their sight and spoke of their sense of injustice as they reflected on their life before sight loss, and their feelings that they were somehow being punished for some previous misdemeanour of which they were unaware.

Some of the younger participants expressed the view that their body, or to be precise their eyes, had unexpectedly let them down. Ollie sums this up when he says: *'I don't think you ever associate sight loss with younger people.'* Nettleton (2013: 65) claims that biophysical changes in the body have significant social consequences; 'if we cannot rely on our bodies to function "normally", then our interaction with the social world becomes perilous [...] and our sense of self may be challenged'. In her reflections on the body in the context of health and illness, Corbin (2003: 258) suggests that the body is not just an object to be adorned and enhanced, nor is it simply a 'physiological organism' performing according to a 'prescribed genetic code'. Rather than being just a container of the self or simply a mediator between the self and the world, it is the 'embodiment of who we are'. Corbin and Strauss (1987) observe that when chronic illness enters an individual's life, it separates the person of the present from the person of the past and shatters any images of self that had been held for the future. This is particularly the case when the illness is severe or debilitating. When chronic illness strikes, the body is perceived as failing in some way (Corbin and Strauss 1987). Disruption occurs because tasks that could be completed previously become difficult or impossible to accomplish following the onset of illness. This results in the person of the past and the person they aspired to be in the future, in whole or in part, being rendered discontinuous with the person of the present.

The comments strike a chord with many of my participants who, in their narratives, reminisced about their past achievements which were now just a distant memory. Similarly, the aspirations they held for the future were now considered unrealistic. A number of the younger participants focused on the activities they could no longer undertake. Jake was aged 15 when he first experienced symptoms of LHON. He recounts how he was no longer capable of undertaking simple activities, such as using a mobile phone or computer:

'It was heart-breaking to be honest. It was a massive blow. Obviously, I just put on a brave face but inside it tore me apart really. I just don't know... it was horrible. It was just weird, really different, and I felt different. [...] I just didn't feel able anymore. I know it sounds stupid, but you constantly think about the things you can't do. I couldn't play football with my mates anymore. I couldn't... the other thing is they tell you that you can't drink, you can't smoke, you can't do all these types of things. Everything was an issue really. Reading I used to love, mobile phone that was a big thing as well. For ages I was trying to find a phone I could use. Obviously, when you are a teenager, it is a big thing. I couldn't use a computer properly. I couldn't watch football on the television. It was just everything, it was just heart-breaking.'

Jake summarises all of the activities, including football, he can no longer participate in because of his physical limitation. His inability to function, as he once did, impacted on his sense of self. When a chronic condition is unexpected, as is often the case with LHON, greater work in undertaking identity reconstruction may be required to come to terms with the disruption (Bury 1982; Whitehead 2006). Jake also highlights the health advice he was given to avoid smoking and drinking. For Jake, going out and drinking with his friends was a big part of his social life. Petersen (2006: 36) suggests that being diagnosed with a genetic condition poses a substantial threat to the individual's sense of self, when individuals are advised to avoid 'risky' activities which can lead to social exclusion.

When changes in bodily function take place, the self becomes 'unsettled and uncertain' (Clarke and James 2003: 1388). Corbin (2003) suggests that, over time, individuals learn to live with an altered body and, once again, take the body for granted and build their self-concepts and identity around what they can do rather than what they cannot do. To what extent this is true in the longer term for my participants will be discussed in Chapter Six. However, in the short term, it was clear that participants struggled to adapt to their altered situation. The disruption Beth experienced was particularly acute in the early weeks and months because she also developed Charles Bonnet Syndrome¹⁷ and experienced visual hallucinations, which

¹⁷ Charles Bonnet Syndrome (CBS) is named after a Swiss philosopher who wrote about his grandfather, who following sight loss, experienced 'visions'; he could see patterns, people, birds and buildings, which were not really there. The main cause of CBS is loss of vision and how the brain reacts to this loss. Exactly how sight loss leads to hallucinations is the subject of continued debate. Current research suggests that when we are seeing real things around us, the information received from our eyes stops the brain from creating its own pictures. When a loss of sight occurs, however, the brain is not receiving as much information from the eyes as it used to. The brain can sometimes

have become less frequent as time has passed. Beth explained that the hallucinations were extremely frightening as she was unable to distinguish what was real from what was not:

'Because of this condition, I thought I was going daft, I started suffering hallucinations. I can see people. I didn't know who was real and who wasn't. I actually see people in the bedroom sometimes; I am frightened to go to bed. In fact, the other week, it got to a stage I didn't know what I was going to wake up to. My bedroom wasn't my bedroom. One day—you might laugh—I had five naked men in the bedroom. My bedroom has been an operating theatre, it's been a hotel room and I haven't been able to get out of there. The bedroom is completely different, not my bedroom and one night, I couldn't get out of there. I was all panicky, I didn't know where I was. The other week I phoned my dad at quarter to six in the morning. He came around because I couldn't find the bedroom door. I couldn't get out of my room. The lady across the road came over one day to see how I was. She had three girls with her. I thought they were real I said "who are these girls you have got with you?" and she said, "I haven't got anybody with me, I am on my own".'

Beth can no longer trust what her eyes are telling her. Corbin (2003) highlights how people learn to trust their bodies and the sensations that it sends. Using the example of placing a foot on the ground, Corbin suggests that the body sends a message that the pavement is even or uneven. As result of the message sent, the necessary adjustment is made. We, as individuals, know that certain experiences are 'real' (2003: 258) because we have learned to trust the messages our bodies send. Beth was no longer able to rely on what her senses were telling her. She could not trust the signals being sent to her brain and has had to adapt her perception of the world accordingly.

Bury (1991) notes that the loss of confidence in the body's ability to function as it once did leads to a loss of confidence in social interaction. Coming to terms with limited sight proved to be overwhelming for many of the participants who felt that it was easier to shut the world out by participating in what can be described as self-imposed exile, often locking themselves away in their bedroom and keeping friends and family at a distance. Charmaz (1983) has identified that following the onset of

fill in these gaps by releasing new fantasy pictures, patterns or old pictures that it has stored. When this occurs, the images stored in the brain are experienced as hallucinations. CBS tends to begin in the weeks and months following a deterioration in eyesight (RNIB 2018).

chronic illness, individuals recognise that they can no longer undertake the activities they once enjoyed in the past. This leads to a restricted life as they choose to withdraw rather than allow friends and family to see them in their altered state. This was certainly true for many of my participants. David was diagnosed when he was 23, and at the time, was in the final year of his degree. He explained that his reaction to his sight loss, not untypical of other participants, was that he felt he needed time to himself:

‘I needed some time to come to terms with this [sight loss]. I thought I am going to need some time to myself. I did kind of end up having that bit of time to myself. I kind of pushed them [family] and the world away a little bit for a while.’

Eventually, after having time to himself, David returned to University to complete his degree. Tim, who was diagnosed when he was 23 in 2013, also talks of avoiding his friends:

‘I certainly pushed my friends away because you know what 23-year-old guys can be like. No one knows how to deal with those emotions and fears. The only way you deal with it is by taking the mick out of each other and, at the time, I hadn’t really accepted it and I couldn’t be dealing with that.’

Ollie, who experienced suicidal thoughts during the early months following his sight loss (as highlighted earlier in this thesis), also withdrew from the outside world:

‘When I left [name of university], it was three months of not really leaving the house. You just don’t want to let anyone know what you have become. You’re very embarrassed about your condition. You’re very insecure and the people that know you very well will see how much it affects you, how much you have changed.’

Ollie’s perception is that he feels his identity has changed following his sight loss and that his friends will see him in a different light. The onset of chronic illness has been described by Little *et al.* (2003) as an ‘extreme experience’ and one that challenges the individual’s sense of identity in all its elements because it ‘leaves no aspect of identity untouched’ (2003: 76). Similarly, Bury (1991) claims that chronic illness represents an assault not only on the person’s physical self, but also on their sense of identity. In the following extract, Adam explains how he felt different after he lost his sight and refers to the reaction of his friends:

'I was 14, I was free spirited. I used to have a credit phone, £20 would last me six months because people wanted to ring me, they wanted to text me, they wanted to come around. That all died very quickly:

"You are not the same, are you? No, so we don't really want to be around you.

I wouldn't be different if you didn't treat me differently".

I quickly realised that you can't ask 14-year-old kids to understand. They have a spot on the end of their nose and it's like the end of the world. Having a friend who has just lost 95 per cent of his sight overnight... I had friends who didn't understand it. A lot of people do get lucky, I have spoken to a couple of my [visually impaired] friends and their friends stuck by them and were a bit more understanding. Mine just went "You are not what you were, go and stuff yourself". They didn't want to know.'

Being perceived as different by friends following sight loss was also present in the narratives of other participants who indicated that friends disappeared following sight loss. Beth was both upset and angry that one of her oldest friends now avoids her: *'one of my friends, I had known her for 30 years and, you know, I saw her twice last year. I was so upset by it; I don't think she could cope with it (sight loss). She avoided me, she never rang'*. Amy, who was 18 years old at the time of her sight loss, also talks of losing friends: *'I lost a lot of my friends because they can't seem to accept that I have got something wrong with me. ...like I am registered blind, or I have got an eye condition. They are like "we can't speak to you now"'*. Ollie, whilst acknowledging that he also lost friends after his sight loss, also has a good group of friends who have continued to support him:

'You lose friends. You really do understand who your friends are. A lot of people just don't know how to react [to sight loss], so the best thing you can do when you don't know how to react is not react at all. My friends dropped out of my life. I had two or three really good friends and very quickly... they haven't spoken to me since. On the converse, you have friends who will stick with you through thick and thin. They were absolutely brilliant; it is six of one and half a dozen of the other really.'

Beth, Ollie and Amy, consistent with the sight loss literature (Davis 1961; Scott 1969), believe their friends did not know how to react to sight loss and, so, chose to avoid them. A number of the younger participants also referred to their initial early fear that their sight loss would stop them from finding a girlfriend or boyfriend. The fear of rejection prompted George, who retains a good level of residual vision, not to

tell his girlfriend that he was visually impaired. *'I didn't tell her for a year. She was living in [city] and I would go up on weekends and I was literally living my life on edge because I was trying to do everything without even showing her that I had bad sight. In the end, I had to tell her'*. When the relationship ended, George, resolved that he would tell his next girlfriend straight away that he is visually impaired. George is now married with two children. Not all participants have been fortunate in finding a partner. Declan, who was aged 12 when he was diagnosed with LHON, is now in his early twenties and believes his sight loss has prevented him from finding a girlfriend: *'I have never had a relationship because of it [sight loss]. I feel like no one wants to be with me'*. Unsurprisingly, the majority of the participants spoke of their initial reaction to sight loss in very negative terms. However, for a few participants, receiving the diagnosis was viewed as having a positive effect on their lives. David, diagnosed when he was 23 years old, described himself as lacking in confidence before his sight loss, but now feels that sight loss has helped him gain in confidence: *'I feel losing my eyesight helped me gain in confidence, I think it pushes you, it's either sink or swim'*. I also interviewed David's mother, Marion, who whilst acknowledging that David's sight loss was a steep learning curve for the family, also highlighted positive aspects: *'My son has certainly ...it sounds clichéd but... gained a vision through losing his vision. Before he lost his vision, we used to say when is he going to grow up and realise there is a world out there that he has got to contribute to'*. For one of the other younger participants, Jason, who was initially disbelieved by the ophthalmologist, who thought he was making-up his symptoms, receiving a diagnosis after 14 months had a positive effect upon him:

'It had a profoundly positive effect, I started to try a bit harder. Alongside all the medical stuff, you then kick into the educational system and assessing your needs. By now you are a special needs kid. They are testing you. I think the difference for me was I started to get my head into stuff. I remember sitting in a room for basically most of the afternoon. My mum was present whilst they tested me on English and they tested my maths and everything else and they started doing IQ testing. I think that's when they started to click that I wasn't thick, I was lazy (laughs) because, as I say, during this period, I had started to read more and work.'

(Jason, diagnosed aged 13 in 1990)

Jason, reflecting on his previous identity, described himself as lazy and underachieving: *'I was a very different child in terms of personality to the person I*

grew into after it (LHON). At school, I was a fad kid so after a month, I would then get bored and stop trying'. Jason's comments mirror the view expressed by Frank (1993: 42) using the analogy of the 'phoenix' rising from the ashes. Notwithstanding being catapulted into a critical situation, individuals in talking about their chronic illness present their experience as a life changing event. Charmaz (1987: 296) also talks of people, in coming to terms with chronic illness, constructing a new identity—a 'supernormal self', which will be discussed in more detail in Chapter Six.

Re-conceptualising biographical disruption

Bury's conceptual framework has been omnipresent in the medical sociological literature for over 30 years, having been both adopted and extended (Carricaburu and Pierret 1995; Locock *et al.* 2009; Larson and Grassman 2012; Saunders 2014). However, it has also been critiqued. Williams (2000: 51) suggests that biographical disruption is grounded, for the most part on the adult experience of illness—thus neglecting congenital and childhood conditions which he argues are 'integral to an individual's biographically embodied sense of self'. In their study exploring responses to the onset and impact of stroke in older age. Pound *et al.* (1998: 490) consider that studies which have adopted Bury's model are based on the 'unspoken assumption that illness enters lives which have—until then—been untouched by crisis or struggle'. For these authors, biographical disruption is not an inevitable consequence of chronic illness when experienced in later life; once individuals have survived into their 70s and beyond, their life experience equips them to deal with crises and adapt to their new situation, accepting chronic illness as an inevitable consequence in old age. Williams, (2000), suggests that when chronic illness is viewed against a background of general hardship and adversity, it may be perceived as 'biographically *anticipated* rather than a *disruptive* (i.e. unanticipated) event' (2000: 51, original emphasis). This view was subsequently endorsed by Faircloth *et al.* (2004: 245) who also argue that the onset of sudden illness such as stroke is not always perceived as a disruptive event, but instead may be viewed in terms of a 'biographical flow': once mitigating factors, including age and previous illness experience, are factored into the equation illness, 'melds into an enduring chronic illness narrative, part and parcel of biography'. Two of the participants who were the oldest at the time of their LHON diagnosis articulate very different experiences

following their sight loss. Firstly, Sandra had previous experience of being diagnosed with glaucoma, a condition her father had also experienced:

[I] feel almost cheated because if you say “Okay I have been doing all the right things with my glaucoma” ... I had lived with the fear of anything to do with sight for a long time you know. Everyone has a fear. Lots of people have a fear of cancer or whatever it is. This had been my big one [fear of sight loss]. I have been doing all the right things, I have been doing exercise, eating healthily, absolutely compliant with the medication down to almost the exact minute of the day. I know there are plenty of people who forget medication, but I never do and then something else comes in from left field and, you know, completely unexpected. It is that feeling of being cheated.’

(Sandra, diagnosed age 69 in 2015)

Sandra’s history of glaucoma did not prepare her for experiencing further sight loss. As with Stuart (discussed above), who felt he worked hard and did not deserve to lose his sight, Sandra also talks about living a healthy life, and feels cheated that she has lost her sight. Charmaz (1995: 66) posits that when ‘wholly unanticipated, even middle-aged people may view their bodily changes with a sense of betrayal’. In contrast to Sandra, Gerry, who was diagnosed aged 62 and who had previously enjoyed good health with no history of sight problems, explained:

‘I am very pragmatic about things. Once I had got the diagnosis... my view on things is that a lot of people go around with the attitude “why did it happen to me?” On the other hand you can say “why shouldn’t it happen to me?” Once it’s happened, you might as well accept it and get on with it and try and make the best of it.’

Gerry talks of accepting his sight loss and moving on. In displaying a stoic attitude towards his sight loss, he may be said to be adopting the ‘tough-men stereotype’ (Garcia-Calvente *et al.* 2012: 920) by hiding his true feelings because he may feel that to do otherwise would display vulnerability. Charmaz (1995: 268) has also noted that ‘illness can reduce a man’s status in masculine hierarchies, shift his power relations with women and raise his self-doubts about masculinity’. As the interview with Gerry progressed, he also referred to experiencing suicidal thoughts in the early stages of his sight loss:

‘I was feeling low, I did go through a phase where... I don’t think particularly seriously... where I was thinking I might as well kill myself,

what's the point of living if I can't do these things, I can't get out, I have lost all my independence, I feel like a prisoner. What can I do? What's the value in my life...?'

(Gerry, diagnosed aged 62 in 2014)

Gerry focuses on his feelings of loss including the loss of independence, explaining that he has to rely on his wife to help him with everyday tasks. Just after his sight loss, Gerry did not have the confidence to go out on his own so again he was dependent on his wife to accompany him. However, if she was busy, he would have to stay home, and, thus, he described how he felt like a prisoner. Corbin and Strauss (1987) take the view that for some people, chronic illness becomes the focus of their life (Sandra). Yet others, like Gerry, are able to integrate their illness into their life as it 'remains as part of the texture of a biography—something to be managed and taken into consideration but certainly not the only aspect of life (1987: 251).

Telling the family: disclosing genetic test results

Having considered the emotional impact of the LHON diagnosis on affected participants, I now consider the complex moral dilemmas negotiated within families when deciding to communicate genetic information to members of the wider family who may also be at risk of developing the condition. Mothers have to make difficult decisions regarding whether to tell the siblings of an affected child that they may also lose their sight. Previous research (McAllister *et al.* 2007) suggests that being diagnosed with a genetic condition is viewed as doubly emotionally burdensome by the proband [initial person to be diagnosed], who is faced with the challenge of coming to terms with the diagnosis, and also with the dilemma of whether to deliver 'bad news' (d'Agincourt-Canning 2001: 240) to other family members that they may have inherited a genetic condition. Scholars have suggested that genetic information belongs not only to the proband, but also to other members of the biological kinship as the information may have implications not only for their long-term health, but also be relevant in reproductive decision-making (d'Agincourt-Canning 2001; Forrest *et al.* 2003).

In their research with patients at risk of hereditary breast and ovarian cancer and Huntington's disease, Forrest and colleagues (2003) argue that there is a moral duty to disclose test results; failing to disclose such information to relatives potentially

deprives them of the opportunity to make autonomous health decisions. Providing wider family members with genetic information may encourage carriers to engage in health surveillance and possibly avoid exposure to triggering mechanisms. This is particularly salient for my participants, both affected and carriers, who are encouraged to avoid smoking, limit alcohol consumption, avoid contact sports, and exposure to toxins (Yu-Wai-Man *et al.* 2014).

Disclosing genetic information raises complex social, ethical and legal issues regarding not only whether there is a duty to disclose genetic information, but also whether family members have a right *not* to know (Hallowell *et al.* 2003; Adorno 2004; Arribas-Ayllon *et al.* 2008). When making the decision to inform other family members, questions arise as to whether to disclose the information, how much information to disclose, the potential impact of the information, deciding when the best time would be to disclose, and which family members need to be told (Forrest *et al.* 2003; Hamilton *et al.* 2005). Gaff *et al.* (2007) suggest that where the proband does not immediately communicate genetic information to their relatives, they are likely to be undergoing a period of deliberation in which they balance a feeling of obligation to pass on information against not wanting to cause alarm or distress. Forrest *et al.* (2003) suggest that disclosing genetic information may be viewed as a process as opposed to an act. The authors identify two broad categories of disclosure which they define as 'pragmatism' and 'prevarication'. The pragmatists are perceived as being pro-active in a practical way in disclosing information to other family members.

In contrast, the prevaricators described waiting for appropriate opportunities to bring up the subject but, as the authors highlight, it could take months or years to find the 'right moment' (Forrest *et al.* 2003: 321). In my study, at the time of diagnosis, several of the affected participants were children or young adults, and the decision to tell other family members was entrusted to their mothers. This finding is not unexpected given the previous research which highlights that in the Global North, women are traditionally considered to be the 'gatekeepers' and 'transmitters' of health information (Richards 1996; d'Agincourt-Canning 2001; Arribas-Ayllon *et al.* 2008).

The participants in my thesis displayed both pragmatism and prevarication. The adult women participants (carriers and affected) displayed pragmatism; as soon as they were aware of the diagnosis, they decided to contact immediate family members. When those family members also had children, this was considered to be imperative. It was not apparent from the interviews whether they had considered the impact the information would have on other family members. There was little, if any, thought given to whether those relatives would want to know or, indeed, the psychological impact of receiving the information. For a number of the participants, disclosing the information was prompted by a sense of duty, mirroring d'Agincourt-Canning's (2001) findings that her participants felt a moral responsibility to safeguard and inform other family members irrespective of the impact the information would have or whether the relatives would wish to know. When Sarah's son was diagnosed, she adopted a proactive approach, disclosing the diagnosis to her sister who has two young sons and also arranging for her and her sister to attend genetic counselling:

'As soon as we knew it was a mitochondrial disease, we realised what the implications were. They did tell us at one point "you would have passed it on to your son". I said "how can I have passed it on to my son, because my mum never had anything". They said it doesn't always come out. I then had to tell my sister that she and her boys are involved in it. She was like "no, no". We went for genetic counselling, me and my sister. We had a blood test taken and they said it won't be worth testing my daughter or my sister's boys because they have got the gene and all it would do is hinder them if they ever needed to get any insurance by getting it put on their record that's what they have got. Obviously, if I and my sister have got it, then the children have got it, there is no doubt about it. They don't need it actually put in writing, it is not going to help anybody.'

In contrast, Sandra waited several months trying to find the right time to tell her two daughters who are of child-bearing age:

'Inevitably, I had been putting it off and putting it off. One lives in [city] and the other in [city]. I wanted to do it at a time when they were both together and also a time when their partners were there to support them. There hadn't been many opportunities. I was going to do it in the November, but there wasn't a space or time in which we could all talk and so it wasn't until my birthday party (December 2015). They were much more grown up about it than I was. I suspect that they had probably worked out that it was something of this sort'.

One of the affected adult males, John, has delayed discussing his condition with other family members:

'My mother had two sisters, I have four female cousins, so they haven't passed it on to their children at the moment. So, my mother has a brother, but I don't see the brother due to a family dispute. The two sisters, one has passed away now, but both had two daughters each. So potentially it is still progressing through their side. So, you know, possibly they should be made aware of it. I could certainly contact them and make them aware that this goes through female genetics and so potentially because my mother must have got it from her mother, their mother is probably carrying the gene. I don't know what triggers the gene, you see. With me, they thought because I was drinking a lot that it could have triggered the Leber's, you see. It might have stayed dormant. Unfortunately, it didn't. I might not have been aware I had it myself (*laughs*)'.

(John, diagnosed aged 42 in 2008)

John displays signs of prevarication and, although diagnosed in 2008, he has not had a conversation with his female relatives. In part, this is due to his belief that it was his alcohol consumption which triggered the condition. He considers it unlikely, therefore, that his relatives will be affected. John also refers to a rift in the family which has precluded him from disclosing his condition to his uncle. Previous research (Forrest *et al.* 2003; Hamilton *et al.* 2005) suggests that the decision to disclose genetic information is influenced by pre-existing relationships within the family, cultural factors, and the perceived vulnerability and receptiveness of the individual receiving the information. This was apparent in my research with mothers who were reluctant to inform unaffected siblings that they could develop sight loss because they were uncertain of their reaction. The following extract illustrates a typical response from Gwyneth, a mother, whose elder son was affected aged 12. Her younger son is now aged 14, and she has not discussed (and does not intend to discuss) the possibility of developing LHON with him:

I have spoken to his school and explained to them that he could develop it [LHON]. They are really good and said they wouldn't say anything, but they would keep an eye on him.

Why have you decided not to tell him?

He seems for a young boy to be quite sensitive. I think he would dwell on it and think about it a lot. I wouldn't really want to worry him for something that might not happen.

Did you communicate the results to other family members?

Yes. I told my niece because she has got boys. She said she would rather not know. She said if they have got it, they have got it. She would rather not think about it.

Gwyneth faces a dilemma which was articulated by other mothers in my thesis that there is a great deal of uncertainty surrounding which family members will potentially develop sight loss. Gwyneth does not want to affect the way her younger son lives his life by telling him that he may lose his sight—an event that may not happen. In her research with families affected by Huntington's Disease, Skirton (1998: 107) suggests that in disclosing genetic inheritance, there may be a critical age at which this should be undertaken as the 'disadvantage of delay may outweigh any benefit'. Adam, explained that his mother was aware that she carried the LHON mutation, having been tested as her three uncles had lost their sight. Adam's parents decided not to tell him that he was a carrier as they did not want to affect how he lived his life. When Adam was aged 14, he developed problems with his sight. During an appointment with an optometrist, the family history started to emerge.

I knew I had uncles in America [who were blind] ...but it was never really mentioned. It all came to a head when I had my optician's appointment. My Mum and Dad knew that I could have an eye condition. They [uncles] all knew about it. My Mum had a test to determine whether I was a boy or a girl. Apparently, it was the "done thing" then. If you have a boy, you terminate the pregnancy because he could go blind.

Did your mum know how LHON is inherited?

Yes, she was fully aware. So was my dad before he married my mum and had me. They kept it to themselves because they didn't want to affect how I lived my life: "I don't want to do this just in case I lose my eyesight". The dream was to play for [professional football team] in goal. My mum said that when she was 16, her and her sister had a test because they thought they could be affected.

At the time, that Adam started to experience sight loss, he was being evaluated by a football scout on behalf of a professional football team. On the day the football coach was watching, Adam conceded five goals—which was unusual and was the first indication Adam had that there was a problem with his sight. I asked Adam if he understood the significance of his diagnosis:

I was fully aware. You have got your Mum and Dad saying to you “We had you because we wanted a kid”. I want my frigging eyesight back, but it isn’t going to happen. I can’t read a newspaper; I have to get magnifiers for everything. I don’t bump into things as much now but at the start, I was a bumping into everything, hitting my head on things. It was horrible, really horrible, and it was like “what’s the point?” I am not going to save the world. I am not going to do anything magnificent, so what was the point in having me for this?

What were your feelings towards your parents after your diagnosis?

I resented my parents something shocking because it was, like, they knew about it. Other people who I have spoken to with this eye condition, their parents didn’t know. I had two parents looking me straight in the face saying “We knew about this, we love you.” I can’t see because of you two. I have had my eyesight taken away from me and you did it because you knew about it?”

Adam’s relationship with his parents remains difficult, firstly, because he believes they were selfish when they decided to have him, knowing that he could lose his sight. Secondly, he believes they should have told him that sight loss was possibility, so that he would have had time to prepare himself. In research undertaken into diverse genetic conditions, McAllister and colleagues (2007) note that individuals, when confronted with a genetic diagnosis, may express anger towards the person who provides them with the information, but also towards someone for not disclosing the family history.

Previous research suggests that the decision to tell children that they may be affected by a genetic condition is considered to be the responsibility of the parents (Forrest *et al.* 2003; Keenan *et al.* 2005). However, when the parents make the decision not to disclose genetic information to their children—in some cases, even when they are adults—that has the potential to engender intense emotional reactions from relatives who feel they cannot tell their cousins, nieces and nephews about their genetic status (Green *et al.* 1997; d’Agincourt-Canning 2001; Gaff *et al.* 2005). This was apparent in my thesis with Beth, who expressed her frustration that her aunts did not want their children to know about the inheritance of LHON:

‘My Mum and Dad were devastated but it has split the family up a bit really. My Mum’s youngest sister has two sons, the eldest one is in his 30s and the younger one is in his 20s. She has never told them about this condition, probably because she has wanted to protect them. Some

people have said maybe it's a good thing, she has done the right thing, but other people have said she should have told them.'

Beth expressed her own view that her aunt should have told both of her sons. She then recounted an incident when the younger son, although not experiencing symptoms, believed he was losing his sight and experienced a problem with his mental health. Beth went on to say: '*we believe she still hasn't told the two sons. They don't know anything about my sight going and she has gone to great lengths [to hide this] ... she has put a bar on the telephones, so I can't ring the house*'. I asked Beth why she thought her aunt had not disclosed the diagnosis:

'She is frightened of it happening to her, isn't she? I think she is in denial. One day I said to her "You haven't told him [younger son], have you?" She said "No, I haven't". I said "Why, would it tip him over the edge?" and she said "Yes". So now he is sort of back on track, she doesn't want anything to disrupt that. I won't be there to support her if anything happens because I have had no support off her at all.'

Having discussed the factors that influence the disclosure of genetic test results to the wider family, I now consider the reaction of mothers on learning of their child's diagnosis, and how they negotiate feelings of guilt.

Reactions of mothers to the diagnosis

All mothers, on receiving the news, of their child's diagnosis, described experiencing feelings of devastation and sadness. The mothers mourned the loss of their child's future expectations, believing that following the diagnosis, their child's life would be over and that they would become dependent on their parents for the rest of their lives. Sarah, whose son Tim was diagnosed when he was 22 years old and living at home, captures this view:

'As far as we were concerned, that's it. Tim's life was over, our lives were over. We would be his carers; he is going to be completely blind, he is going to have a white stick, a guide dog. We were like this is how it is going to be.'

Sarah's comments resonate with the research of Locock *et al.* (2009) exploring biographical disruption within the context of the terminal illness, Motor Neurone Disease (MND). The authors suggest that, on receiving the diagnosis, participants believed they had been given a 'death sentence' (2009: 1043) and that their life

was over. Sarah feels that not only was her son's life over, but that her and her husband's lives were also over, as they perceived a future where they would look after their dependent son. Expressing similar sentiments, Jacqui, whose son Stuart was diagnosed in 2013 aged 23, explained that, fearing he would never live independently, they built an annex for him:

'At the back of the house we had a shed and a toilet area. We converted that into an en-suite for him. He has got his own space. It's outside the house so he has got his own separate building. He has got his bedroom in there.'

Both Tim and Stuart are now in relationships and have left home. Gloria's daughter Amy lost her sight when she was 18 and, at the time, did not have a boyfriend. Amy became what Gloria describes as '*clingy*' and did not like being left at home when her parents went out. Gloria, reflecting on the early months following the diagnosis, recounted her thoughts:

'I was thinking how is she ever going to meet anyone and coming out with me and my husband. How are we going to find somebody for her, who is going to have her now she has lost her sight? All these sorts of things were running through your mind.'

Amy is now in a relationship, and although still living at home, has become less dependent on her mother. When I interviewed Amy, she described how, on being introduced to Amy's boyfriend, her mother had been over-protective: '*It took her a very long time for her to accept someone else looking after me. Not looking after me, but someone else to sort of look out for me. Bless her*'. A number of the other affected participants also referred to their mothers becoming very protective of them which, at times, they found difficult to cope with. This may in part be attributed to the guilt that the mothers experience when being told they have transmitted the condition to their children. Joyce, whose son was affected when he was 13 years old, spoke of her guilt and how, on occasions, she '*fusses over him*' which causes arguments between them as her son gets irritated:

'It's just the guilt doesn't go away and it's probably more difficult because when I try to make sure he is alright he gets really annoyed with me. He doesn't like it when I fuss because he says he is fine.'

Joyce is one of the few mothers who was offered genetic counselling and, although she attended appointments for three months, and did not know of her carrier status, continues to feel guilty. Despite having been unaware that they were carrying the LHON mutation before their children were born, mothers with affected children articulated feelings of guilt, and self-blame when given the diagnosis. This is consistent with previous studies that have explored parental self-blame and guilt when children are identified as carriers or are affected by a genetic condition (Chapple *et al.* 1995; Arribas-Ayllon *et al.* 2008). The mode of inheritance has also been reported to have a major impact on feelings of guilt and self-blame experienced by female carriers of sex-linked conditions in contrast to autosomal disorders (where both parents have a copy of the mutant gene) (James *et al.* 2006; Clarke 2016). There is a scarcity of research into mothers' emotional responses to transmitting mitochondrial conditions to their children. In their research into genetic counselling strategies for maternally inherited mitochondrial disorders, Poulton *et al.* (2017) claim that the uncertainty surrounding the diagnosis and prognosis of mitochondrial conditions results in feelings of psychological distress, including feelings of blame and guilt. The following three extracts are typical of the responses of mothers in my research when discussing transmitting LHON to their children. Catrin says:

'I felt pretty shit to be honest with you (*laughs*). You know throughout [son]'s life, we had always encouraged him to try and do well at school and get a good job and everything else. To be honest with you, that all came crashing down. He wanted to travel the world, he wanted to have a nice car and everything and then, all of a sudden, all those opportunities seem to have been taken away from him. To be honest with you, I tried to take every day as it came and support him, and to a degree, keep my feelings to one side because he was the most important person. I do have the odd moment where I think "this is my fault".'

In this extract, Catrin expresses her feelings of self-blame by focusing on the opportunities her elder son will now miss out on and the life he would have had. Catrin also has a younger son and is acutely aware he may also lose his sight: '*I now have [younger son] coming up to GCSEs and, you know, I can't let my guard down really because if he thinks that I am worried about him, he might not try as hard at school.*' Catrin's guilt is exacerbated because she has not told her younger son that he may

also lose his sight. Catrin referred to walking on eggshells as she did not want her anxiety to be visible to her younger son in case it had an impact on his upcoming exams. In the following extract, Sarah talks about her feelings of guilt and how she has struggled to come to terms with her son's sight loss and blames herself for it:

'If [son] hadn't have had me for a mother, he wouldn't be blind, but then he wouldn't be here (*laughs*). Realistically, you think I am talking nonsense, but it is still a nagging feeling at the back of your mind. If he had got anything else that wasn't related to something I had given him... if he had picked up an illness, he had lost his sight through contamination of dirty water or something, it would be nothing to do with me. He would still be my son that has lost his sight and I would still have to be dealing with it and I would still be there for him. But it wouldn't have been through something I have done to him. I know there are lots of hereditary things and some of them can be a lot worse than this. But it [the thought] is still there, unfortunately.'

Gwyneth also explains her sadness that she has transmitted LHON to her son and explains that her son holds her responsible for his sight loss, directing his anger towards her:

'I did feel sad that this had happened. I did feel guilty, really all the stress of it all as well. I had a lot going on with [son] at the school and then...he became very...he was becoming a teenager as well. He became very angry and bitter and nasty towards me. He would say "it's your fault, you and your family". He became very nasty, quite aggressive and hostile. He was saying "you should never have had me". I did feel guilty and things. It was very hard; he even still blames me now. He will bring up things like "you didn't understand what sort of childhood I had, I was bullied". Obviously, I think some children would make comments about his sight and things. They would call him a blind whatever. He still brings up every now and again "you never wanted me, you don't realise what a hard life I have had".'

Gwyneth has to deal with her own sadness and guilt which she refers as overwhelming at times, particularly when her son blames her for his sight loss. Other participants have also spoken of their anger, and how on occasions this has been directed towards their mother. Amy, discussed above, in addition to inheriting LHON from her mother has also inherited other conditions ¹⁸for which she will have to take medication for the rest of her life:

¹⁸Although LHON is a condition predominantly associated with sight loss, a small group of people also experience what has been referred to as "LHON plus" (Yu-Wai-Man *et al.* 2011) including

‘Sometimes I still blame her [mother], which sounds horrible. I don’t blame her, but when we sometimes have an argument, I will say “it’s your fault”. She hasn’t just given me the sight problem. It’s the other [health] problems as well. Then I make her cry and that’s awful. It’s not her fault. She didn’t mean it to happen.’

(Amy, diagnosed aged 18 in 2011)

Similarly, Beth also described how, at times, she also blames her mother: ‘*sometimes I am so angry with her [mother], I say it’s her fault and... I will say it more so when I have had a drink.*’ Later in the interview, Beth acknowledges that she knows that her mother would not have realised she was a carrier: ‘*It is something they didn’t know about. She couldn’t have known, but I think she feels... she would go to the ends of the earth to try and get my sight back.*’ Jake sums up the view of many of the participants when he says: ‘*It’s not at all my mother’s fault. It’s no one’s fault really. It’s just one of those things. My mum didn’t know.*’ With one notable exception, Adam, whose mother was aware of her carrier status, the other participants realise that their mothers did not know they would transmit LHON when they decided to have children. In Chapter Seven, I will discuss how females who are aware of their genetic status negotiate reproductive decision- making.

Summary

This chapter began by reiterating that LHON is a chronic condition —it is of long-term duration and currently has no cure. In exploring participants’ initial reactions to their unexpected diagnosis, I used Bury’s (1982) conceptual framework of biographical disruption. As explained, although LHON can affect people at any age, it predominantly affects young men who, prior to their diagnosis, were fit and healthy, which made it more difficult for them to understand why they had lost their sight. Unsurprisingly, and consistent with the sight loss literature (Kübler-Ross 1969), participants spoke of grieving for the loss of sight as they experienced a complexity of emotions including denial, anger and depression, which led to suicidal thoughts. A number of the participants felt that their body, or more precisely their eyes, had let them down as they struggled to undertake simple tasks, including using

symptoms of cardiac conduction defects [heart’s normal rhythm is disturbed] and neurological abnormalities (including ataxia).

a mobile phone or making a drink. It was in these moments of frustration that participants experienced a loss of self-esteem and asked themselves 'Why me? What have I done to deserve this?' The loss of self-esteem has repercussions for their identity work as participants come to terms with who they now are and how they will be perceived by others. As Green *et al.* (2002: 259) suggest, '[a]t the level of self-identity, vision loss potentially disrupts a secure sense of self in the world, with a new identity having to be incorporated into biography'.

This chapter has also discussed the dilemmas that individuals face when making decisions to disclose genetic test results to other family members who may be at risk of sight loss. Given the ages of the majority of participants when they were diagnosed, responsibility for communication of genetic test results to the wider family was left to the mothers. However, one of the most challenging decisions negotiated by mothers was whether to tell unaffected siblings, that they were at risk of losing their sight. Decisions not to disclose information was grounded in the belief firstly, that the sibling would not be able to cope with the information. Secondly, given the uncertainty surrounding the inheritance of LHON there is a possibility that the siblings will not become affected. Finally, mothers were concerned that the information would affect how they lived their life. In the final part of this chapter, I explored notions of guilt and blame. Notwithstanding that they were unaware of their carrier status when they decided to have children, all the mothers interviewed expressed feelings of guilt. Similarly, their children also conceded that they do not blame their mothers as they were unaware of their carrier status, although they did admit that, in moments of frustration and anger, they had expressed the view to their mothers that they were to blame.

In the following chapter, I consider the longer-term implications of sight loss for participants and how, over time, they adjust to this.

Chapter Six: “I Just Want My Life Back”: Living with Leber Hereditary Optic Neuropathy

Introduction

In previous chapters, I have described participants’ chronic illness trajectory from experiencing symptoms of sight loss, through to the challenges encountered in receiving a diagnosis and their first reactions to the irrevocable loss of sight. In this chapter, I explore the longer-term disruption for participants and their families of living with Leber hereditary optic neuropathy (LHON). The chapter will be presented in three sections. In the first section, I build on the previous discussion around the physical limitations imposed by sight loss to consider the mundane everyday reality of living with LHON, highlighting the changing nature of relationships within the family following sight loss. While family members are the primary source of practical help by providing care and emotional support following sight loss, at times, the transformation of those relations causes friction between family members as they adjust to the changing dynamics of their relationships. A number of participants referred to no longer being able read documents, watch television, go shopping or cook a meal. They rely on others (parents, partners, and friends) to assist them in such tasks. These changes influence how individuals view themselves and how they think others perceive them, shaping their social relationships and their identity (Charmaz 1983; Nettleton 2013).

In exploring the on-going daily challenges of living with LHON, I again draw upon Bury’s (1982) conceptual framework of biographical disruption. Taking Bury’s explicit reference to biographical disruption as an event occurring at the *onset* of chronic illness, as a point of departure suggesting that his conceptual framework is equally relevant in exploring the longer-term implications of living with LHON; participants reported experiencing repeated incidents of disruption *throughout* the trajectory of the condition, irrespective of the age of onset. My positioning is not unique; Larsson and Grassman (2012) adopted Bury’s conceptual framework when exploring, over a 30-year period, the narratives of chronically ill people who had lived with sight loss from childhood, adolescence and early adulthood. Mirroring findings in this thesis, rather than experiencing a single event of disruption at the

onset of symptoms Larson and Grassman's participants experienced repeated disruptions which shaped their lives.

In the second section, I consider whether the passage of time has changed my participants' attitudes towards their sight loss. At the time of interview, participants had experienced sight loss for at least 12 months, but some were 'seasoned professionals' (Williams 1984: 176) having been diagnosed over 20 years earlier. I explore how (un)successful participants have been in regaining a sense of ordinariness (McLaughlin and Coleman-Fountain 2018) and a sense of normality (Davis 1995; Buse and Twigg 2018) by undertaking biographical repair work (Locock *et al.* 2009), and in so doing, have 'become whole again' (Corbin and Strauss 1987: 264) by 'restoring their former self-images' (Charmaz 1987: 283).

In the final section, I consider the complex inter-relationship of sight loss, disability and stigma, by exploring the dichotomy between the medical and social models of disability. Whilst the disability movement has pushed forward the agenda that promotes a more inclusive social model of disability, it is also evident that the visually impaired are still subjected to discrimination, marginalisation and stigmatisation. This is not limited to micro-level social interactions, but also occurs at the macro level in the built environment where public spaces are designed in such a way as to potentially exclude those who have a disability. Throughout this chapter, it is evident that participants have an overwhelming desire to undertake ordinary everyday tasks and regain a sense of what they perceive as "normality".

The everyday reality of living with sight loss

Sight loss is a life changing, unique, and variable individual experience. The majority of the participants were fit and healthy before and after their diagnosis. However, four women (Laura, Moira, Mary and Sian) in addition to their sight loss also experience multiple sclerosis-like symptoms (MS).¹⁹ For these four participants, their embodied experience of sight loss differs from that of other participants, as they overcome the challenges of sight loss but also cope with debilitating illness

¹⁹ LHON has been associated with multiple sclerosis-like (MS) symptoms in females who carry the m.11778G>A mutation (Harding *et al.* 1992).

(including prolonged bouts of pain, muscle weakness and sensations of tingling and numbness).

In this thesis I draw on participant interviews, highlighting not only commonalities but also the variations in subjective experiences between individuals and also the ‘fluctuations that occur *within* the individual’s experience’ (Warren and Ayton 2018: 142, original emphasis) of living with LHON. Focusing on everyday mundane activities—by which I mean the ‘mundane, familiar and unremarkable *stuff* of everyday worlds that is the routine, repetitive, and rhythmic reproduction of social life’ (Thomas and Sakellariou 2018: 6, original emphasis)—allows the identification of recurring themes within the data including frustration, loss of independence and social isolation confirming the findings of the wider sight loss literature (Lamoureux *et al.* 2004; Vu *et al.* 2005; Senra *et al.* 2011; Stevelink *et al.* 2015a). Here, I am referring to the taken-for-granted activities that are reproduced at the same time in the same places every day (Scott 2009). It is only by observing the minutiae of the everyday that we see how social life is produced and reproduced in the context of living with a disability. These every day activities include going to work, continuing with education, going to the gym, or having a drink with friends. By exploring the micro-encounters of the visually impaired, I highlight the changing nature of the relationships between partners, children, friends and work colleagues.

I commence an exploration of the everyday by drawing on the interviews of the younger participants, many of whom, at the time of sight loss, were either in their early teens preparing to go to university or in their twenties and had already commenced studying for their degree. Ollie, a keen sportsman, was diagnosed with LHON when he was 18. Prior to his diagnosis, he had devoted considerable time and effort gaining the necessary examination grades and undertaking extracurricular activities to secure a place at a prestigious university. Within a few months of commencing his degree, Ollie’s sight deteriorated, and he was faced with the reality of no longer being able to continue with his studies. Ollie explains his frustration when he realised his dream was no longer a possibility:

‘I asked the doctor what that [diagnosis] really meant and he said “Well it’s going to have huge ramifications on your life style” and those words stuck with me. In terms of my career, it was over really before it had started; a

[degree] is obviously something you don't fall into. I had been preparing for that for about eight or nine years.'

(Ollie, diagnosed aged 18 in 2009)

For Ollie, his diagnosis had profound repercussions for his future plans, forcing him to reconsider not only what he was going to do with the rest of his life, but also how he was going to live it (Petersen 2006). Finding himself in a similar situation, George experienced sight loss in his teens whilst studying for his 'A' levels.²⁰ When he was applying to join one of the Emergency Services. He spoke of his disappointment that he would no longer be able to follow his chosen occupation:

'I couldn't follow the career path that I always wanted to do. I wanted to become a [occupation]. People were saying, "You can still join and do admin". I could do admin anywhere, it's not the same. Although I wasn't different, it was just that I had this eye condition where it was quite difficult for me—well, impossible—for me to do the things I have always done.'

(George, diagnosed aged 16 in 2004)

George, who was 26 at the time of interview, explained that following his sight loss, he did not go on to higher education nor has he secured employment since leaving school. He spoke of his frustration of applying for jobs but not being shortlisted for interview: *'it's just finding that employer who will employ you to give you a chance, give you an opportunity'*. Later in his interview, George returned to the subject of being unemployed and how this impacted on his life: *'even now 12 years down the line, I still don't know what I would like to do with my life'*. The RNIB (2017) report that those who are registered as severely sight impaired (previously blind) or sight impaired (previously partially sighted) are significantly less likely to be in paid employment than the general population or other disabled people.

For many of the participants, whether to disclose their sight loss to a prospective employer caused a dilemma and echoes the wider disability literature (Barlow *et al.*

²⁰ The "A" level examinations are taken in the UK (excluding Scotland) in a specific subject. The "A" level is usually taken between the ages of 16 and 18 years old.

2007). Ross was diagnosed in 2003 aged seven. He was asked whether he discloses his sight loss to prospective employers, to which he replied:

‘No, I don’t, I can use a computer okay I can get to and from work. I don’t think they would need to know, If they asked me, I would tell them. I think if I told them it would be a negative, I wouldn’t want to give them a chance to...I don’t think that most employers know enough about it really. They would just make assumptions on things like that’.

Similarly, Jason, who is currently in employment, also explained:

‘At interviews, I have never quite worked out whether to hold it [sight loss] out upfront and say, “by the way, this is this” and then you can’t ask any questions from an HR perspective. Or do as somebody else told me which is completely hide it as much as possible to the latest point in the interview process; by then, they have fallen in love with you before they have seen the issue and therefore they are sold on you, they will see it as less significant.’

(Jason, diagnosed aged 15 in 2004)

Douglas *et al.* (2009) in a review of the rates of employment among working age people who are registered as severely sight impaired or sight impaired found that only 33 per cent were in employment. This figure is significantly less than those people without a disability, or people with other disabilities. In contrast to George, Ollie returned to university to study a different discipline. He was awarded a First-Class Honours Degree, after which he passed his Professional Examinations and is now employed in a large city firm. The factors that influence why some participants might be considered to be successful in a ‘mobilisation of resources’ (Bury 1982: 170) to overcome adversity, while others do not, will be discussed later in the chapter.

Unsurprisingly, sight loss has had a significant impact on all participants, making it more difficult for them to function and navigate in a world that, at times, is not compatible with sight loss. In the following extract, Ollie echoes the sentiments of many of the other participants:

‘Visual impairment affects everyone who has it incredibly—socially, in the ability to independently plan and travel journeys, in the ability to see what people are saying, lip reading, and the ability to use technology, in the ability to communicate and make food and dress yourself in the morning. It affects every single aspect of one’s life.’

The considerable body of literature exploring the consequences of chronic illness (e.g. Charmaz 1983; Bury 1988, 1991; Sanders *et al.* 2002; Lowton and Gabe 2003), highlight the impact symptoms have on routine body maintenance activities which can only be accomplished with the assistance of others, such as bathing or going to the toilet (Barnes and Mercer 2010; Thomas and Sakellariou 2018). Ollie describes the daily frustrations and challenges of living with sight loss: *'being able to put a piece of toothpaste on your toothbrush is difficult (laughs)'*. He also explains how difficult it is to get dressed in the morning: *'you try and put your socks on, you put one down, and then you can't find it'*. After his sight loss, Ollie claims that every aspect of his life has been affected and that dressing and getting ready for work now takes so much longer than before. In their research, Hansen and Philo (2007) also allude to this point, suggesting that taking longer than normal to complete tasks denotes, and reminds them of, their bodily difference.

Ollie also highlights another issue identified by participants: sight loss makes it impossible for them to read facial expressions or recognise people. Baus (1999), who is a visually impaired therapist working with blind organisations, highlights the difficulties encountered in social interactions, including not being able to make eye contact or observe body language. One consequence of not recognising friends or indeed being aware of the presence of others in a social situation is a loss of social skills, a recurring theme in the interviews mirroring the wider sight loss literature (Owsley *et al.* 2006; Stanford *et al.* 2009; Bennion *et al.* 2012; Ferguson and de Abreu 2016). As Paul put it:

'The most frustrating thing for me was struggling to recognise my friends. Not only is it embarrassing, it is also isolating.'

(Paul, diagnosed aged 17 in 2011)

Goffman (1956, 1959, 1963) has written extensively about how people can experience embarrassment in social interactions. He suggests that embarrassment arises when an individual in attempting to project a single integrated self is thwarted when 'somehow confronted with another self which, although valid in other contexts, cannot be [...] sustained in harmony with the first' (1956: 269). For Paul, his embarrassment is heightened because his friends had previously known him as fully sighted and now he is visually impaired, changing the nature of his

relationship with them. Similarly, Declan, who was diagnosed with LHON when he was 12 also talks of his former friends who now pass him in the street and do not acknowledge him: *'If I am walking down the street like they think I don't see them, but I do. When people walk past me that I know I think, "Why have you done that?" They walked past me and not said nothing'*. Paul also refers to the isolation he experiences since his sight loss. Living an isolated existence is often linked to spending protracted periods of time alone within the home; it can also be experienced in public spaces. One of the other participants, Andy, who described himself as an extrovert before his sight loss, explains his frustrations when in social situations:

'I would always be that kind of social person so if I went into a place, I would always say hello to everyone I knew and be the first to see someone I knew at the other end of the bar. Whereas I can't do that now and so some people, even now, think I am not very sociable. They sort of think "He's ignoring me", whereas I would normally be that person that would be out there welcoming, seeing everyone, so that is probably the biggest frustration.'

(Andy, diagnosed aged 20 in 2008)

Jake, who was diagnosed with LHON when he was 16, became very emotional when he confirmed that he had not seen the faces of his wife and children:

'Not being able to see peoples' faces, that's a massive frustration. Not being able to read body language that's another big one. I have two young children and a wife that I have never seen their faces. Certain things... like I didn't see my wife in her wedding dress. Sometimes I think it's horrible really, it's torture.'

Later in the interview, Jake spoke of his anxiety that his sight loss has practical consequences for his children:

'I take my kids out on my own but it is proper nerve-racking sometimes. If one of them runs off, I am screwed really. I don't really want to grab other people's kids and things like that.'

(Jake, diagnosed aged 16 in 2005)

In his interview, Ollie refers to his eyes as *'the main source of knowledge acquisition and without your eyes; you realise everything is just so much more difficult.'* A similar sentiment was expressed by Tim who was diagnosed in 2013 at the age of 23: *'obviously, vision is... it affects everything in life. We live in a visual world, so everything*

suddenly became a struggle'. Here, Ollie and Tim tap in to cultural representations that privilege sight over the other senses (De Leo *et al.* 1999). The Global North is said to be dominated by ocularcentrism (Måseide and Grøttland 2015); the loss of sight can cause suffering far greater than that experienced from other forms of sensory impairment (Abolfotouh and Telmesani 1993; De Leo *et al.* 1999). In their study of retinitis pigmentosa (a condition that, similar to LHON, currently has no cure)²¹, Hayeems *et al.* (2005) define suffering in the context of sight loss as:

'A state of distress that occurs when person's integrity or life plan is threatened disrupted or burdened; it lasts until integrity is restored either by eliminating the threat or adjusting to it' (2005: 615).

Within the sociological literature, the theme of suffering has been extensively considered by Charmaz (1983). Rejecting the narrow medicalised view of chronic illness which focuses on suffering caused by physical pain, Charmaz turns her attention to the destructive effect of chronic illness on the self and, in doing so, posits that the self is 'fundamentally social in nature [...] developed and maintained through social relations' (1983: 170). Charmaz explains that individuals possess a 'self-concept' grounded in the '*organisation* of attributes that have become consistent over time' (1983: 170, original emphasis). Chronic illness is problematic, firstly, for the organisation of the self as the images the chronically ill present to others may be 'inconsistent with their core self-concepts' (1983: 170). Secondly, the ill person presents self-images that may be found to be 'wholly incompatible with the individual's criteria for possessing a valued self' (1983: 170). Charmaz described the process as a 'loss of self' defined as 'the crumbling away of former self-images without simultaneous development of equally valued new ones' (1983: 168). The loss of self leads individuals to 'question their own self-worth and view their developing limitations as losses' (1983: 169).

Four themes are identified by Charmaz as contributing to a loss of self. Firstly, living a 'restricted life' (1983: 172); individuals become aware that the symptoms of their illness impacts on their ability to undertake activities they previously valued and

²¹ Retinitis pigmentosa (RP) is an inherited eye condition that affects the retina at the back of the eye and, over time, stops it from working properly. RP is characterised by a gradual but permanent loss of sight by affecting the peripheral and central vision. The changes in vision occur slowly over a period of many years. There is currently no cure for the condition (RNIB 2017).

enjoyed. Withdrawing from society becomes preferable to allowing people to see them in their altered state (see also Locock *et al.* 2009). Secondly, leading a restricted lifestyle results in 'social isolation' (Charmaz 1983: 176). Charmaz suggests that the experience of being 'discredited, embarrassed, ignored and otherwise devalued [...] contributes to the growing isolation of ill individuals and to their subsequent reappraisals of self' (1983: 177). Thirdly, experiencing a loss of self may result from 'discrediting definitions of self' (1983: 181), including 'those arising in interaction with others' and 'those developing out of unmet expectations of the ill person' (1983: 181). Finally, 'becoming a burden' (1983: 188) is associated with a loss of self as the chronically ill become more dependent and immobile, thus leading to a reliance on family, friends and work colleagues.

The themes identified by Charmaz resonate with the interviews undertaken with participants in this thesis. Beth, who was diagnosed aged 51, represents a marginalised group in the LHON clinical literature (as being both a woman and older); she is also typical of the older participants who have struggled to come to terms with their sight loss. Beth has a more restricted lifestyle, since losing her sight, relying on her elderly parents to help her with daily tasks. Prior to her sight loss, Beth described herself as very independent with a wide social circle of friends. She also had a very successful career travelling the world. When I interviewed Beth 14 months after her diagnosis, she talked about her life, using the metaphor of 'living in a nightmare' and spoke of her life being snatched away:

'It's been an absolute nightmare. It still is. It's been terrible. It's like somebody has come and snatched away my life and I just want my life back, I want my independence back. I can't just jump in the car and go to the shop. I can't work, I can't drive, I can't read, I can't write, I can't watch the television.'

(Beth, diagnosed aged 51 in 2015)

In her interview, Beth articulates her feelings that the life she once knew is no longer available to her. Beth is registered as severely sight impaired and is no longer able to drive,²² which she associates with a loss of independence. In the Global North,

²² In the UK, once registered as severely sight impaired, participants are prohibited by the Driver Vehicle Licensing Agency (DVLA) from driving.

independence and self-reliance are highly valued and therefore not being able to fulfil a socially expected role poses a threat to the individual's self-esteem and sense of self (Nettleton 2013). Charmaz uses the example of driving as indicative of leading an independent life:

'knowing that one can no longer drive or function in other ways that symbolised independence prompts the realisation that life is becoming increasingly restricted as previously taken-for-granted activities become precluded' (1983: 172).

In the wider sight loss literature (Green *et al.* 2002; Horowitz 2003; Khadka *et al.* 2012), the ability to drive is not only associated a sense of autonomy and independence but is also considered to be a 'symbol of normality' (Green *et al.* 2002: 259). No longer being able to drive is one of the most feared losses associated with sight loss (Horowitz 2003). Beth is wistful when she reflects on her inability to drive and the impact on her participation in activities previously enjoyed:

'I would come home from a trip; I would have a couple of days off. I used to be able to get in the car—I would be meeting with friends. Those days off were quite full.'

Beth describes a very active life and one that revolved around her ability to drive and visit her friends. This life is no longer available to her as she is now reliant on her friends to visit her. The emotional effect of no longer being able to drive was present in a number of the other interviews irrespective of age, as the following extract illustrates:

'I remember at the time [of diagnosis] asking would I be able to drive. He [consultant] said "no". I was fine while I was there [eye clinic] but I remember in the car journey home with my Dad, I just cried my eyes out for 10–15 minutes with the thought of not being able to drive. It gives you independence. It's quite a big thing, driving.'

(Andy, diagnosed aged 20 in 2008)

Andy explained that he lives in a small rural community where public transport is unreliable and he learnt to drive as soon as he was able to apply for a provisional licence. Kate, who had been driving for many years before her diagnosis, referred to not being able to drive as a 'cross to 'bear':

'The only thing that gets me down sometimes is not being able to drive. It is a life changing disease. That's been my biggest cross to bear really... is not being able to drive because I would think nothing of jumping into the car and driving to the shops. Now I rely on public transport; I can't see signs at the railway station or I can't see bus numbers. Life is much harder.'

(Kate, diagnosed aged 48 in 2012)

For one of the older males, Gerry, his refusal to acknowledge his failing eyesight caused arguments with his wife:

'I do remember some nasty arguments between my wife and I because she was trying to do the best for me, but you don't, as the one with the problem, necessarily see that what someone is trying to do is the best for you. You see them as interfering and manipulating and things, particularly when it came to the point where she thought I wasn't safe to drive, whereas I thought I could still see enough to drive at that time. But I did follow her guidance and did stop at that point.'

(Gerry, diagnosed aged 62 in 2014)

Gerry's reluctance to give up driving epitomises his desire to 'hold on to normality' (Locock *et al.* 2009: 1052). The younger participants were also reluctant to give up driving. One of the mothers, Catrin, confirmed that, notwithstanding his deteriorating eyesight, her son (aged 20 when he started experiencing problems with his sight) refused to stop driving. She explained:

'We had to stop him driving and I think we did have a bit of unhappiness when he realised he couldn't drive anymore, I think there was a bit of anger and frustration. He was sulking around; he lashed out a few times. He was stomping around slamming doors and things like that. I think he punched the wall and put a hole in it at one point. Boys his age... they are all driving: it's not the end of the world, but it's the end of the world as you know it in your eyes.'

The point Catrin alludes to is that, for younger people affected by LHON, holding a driving licence is viewed as a 'milestone, a symbol of independence' (Rosenblum 2000: 434) and an indication that adulthood is approaching. George's comments are typical of the younger participants: '*when you meet a woman, you think they want you to drive, especially when you are younger, because it's cool to have a car when you are 19 or 20*'. The reluctance to give up driving was linked for many of the participants with a desire to not only hold on to their independence, but also linked

to frustration at having to use a public transport system that is not designed for the visually impaired. Sarah, whose son had to sell his car after his sight loss, claimed:

‘He can’t use a bus, it’s impossible—he can’t see the bus coming, he doesn’t know to put his hand out and he can’t see the number on the bus. That’s impossible for him.’

Andy travels extensively on both the underground and overland trains for his job, and says that this is one of the main causes of frustration for him:

‘When I am travelling on a train and they don’t announce stops and things like that. They do have them all but its reliant on the drivers to switch them on or it might get out of sync, so it might be one station wrong. In terms of mainline trains as well, then sometimes they may announce it [train], sometimes they may not. The capability is there or if you get given information they say, “It’s Platform X or Y, it’s just over there”. Then you find you’re actually on a totally different platform. It can be really frustrating.’

In an attempt to minimise the impact of sight loss on their daily lives, participants like the young ex-service personnel in the work of Stevelink and colleagues (2015a: 1), who experienced sudden sight loss whilst deployed in Iraq and Afghanistan, adopted a variety of “coping strategies” including learning new skills (touch typing), goal setting (finding employment) and utilising low vision aids. Andy, referred to above, overcomes the problem of getting off at the wrong tube station by: *‘double checking or counting [the stations] in my head or I will know the surroundings and know the door should be on the opposite side or whatever’*. Whilst participants in this study adopted coping strategies to help with orientation and navigation, they also explained that getting around on foot in a built environment that is designed for those who do not have a disability could be problematic and, at times, precarious, particularly when using pedestrian crossings or avoiding inanimate objects including bollards, lampposts and, on occasions, badly parked vehicles. Charmaz (1983: 174) suggests that the lives of the chronically ill are more restrictive than they need to be as the ‘world is set up for the healthy and able’. This point is also alluded to by Green and colleagues (2002) in their study of glaucoma, highlighting that we live in a ‘social environment that systematically excludes those with impaired vision’ (2002: 259). The exclusion of the visually impaired from public spaces will be discussed in more detail later in this chapter. Beth described her

experience of being injured when she attempted to walk to her parents' house, located in the next road. She tried to avoid a car parked on the pavement:

'I ended up in A&E... a car was on the pavement and I moved over, and I went over a neighbour's wall. My leg was cut open to the bone. For six weeks I had to go to the hospital three times a week to have it dressed. During that time, I couldn't even get to my Mum's on my own. I felt like a prisoner in my own house. I had to rely on someone to come for me.'

One repercussion of her accident is that Beth no longer has the confidence to go out on her own. Throughout her interview, she spoke of becoming a burden on her elderly parents who take her shopping, to social engagements, and to medical appointments. Beth spoke movingly of her parents, both of whom are in poor health:

'My Mum and Dad ... my Dad's coming up to 86, my Mum's is 82 and they have already gone through it [LHON diagnosis] once [with brother] and, for it to happen a second time, it's unthinkable really. They are absolutely devastated (*becoming emotional*), and they have to do those things for me.'

Beth is conscious that her parents have become her carers. Later in the interview, she reflected on what would happen to her if her parents were no longer able to help her:

'I try not to think about the future because if I do, I would go under. I try and get through each day as best I can. I can't think about the future without my Mum and Dad. I honestly don't know how I would cope. If I am perfectly honest with you, I would rather go before them. I have got to try and get through each day at a time. Sometimes, I get up in the morning and I can't wait, I am counting the hours until I go to bed again. If my parents weren't here, or something happened to them, I really don't know what I would do.'

Beth talks of counting down the hours before she can go back to bed. She also stated that on occasions, she will not get out of bed:

'Some days, I don't want to get out of bed. Some days, I can't see a reason to get out of bed. The other week, I was in bed for 24 hours and it's not good.'

A similar experience of staying in bed for prolonged periods was also described by one of the other older female participants, Sandra, who was diagnosed with LHON when she was 69:

'If I feel like lying in bed all day which I quite frequently do... I can't be bothered to do anything because I have a feeling there is nothing to do. Sometimes, it can go on for days and sometimes, it's just the odd day.'

(Sandra, diagnosed aged 69 in 2015)

Beth and Sandra describe living an isolated and, at times, lonely existence, with few friends calling in to see them. Social isolation has been linked to undermining self-esteem (Whitehead 2006). Sandra commented: '*there are people who you might have thought would have made a little bit of an effort and popped round*'. She also went on to comment that '*I am not imprisoned in the house but there is the feeling that a lot of the time I am*'. Both participants also report being prescribed medication for depression. This is consistent with the wider sight loss literature that confirms experiencing sight loss later in life can result in feelings of anxiety, depression and isolation (Brody *et al.* 2001; Wong *et al.* 2004; Mogk 2008; Stanford *et al.* 2009; Hersh 2015). Beth describes a lifestyle that is dominated by her sight loss. She recognises that it is not healthy to stay in bed all day, but she struggles to find a way out of the downward spiral she finds herself in; she does not contemplate her life improving. Beth's comments are mirrored in Frank's (1995: 97) 'chaos narrative'—individuals are unable to interpret and make sense of their illness, and so there is no prospect of life getting better. In contrast, research undertaken by Moore (2000) with older women (63 to 85 years old) diagnosed with macular degeneration,²³ found that all their participants were able to maintain a positive outlook, notwithstanding the knowledge that their sight would not improve. In fact, the participants were able to 'move beyond the difficulties of the visual loss experience to create new and unique ways for approaching life' (2000: 578). One explanation for the difference in attitude may be attributed to the fact that Age Related Macular Degeneration (AMD), similar to rheumatoid arthritis, is considered to be an older person's condition and, as such, is not totally unexpected. Therefore, the women in Moore's study are likely to be able to adjust more easily to their sight loss.

When I interviewed Beth and listened to her talk about her present and future life, I had a sense that she is experiencing emotions more acute than biographical

²³ The participants had experienced visual loss in the first eye between 6 months and 20 years; and in the second eye 6 months and 12 years).

disruption—for her, life has ended. In their research with patients diagnosed with the terminal illness motor neurone disease, Locock and colleagues (2009: 1047) noted that whilst biographical disruption was present in their data, another strong theme emerged which they refer to as ‘biographical abruption’. Biographical abruption signals a sudden ending, a breaking off of life; the diagnosis was viewed as a death sentence—life was over, and participants were denied a future. Similarly, Beth describes her life as ending, as the following extract demonstrates:

‘I have thought about suicide. I wish I was dead. Some days I have thought about it, I really have. I have thought “What have I got to live for?” It has been sprung on me and it’s like my life has been taken away. I don’t feel any better now really than I did last year.’

In their research with elderly participants (aged 60–85 years old) diagnosed with Age Related Macular Degeneration (AMD), Wong *et al.* (2004) also report that suicidal thoughts were voiced by women who had lived with the condition for several years. In the previous chapter, I discussed how a few participants expressed suicidal thoughts at the onset of sight loss. However, for most participants, such thoughts subsided as they began adjusting to their new circumstances.

Beth is not typical of other participants as she continues to suffer with her sight loss. In her research, Charmaz suggests that, for her participants, the ‘language of suffering’ is the ‘language of loss’ (1983: 191). They rarely talked of their suffering in positive terms or of ‘gaining heightened consciousness of the world’ (1983: 191) as a result of their chronic illness experience. Charmaz’s participants in their suffering experienced ‘heightened self-concern about the person they see themselves becoming and about valued self-images from the past which they have lost, sometimes irretrievably’ (1983: 190). Beth’s social life revolved around her work colleagues and she is still suffering as she struggles to reconcile her former life as a successful career woman with her present and (potentially) future life, which she sees as living an isolated and dependent existence. Her sudden sight loss in short, has deprived her life of ‘meaning, structure and purpose’ (Clarke and James 2003: 1390).

Participants referred to their dependency on family support to help them with everyday tasks. Providing care—whether that be cooking, helping someone to dress

or assisting in more intimate activities such as bathing—has the potential to alter the nature of family relationships, introducing new forms of engagement (Warren and Ayton 2015; Thomas and Sakellariou 2018). Ollie, (discussed earlier) explains that it can be the small tasks that prove the most frustrating and difficult for the visually impaired to accomplish:

‘I rely on her [wife] for a lot of things in terms of being able to wash my own clothes and put the dishwasher on and things like that are quite difficult. Cooking is very difficult; ironing is impossible. There are things on a day to day basis.’

Similarly, George (discussed earlier) refers to the ordinary, mundane tasks that he finds difficult to accomplish and how reliant he is on his family and friends to help him:

‘Like people don’t understand every day is a challenge, making a cup of tea is a challenge, having a shave is a challenge, getting dressed is a challenge, like colour co-ordinating and things like that, like you don’t realise half the time how much you do rely on your family and your friends and things like that.’

One of the other participants, Laura, indicated, that she relies, at times, on her fully sighted daughter aged 13. Laura describes herself as having Harding’s disease [diagnosed with LHON and multiple sclerosis]; she is unable to walk very far and experiences a tingling sensation in her legs and hands. Laura is severely visually impaired confirming that: *‘I can see objects, but I can’t read any writing unless it is very large but even then, not really’*. Laura is dependent on her daughter to help her with daily tasks and reflected on the reversal of their roles:

‘I was her carer and, all of a sudden now, she is my carer. If there is something on the floor, I can clean it. But I just get her to check it. Sometimes serving dinner up... that’s an everyday thing... I can’t quite see what I have served up, if I have served it up equally or if it’s cooked.’

(Laura, diagnosed aged 48 in 2012)

Laura spoke of her feelings that she is not fulfilling the role of a good mother. She spoke of feeling guilty that her daughter is having to take care of her and commented that she tries to ensure that her daughter leads a relatively normal life. In her research with mothers diagnosed with HIV, Wilson (2007) also highlighted that many of the mothers made strenuous efforts to ensure their children enjoyed a

normal childhood in spite of the symptoms of their illness and the dilemmas encountered in attempting to achieve this aim. Laura's brother, who is single, following his LHON diagnosis has become an alcoholic and has repeatedly attempted to take his own life. Laura expressed her concerns, that without her daughter, she may also have descended into alcoholism: *'I have had some very dark days, I won't deny it. I have drunk a lot of wine'*. Laura was asked whether there were days when she didn't want to get up:

'No. I did get up with a hangover and just carried on. When you have got a child...my brother he just goes to the pub every day, but when you have got a child you can't do that. I think my saviour has been my daughter, life would have been very different without her.'

Bury (1982: 169) suggests that chronic illness brings into sharp focus the nature of the relationship between individuals, their families and allies 'disrupting normal rules of reciprocity and mutual support. Conrad (1987: 15) also makes the point that families are 'pivotal actors' in the world of chronic illness, and whilst providing valuable support systems, may also feel overburdened by the demands placed upon them (Charmaz 1983; Strauss *et al.* 1984; Conrad 1987). Catrin, one of the mothers, explained how her son's loss of independence has impacted on her own life. The family live in a remote location with sporadic public transport which means she has to drive him everywhere:

'Today I had to pick him [name of son] up from someone's house and take him to work. It's not just a quick matter of just picking him up and dropping him off. He wants to go to the supermarket to get some lunch and then that turned into another half an hour of him wandering around saying "I will have this, I will have that". It's almost as if he is back to being a toddler again trailing around behind me at times. I have lost a part of my own life really. Everything that [name] arranges has an impact on me because he needs to be taken to and from places.'

Catrin feels that she has lost a part of her own life and articulates her exasperation that her daily routine is now organised around her son. As Friedson has pointed out, the 'chronically ill person who "expects too much" or "makes too many demands" is likely to be rejected by others' (Friedson 1970: 235). Catrin faces a dilemma, in that she does not want to see her son struggle to undertake daily tasks. However, she also explained she is trying to encourage her son to be more independent as she is concerned for his future when she and her husband are no longer there to help him:

‘We still get very frustrated with each other because what he thinks of me as being awkward, I am trying to help him, like, instead of getting up and getting everything for him, I will say “right, it is on the shelf to the right.” Because I don’t want to be jumping up every time he wants something. He thinks I am being awkward by not getting it for him. But there is going to come a point in his life when he is going to have to live alone and he won’t have someone there doing everything for him.’

Later in the interview, Catrin voiced her biggest fear that her younger son would also lose his sight. This was echoed in the interviews of the other mothers who also have children who are currently asymptomatic, for example Sally, who was diagnosed with LHON when she was 29 years old (having previously been misdiagnosed), explained that her younger son was diagnosed when he was seven years old. The older son started experiencing symptoms when he was studying for his ‘A’ levels. Sally reflected that as the years passed, the family thought the older son would not be affected: *‘we thought we had got away with it’*. This family’s experience highlights the extremely unpredictable inheritance of LHON and this will be discussed in more detail in Chapter Seven.

Having focused on the consequences of sight loss for my participants, the next section considers the biographical repair work undertaken by participants and whether they have come to terms with and ultimately accepted their sight loss.

Triumph over adversity: reconstructing the self

In this section, I turn attention to the biographical repair work undertaken by individuals in an attempt to come to terms with their chronic illness. In mobilising resources (Bury 1982), including rallying support from family and allies (Corbin and Strauss 1985), participants strive to reconstitute the self. Bury observed that chronic illness necessitates a fundamental rethinking of an individual’s biography and self-concepts if the chronically ill are to successfully adapt to their condition. In his research with patients diagnosed with rheumatoid arthritis, Williams (1984) also illustrates how his participants attempt to repair the threat to their biography by reconstructing accounts of the cause of their illness and, in so doing, make sense of why they had developed arthritis which was experienced as a profoundly disruptive event in their lives.

Similarly, participants in this thesis searching for meaning to their diagnosis reflected on their previous lifestyle. Declan explained: *'I did play rugby for a local team; I think it was when I was playing rugby for the school when my head was stamped on. They think that's what could have triggered it because it was around about the same week I can remember sort of noticing things'*. Other participants referred to the levels of their smoking and alcohol consumption (both associated with the onset of LHON) in an attempt to rationalise their diagnosis.

In her early research, Charmaz (1983) suggests that no positive benefit was derived by her participants from their chronic illness experience. However, in her later work, Charmaz (1991: 4) posits that for her participants, the loss of self is not a permanent state and that the chronically ill learn to 'live with their illnesses rather than for them [...] often, they try to keep illness at the margins of their lives and outside the boundaries of their self-concepts'. Charmaz also suggests that for the chronically ill, adapting to chronic illness shades into acceptance (1995: 657). I will explore how (un)successful participants have been in adapting to sight loss and how, in the process, they are able to reconstruct a self.

The narratives of Beth, and to a lesser extent, Sandra, suggest that their lives continue to be typified by Frank's (1995) chaos narrative—life will not get better. This is because their lives are dominated by overwhelming despair, making it impossible for them to push their sight loss to the margins of their lives. However, Beth and Sandra are not typical of other participants, some of whom refer to experiencing "good days and bad days". This is consistent with research by Charmaz (1991: 51) who claims that a good day is typified by 'minimal intrusiveness' of the illness and a bad day by 'intensive intrusiveness'. One illustration is provided by Jake, who was interviewed when he was 27. Having lived with LHON for 11 years, he sums up the feelings expressed by other participants:

'I have good and bad days. It's just like you can't let it win but, deep down, I can't let on to people that I am struggling. So, with my job, I travel quite often. I go to [city] probably once or twice a week and, if I get lost or someone is funny with me or I bump into someone or they are rude to me or whatever or I can't do something, that's when it sets it off. If I have a crap day... and then I go to bed that night feeling crap and then I wake up and it's a new day.'

Jake seeks to hide a bad day—which for him is characterised by bumping into inanimate objects or not being able to undertake simple tasks—from his family and friends. For Jake, the next may be a good day. With the possible exceptions of Beth and Sandra other participants, such as Jake, were not able to predict in advance the day they would have, but instead found that their day was shaped by their social and environmental interactions. What is also evident is that participants on occasions experience days that are neither good nor bad. Tina, who was diagnosed when she was 30, is in full-time employment and is typical of a few of the participants in that she explains: *‘I sometimes have my grumpy days’*. Tina claimed that a grumpy day can occur even when nothing out of the ordinary has happened to trigger it. This is consistent with research undertaken by Warren and Ayton (2018) with participants diagnosed with Parkinson’s disease. The authors challenge the binary approach of the good and bad days scenario by highlighting that their participants experienced “Parkies” days, which are neither good nor bad, but are characterised by ‘a sense of ennui, in which they feel devoid of all emotion’ (2018: 152). For participants in this research, the level of residual vision may have an impact on whether they experience a good or bad day, and how successful they are in minimising the intrusiveness of sight loss in their daily lives. This point is illustrated by two brothers, Ross and Paul, whose mother, Sally (discussed earlier), is also visually impaired. Ross was diagnosed with sight loss when he was seven years old but experienced some visual recovery when he was 14 (although he remains eligible to be registered as severely sight impaired).²⁴ His brother Paul experienced sight loss when he was 17 years old. The brothers have very different experiences of sight loss as Ross considers that the level of his residual vision allows him to lead a relatively normal life:

‘My sight has improved. I can do pretty much anything I want. It is only in tough light conditions or some days are worse than others. I am pretty good. I know my brother’s is a lot worse and my Mum’s is a lot worse. Mine is the best sight out of the three of us. It has never really stopped me from doing anything like it seems to stop my brother. I can always find a way around things.’

²⁴ Evidence suggests that there is more likelihood of spontaneous visual recovery if the onset of LHON occurs at a younger age (Riordan-Eva *et al.* 1995).

(Ross, diagnosed aged 7 in 2003)

In contrast, Paul, whose peripheral and central vision are worse than his brother's, indicated that following his sight loss, he dropped out of his 'A' levels, returning to school nine months later to take his exams. He is now in the final year of university, but says he is struggling to cope:

'Every day I am having to work twice as hard as everybody else. I still don't know what I am going to do next year. I just don't know what's coming next. Almost as soon as I step out of the door to when I come back there is something that I can't do. I feel like I have been in limbo for the last five years, or however long it's been.'

(Paul, diagnosed aged 17 in 2011)

The story Paul tells is consistent with Charmaz's (1991) description of a 'bad day'. For Paul, his sight loss is highly intrusive. When I interviewed him, he was due to sit his finals the following year, but he cast doubt on whether he would actually attend the exams: *'I don't really care either way. I almost think there is not much point'*. The two brothers recount very different experiences of adjusting to their sight loss. Whilst the level of residual vision appears to be significant, other factors which also seem to be relevant are the age and the level of educational attainment at the time sight loss is experienced. Ollie believes that his educational background and work ethic were instrumental in helping him resume studying after his sight loss:

'I think educational background is definitely a factor, but then you have to see what really feeds into higher educational achievement. So what feeds into higher educational achievement is a good work ethic, probably a supportive family, probably a family with the resources financially and emotionally to help you through trauma. Then if you look at the aspects of someone who hasn't achieved, this is a generalisation, but these are factors I suspect and someone who hasn't achieved academically, and I suspect they haven't got the network to support them, they may be from single parent families. They may be from a lower income band.'

My research also suggests that the assistance of a mentor can be a highly influential factor. As Jason commented: *'in your life you meet unexpected angels, people who come in for a moment and do something significant to steer you in the right direction. I am privileged enough to have met a few'*. Jason explained that from the age of 12,

he had wanted to work in a prestigious city firm, he recounted how his father arranged for him to speak to someone:

'My Dad arranged for me to go and see somebody [senior figure in the firm]. He was blind since he was eight, and he said to me "In the kingdom of the blind, the one-eyed man is King". What he was saying to me was "you are the only person that can sort yourself out".'

Ollie also refers to an 'angel' who inspired him to change his degree course. He explained that, initially, it was his teacher who was inspirational in getting him to return to university:

'I just turned up [at school] one day—and I said this is what happening to me. He said, "Everything is fine apart from your eyes, let's start running". So we used to get a pair of women's tights, we used to tie them in a band, and he basically used that as a guide rope and we used to run. I think that teacher was very instrumental in getting me back into education.'

Later in the interview, Ollie explained that after he completed his degree, he spoke to a number of charities to help find employment:

'I asked them "What can I do?" "You can do some piano tuning, or you can do some basket weaving." I was not really interested in that. I had to take matters into my own hands and the only blind person I really knew in the public sphere doing a good job was David Blunkett, the Home Secretary at the time. I wrote to him and said "Dear David, I have recently lost my sight as of a year ago. I would like to see how a blind person operates in the work place". He was like "Of course, come in". I went to the House of Commons and followed him around for two days. I was absolutely in awe of this blind man who could rise to the top of the country and run it.'

Both Ollie and Jason successfully completed their degrees and professional examinations and now work in the city. In striving to return to employment, find employment, or continue with their education, participants spoke of wanting to maintain a 'sense of ordinariness' (McLaughlin and Coleman-Fountain 2018: 68) by undertaking the mundane taken-for-granted tasks that they had accomplished prior to their sight loss. LHON ruptures everyday life and participants sought to repair the rupture by resuming (subject to the limitations of sight loss) their previous lives and regain a 'sense of normality' in the face of that disruption (Gregory 2005; Buse and Twigg 2018; Coleman-Fountain 2018). One everyday taken-for-granted task that is said to typify the ordinary is that of dressing. In their research, Buse and Twigg

(2018) explore how people diagnosed with dementia and their carers are able to restore continuity or 'ontological security' (2018: 23) by focusing on dressing practices. The authors suggest that dress, and dressing, can support a sense of '*feeling normal* through the maintenance of clothing and routines which sustain biographical continuity at an embodied and tactile level' (2018: 33, original emphasis) and, so manage the disruption caused by the condition.

Participants talked of wanting to ensure they were dressed appropriately. Mary (diagnosed with LHON and MS) explained that she cannot see colours and relies on her daughter to arrange her wardrobe:

'she puts the darker things on one side of the wardrobe and the lighter things on the other side. I have two wardrobes, one in my bedroom with my winter things in and the other one is in another bedroom with my summer things in.'

By asking her daughter to arrange her wardrobe, Mary maintains a level of independence and 'continuity of identity and self-expression' (Buse and Twigg 2018: 19) as she is able to ensure that her clothes are colour co-ordinated and that she is wearing the appropriate clothes for the seasons. The ordering of the wardrobe has also been considered to be a 'strategy for the ordering and management of our everyday lives' (Woodward 2007, cited in Buse and Twigg 2018: 23). For Mary, dressing appropriately allows her to manage the public perception of herself and 'pass' (Goffman 1968: 92) as normal.

For a number of participants, undertaking ordinary tasks required the use of assistive technologies. As observed in the previous chapter, participants recounted how they were not able to use their mobile phones or their computers. However, after undertaking extensive retraining, they were able to use their devices once again. This is consistent with the research undertaken by McLaughlin and Coleman-Fountain (2018) with children diagnosed with cerebral palsy. The authors described how a lack of dexterity made it problematic for their participants to perform daily activities but, after undertaking training, they were able to complete difficult tasks. The authors identified that the impetus to undertake training was the need to feel independent rather than dependent on others. This again resonates with many of the participants in this thesis who underwent intensive rehabilitation, so they could

use magnifiers to read documents and use software packages on computers which enabled them to resume their degree courses or return to their previous employment. One of the activities that personified ordinariness for participants in this thesis was the ability to cook. Laura, who loves to cook, is able to continue to do so: *'I do have a speaking thermometer—I haven't poisoned anyone yet'*. Catrin spoke of her son's culinary expertise, explaining: *'He enjoys cooking, we did buy him a couple of gadgets to help him, although I might have to go and check if his chicken is cooked (laughing)'*. For other participants, being able to undertake sporting activities, including joining a visually impaired running club or playing visually impaired football, also helped them to feel a sense of normality.

Striving to return to normality and resume their life much as it was before sight loss was evident in the interviews. This parallels Charmaz's research where she suggests a hierarchy of identities, including the 'restored self' (1987: 301), which sees individuals strive for a self that is the same as before, picking up their previous life and re-establishing a sense of themselves. Ross and Ollie may be said to typify the restored self. A few of the participants spoke of the activities they had undertaken and goals they had accomplished after sight loss, which they would not have even considered before. By attempting to excel and compete and take on new challenges, they adopt what Charmaz refers to as a 'supernormal self' (Charmaz 1987: 301). This supernormal self is alluded to by Jason:

'I have flown off the Orbit [large steel sculpture] in the Olympic Park. I have abseiled off Battersea Power Station before they started to renovate it. I have cliff jumped from Victoria Falls in Zambia, horse riding in Antigua, skiing in Canada and France, and quad biking.'

(Jason, diagnosed aged 15 in 2004)

Amy spoke of her passion for driving and, with the help of a local charity, was given the opportunity to drive fast cars notwithstanding her sight loss:

'I love cars, fast cars—Ferraris, Lamborghinis. I have with a couple of charities got to drive a Mini Cooper around a track, which was really fantastic and good fun. I have done quite a bit: I have walked on a fire, I have done abseiling. I have done loads.'

(Amy, diagnosed aged 18 in 2011)

In adopting a “supernormal identity”, Charmaz (1987:298) suggests that individuals avoid interactions with others who have a similar illness or disability. Sandra illustrates this point when recounting that she had been invited to attend a meeting with a sight loss charity: *‘the thought of being at a gathering of people with varying degrees of sight loss did not appeal to me in the slightest. I felt that I didn’t belong there’*. Later in the interview, she explained that she avoids the use of a white cane because she does not want to be identified as visually impaired: *‘I would prefer not to use a cane. I know everyone would prefer not to, but you know what I mean. I would find it ...I would have to put a paper bag over my head’*. For participants, not associating with other visually impaired individuals resonates with the wider sight loss literature (Southwell 2012) and appears indicative of distancing themselves from the visually impaired community. The use of the white cane, for several of the participants, was identified as a contentious issue and associated with both “felt” and “enacted” stigma (Scambler and Hopkins 1986: 33). This will be discussed in more detail later in this chapter.

Another contentious issue that divided opinion was the concept of “acceptance”. A few of the participants in this thesis have attended counselling for their sight loss, including the use of cognitive-behavioural therapy (CBT). When participants described their feelings at the onset of their sight loss (Chapter Five), they referred to the Kübler-Ross’s (1963) five-stage grief model (discussed in Chapter Two), ultimately arriving at the final end stage of “acceptance”. Both Beth and Sandra, who are in their fifties and seventies, respectively, referred to their inability to “accept” their sight loss. Beth indicated:

‘I don’t think I will ever accept it. It’s like I can dream about the past and about my job and I know that I will never do it again’.

Sandra similarly expressed the view: *‘I think the problem with acceptance is that it can almost mean giving up hope. I know that they are not mutually compatible, but they do appear to be.’*

Similarly, Ollie, who is now in his twenties, also claimed that he did not envisage a point in the future where he would accept his sight loss:

'I don't think I will ever accept it. I will never be able to accept on paper an 18-year-old person in excellent physical condition losing their sight. I don't think I will be able to accept that. I have come to terms with what my life is now, and I will appreciate all the benefits that have derived and all the things that I have done. I have come to terms with that and I appreciate that. But emotionally, I would never want to go through that or ever wish that on anyone because, if someone had told me how hard it was to recover from sight loss, I wouldn't have done it. Because there is no manual that is written on how to deal with sight loss in an 18-year-old person.'

A similar view was expressed by Adam:

'I will never accept it [sight loss]. How can you accept having something you were born with taken away from you? I would say I have got on with it, I am getting on with it, and I am putting up with it. There is always that day that I hope to God that somebody says to me "we can sort your eyes out". I doubt it will happen in my lifetime.'

In contrast, Jason viewed sight loss as a life—affirming event, suggesting that he put more effort into his academic studies. Jason adopts a different attitude to Ollie and Adam when discussing acceptance: *'I have accepted it [sight loss]. I am so grateful for what it [sight loss] did for me. I don't believe that lazy kid was capable of what I have achieved'*. Jason's comments are consistent with Petersen's (2006) research with people diagnosed with diverse genetic conditions, highlighting how his participants spoke of the valuable lessons they had learnt and their positive experiences since being diagnosed. Andy's comments that he has not only accepted his sight loss but is moving on with his life: *'I have only just over the last three years accepted it and I not [only] accepted it but have moved on from it'*. Other participants, whilst they did not feel that they have reached the point of "acceptance", did consider they had reached an accommodation in that they occupy the middle ground, expressing their gratitude for the things they experienced before sight loss, and that life could be much worse. Adopting this attitude is consistent with the research undertaken by Moore (2000: 578), whose participants talked in terms of being 'blessed' with some residual vision. George, who after his sight loss attended a residential sight loss college, reflected on his experience of meeting other visually impaired students:

'There were young girls and boys there [college] who had been born blind and deaf. At least I had 16 years where I have got memories—like how do you explain what the sky looks like to someone who has never seen it, or

a star? You can give someone an object to hold and they... it's tactile but how do you explain what the sea is like or grass? You can't, or colours. So, I have had that, and I am quite grateful for that.'

Expressing a similar view to George, Amy also voiced her gratitude that her life could have been much worse: *'There are other people out there worse off than me. I could have cancer and die. I haven't got that, I have got my life. I am a still out there.'* These two accounts echo previous research on the narratives of those living with chronic illness (Petersen 2006). One of the older female participants, Mary, who has been diagnosed with LHON and multiple sclerosis (MS), has a younger sister, Sian who had coped with symptoms of MS for many years. In 2015 Sian began to experience problems with her eyes and was subsequently diagnosed with LHON at the age of 48. Both sisters adopt a pragmatic view of their sight loss. Mary was particularly stoic:

'Everyone says how do you cope? I just get on with it. My motto is you can't do nothing about it. There is no point sitting in the corner moaning, as no bugger listens. You know what I mean? Everyone sitting moaning "look at me, look at me". There is no good sitting in the corner saying woe is me. It doesn't do anything for you, does it?'

(Mary, diagnosed aged 31 in 1992)

In adopting the 'mustn't grumble' (Barnes and Mercer 2010: 54) approach, Mary presents to the outside world an account of her sight loss which, at times, is inconsistent with her 'private' thoughts (2010: 54). As the interview progressed, Mary began to explain a little more about her symptoms:

'[T]he tips of my fingers are always numb. I can't pick anything up... my extremities.... It is awkward to describe the numbness because it is always there. My arm is always numb. I have got a heavy leg on the right side.'

Later in the interview, Mary said that her sight loss did get her down: *'I am a bit pissed off [...], I think why me?'* In their research with participants diagnosed with osteoarthritis, Sanders *et al.* (2002) also claim that their participants presented a sense of resignation about their symptoms. However, participants also conceded that the consequences of their illness had a major impact on the way they lived their daily lives. Mary explained that, at times, her inability to do things for herself intrudes into her day: *'I can't drive; now I have to wait for someone to take me*

shopping. I was going shopping for an outfit for a wedding: my daughter had to come with me. She had to go on the floor to put my foot into the shoe. I couldn't feel where the shoe was.' Before her sight loss, Mary was the 'one taking care' of her family, doing the shopping, cooking and cleaning, which typifies the traditional gendered roles within the family. However, her husband now has to perform the household tasks which has disrupted both of their lives.

I have discussed the longer-term consequences of living with sight loss experienced by participants. I have also described how the support of family, allies and mentors together, with use of assistive technologies helps participants in undertaking ordinary mundane everyday tasks. In accomplishing such tasks, the ordinary participants move towards reconstituting the self and regaining a sense of normality. In the next section, I will consider notions of stigma and disability within the context of sight loss.

Sight loss, disability and stigma

In the final part of this chapter, I consider conceptualisations of disability and stigma within the context of living with sight loss. Some of the participants claimed that they had experienced stigmatising attitudes since the loss of their sight. This was particularly evident when they were using the white cane—a powerful symbol of disability (Southwell 2010). I begin the discussion by considering the relationship between impairment and disability which remains contentious within the disability literature, with debates focusing on the dichotomy between the medical and social models of disability (Barnes 1990; Oliver 1990). Criticised for predominantly adopting the medical model (Oliver 1990; Pfeiffer 1998, 2000; Bickenbach *et al.* 1999), the WHO (1980) International Classification of Impairments, Disability and Handicaps (ICIDH) defines impairment as 'any loss or abnormality of psychological, physiological, or anatomical structure or function (Pfeiffer 1998: 504). Disability is defined as 'any restriction or lack (resulting from an impairment) of ability to perform an activity in a manner or within the range considered normal for a human being (Pfeiffer 1998: 504). A handicap is defined as 'a disadvantage for a given individual, resulting from an impairment or a disability, that limits or prevents the fulfilment of a role that is normal (depending on age, sex, and social and cultural

factors) (Pfeiffer 1998: 504). Hansen and Philo argue that the medical model of disability invites a 'personal narrative of "tragedy" followed by "heroic" efforts at self-adjustment' (2007: 494). Many participants in my study expressed their irritation when others treat them in such a way (see below). Within the context of the ICDH, LHON is a condition that causes impairment (deterioration of the retinal ganglion cells), resulting in disability (severely sight impaired) and handicap (restrictions in full participation in social roles).

Bury (2000: 1073), who was one of the three original authors of the ICDH, asserts that the classification was an attempt to move away from a 'narrow medical model of health and disease—one primarily concerned with body systems and aetiologies—to one which recognised the *consequence* (original emphasis) of health-related phenomena'. Amid criticisms of the ICDH, including a suggestion that it advocates eugenics and euthanasia (Pfeiffer 1998), Bury contends that the ICDH has made a 'positive difference' (2000: 1073) to people living with a disability. Notwithstanding his assertion, the ICDH was subsequently revised and replaced by the ICDH-2, The International Classification of Functioning, Disability and Health (WHO 2000). Although again not without its critics (Pfeiffer 2000; Imrie 2004), the rationale of the ICDH-2 moves away from disability as a consequence of disease towards the social model, which distinguishes disability and impairment, acknowledging that 'disability is located within oppressive and excluding environments' (Ryan 2005: 292) or, as Oliver (1996: 35) has pointed out, 'disability has nothing to do with the body but everything to do with society'.

The extent to which the ICDH-2 has integrated a social model of disability remains contentious, as critics suggest that the model medicalises disability and, so, allows medical professionals to make judgements on the quality of life of individuals with a disability who, apart from their disability, may be otherwise healthy (Pfeiffer 2000; Green *et al.* 2002). This is particularly pertinent for LHON where many affected individuals are young, fit and healthy at the time of their diagnosis, and remain so after their sight loss. The disability movement have been instrumental in moving debates away from focusing on the medical model of disability towards an examination of a society that promotes disabling attitudes through social structures including cultural frameworks which privilege able-bodied normality. In her book

Songs at Twilight (2011), Susan Dale, a visually impaired sight loss counsellor, suggests:

‘The attitudes society has towards blindness are socially constructed, thus within a world where the majority are sighted social interactions have developed to consider vision as “normal” and those of us who have different vision as “abnormal”’ (2011: 1).

If Dale’s assertion is correct that people living with a disability are perceived as abnormal, and people who do not have a disability are normal, we can ask what does it mean to be ‘normal’? Who, or what, determines the parameters of normalcy? Davis (1995: 23) suggests that we live in a world of norms where we strive to be normal or, indeed, try to avoid being placed in the normal category. He further suggests that to understand disability, it is necessary to return to the concept of the ‘the norm, the normal body’. By focusing not on the construction of disability, but instead on the construction of normalcy, Davis argues that the “problem” is not the person with disabilities; the problem is the way that normalcy is constructed to create the “problem” of the disabled person’ (1995: 24).

Many participants in this thesis did not consider themselves to have a disability. Adam’s view is typical of other participants when he says: *‘I think of myself as completely normal apart from the fact I can’t see very well’*. Barnes and Mercer (2010: 49) suggest that individuals who acquire a stigmatised condition in adulthood (as opposed to congenital impairment) are likely to be more resistant to identifying as a person with a disability because of ‘their prior assimilation of a personal tragedy perspective on impairment and disability’. Adam later explained that: *‘for a long time I was, like, I am not going to go out with a blind woman, never going to do that, it’s not going to happen. I am not going to have any blind friends, I am normal’*. Although participants did not consider they had a disability, a recurring theme in the interviews was a desire, like Adam, to be perceived as normal. This was apparent in the interview with Declan, aged 22. During my interview with Declan, he unexpectedly jumped out of his chair and, standing in front of me, asked *‘Do I look right? Am I looking right? [pointing at his clothes.] Was I walking alright? [coming from the clinic waiting area.] Do I look normal to you?’* For Declan, the way in which he walks and being dressed appropriately is crucial in managing the ‘public

presentation' (Entwistle 2000: 11) of himself as someone who is 'normal' resonating with the work of Buse and Twigg (2018) who highlight the importance of dress in managing the visibility of disability in the public realm.

Declan raised the issue of 'looking normal' when we were discussing an unrelated topic. I asked Declan what had prompted his questions, to which he replied '*I just want to be normal really. I just want to feel normal because I am still so young. I just know that I am not like everyone else*'. Davis (1995: 24) suggests that people have an 'inherent desire' to compare themselves to others. In Declan's case, he expressed feelings of inadequacy because he believed that when he compared himself to others, he did not meet their expectations. Declan's sight loss has had an impact on his sense of self as he considers that, notwithstanding his attempts to be perceived as normal, he does not actually believe that he is normal. As the interview progressed, it became much clearer why Declan felt he was not perceived as normal. He explained that after his sight loss, he initially stayed in mainstream education but, during this time, he was bullied by other children in the class: '*they called me blind this and blind that. You can't see this, and you can't see that. I had a teaching assistant and they all said I was thick*'. Declan's comments are consistent with McLaughlin and Coleman-Fountain's (2018: 68) research with children with cerebral palsy who, having been subjected to name calling, being the subject of jokes and physical bullying, realised at a young age that 'others did not treat them as belonging in their social worlds'. Declan was later placed in a unit within the school with other children who had severe learning disabilities. He described how his teachers had low expectations of what he as a visually impaired student, could achieve:

'I noticed how they treated people with a VI, they were treating them as if they were babies. As if you had something...they would treat them like, I don't like to say it, but like people treat Down syndrome people and people like that. Obviously, you can tell by me, the way I am speaking to you that I am not really slow [...], they would drag you around by your arm, and if I wouldn't go, like, they would grab me. They would try and guide me as well; I don't need it. I was trying to be as normal as possible.'

(Declan, diagnosed aged 12 in 2006)

In his study of blindness in America, Scott (1969) identified that children and young people who attend specialised rehabilitation units are taught to adopt the stereotypical 'blind' role in everyday social interactions. This is not only contrary to the attitude and behaviour of the wider visually impaired community, but it also renders the severely sight impaired 'seriously maladjusted to the outside world' (1969: 199). Declan's residual peripheral vision negates the use of a white cane. He felt it was unnecessary for the teachers to try to guide him, not only because he perceived this as a threat to his mobility and his independence, but also because it hindered his attempts to be considered normal. Two of the other participants in this study, Ollie and Jason, were particularly vociferous in sharing Declan's view that the visually impaired can be held back by low expectation of their ability. Both participants considered that the visually impaired do not always reach their full potential because of society's deeply embedded discriminatory beliefs. As Ollie explained:

'I think the greatest hindrance to a lot of VI people now is that they are really held back by the beliefs and the intrinsic and deep embedded discriminatory views that some people have. I am not really one to go and beat the left-wing disability drum which is disability rights. Of course, it's incredibly important and we are not going to move on without that, but I also take the view that no one should view me any differently. I should be doing all the work that all the other employees do here, and I will tell someone if I can't.'

Jason also expressed his frustration that the expectations placed on the disabled are low:

'The thing I hate seeing is the carers, parents, do-gooders, and well-wishers who restrict you because their expectations are defined by what's in the ignorant social understanding of what it means to be VI. In other words, you don't know anybody who is blind, you have no experience other than seeing somebody on the street with a dog or a cane, and your concept of blind is black, no vision whatsoever, can't make a cup of tea without needing help and therefore how can they possibly have a career. So, if your expectations are down there, then this person you are their moral support and then their emotional net to catch them if that's your expectations of them, that they won't do much; you have written them off and I think out of the restrictions on us, the expectations are so light and that can ripple into the expectations of yourself.'

The point made by Jason, that the visually impaired may have low expectations of their own ability has also been observed by Goffman, (1968: 18) who suggests that

stigmatised individuals are acutely aware of what others see as their failings and, on occasions, acknowledge (albeit briefly) that they do 'indeed fall short of what they really ought to be'. A similar point is made by Scott (1969), who considers that the visually impaired are acutely aware that they possess a spoiling attribute, which comes to the fore when in the company of the sighted:

'The stigma of blindness makes problematic the integrity of the blind as acceptable human beings because those who see impute inferiority, the blind man [woman] cannot ignore this and is forced defend him [her] self. If as sometimes occurs, the blind man [woman] shares the values of the sighted, the process becomes even more insidious [...] a man's [woman's] personal identity is under attack from within as well as from without' (1969: 25).

Here Scott is referring to the process of self-stigma which operates at an individual (micro) level and witnesses the stigmatised discredit themselves (Goffman 1968) by adopting society's attitudes to disability, resulting in feelings of self-hatred and shame (Link *et al.* 2001) and having a negative impact on self-esteem (Green 2009). Goffman (1968: 11) posits that stigma sets people apart from "normals"; they are considered to be socially inferior, rejected as 'a blemished person, ritually polluted to be avoided especially in public places'.

Stigmatising attitudes of the sighted towards participants whilst using public spaces has, on occasions, proved problematic. A number of the participants referred to being subjected to comments made by complete strangers who cast them in the role of the tragic figure to be pitied and feared (McLaughlin and Coleman-Fountain 2018). Garland-Thomas (2009: 19), suggests that in '[A]vowing disability as tragic and shameful', we have concealed those living with a disability in secure institutions, special schools and hospitals. So, when we happen to gaze upon the sight of disability, 'we stare in fascinated disbelief' (2009: 20). A similar point is made by Carnevale (2007) who observes that, in public spaces, the stigmatised have their privacy invaded as they are stared at or randomly approached by strangers and, as a consequence, the stigmatised are not 'afforded the social respect that is normally accorded to others' (Carnevale 2007: 10). Fourie (2007) also makes the point that people who have not been exposed to sight loss are embarrassed and not equipped to interact with visually impaired people.

All participants described unwanted public attention as irritating, annoying or patronising. Adam says: *'Some people do have that automatic response of "how do you cope? How do you do this? How do you do that? You are disabled". I know I am disabled, it doesn't mean I am an idiot.'* Jake also voices his frustration at somehow being perceived as less able because of his sight loss:

'Personally, this really winds me up and I would say I probably get this once a week so it's "How do you cope?", "It's amazing how you deal with this" or "I can't believe how you do everything", when really I am just getting on with things. It's just normal really.'

In his research with participants living with disability, Watson (2002) observes that a number of his participants described themselves as normal and refuted suggestions that their disability affected their view of themselves. For his participants, living with impairment was a fact of life and not considered to be important for their sense of self. This echoes Jake's view of himself as he feels normal, considers himself to be normal, and wants to be thought of as just a 'normal lad' (McLaughlin and Coleman-Fountain 2018: 71) undertaking normal activities — the same as everyone else. Watson makes the point that for his participants, it is not their disability that stops them from undertaking activities, but the presence of 'societal and environmental barriers' (2002: 515). This resonates with the views expressed by participants with LHON who also referred to their experiences of being stigmatised and marginalised through the social organisation of public spaces which, at times, make it impossible to undertake normal everyday activities. As Schillmeier (2007a: 195) notes, it is these ordinary acts of everyday life that 'make up the complex and contingent scenarios of disability that create enabling and disabling' practices. The disability studies literature has also highlighted that disability is not a result of individual impairment, but the product of social structures and processes that are potentially disabling (Shakespeare and Watson 1997; Barnes, Mercer and Shakespeare 1999; Green *et al.* 2002). This view is echoed by a few of the participants, including Ginny who strongly refutes the suggestion that she has a disability: *'I am disabled by the way the world works sometimes, but I am not as a person. It is just my eyes don't work too well.'* Ginny claims that within her home, where she knows where everything is, she is completely independent.

However, it is in public spaces that she encounters problems. Imrie (2001: 232) has suggested that:

‘For the disabled people, the physical construction of urban space often (re)produces distinctive specialities of demarcation and exclusion, from the lack of access to public transport systems to the absence of visual clues or guides in towns to enable the vision-impaired people to move with ease.’

The impact of the lack of visual clues referred to by Imrie is demonstrated by Stuart who was diagnosed with LHON when he was 23 in 2013. He explains the problems he encounters when visiting somewhere new: *‘if I am in a new place or go to a new bar and try to find the toilets, I ask someone at the bar and they say they are over there... the problem is getting the right toilet; I have frequently gone to the wrong toilets’*. Toilets are places that are usually identified by a symbol (male or female) on the door, but for the visually impaired, this is meaningless. Stuart explained that it is embarrassing as he only becomes aware of his mistake when the female occupants shout at him to leave. For Stuart and the other male participants in particular, once inside the toilet, they are faced with numerous challenges. For example, the location of urinal or cubical, washbasins, and hand dryers. There is a small body of sociological literature that has explored public toilets as places of exclusion for those living with a disability (Molotch 2010; Slater *et al.* 2017), with Molotch suggesting that toilets become a ‘zone of exclusion’ (2010: 16). Whilst disabled women may be more inclined to ask for help, men confronted with the gendered role of being strong and independent are less inclined to ask for information and help.

One of the other participants, Tim, explained the problems he encountered the first time he attempted to use a cash machine after his sight loss:

‘Accessing my own money is quite a big thing for me because I have been independent with money since I was probably about 10 and that was always a sense of independence. You know, how to control your money, how to have access to your money and not to have to rely on somebody else to put the PIN in and press the button. I just remember thinking “How am I ever going to be able to do anything of any use to anyone when I can’t even get a tenner out of the cash machine?”’

In his research exploring how the visually impaired use money, Schillmeier (2007a, 2007b) suggests that disability is the effect of complex sets of diverse social relations

that link bodies, material objects and technologies. He goes on to demonstrate that money—and indeed accessing money (via ATMs)—plays a pivotal role in being able to participate in daily life and how money technologies make up (dis)abling practices for the visually impaired. For Tim, accessing his own money is associated with leading an independent life. Schillmeier makes the point that cash machines are ‘standardised materialities and technologies’ (2007a: 197) which for people, like Tim are highly problematic to access and use properly in everyday life. The effect of not being able to use money and money technologies, Schillmeier suggests, is that the visually impaired are ‘disabled’ in their social interactions, making them ‘vulnerable, restricted in their mobility and dependent on others’ (2007a:197).

Earlier, I referred to the use of assistive technologies and how they help participants regain a sense of normality by allowing them to undertake ordinary tasks. The use of one piece of equipment—the white cane—has proved to be a contentious issue for participants and will now be discussed in more detail.

The white cane: a symbol of stigma

In Chapter Two, I reviewed the stigma literature (Goffman 1968; Scambler and Hopkins 1986; Link and Phelan 1999, 2001; Scambler 2006, 2009, 2018;) before discussing the complex interconnections between sight loss and stigma. The majority of the participants in this study reported experiencing a degree of stigmatisation, both enacted (overtly discriminatory attitudes and behaviour used towards them) and felt (fear of shame and the fear of being confronted with enacted stigma) following their sight loss. Stigmatising attitudes were particularly evident when participants used a white cane. Notwithstanding advances in technology, the white cane remains the most popular low vision aid (Hersh 2015). It performs a dual function in that it aids mobility in helping people to avoid obstacles, and it is also a familiar symbolic indicator that the individual is visually impaired and may need assistance (Hersh 2015). Southwell (2012: 108) suggests that visually impaired people are reluctant to use the white cane because there is a ‘deep-rooted need to appear “normal”’, with Fourie (2007: 225) explaining that, for him, using the white cane represented a symbolic ‘throwing in the towel’, signalling a loss of ‘normality’, not just for him but to all of those in his world. Avoiding the use of the white cane

allows individuals to participate in what Goffman (1968: 92) refers to as 'passing'; presenting to others as not having a disability and, thus avoiding being stigmatised.

Evident from the participant interviews is the divergence of opinion surrounding the use of the white cane. Some participants are reluctant to use it for fear of being perceived as vulnerable and stigmatised, and not being accepted as one of the "normals". Tim's view sums up the feelings of many participants:

'I carry a cane everywhere I go, but I choose not to use it all of the time. I am lucky to have the choice because that means people aren't judging me before they get to know me. Whether you like it or not, if someone is walking along with a guide dog or cane, people automatically make assumptions about that person before they have even spoken to them. I am able to disclose my disability in certain situations and tell the person what I want them to know about it rather than them forming their own opinion.

However, other participants such as Jason, have embraced its use as a means to achieving a more independent lifestyle:

'The stats around the percentage of blind people that will embrace the stick are so low. I find it liberating, it's sort of a visual cue to sighted people who don't have any training or knowledge or experience of the VI community that this person does have a visual impairment and might need help.'

Ginny, mirroring the wider sight loss literature (Hayeems *et al.* 2005), also explained that learning to use the white cane provided her with a level of independence. Ginny also uses the cane as a 'stigma symbol' (Goffman 1968: 124), alerting others to her visual impairment and signifying that she may need assistance:

'I am so pleased that I learnt to use the cane, it is a great symbol. It explains to someone that I am partially sighted. If you have got the cane as a prop, they are more likely to help'

I interviewed Ginny at the Royal Albion Hospital; it was the first time she had travelled by train on her own. When we met, she was enormously proud of her achievement of arriving at the hospital on time and, as she put it, '*in one piece*'. Ginny explained that, had she not been using the white cane, she doubted whether she would have had the confidence to attempt the journey on her own. For her, the white

cane has made a positive contribution to her mobility and independence. Similarly, Ollie has also embraced the use of the white cane:

‘What a cane does is symbolise the fact that you can’t see very well. It’s not so much a navigation device as a symbolic aid for people to see what you are and that is very useful. The cane is a good piece of technology, it has been used for 250 years now, it clearly works, and it clearly symbolises that you can’t see very well so you may need some assistance.’

The comments made by Jason, Ginny and Ollie demonstrate that, for them, using the white cane alerts others to the fact that they are visually impaired and, as such, may need assistance. However, not all the participants reported receiving positive responses from the use of the white cane. This is consistent with research undertaken by Ryan (2005: 298) with mothers of children with learning impairments. The research highlights how, in order to overcome negative attitudes, the mothers provide an account to others of their child’s impairment or give advance warning by the use of a ‘stigma symbol’, in this case, the use of ‘major buggies’ [large pushchairs]. As a result of these strategies, just under half of the mothers reported receiving positive responses to their accounts or explanations of their child’s learning impairment. Although there were positive accounts of using the white cane, the majority of my participants spoke of their reluctance to use the white cane because they had previously been subjected to unwanted attention in public spaces; this mirrors the experiences of participants in the research of Stevelink *et al.* (2015), who were subjected to verbal and physical abuse. Amy is a reluctant white cane user, as she explained: *‘people jump over mine like a skipping rope, they think I am a slow coach, they are like “move out of the way”. Someone actually stepped on mine and bent it’*. Adam gave up using the cane after initially using it:

‘I think if you do use a stick, people think you are blinder than you are or even more disabled than what you are. I used the cane for a couple of weeks when I went to college. They all jumped round the cane. They were literally doing high jumps over it. I thought “stuff this”.’

By using the white cane, the visually impaired become what Goffman (1968: 14) refers to as ‘discredited’ (their sight loss is known about, or ‘evident on the spot’). Some participants believed that the use of the white cane marked them out as vulnerable or as less able than those who are fully sighted. Jason explained that he

carries a white cane when using public transport and on those occasions strangers approach him: *'some people will come up to you on the tube and say, "Do you need help?" mouthing it loudly and slowly, thinking "are you deaf"? Or am I foreign and thick (laughs)'*. Adam also explained that he has been spoken to as if he has a hearing problem: *'the odd person who has talked to me like I am deaf, or has talked down to me, I have said "No, no, you don't do that anymore"'*. Drawing upon the work of Gowman (1957), Goffman, refers to this attitude as a "gestalt of disability", commenting that the blind are 'frequently shouted at as if they are deaf' (1968: 16). Although Ollie uses the white cane and extolled its virtues, later in his interview, he also conceded that his work colleagues are guilty of enacted stigma:

'I think that people think when you lose your sight and you carry a cane there is a clear deficiency in IQ, so they correlate your sight loss to not having any brains. Still today, people will come up to me and to my secretary and say, "Does Ollie want XYZ?" I will be like, "I am stood right behind her, you can address me", but people will always think that you are inferior due to the fact you can't see very well.'

For a few of the younger participants, the decision not use a white cane is associated with being conscious about their appearance. Catrin explained why her son refuses to use a white cane when potentially he should:

'Image. He is a 23. I suppose it is the image that people see of him. He doesn't want to be known as blind because of the stigma I suppose. People with a disability are seen as not as able.'

The desire to pass as normal raises conflict between the need to use a white cane for safety and the associated social stigma (Southwell 2012), resulting in shame (Fourie 2007). Jake is reluctant to use a white cane: *'I don't use a cane; it's a personal choice really. I should potentially in some areas use one'*. Later in his interview, he disclosed that he had been involved in a road traffic accident:

'I got run over. I was two paces behind my friend. Because I was taking too long, she [driver] tried to swerve round me. I didn't see her coming and she clipped me and ran over my leg.'

Jake's experience is typical of other participants who valued being able to present to others as normal and avoid stigmatisation, (Green *et al.* 2002). However, in his desire to be perceived as sighted, Jake is unable to meet routine expectations of

ability (Green *et al.* 2003) and his refusal to use a white cane had serious repercussions for his personal safety. Two participants, Laura and Jason, explained that they, on occasions, did not use a white cane because they were concerned their children, as opposed to themselves, would be stigmatised. This is described as courtesy or associated stigma (public disapproval as a consequence of associating with a stigmatised individual or group) (Goffman 1968: 43). Laura explained: '*I think when they [children] go to High School, other kids can be very cruel, so I don't use it [white cane] in my hometown*'. Jason, who earlier described the cane as "liberating", later in his interview conceded that he avoids using a cane when trying to protect his children:

'My biggest reason really is how people perceive me in terms of that vulnerability and also when I pick up my children from school. I don't want them—and this is really sad when you quote this as an example—I don't want my children to be picked on at school because their dad has got a disability or is blind. That sounds terrible when I say it but it's the truth really. Children will pick up on anything.'

Southwell (2012) suggests that to overcome stigmatisation, there is a need to promote positive visually impaired role models and undertake a programme of public education. Jake, also referred to the lacuna in public understanding of sight loss:

'I don't think there is enough education about different forms of disability. It is interesting now. So, my children are at nursery and they learn quite a lot around hearing impairment and sign language and things like that, but there is very little done around other disabilities really to make it normal. In school, I never learnt anything about disability. It should be part of science and biology.'

One other problem identified by participants who did not use the cane was being challenged about their sight loss. LHON presents as a 'paradoxical' (Fourie 2007: 222) eye condition as, in contrast to other eye conditions, the eyes look normal, and, although some individuals may be registered as severely sight impaired, the level of their residual peripheral vision allows them to navigate in public spaces without the assistance of a white cane or guide dog. In Goffman's (1968) terms, they are 'discreditable' (their sight loss is not known about). There is a misconception that sight loss means living in a world that is black with no light perception. Schillmeier (2008) suggests that when sight impaired people tell others that they have *some*

residual vision, these claims are dismissed as from the sighted perspective, they look 'blind' to them, and categorise them accordingly. In contrast, participants who retained a good level of residual peripheral vision indicated that they are often challenged by others who do not believe they are visually impaired because they *look* normal. After losing his sight, David attended a college for the visually impaired to which he attributes his determination to be as independent as possible. He also explains: *'I still have got quite a bit of peripheral vision, so I can still see quite a bit'*. He went on to claim that:

'People have accused me of lying about my sight quite a few times because I am quite independent, and my eyes look normal and you know they see me about town avoiding bollards and avoiding bumping into people and things like that. I can fully understand why they accuse me because I didn't understand sight loss until it happened to me.'

(David, diagnosed in 2011 aged 24)

As has been demonstrated by the narratives of participants in this thesis, the white cane continues to be a contentious issue. Whilst it has the advantage of aiding mobility and alerting others that the owner may need assistance, it also symbolises difference and thwarts attempts by those who wish to be perceived as 'normal'.

Summary

In this chapter, I uncover the longer-term consequences of living with sudden sight loss. In observing the minutiae of everyday mundane micro encounters, including maintaining social relationships, continuing to undertake education and employment, I have highlighted the unique and diverse experiences of participants. For a few participants, like Beth and Sandra, they have not adjusted to their sight loss and continue to experience anxiety, depression, and, on occasions, suicidal thoughts. However, the majority of the participants, consistent with Charmaz's (1991) research, experience good and bad days and have been able to continue with their lives—regaining a sense of normality (Davis 1995). Some participants, in reconstructing their former lives, have adopted a 'supernormal identity' (Charmaz 1987: 296) by undertaking high-risk activities, successfully returning to their studies, obtaining First Class Honours degrees, and securing senior management roles within their organisations.

However, I also acknowledge that some participants experience feelings of stigma, which is particularly evident when using the white cane. Nonetheless, other participants (including Ollie and Ginny) viewed its use as a valuable tool—a symbol of their sight loss, inviting others to help when assistance is required. Whilst others refused to consider using the white cane, either because they had direct experienced of ‘enacted stigma’ or ‘felt stigma’ (Scambler and Hopkins 1986: 33). On occasions, their refusal to use the white cane impacted on their safety, particularly when crossing busy roads.

In conclusion, this chapter in focusing on participants lived experience of LHON, has tapped into the wider debates surrounding the medical and social models of disability and how the fear of stigma shapes their social interactions. It has also highlighted how participants strive to regain a sense of normalcy and, in so doing, live an ordinary life.

In the following chapter, I consider participants anticipated futures by exploring narratives of hope within the context of the development of new treatments to restore sight or avoid the birth of children with LHON.

Chapter Seven: Finding a Cure: Narratives of Hope and Reproductive Responsibility

Introduction

In the previous chapter, I considered the longer-term consequences of living with Leber hereditary optic neuropathy (LHON). It was evident from the discussion that a small number of participants claimed to have accepted their sight loss and were not interested in finding a cure, in contrast to the majority of participants who spoke of their hope that new treatments would become available to restore their sight and return them to what they perceived as a 'normal' life. There are currently two clinical trials being undertaken in the UK (the GenSight Gene Therapy clinical trial²⁵ and the Santhera Pharmaceuticals LEROS Idebenone clinical trial),²⁶ which are testing the effectiveness of treatments that potentially stabilise, and/or restore sight following, the onset of LHON. At the time of data collection, none of the participants had been enrolled into the GenSight clinical trial. However, a few were involved in the Santhera LEROS Idebenone clinical trial (hereafter referred to as the LEROS trial).

Drawing predominantly on interview data and some observational data, I locate my claims within the sociology of hope (Petersen and Wilkinson 2014; Brown *et al.* 2015; Petersen 2015). Within the field of health and healthcare, there is a considerable body of literature that has considered the concept of hope (Bernard 1995; Simpson 2004; Wiles *et al.* 2008; Petersen 2015). Hope is said to have a powerful therapeutic effect, which is thought to be an important factor in shaping people's experience of illness (Simpson 2004; Petersen 2015). Where there is no prospect of recovery, Petersen (2015) suggests that hope provides the ability to cope with suffering and the apparent hopelessness of the situation.

²⁵ The GenSight (GS010 AAV2) Gene Therapy Trial is designed to evaluate the efficacy of using a Single Intravitreal Injection for participants who have been affected by the m.11778G>A LHON mutation for six months or less.

²⁶ The Santhera Idebenone trial is a Phase 4 trial evaluating the efficacy and safety of the long-term use of the drug Idebenone for participants affected for ≤5 years with the three primary LHON mutations.

The chapter will be presented in two sections. In the first, I introduce the concept of hope and explore how participants who are enrolled in the LEROS trial construct and negotiate hope to imagine their future biographies. Petersen (2015: 1) observes that the concept of hope ‘saturates both popular and political discourse and is seen to have some essential quality needed to unite communities and achieve change’. Within the LHON community, narratives of hope surrounding the potential benefits of Idebenone have been the subject of a sustained campaign by Santhera to have the drug routinely prescribed by ophthalmologist working in the NHS. By participating in what Novas (2006: 289) refers to as the ‘political economy of hope’, Santhera has been instrumental in mobilising stakeholders including research funders, research institutions, clinicians and patient advocacy organisations to move forward an agenda to establish research centres in the UK and encourage patients to participate in the LEROS trial. I consider how the ‘hype’ (Brown 2003: 3) surrounding the effectiveness of Idebenone has influenced participants’ actions and raised an expectation that Idebenone will restore their sight.

The second section of this chapter explores narratives of hope associated with the use of mitochondrial replacement techniques (MRTs) to avoid the birth of a child with LHON, with the introduction of MRTs raising the expectation that women will be able to give birth to a healthy, genetically-related child. I will consider participants’ experiential knowledge of living with LHON and how their understanding of complex genetic inheritance shapes their decision-making when considering their reproductive options. The use of new genetic reproductive technologies, such as MRTs, is controversial as they are said to devalue the lives of those living with a disability (Shakespeare 1998, 1999, 2011; Edwards 2004; Boardman 2014). The extent to which women who are affected or are carriers of LHON are influenced by such discourses when exercising reproductive choice will also be discussed.

The heartbreak clinic

Hope has been identified as a ‘major motivator of action, as a source of resilience to overcome adversity—in times of [...] limited options and despair’ (Petersen 2015: 1). Within the field of health, hope is delineated as an important element in the

healing process (Wiles *et al.* 2008; Petersen and Wilkinson 2015), with the use of hope scales and indices (Brown 2014) measuring both the influence and the impact of hope (Petersen 2015). When hope is spoken of within the medical setting, Bernard (1995) notes that it is usually in extreme situations, such as when people are faced with a terminal illness or death. However, he suggests hope is equally relevant within the context of living with chronic illness and disability—arguing that such states ‘disclose the very aspect of human existence that gives birth to hope, namely, that human beings are poised on the boundary between finitude and transcendence’ (1995: 38). Bernard (1995: 40) refers to an “‘existential paradox” of living with a chronic illness’. The chronically ill are said to ‘straddle a boundary between hope and despair’. For Bernard (1995: 48), to hope ‘means to project oneself beyond one’s present definition of reality, but with no guarantees against disappointment’.

In the previous chapter, a minority of participants indicated that they had accepted their sight loss and were not interested in finding a cure. Gerry, who was in his early sixties when he was diagnosed with LHON, explained: *‘I am not a cure chaser. I have accepted the fact that I have lost my sight’*. Gerry’s view is consistent with Schillmeier’s (2008) research with visually impaired people in which he observed that not all those who have experienced sight loss want to be cured. After receiving their LHON diagnosis, some participants were registered as severely sight impaired and discharged from the eye clinic without the prospect of any further treatment or a cure. The promises that heralded the arrival of the new genetics are rapidly coming to fruition within the ophthalmology clinic, with the discovery of the genetic basis of LHON and potential cures. However, there remains considerable uncertainty surrounding the prognosis of LHON and, consistent with Locock and colleagues’ (2009) research, this provided a source of hope for people with LHON. Notwithstanding the bleak future suggested by their ophthalmologists, participants continued to hope that their sight would not deteriorate any further or that they may have the mutation that is most likely to have spontaneous recovery. Other participants hoped that they would have the opportunity to be enrolled into clinical trials or that their family members would not be affected by LHON. As Petersen and Wilkinson (2014: 117) reflect, hope ‘has no single defining essence or significance,

but rather is ascribed multiple meanings, articulations, and implications'. For participants in this thesis, hope is multifaceted. Marion, whose son was diagnosed with LHON, explained that whilst waiting for her son's blood test results, she started to consider which of the three LHON mutations would give her son the best chance of restoring his sight:

'I can remember sitting there hoping its 11778,²⁷ that's the most common, so if he has got that, it means he is up for all the research things that are coming along [gene therapy]. The 14484, that's the one where there is the most chance of recovery. That will be good if he has got that.'

Adam, who was diagnosed when he was 14 years old, speaks for the majority of participants who are seeking a cure (and are willing to travel to find it) when he says: '*Hopefully my sight will be back. I am 26 and if I got my eyesight back when I was 36 or 46, I am not bothered. I just want it back as soon as possible. I will go wherever I need to go to get the cure*'. Similarly, Sandra, who was diagnosed in her late sixties, has struggled to cope with her sight loss:

'I would be prepared to go to anywhere if they could do something, you know, some intervention that would do something about this. I am in the fortunate position that I am not rich by any means, but I am not poor either. I have got the resources that I could pay if I needed to. If there were any intervention, I would be on the first plane or train or whatever it was. I would just do it.'

(Sandra, diagnosed aged 69 in 2015)

Sandra is desperate to find a cure for LHON and is prepared to participate in what Petersen *et al.* (2013: 670) refer to as 'medical tourism', a burgeoning trend, one which "[exploits]" 'the high optimism that surrounds new biomedical technologies'. After Amy was diagnosed with LHON, her family searched the internet to find a cure. Having read about stem cell treatment in China, they started fundraising to send her there:

²⁷The m.11778G>A mutation has been identified as the most frequently occurring mutation, accounting for approximately 60–90 per cent of cases, with m.14484T>C and m.3460G>A responsible for 25 per cent and 15 per cent respectively (Spruijt 2006; Martikainen and Chinnery 2015). New LHON mutations continue to be identified and currently there are over 45 point mutations of mtDNA reported (Aune and Walters 2012).

‘The internet was saying about research in China, they even had film bits of people there saying they had used stem cells and that it had started to work, it was a miracle and all this. I remember coming to the hospital and having an appointment with [ophthalmologist]; he said, “whatever you do, don’t waste your money on any of it”. He said “it’s all a rip off; people are going over there paying thousands of pounds. People are being injected with stuff, they don’t know what they are being injected with, and actually, it does absolutely nothing”.’

(Amy, diagnosed aged 18 in 2011)

The increasing popularity of medical tourism has prompted influential science bodies (including the International Society of Stem Cell Research) to raise concerns that private providers are profiting from clinically unproven and potentially harmful treatments (Petersen 2015). Petersen and Wilkinson (2014: 3), suggest that hope is increasingly being linked to the development of new technologies, particularly those that ‘promise to offer cures, [and] alleviate suffering’. For many participants the motivation for participating in clinical research was the hope that their contribution would lead to finding a cure not only for themselves, but also for their families and the wider LHON community, consistent with Herbrand and Dimond’s (2017) research with 21 women at risk of transmitting a mitochondrial condition to their children. The authors discovered that only a minority of the women who supported the introduction of MRTs were intending to use the techniques themselves. The majority of the women who were not intending to use the techniques nonetheless supported their introduction from a desire to provide hope for their families and hope for wider society.

Tina was diagnosed with LHON when she was 30 years old and claims she has accepted her sight loss, but as she and her brother have one of the rarer LHON mutations, she agreed to participate in clinical research to find a cure, not for herself, but for others:

‘It would be nice for other people to have a cure. I am not bothered for me. I am happy with the life I have got. I want to help other people understand it [LHON] because in my case, I do have a very rare mutation. My brother did get his eyesight back after 5 or 6 years. He is very unusual, being that we are related and we have the same mutation. If my eyesight did come back, then at least they have got the stuff from now, so it would be a good comparison for them. There is no guarantee that it will come back. I know that.’

(Tina, diagnosed aged 30 in 2013)

Laura is also participating in clinical research and summed up the feelings of a number of the participants:

'I am hoping they are going to come up with a cure for Leber's, I really am. I think of all those young boys, I saw them at [name of research centre], it is heartbreak clinic. I have heard young boys telling them [ophthalmologists] where to go, swearing at them. I have seen them coming out slamming the door and then trying to find their way round the wall trying to get out, falling over chairs and they are upset, and they are crying, it's heartbreak clinic. If they could just come up with a cure for these young boys.'

(Laura, diagnosed aged 48 in 2012)

For other participants, although they were hoping for a cure, they adopted a pragmatic attitude, accepting that this may never happen. Jake, who lost his sight when he was 15, acknowledges:

'Hopefully they can find a cure for my eyesight. I am not going to bed every night praying for a cure. I am accepting that potentially, probably most likely, they will never... I will never see properly again for the rest of my life.'

(Jake, diagnosed aged 15 in 2005)

Two male participants, David and Declan, referred to winning the lottery when discussing the possibility of finding a cure. David, who was diagnosed when he was 23 years old, says:

'I guess there is hope in the back of my mind. It's more like thinking of it as winning the lottery. It's what you would do if you won the lottery. You have that little hope in your mind where if I won the lottery I would do this but, at the same time, it's also knowing that it might not happen.'

(David, diagnosed aged 23 in 2011)

Similarly, Declan who experienced sight loss when he was 12, expresses how he would feel if his sight was restored: '*It would be like winning the lottery. It would probably be better than winning the lottery, better than winning the Euro Millions*'. Ollie, who experienced sight loss during his first year at university, indicated that as he has learnt more about LHON, he has become more realistic about the prospect of a cure:

‘I am actually very sceptical about finding a cure. I understand it is a whole body condition; it is a mitochondrial DNA condition. I understand the real limitations of finding a cure’.

(Ollie, diagnosed aged 18 in 2009)

One treatment potentially offering hope to participants that their sight will be restored is Idebenone, an antioxidant drug that, it is claimed, stabilises sight loss and may for some restore sight. Notwithstanding European Medicines Agency²⁸ approval under exceptional circumstances for the treatment of patients affected with LHON, Idebenone is not routinely prescribed for use in England and Wales.²⁹

A number of participants are currently enrolled into the LEROS trial which evaluates the efficacy and safety of Idebenone. The possibility of finding a cure is a powerful incentive to participate for those who are desperate to have their sight restored. I interviewed Dr Urquhart, a representative of the pharmaceutical company developing Idebenone. I asked him to explain what Idebenone actually does and, in response, he stated that: ‘*We [pharmaceutical company] are going to argue that it actually can, in some patients, improve sight*’. He went on to explain how this was possible:

‘Idebenone doesn’t correct the genetic problem, but it bypasses the problem. It takes electrons that normally get transferred from complex 1 to complex 5 to make energy. Think of it as a factory: the factory has five steps and you need to go through five steps in order to produce energy. Step one is the one that doesn’t work—imagine all the electrons, all the parts that are supposed to go to step two, falling outside of the machine at step one. Idebenone soaks them up and takes them to step two or three, it is as simple as that. It is molecular in design; its chemical characteristics allow it to soak up the electrons that are escaping from complex 1 and transfer them to complex 2 and 3. Taking Idebenone allows the mitochondria to become functional again, but it is not a steroid, it doesn’t make them work better. We are not claiming that it works to 100 per cent. It just gets things moving again. By doing that, it restores energy to the RGCs [retinal ganglion cells] and, therefore, allows these electrical

²⁸ The EMA produces evaluations of pharmaceutical products which are used by the European Commission to decide whether a medicine can be authorised for marketing in the European Union (EU). Without EMA approval Santhera are not able to market Idebenone until they receive a marketing authorisation from the European Commission.

²⁹ However, Idebenone was approved in May 2017 by the Scottish Medicines Consortium for use in Scotland.

impulses, the light that you see in your eye, to be converted to electricity to be transmitted to the brain.’

(Dr Urquhart, Pharmaceutical Company Representative)

The claims made by Santhera Pharmaceuticals that Idebenone has the potential—in some patients—to improve sight have divided the scientific community (Carelli *et al.* 2011; Lyseng-Williams 2016; Yu-Wai-Man *et al.* 2017).³⁰ In evaluating the veracity of the claims, participants are faced with traversing ‘regimes of truth and regimes of hope’ (Petersen *et al.* 2013: 671). A previous clinical trial evaluating the safety and efficacy of Idebenone, (RHODOS),³¹ failed to show a benefit for its pre-specified primary end point,³² however it did show a benefit for three secondary end points. Dr Urquhart, without prompting, confronted the issue of the criticism levelled at Idebenone from the scientific community:

‘The RHODOS study didn’t meet its primary aim, but it tended to be positive on a secondary main outcome. There were three secondary outcomes, and as you go down the secondary outcomes the P value got stronger and stronger but didn’t quite meet significance until the last one. The shortfall is when the RHODOS study was published the first perception that clinicians had was it [Idebenone] didn’t work. When we looked at the data we felt strongly that the trend was an indication that there was efficacy, and then what we did was we did some additional work to try to prove that.’³³

³⁰ Yu-Wai-Man *et al.* (2017: 42) reviewed of the use of Idebenone in the treatment of the m.11778G>A mutation. Their findings suggest that overall: ‘the current body of evidence does support a visual benefit with Idebenone in acute LHON, but only in a subgroup of treated patients and importantly, it is not possible to predict who will respond. Furthermore, it should be stressed that there is no solid evidence base to guide the optimal dose and duration of treatment, and it remains debatable whether Idebenone has any beneficial effect once optic atrophy has become established in the chronic phase of the disease’.

³¹ RHODOS was a multicentre, 24 week double-blind, randomised placebo-controlled trial that enrolled 85 patients with a confirmed diagnosis of one of three primary mtDNA mutations (Yu-Wai-Man *et al.* 2014).

³² At the end of the 24 week trial, the primary end point (best recovery of visual acuity) in the group taking Idebenone was not statistically significant compared to the placebo group. However, Idebenone displayed an overall consistent trend (compared to the placebo group) with regard to providing protection against future vision loss.

³³ In the follow-up RHODOS trial, the beneficial effect of 24 weeks of treatment with Idebenone persisted even after treatment was discontinued (Klopstock *et al.* 2013). Similarly, in a retrospective study of 103 patients with LHON, 44 patients with sight loss of one year’s duration or less were treated with Idebenone and followed for a period of at least five years. A higher proportion of those patients treated with Idebenone recovered vision. The contributing factors included the early initiation of treatment and prolonged treatment (Carelli *et al.* 2011; Yu-Wai-Man *et al.* 2014).

Using the ‘positive’ secondary outcome data from the RHODOS trial, and its follow-up trial, Dr Urquhart suggested that there is evidence to support the expectation that Idebenone will provide a future benefit for the treatment of LHON. Relevant to the development of both Idebenone and MRTs, there is a small but influential body of literature that has explored the ‘sociology of technological expectations’ (Hedgecoe and Martin 2003; Brown 2005; Novas 2006). Borup *et al.* (2006: 286) suggest that technological expectations, or promises, visions and imaginaries can specifically be described as ‘real-time representations of future technological situations and capabilities’. Petersen (2015: 12) distinguishes the concept of hope from that of expectation, suggesting that:

‘Expectations tend to involve implicit estimations of probabilities [...] based on empirical research (e.g. lab-based experiments, double-blind trials). [...] positive outcomes may provide sufficient empirical evidence to support the expectation of similar outcomes for other endeavours in the future. In this case, ‘expectation’ may equate with *prediction*’ (original emphasis).

Whilst I was in Dr Penvenen’s clinic in the Royal Albion Hospital, I asked whether Idebenone was an effective treatment:

‘My rule of thumb is that 1 in 3 people will benefit from taking Idebenone. I have a lot of difficulties with a lot of the claims that have been put forward which do not reflect my clinical experience. From my perspective some people believe its rubbish and it doesn’t work. My personal feeling is that the drug actually can have a modulatory effect. The bottom line is we need something better really. This is the only thing we have now and even though this is the only thing we have now, we can’t even prescribe it.’

(Dr Penvenen, Clinician Scientist and Neuro-ophthalmologist)

Dr Penvenen suggests that some of the claims being made by Santhera are not consistent with the clinical experience of genetic ophthalmologists. As Idebenone is not routinely available on the NHS, participants who are not enrolled into the LEROS trial and wish to take Idebenone may buy it over the internet from the US. However, it is expensive, with participants explaining that it can cost up to £250 a month. Alternatively, participants can request that their ophthalmologist completes a named-patient request, thus authorising the drug to be prescribed by the NHS. When Marion’s son was diagnosed, she asked the ophthalmologist to prescribe

Idebenone: *'He [ophthalmologist] laughed and said it's no good. You can buy CoQ10 [health supplement] over the counter in health stores, it will probably be as good'*. Like many of the participants, Marion obtained Idebenone over the internet. Dr Urquhart spoke later in the interview of his concerns that people who order Idebenone over the internet may not be receiving Idebenone:

'Imagine you are a patient, you go to your physician and they say yes, I have heard about Idebenone, but I don't think it works. The patient has to go away thinking what do I do now? They go on to Facebook, Twitter, they hear about Idebenone being available in the US, they go to a website and they order it. What they don't know is what they are getting. These are not pharmaceutical grade products—some claim to be, some not. These compounds have not been tested in a formal clinical trial, they are not compounds that go through GMP [Good Manufacturing Practice] which is a global standard for pharmaceutical companies and the Health Authorities. When they approve a drug, they approve a drug based on the data done in clinical trials, toxicology, safety and manufacturing, so we have very strict regulations about quality standards and manufacturing, which these internet suppliers don't have to go through.'

Two of the female participants, Sandra and Beth, also ordered Idebenone over the internet and asked their respective ophthalmologists to test the drug, confirming that they had indeed received a product of pharmaceutical quality. Sandra and Beth have invested both emotionally and financially in the treatment. For Sandra, having hope that her eye sight will improve is what keeps her going:

'Having heard about it [Idebenone], we went and ordered it [from the internet] straight away. I want to cling on to that one little hope that I am investing all this money in Idebenone. I have been taking it for quite a while. I think there has been a marginal improvement in colour perception and although the general fogginess is no better, but black and white seems to be a little better'.

Taking Idebenone gives Sandra agency to take control of her own destiny and imagine a future self—one with improved sight. Beth has been using Idebenone for 11 months. In the previous chapter, Beth stated that she was not coping with her sight loss and, on occasions, spent days in bed. I interviewed her on two occasions. On the second occasion, she indicated that at her last sight test:

'There was a slight improvement and some of the central vision had come back in the right eye and just a slight improvement in the left. Today all these tests they have done show that there has been a bit of an improvement. I had given up all hope'.

The slight improvement in her sight has given Beth some hope for the future, whereas previously, there was simply a sense of hopelessness, as she moved between feelings of hope that her eyesight would be restored, and despair that it may deteriorate further. However, it is increasingly being recognised that hope can be problematic. As Petersen and Wilkinson (2014: 116) suggest, hope can be “misguided” or “false”—based on faulty premises or no evidence’. Marion explained that, when her son David was diagnosed, she researched possible treatments for LHON: *‘I was in the position where I could drop everything and spend thousands of hours on this’*. Marion, persuaded by the ‘hype’ (Brown 2003: 3) surrounding the effectiveness of Idebenone, was encouraged to buy the drug over the internet. Later in her interview, Marion reflected her current view of the drug: *‘We have lost faith in Idebenone. As far as we are concerned, my son took it religiously for three years and then stopped taking it because we believe it made no difference’*. Marion was deeply disappointed that Idebenone did not live up to her expectation that it would improve her son’s eyesight. Notwithstanding Marion’s view of the effectiveness of Idebenone, based on the secondary outcomes of the RHODOS trial, Santhera have been successful in attracting research institutions, funders, clinicians and patient advocacy groups to form alliances or participate in what Novas (2006: 289) refers to as the ‘political economy of hope’. In interview Dr Urquhart explained in more detail how Santhera have sought to raise awareness of LHON amongst health professionals, many of whom (as observed in Chapter Four) do not include the condition in their differential diagnosis:

‘For us, traditionally, as a pharmaceutical company we try to work with organisations and individuals so what we have is an opportunity at international congress to put on symposia. We are, of course publishing our data all the time. We have a highly specialised and very scientific sales force. We talk to a lot of doctors about disease. We also think about online; Medscape is quite popular. We are working with the Royal Colleges in individual countries to see what we can do from an educational point of view. We would love to work with the Royal College of Ophthalmology to put together an educational [service]. We work with a lot of patient advocacy groups because the problem isn’t just on the physician side. It is also making people aware who have Leber’s that they have Leber’s... that they should talk to their doctor about treatment options. We are putting together a list of expert centres, we are putting them on websites, so we can share them with advocacy groups, we can share them with... You know, at the moment, there is no professional

organisation for LHON. There are small groups. What we are trying to do is, in some countries, we are helping to fund expert centres.'

In 2016, Santhera was instrumental in bringing together leading experts from Europe and North America to produce a consensus statement for the clinical and therapeutic management of LHON based on the current evidence available (Carelli *et al.* 2017). Dr Urquhart alluded to the targeting of patient advocacy groups, referring to the pivotal role they also play in raising the profile of LHON and, thereby, Idebenone as a treatment option. Petersen (2015: 10) notes that the:

'Intermingling of the hopes of patients with the promises of new biomedical technologies [...] contribute to the economy of hope for research through lobbying for research into a particular condition and working with policy makers and other influential actors to help bring about some hoped for clinically beneficial outcome.'

I interviewed Adrian, a representative of a patient advocacy charity based in the UK providing information and support to families coming to terms with a diagnosis of LHON. He described how his organisation had previously been actively involved with the European Medicines Agency. He explained that, although sceptical of many of the claims being made in relation to Idebenone, his organisation was now advising the pharmaceutical company which produced the drug. He explained his rationale for collaborating with them:

'I have actually taken the view now that it's more important that I get involved with the pharmaceutical companies than it is with the regulators, because the EMA [European Medicines Agency] approved Idebenone but it is not available on prescription in the UK and it is going to be difficult to see how it will be available in the foreseeable future, notwithstanding the fact it is approved. I can probably be more effective trying to convince the pharmaceutical companies to do their jobs more diligently than I can with the regulators. They are concerned about delays in diagnosis; I think they feel the sooner the diagnoses are made, probably the more effective their treatment will be and, in fact, they were working with us to try and put together some materials that could be used to educate professionals. Now we are starting to get a little bit more involved as a society with them.'

(Adrian, Representative of Patient Organisation.)

Evident from Adrian's account of his organisation's involvement with the pharmaceutical industry is the desire to speed up the process by which Idebenone is routinely prescribed in the UK. The organisation is also fulfilling a political role, as

Adrian explained that his organisation had recently successfully submitted written evidence to the Scottish Medicines Consortium on behalf of the organisation's members.

In the next chapter section, I explore the narratives of hope associated with mitochondrial replacement techniques to avoid the birth of a child with LHON.

Reproductive technologies and reproductive risk

In this section, predominantly drawing on interviews with women, I consider how hope is constructed and negotiated in the context of the development of new developing reproductive technologies to avoid the birth of a child with LHON. I interviewed 11 affected women, and six mothers (carriers). I also interviewed a woman (carrier) who, at the time of interview, was considering her reproductive options. Whilst a number of the women were no longer of child-bearing age, they expressed their concern for other relatives at risk of passing LHON onto their offspring. A few women raised the subject of what they refer to as 'three-parent' babies. In the following discussion, I use the term mitochondrial replacement techniques (MRTs) when discussing the techniques developed to avoid the birth of a child with LHON.

Mitochondrial replacement techniques

A woman at risk of passing LHON onto her offspring is presented with a number of difficult reproductive decisions due to the incomplete penetrance of the condition. Firstly, she may wish to avoid the risk by using a donated oocyte. The donated oocyte is fertilised with the partner's sperm and the resulting embryo is transferred to the uterus of the mother who will give birth to the child. There is also the option of adopting a child. Neither of these options is appropriate for a woman who wishes to give birth to a genetically related child. Prenatal diagnosis for LHON after conception is a possibility where the pathogenic mutation has been identified. However, this technique is not appropriate for women who carry a homoplasmic mtDNA mutation as chronic villus sampling (CVS) or amniocentesis will not assist in assessing the risk of recurrence. Similarly, in heteroplasmic mtDNA mutations using, CVS and amniocentesis to test the foetus will not accurately predict the level of the mutation

in the tissues that are most susceptible (retinal ganglion cells) to LHON (Poulton *et al.* 2017).

A possible solution for a woman who wishes to avoid passing LHON onto her offspring, but also wishes to give birth to a genetically related child, is to use mitochondrial replacement techniques (MRTs). There are two available techniques licensed (in 2015) for use in the UK, the first of which is maternal spindle transfer (MST). This is where the nucleus of the mother's oocyte is transferred into a healthy enucleated donor egg which has healthy mitochondria. The reconstructed oocyte is fertilised and transferred into the mother's uterus. The second technique—pronuclear transfer (PNT)—takes place after fertilisation, following which the embryo is transferred into the mother's uterus (Bredenoord and Braud 2011; HFEA 2011, 2014). The techniques are controversial as they require germline modification, which means that the donor's mitochondria will be passed down through future generations. In 2012, the UK Human Fertilisation and Embryology Authority (HFEA) convened a number of public consultation workshops on mitochondrial donation.³⁴ Oral and written representations were received from a diverse group of stakeholders, including pro-life groups who tapped into analogies and metaphors from popular culture and science fiction (e.g. designer babies, 'the slippery slope', eugenic consumerism, and 'Frankenstein's monster') to argue against the introduction of MRTs. In contrast, mothers who had experience of a child dying as result of mitochondrial disease actively sought to have the new techniques licensed for use (Herbrand and Dimond 2017). The social, legal and ethical repercussions of allowing germline modification have been extensively discussed by scholars (Bredenoord *et al.* 2011; Jones and Holme 2013; Dimond 2015; Wrigley *et al.* 2015; Newson and Wrigley 2017; Scully 2017). Later in the chapter, I consider whether women in this research were influenced by the ethical debates surrounding the use of MRTs when making reproductive decisions.

³⁴ During July to December 2012 the HFEA conducted a comprehensive public dialogue and consultation exercise involving a survey, focus group, workshops, an open consultation questionnaire and open consultation meetings to consider the ethical and social issues surrounding the use of mitochondrial replacement therapies (Department of Health (DOH) 2014).

Reproductive responsibility

The knowledge that LHON is passed on maternally provided certainty for the men in the study. During his interview, Gerry talked about his initial concerns for his daughter and grandchildren; his comments are typical of the other men:

‘When we heard it was a genetic condition, obviously, the first concern was whether there was possibility it could have been passed to my daughter. So it was a relief to find out that I couldn’t pass it on, it was a purely a female mitochondrial condition.’

One of the other participants, Jake, who experienced sight loss when he was 16 years old, was now aged 27, married, and keen to start a family:

‘I have always really wanted children and they told me you can’t pass it on. That was important to me because if I could pass it on, I wouldn’t have children. That was something I needed to make sure of.’

For women who were still of child-bearing age, the knowledge that they would pass LHON to their children led to a dilemma: should they conceive naturally and negotiate the uncertainty of whether their child would develop sight loss—or use MRTs to avoid passing LHON to their offspring? There is a body of sociological literature that has considered reproductive decision-making within the context of genetic inheritance (Kay and Kingston 2002; Etchegary *et al.* 2008; France *et al.* 2011; Boardman 2014; Pond and Dimond 2018). In their research with women carriers of X-linked conditions, Kay and Kingston (2002: 170) identified a number of factors that influence reproductive decision-making, including ‘the actual risk of the condition being inherited [...], perceptions of the consequences of raising an affected child and personal experience of the condition’. Similarly, Pond and Dimond’s (2018) research, with five mothers of children with undiagnosed developmental delay, identified five factors that influence reproductive decision-making: (1) future uncertainty; (2) perceptions of risk; (3) the potential impact a child with LHON would have on their current children; (4) expectations of a family; and (5) the desire for another child. To what extent these factors are evident in the interviews with women in this study will be considered later in the chapter.

One of the affected women, Tina, who experienced sight loss when she was 30 years old, has decided not to have children as she does not want to take the risk that her

child would develop sight loss: *'When you are young, you think "I would like to have kids". On the other hand, I know I can kill the gene by not having them'*. Similarly, one of the other affected young women, Amy, who was diagnosed when she was 18 years old, faces a dilemma as she would like to have children but also wants to eradicate LHON: *'I want to do everything to stop the gene. It's an evil condition'*. These views echo the sentiments expressed in Petersen's (2006: 40) research with people living with diverse genetic conditions. He observed that concern for future generations, and taking responsibility for the health of others, precluded his participants from having children, presenting themselves as 'moral, responsible citizens' and, thus, placing the health and wellbeing of any future children above their own desires to be mothers. Arribas-Ayllon *et al.* (2008) suggest that the rise of genetic testing and genetic counselling has led to an increased awareness of the risks associated with passing on a genetic condition. However, one of the recurring themes in the interviews with the women was the lack of availability of genetic counselling following the LHON diagnosis.

The aim of genetic counselling is to provide up-to-date and accurate information to people who are affected by, or are carriers of, a genetic disease, in respect of their prognosis, recurrence risk for the wider family and reproductive options (Poulton *et al.* 2017). Genetic counselling for women carrying mtDNA mutations is complex and influenced by a number of factors including the specific mutation, homoplasmy, heteroplasmy and disease penetrance (Thorburn and Dahl 2001; Poulton *et al.* 2017). A number of the women explained that they had not been referred to genetic counselling following their child's diagnosis, with the lack of information leading them to research LHON using the internet. However, mirroring research by Whitmarsh *et al.* (2007), they found the information was depressing and alarming. As Sara related: *'I read the first two lines on Wikipedia, I was absolutely horrified, it was talking about the most sensational stories and worst case scenarios, I haven't Googled since'*. Notwithstanding the difficulties experienced by participants in obtaining accurate information, they all understood that, if they conceived naturally, they would pass LHON to their own children. There was, however, confusion surrounding the carrier status of other relatives. This became evident during the interview with Gwyneth (carrier) whose son, Declan, has been diagnosed with

LHON; she understands that her younger son is also at risk. Gwyneth's brother (now deceased) also experienced sight loss and Gwyneth suspects he had LHON, but it was never discussed within the family. Gwyneth also has a sister and a niece; the niece has two sons, both born after Declan's diagnosis. I asked Gwyneth if, given the known family history, her niece had undertaken genetic testing and counselling before she decided to have her children. It was at this point that Gwyneth appeared to become confused³⁵ in respect of her niece's carrier status:

'No, she doesn't need to. She wouldn't have LHON because she is my sister's daughter.

If you and your sister have the same mum, it's likely your sister also has LHON and she has passed it to your niece.

Then it can be passed to her?

You passed LHON to your sons, your sister is likely to have passed it to your niece.

(sigh) It is so complicated.

Was the inheritance of LHON explained to you?

I think so, I had one genetic counselling session, but it is so difficult to keep it all in here (points at her head). My sister and niece have never been offered any blood test or counselling.'

For Gwyneth, there is ambiguity surrounding the status of her sister and niece. She found that understanding complex genetic information following one genetic counselling session was challenging. Her sentiments resonated with Catrin (carrier), whose son was diagnosed with LHON; she also has a younger son who, again, she knows it is at risk. Catrin has a sister, and a niece. Catrin realises that her niece may pass LHON to her children, but explained that the family find it difficult to comprehend which family members may go on to experience sight loss:

'You know, for the lay person, all the genetics background is quite difficult to understand sometimes, you know when they go on about chromosomes. It is quite difficult. From my understanding, my niece, she is 20 at the moment, may be eligible for the three-way children.

You mentioned the three-parent baby?

³⁵ During the interview with Gwyneth and a few of the other women, I found myself in the unusual position of having to explain the inheritance of LHON. This was an example of one of the 'ethics in practice' situations that I referred to in Chapter Three.

Yes, it is taking an egg from a woman then replacing the bad cells with the good cells or something like that.

Would you have considered using it?

Possibly, yes.

Would you have had children if you had known about LHON?

If I am being honest, no, I wouldn't. I wouldn't have had them because seeing what [son] is going through, that really upsets me (becoming emotional).'

Catrin is aware of MRT potentially being available for her niece; she also indicates that she may have used MRT had the techniques been available when she decided to have her children. One of the other issues present in the interviews was the most appropriate time to provide genetic counselling. Amy remembers being in the hospital and a genetic counsellor coming to see her to talk about LHON: *'I was offered genetic counselling, but I didn't accept it at the time. I wasn't ready to talk about it'*. At the time of her sight loss, Amy was not in a relationship and, so, felt it was pointless to have genetic counselling at that stage. Ginny recounts a similar experience when she and her boyfriend were offered genetic counselling:

'We had genetic counselling when we were 18 which was very.... I don't think we were even thinking about getting married, so it seemed a bit of a ridiculous conversation. We hadn't really found out that much about each other.'

Echoing previous research (Parsons and Atkinson 1992; Cox and McKellin 1999; Petersen 2006), participants made the effort to become better informed in respect of the inheritance of LHON when they or their daughters reached a 'critical juncture' (Cox and McKellin 1999: 628) in life, for example, when they met a potential partner or were considering having children. Amy is now in a serious relationship:

'I have recently asked for genetic counselling; it is because of my partner. We are going to have children. I have asked for it now. But it needs to be talked about. It is very complicated, and I don't want my children growing up with the same condition as me. I know that sounds terrible, but hopefully they can do something to not let it happen to them.'

Amy faces a dilemma: she wants to eradicate LHON, but also would like to give birth to a child that is genetically related to her. She has recently asked to be referred to a genetic counsellor to find out her reproductive options. There was considerable UK

media coverage of the mitochondrial debate in the lead up to the Parliamentary vote in 2015, though Amy claimed not to be aware of the techniques. Similarly, other women also stated that they had not heard of MRTs. Consistent with Herbrand and Dimond's (2017) research, this may be attributed to the fact that a number of women were no longer of child-bearing age, not intending to have further children, or did not want to have children. Of the remaining women only four (Marion, Naomi, Ginny and Sally) felt confident in discussing MRTs in any detail. Drawing on the interviews with these four women, I explore how reproductive responsibility is negotiated within families at risk of passing a genetic condition to their offspring. Focusing on reproductive decision-making, I discuss whether the four women are influenced by the ongoing ethical debates surrounding the use of MRTs and whether in imagining their child's future (France *et al.* 2011) their experiential knowledge of either living with sight loss, or living with an affected relative, influences their decision to conceive naturally or use the techniques.

Firstly, I talk about Marion, now in her mid-fifties, and who was unaware of her carrier status until her son David was diagnosed. In her interview, she spoke of her concern for her daughter, Naomi (carrier), who is in a long-term relationship, and although she has often spoken of having children, has yet to do so. Secondly, I discuss Naomi, who is well informed in respect of the ethical debates surrounding the use of MRTs. Naomi, together with other family members, attended a genetic counselling appointment after her brother was diagnosed. At the time of interview, Naomi and her partner had asked to be referred to a genetic counsellor to discuss their reproductive options. I then consider Ginny who experienced sight loss when she was 18. Knowing that she would pass LHON to her children, Ginny and her husband decided to conceive naturally. Finally, I discuss Sally, who experienced sight loss when she was seven. Sally was initially misdiagnosed and only became aware of LHON after her youngest son was affected when he was also seven.

Marion

I begin the discussion with Marion who, in addition to David and Naomi, has two other children. During my interview, she raised the subject of Naomi not having had children yet, which had clearly been troubling her for some time. Marion explained

that Naomi had been in a stable relationship for some years and worries that her carrier status is deterring her from having children. She explained:

'My daughter spoke to her partner and he said it [LHON] didn't matter. Now we are three years down the line and she still hasn't had children and I know it's something she worries about. I said to her before "why don't you just go ahead and have children? The only difference between you and me is that you know about it and I didn't know about it".

Would knowing about LHON have made a difference to you?

(long pause) That's an interesting question. I... (sigh) I feel desperately sorry for my daughter because I know it makes a difference to her. As to whether it would have made a difference to me, I would like to say no but I don't know. I certainly wouldn't wish any of them [children] away.

Do you think the diagnosis is stopping your daughter having children?

(sigh) I don't know. I keep saying to her [daughter] other people are having children, they don't know what the odds are for them, the child could be born with hereditary cancer or hereditary you name it... just because we happen to know what the odds are for us.'

Before her son received his formal diagnosis, the family had been given an indication that it could be LHON. Marion had researched the condition on the internet and realised that the condition was maternally inherited. She explained that Naomi attended the appointment with the genetic neuro-ophthalmologist to receive confirmation of her son's blood test. The family also attended a genetic counselling appointment. Given that Naomi has not yet started a family, she reflected on whether it was the right decision to take her daughter to the appointments:

'I know she was going to have to hear it sometime. How can you sweeten the pill? Should I have shielded her at the time do you think? It's a complex one. She has been with her partner for a long time. Bearing in mind she is [age] now, I know she has thought about it [having children] and we have talked about it. Knowing her as I do, I feel that it's stopping her from getting on and she has talked about adopting a child.'

Marion spoke of the turmoil she experienced at the time of her son's diagnosis. Mirroring Hallowell and colleagues' (2003) research, Marion wanted to protect her daughter from unnecessary distress, but by the same token, she believes that her daughter had the right to know the inheritance of LHON to make an informed reproductive choice. Six years after her son's diagnosis, echoing previous research (Arribas-Ayllon *et al.* 2008; Chapple *et al.* 1995), Marion, still experiences feelings of

guilt and self-blame that she had passed LHON to him. She also feels guilty that her daughter has not yet had children, and she believes it is because of her carrier status. Later in the interview, Marion acknowledged that her daughter would have discovered that she has inherited LHON and would pass it to her children. However, she still experiences conflicting emotions as to whether it was the right decision to take her to the appointments:

‘I am asking for the impossible, aren’t I? Because she is an intelligent woman, she would have realised she will pass it [LHON] to her children. We are very interested in the three-person IVF. We went to all the consultation workshops which were very interesting, especially hearing the religious side of it. I thought “where is your human compassion in all this?” On one platform, you have a woman talking about her child dying in front of her and a slight alteration in her mitochondria making a change to this. The attitude [of others] is unbelievable.’

Hallowell *et al.* (2003: 77) suggest women, in delivering bad news, to family members draw on different rhetorical strategies to balance responsibility and autonomy. One strategy is to draw on ‘discourses of hope’ associated with new technology breakthroughs such as MRTs that promise to eradicate disease. Marion who continues to feel guilty that she has passed LHON on to Naomi encouraged Naomi to attend the HFEA public consultations workshops, in the hope that she would become better informed about the possibility of the use of MRTs. Towards the end of the interview, Marion again returned to the subject of Naomi not having children and suggests that it may have been better for Naomi not to know of her carrier status and to have been in the same position as she was when she had her children:

‘I keep saying to my husband I wish she didn’t know. I wish she didn’t know that she is a carrier for LHON and that she will definitely pass it on to all of her children. I just wish she was in the same situation that I was when I had my four children in that we didn’t know anything about it.’

Given Marion’s comments, I asked her if she would have preferred her daughter to have had children before her son David’s diagnosis:

‘Yes, that would be it.. we all had genetic counselling following my son’s diagnosis. My daughter was told by the genetic counsellor that she could be eligible for sex selection. What is the point of that? So, you can have a

girl who will pass it on to all subsequent generations or you have a boy who is more likely to get it, so it's Hobson's choice really.'

Evident from Marion's interview is that she continues to try and reconcile her feelings of guilt that she has passed LHON to her children—even though she was not aware of her carrier status when she conceived her children. She also continues to be conflicted as to whether it was the right decision to take Naomi to the appointments.

Naomi

I later spoke to Naomi who, together with her family, attended an appointment with a genetic counsellor where her reproductive options were explained to her:

'The genetic counsellor said I would pass it [LHON] on but there are some options. Some people might go for sex selection to have a female [child] with less chance. She mentioned at that point about the three-person IVF. I have kept an eye on what's been going on in the media. Also thinking about children, I am going to have to think about it. I am not getting any younger and I do want to have children. It is a bit of a dark cloud I feel like I have to think about but don't really want to. My partner says why not just get IVF? It's not as simple as that. It can be painful and traumatic on couples if it doesn't work. I am not sure that I want it, even if it is available right now, I don't know if I would want to be a guinea pig. I think that having children is a bit of a lottery anyway.'

At the time of the interview, Naomi, was waiting for an appointment with a genetic counsellor to discuss the possibility of being eligible for MRTs. She has spent considerable time reading the scientific literature following the media debates at the time of the Parliamentary vote to legalise MRTs in the UK. Naomi suggests that having children is a lottery and I asked her, given this view, if she would consider having a child naturally:

'That's kind of what I am thinking, but I don't know. It is something my partner and I are not talking about very often. But are thinking about it and trying to figure it out. I think going to genetic counselling will give us something to chew on. So we can weigh it up. I am not a fan of the slippery slope argument. As humans, we are sensible enough to not go down the slippery slope. I feel quite angry that people's religious views play a bigger part than they should in issues like this. I think for me when I was reading about it at the time, there are lots of mito [mitochondrial] conditions that are far worse, for want of a better word, where women just can't have babies or they do, and the baby dies before it is a year old.'

If that is what I had then it would be a much simpler choice to have the three-person IVF. I think the media are making the mistake that it's three-parent IVF that annoys me. It's clumsy and it's stupid and I think it is nobody's business stopping people's access to these things.'

Whilst Naomi refers to the ethical debates surrounding the use of MRTs, she was firm in her view that she was not influenced by such rhetoric. Naomi suggested that she and her partner have a tendency to avoid making decisions. Pond and Dimond (2018) suggest that, for some couples, they never reach a consensus, or if they do, this may change over time. The authors suggest that the decision to have a child is influenced by perceptions of risk and personal experience of the genetic condition. As the interview progressed, it became apparent that Naomi's decision to conceive naturally was being influenced not by the ethical debates, but instead based on how well she perceived her brother had coped with his sight loss. I asked her if this was the case:

'There is an element of that. My brother has dealt with it very differently to the way I would deal with it. I would hate that if I couldn't see. I think having a child and the child knowing that we had chosen to have a baby knowing that they may go blind, I wonder what the child would think of that.'

Naomi reflects on how her child would react to losing their sight knowing that their mother was aware that she would pass LHON to them. In the case of Adam, one of the participants, discussed in Chapter Five, his parents were aware of his mother's carrier status and chose to have a child naturally. Adam lost his sight when he was 14 years old, and now 26 years old he and remains extremely angry that his parents chose to have him:

'Since what's happened to me, I am a big believer now that if you have got a genetic condition and there is no way you can pass it on, fine, have a child. If you are going to pass it on, don't do it, there is no point. I still get moments now when I am thinking I am blind, and I shouldn't be because that's what I went through: "I shouldn't be like this".'

Adam's relationship with his parents remains uncomfortable; he is unable to forgive them since he believes that they were selfish and had a child despite knowing that he would lose his sight.

Ginny

Ginny experienced sight loss when she was 18 years old. Knowing that her children may also lose their eye sight, she decided to conceive naturally. Ginny's decision is influenced in part by her brother, who is now in his early thirties and has not experienced problems with his sight. Ginny has two sons, and at the time of interview, was considering having another child. Although MRTs were not available when Ginny decided to have her first two children, there were other options (adoption and oocyte donation) which Ginny and her husband chose not to use. I asked Ginny if she had genetic counselling before having her children:

'We had genetic counselling—they gave us the 50% chance that a boy would be affected and 10% that a girl would be affected. You just think it's 10% and I have still done it [lived life], so anything is possible. We were of the opinion, yes, it could happen, but it's not the end of the world. We feel very much that there is a genetic load that you bring into this world that gets passed down from generations of your family. There are plenty of other things in our family, like cancer and heart disease, that I would want them to have less [than LHON] in some respects.'

Ginny expressed thoughts that were present in a number of the other participant interviews that LHON is not a life-threatening condition, which is consistent with Petersen's research (2006). Ginny plays down the significance of LHON on her life, indicating that she has still managed to live her life the way she wants to. At the time of interview, Ginny was in the process of returning to further education, having given up her studies because of a lack of support from her teachers when she was first diagnosed. Ginny has recently celebrated her 15-year anniversary of sight loss:

'We had a 'balls to blindness' dinner with all my family. We had meat balls, then we had melon balls and grapes, and then profiteroles in the shape of eye balls. It was an "up yours", you know "look at me now, you might have thought you beat me 15 years ago". I would love to go back and see the teacher who said I couldn't do it and say "remember me?"'

Ginny has taken what was evidently a traumatic experience in her life and reframed it in a positive light. I asked Ginny if, when she made the decision to conceive naturally, she had discussed her options with other family members.

'No. It was our decision to make; she [mother-in-law] didn't like it though. I have as little to do with her as possible (laughing). We don't get on, shall we say. She has lots of opinions about me.'

Ginny believes that her mother-in-law would have preferred her son to be married to someone who did not have sight loss. She also explained that her mother-in-law is aware that Ginny has transmitted LHON to her children and was disapproving of her decision to conceive naturally, consistent with Parsons and Atkinson's (1992) insight that women at risk of transmitting Duchenne Muscular Dystrophy faced hostility from their future mothers-in-law who considered it inappropriate to take the risk of transmitting the condition to offspring. I asked Ginny, given her mother-in-law's concerns, if she had considered any other reproductive options:

'It didn't even cross my mind at that point. If my Mum could have had that information that long ago and decided to have me, and not my brother, and then I would still have a problem. There is no guarantee if I had girls, they would pass it on.'

The point Ginny makes is that there is no guarantee that a female child will not experience sight loss or that a male will become affected. Although the scientific literature suggests that affected females are more likely to give birth to children who will also be affected (Van Senus 1963; Harding *et al.* 1995; Newman and Biousse 2004). Ginny reiterated that she has coped very well with her sight loss: '*It doesn't affect my daily life, I know my way around and I also have somebody to help me*'. I asked Ginny how she would feel if her sons developed sight loss:

'I would try to do my best to make sure they have positive role models around them and hope that I am one of their positive role models, that Mummy did it, she still does everything else, and go from there'.

As the interview progressed, the subject of Ginny having another child was raised. Given that MRTs are now licensed for use in the UK, I asked her if she would consider using the techniques:

'It's a personal preference thing. If people want to do that and make sure their children are without this, that's their opinion, their choice. To me, where do you draw the line? What else do you take away, where are we going with this? It will be almost like a form, there will be a tick box: you can get rid of Leber's and maybe diabetes and cancer, what would you like us to get rid of in your child? That's not nature because that's not the way it works. We are going to end up with people who are never poorly or then maybe they will live for an awfully long time. To me, where does the line stop?'

Ginny did not indicate that her decision to conceive naturally was being made on religious grounds. However, her decision is influenced by the ethical arguments surrounding the use of MRTs. Ginny refers to having a tick box where genetic conditions can be removed, or characteristics altered. One of the enduring arguments associated with the use of MRTs is that they will result in a 'slippery slope' to the modification of the nuclear DNA and the birth of 'designer babies' (NCOB 2012: 52). Ginny also refers to what Buchanan (1996: 28) describes as the 'expressivist objection', which suggests that using reproductive technologies to 'correct, ameliorate, or prevent genetic defects expresses (and presupposes) negative, extremely damaging judgments about the value of disabled persons'. Whilst Ginny is influenced by this argument, it is also evident that her view of reproductive technologies is grounded in her own 'embedded experiential knowledge' (Abel and Browner 1998, cited in Boardman 2014: 137) of living with sight loss. Boardman (2014) argues that experiential knowledge in the context of reproductive decision-making is increasingly being recognised as an important source of knowledge utilised by prospective parents when evaluating reproductive risks and exercising informed choice. Whilst Ginny uses her own experience to inform her thinking, Naomi uses her 'empathetic knowledge' (Abel and Browner 1998, cited in Boardman 2014: 137) of the way her brother has coped with his sight loss. For Ginny and Naomi, their experiential knowledge is highly influential in their decision-making around reproductive options. Ginny, later in the interview, talked of her experience of joining a Facebook support group for LHON and how she had frequently contributed to the threads. However, following a number of adverse comments, prompted by her decision to conceive naturally, she now describes herself as an observer. I asked Ginny to expand on the comments that had been posted:

"That they would never give this to their child. It is the worst thing in their life. They would never accept it, would never move on and they feel guilty now because they have got children and if they knew about the condition, they would not have had them. I have now become this quiet observer, I don't often offer up my opinion. In no way do I think they are wrong, but it's not quite the way I see the world.'

Sally

One of the other affected mothers, Sally, experienced sight loss when she was seven years old and did not receive her LHON diagnosis until after she had her two sons, both of whom have experienced sight loss. When talking about her decision to have children, Sally was very emotional that she was deprived of the opportunity to make an informed choice. I asked her whether, if she had been aware of LHON, she would still have conceived naturally:

‘Yes (long pause), I don’t know. It’s really hard because they are here. You are talking about two individuals. You are talking about the possibility of having a child with a condition. I feel quite resentful about it all really, the lack of being told anything.’

As the interview continued, it became apparent that Sally’s embodied knowledge of growing up with sight loss has been influential in the way she has lived her life. Sally explained that she attended a special school:

‘All my mates went to the school down the road. I was different; I went to this special school. I refused to go on the school bus because it was what everybody called the brown bus, the special bus. It’s horrific really but you know it was the special needs bus. People had tokens for bus fare. You were just made to be very, very different.’

Sally suggests that if she had known of her diagnosis, she would have been in the position to imagine what kind of future her two sons would have. Given her own experience of growing up with sight loss, she may have decided not to have children. She feels angry and frustrated that the choice whether or not to have children was taken away from her.

The affected women who were also mothers expressed strong views that they wanted their daughters to receive genetic counselling before they themselves decided to have children. Downing (2005) suggests that people who share a genetic risk with other relatives feel an obligation to help them in making reproductive choices by encouraging them to participate in genetic testing. Moira, who has MS-like symptoms and LHON, and experienced sight loss when she was 26 years old, is typical of these participants. She has four daughters (one of whom has also lost her sight) and 10 grandchildren. In her interview, Moira expressed her regret that, knowing that she had lost her sight, her daughters did not have genetic counselling:

'None of them had any counselling. They all knew [about LHON] and they said to me "Mum, we made our decision, it was our choice to have our children". Now with the three-parent baby... it's too late for them because they have children... but their children, if they want to get rid of LHON, they can.'

Moira believes that her daughters chose not to undertake genetic testing because their perception is that she has coped with her sight loss. However, it seems that Moira would dispute this view, and refers to her grandchildren having the opportunity to use MRTs to avoid passing on LHON. One of the other affected women, Mary, recounted a similar experience with her children who were also reluctant to consider genetic testing and counselling. Mary and her sister, Sian, also have MS-like symptoms associated with the LHON m.11778G>A mutation. For this family, there is heightened uncertainty as to whether it is MS or LHON that is responsible for their sight loss. Mary developed sight loss in the early 1990s after the birth of her youngest daughter and, because of her symptoms (including lack of sensation in her fingers and numbness in her feet), she was given a diagnosis of MS. Mary also experienced seizures for which she was given medication. However, Mary's eyesight continued to deteriorate and, in 2010, she was diagnosed with LHON. Sian, following the sudden onset of pins and needles and numbness in her feet, was diagnosed with MS after an MRI scan and lumbar puncture. The two sisters are concerned that their children and grandchild may also lose their sight. Mary has two daughters, who thus far have refused to participate in genetic testing and counselling. Mary's youngest daughter is now thinking of having children:

'I think I might push it now. Get the two of them to go and have it [genetic testing]. To actually sit down with a geneticist and to go through it with them. It's not sinking in with them. I can tell my children until I am blue in the face. But for their peace of mind, it would be nice to know. They might get problems in the future.'

Why don't they want to be tested?

I think it is because we said they have got it. We have said this gene is in your body as well. Gran has passed it down to me and I have passed it down to you. They didn't go [for blood test] because they don't like needles (laughs). My older daughter has got two boys, one is six and the other one is 18 months.

Your daughter knew she was a carrier when she had the youngest child?

Yes, but then again, I don't think anything would have stopped her. If she wanted to have a kid, she would have had one. We said "you could end up like me or it could bypass you lot altogether. You could be like Gran because my mother has got nothing wrong with her apart from old age".

Moira, Mary and Sian's experience of their children refusing to participate in genetic testing is mirrored in Dimond's (2013) research with adult patients living with mitochondrial disease. Dimond claims that the participants attributed their children's reluctance to be tested to not wanting to assume an 'illness identity'. Instead, they made the decision to 'get on with life' (2013: 6). Moira also has a diagnosis of LHON and MS-like symptoms; during her interview she expressed her confusion as to whether she has MS with sight problems or LHON with MS-like symptoms. Moira when attending appointments, makes a point of asking for clarification of her symptoms:

'I always ask [clinicians] "have I got LHON with MS symptoms, or have I got MS?" They [clinicians] say I will never have full blown MS. My daughter is crippled with it. What is that? Definitely MS? And yet they say MS isn't hereditary. I don't understand it. MS is not hereditary but LHON is, so what is the connection between the two?'

Moira expresses her frustration that she cannot be given a definitive answer to her questions. Stivers and Timmermans (2016: 202) note that genetic testing does not 'always produce causal variants but often reveals ambiguous findings that have the potential to exacerbate rather than ease the patient's diagnostic uncertainty'. Mary and Sian had been referred from their local hospitals to attend an appointment with Dr Morgan in St Tristan's Hospital. I spoke with the sisters prior to the appointment, and they explained that they were hoping Dr Morgan would be able to provide some certainty in respect of the prognosis of their condition. During the appointment, Mary raised her concerns that her daughters and grandchildren may also lose their sight. Dr Morgan explained the uncertainty for patients diagnosed with LHON and who also experience the MS-like symptoms:

Dr: There is not much you can do other than reassure them [daughters] and hope for the best. But you know they could all be lucky. They, in fact, have got to a stage where they are in their mid-20s to early 30s, they seem okay. So maybe things will be okay.

Mary: But the thing is we are looking at my mother, she is 83. There is nothing wrong with her.

Dr: This is where the uncertainty comes in. They all carry the mutation in the mitochondrial DNA, but only some people develop the condition. That's partly bad luck. You have been unlucky, both of you. In some respects, you are more closely related to each other than say your grandchildren are to you, obviously you are sisters. There is something that has clearly triggered the MS-like illness in yourselves that may not carry through to the other people in your family.

Mary: It [sight loss] could have been something specific just to us with the MS?

Dr: If you look at the MS and the causation of MS, of course we don't know... but there could be environmental triggers, there could be things in your childhood, there could be... something that may have affected you together. It could be lots of things; no one has actually found a trigger. It could be even past infections. The one certainty is you have the visual problem and obviously you also have the other MS symptoms. It's a really difficult conundrum. Obviously, something in females, whether it is hormones, whether there are other aetiological factors which might cause it and predispose women to be more likely to get it. But again you are dealing with risks, you are not dealing with certainties.'

Mary and Sian have a degree of certainty as they know they have the m.11778G>A mutation and they also know that they experience MS-like symptoms. However, the cause of their symptoms remains unidentified which leaves them living with uncertainty. After their appointment with Dr Morgan I interviewed the sisters, who expressed their disappointment that they had not received the answers they were looking for. Sian captured the thoughts of the sisters:

'Whenever I go anywhere, its well it's probably to do with the MS. Nobody seems to want to commit themselves. Like this morning the consultant hasn't told me anything that I didn't already know. I saw the pictures [OCT and retinal imaging] which were nice because I haven't seen any pictures before.'

Stivers and Timmermans (2016: 201) suggest that diagnostic uncertainty 'occurs when available information is limited or characterised by probability, ambiguity, or complexity and may lead to confusion, anxiety, and indecision'. Dr Morgan attempts to provide some certainty for the sisters, by suggesting that they may have experienced an event in childhood, which is unlikely to be replicated for their children and grandchildren. Dr Morgan aware that the two sisters were disappointed, that they had not been given more certainty in respect of whether

their children and grandchild would be affected suggested to me: *'uncertainty is not a concept that patients very easily accept. The fact is people expect there to be certainty, they expect there to be certainty around treatments, and what is going to happen in future, and that isn't always so.* During the appointment, the sisters continue to experience anxiety because they do not know whether their extra-ocular symptoms will deteriorate over the course of their lives. However, consistent with the findings of Latimer *et al.*'s (2006) research in the dysmorphology clinic, Dr Morgan asked the sisters if they would like to continue to attend the genetic ophthalmology clinic, so that their condition would continue to be monitored.

Summary

I began this chapter by locating participants' claims within the sociology of hope. Participants spoke passionately of their hope that new treatments would be available to restore their sight or avoid the birth of children with LHON. A number of participants have invested emotionally and, in some cases, financially in the potential for one treatment—Idebenone—to improve their sight. It is evident that the pharmaceutical company developing Idebenone have participated in the 'political economy of hope' (Novas 2006: 289) and strategically brought together key actors (including research funders, clinicians and patient advocacy groups) to raise awareness of LHON and in so doing to accomplish their agenda of recouping their investment in the development of the drug by having it approved for use by the NHS in the UK. I discussed how the 'hype' (Brown 2003: 3) surrounding the claim that Idebenone has the potential to restore sight has divided the scientific community (Carelli *et al.* 2011; Lyseng-Williams 2016; Yu-Wai-Man *et al.* 2017). Similarly, participants were also divided on the effectiveness of the drug. Beth and Sandra felt it had helped improve their eye sight. In contrast, Marion spoke of her disappointment that her son had used Idebenone for several years without an improvement in his visual acuity.

In the second part of the chapter, drawing predominantly upon the experiences of four women (Marion, Naomi, Ginny and Sally), I discussed narratives of hope surrounding the introduction of mitochondrial replacement techniques (MRTs) to avoid the birth of a child with LHON. I explored to what extent the women were influenced in their reproductive decisions by the ethical arguments that the

techniques would create a 'slippery slope' to 'designer babies' (NCON 2012: 52). Naomi, who is a carrier, supported the use of MRTs. In contrast, Ginny, who was affected when she was 18 years old, raised her concerns that in choosing to use MRTs, this devalues the lives of those who live with a disability. Sally suggests that she may have considered the use of MRTs had the techniques been available when she conceived her two sons. I would suggest that these women were more influenced by their experiential knowledge of either being affected (Ginny and Sally) or, in the case of Naomi, living with a relative who has been affected. Sally describes her experience of growing up with sight loss as traumatic. In contrast Ginny views her sight loss in a positive light, claiming that it has not stopped her from doing the things that she wants to do. Similarly, Naomi considers that her brother has coped extremely well with sight loss.

I also considered the extent to which women affected by LHON were keen to persuade their children, particularly their daughters, to undertake genetic testing and genetic counselling before having children of their own. Consistent with Dimond's (2013) research, there was a great deal of reluctance by women who are carriers of LHON to be tested, which in part, is attributable to adopting a philosophical attitude that if their children are affected, they will deal with it. It was evident that the children of affected women, using their experiential knowledge, decided that their mothers had coped well with sight loss and so they did not perceive having children who may develop sight loss as an issue.

In Chapter Eight, I provide an overview of the thesis and bring together the themes that have been identified in the empirical chapters. I will also consider how the experience of living with sudden sight loss can contribute to wider debates around living with chronic illness.

Chapter Eight: Discussion

In the preceding chapters, I identified Leber hereditary optic neuropathy (LHON) as an under-researched mitochondrial condition, suggesting that people living with LHON constitute 'missing voices' (Lawton 2003: 36) within the sociological scholarship. To address the gap in the literature, I have drawn heavily on interview data and some observations to explicate the lived experiences of men and women affected by LHON, their mothers who carry the LHON mutation, and the health care professionals who diagnose, treat and seek to find a cure for the condition. I have discussed, within the four empirical chapters, participants' chronic illness trajectory (Glaser and Strauss 1964), looking at their past, their present and their imagined futures, with trajectory seen in terms of Conrad's (1987: 10) comments that trajectory 'encompasses process and change but does not assume linearity or orderliness'. On the contrary, mirroring Yoshida's (1993: 222) research with people who experienced traumatic spinal cord injury, participants in this thesis, over a period of years, move back and forth between experiencing 'non-disabled and disabled aspects of self'.

This chapter will begin by unpacking three of the major themes emerging from the research: (1) professional knowledge, uncertainty and contestation: (2) normalcy, stigma and the everyday experience of illness: (3) imagined futures. These themes evolved from exploring key sociological and disability studies scholarship, and the formulation of an over-arching research question outlined in Chapter One:

What are the subjective experiences and perceptions of people affected by LHON, and how are relationships within the family renegotiated following the diagnosis?

Moreover, the themes were informed by four subsidiary research questions:

1. What are the challenges presented to genetic ophthalmologists in the diagnosis and treatment of LHON?
2. To what extent does LHON disrupt an individual's biography and what are the implications for their identity and sense of self?
3. How do people with LHON imagine their future?

4. Are women influenced by their experiential knowledge of LHON when making reproductive decisions?

In discussing the three themes, I suggest how the experiences of participants both coalesce and on occasions diverge from classic and contemporary social scientific accounts of chronic illness and disability. From here I am able to justify why I have both adopted and extended the work of leading scholars, including Bury (1982) and Charmaz (1983, 1991). In the final part of this chapter, I will ponder what the future holds for people with LHON. Specifically, I consider the challenges and hardships likely to be encountered by visually impaired people (and disabled people more generally) going forward, including navigating the context of austerity politics which has provoked drastic cuts to the Health and Social Care budget,

Professional knowledge, uncertainty and contestation

Drawing on interview data and several observations, I capture how LHON is characterised by uncertainty and contestation surrounding the diagnosis and prognosis of the condition. Consistent with other rare conditions (Rare Disease UK 2016), participants often experienced a protracted and contested quest to find a diagnosis. In Chapter Four, I suggested that diagnosis is an interpretive process of identifying and classifying disease which takes place within the doctor/patient consultation—in which the doctor calls upon an agreed stock of knowledge to label a condition and provide treatment (Jutel and Dew 2014). However, I also argued that diagnosing LHON is not confined to such an interaction. Indeed, I suggest that the identification and categorisation of LHON parallels previous research in the dysmorphology clinic, for example where accomplishing a genetic diagnosis is located at the intersection of the clinic and the laboratory (Featherstone *et al.* 2005; Latimer *et al.* 2006; Latimer 2013). The clinician's expert gaze is utilised to identify the characteristic symptoms of the condition, by calling upon not only repertoires of evidence (slit-lamp examination, OCT, MRI) but also their clinical judgment. The use of innovative molecular genetic technologies, (including next-generation sequencing) provides confirmation of the LHON mutation. However, it is the clinician's expertise that remains central to the diagnostic process. I have highlighted how LHON is an elusive condition and symptoms are, occasionally

misinterpreted and—it is in these moments —where clinicians cannot ‘transform patient problems into solvable problems’ (Berg 1992: 172) that people with LHON are reconfigured as ‘bad’ patients (Jeffrey 1979). Care is downgraded with patients categorised as time wasters, who are denied access to medical treatment and ‘disposed’ of by being discharged from the clinic (Latimer 1997, 1999). The repercussions of not receiving a timely diagnosis are significant as participants are denied access to additional resources to support their return to employment and education, obtain welfare benefits and secure referrals to specialist services (including genetic counselling).

When I interviewed people with LHON, I did not anticipate that the process of diagnosis would dominate their narratives. In unpacking their accounts, I identified three areas of concern in the diagnosis and treatment of LHON. Firstly, LHON is rarely observed in clinical practice, and therefore specialist registrars (and sometimes consultants) do not include it in the differential diagnosis. When women, young children and older people present in the eye casualty clinic with bi-lateral sight loss, ophthalmologists discount LHON as a possibility as they believe the condition is the preserve of men in their early teens and twenties. I subsequently argue that the ophthalmic training curriculum is inadequate in training ophthalmologists to recognise rare inherited conditions in clinical practice.

Secondly, it is suggested that in the UK doctors are trained to use diagnostic tests in a linear way; only after a test has proved negative is the next test ordered. This is problematic for LHON, as the clinical evidence (see Chapter Four) suggests that new treatments such as Idebenone and gene therapy are likely to be most effective when administered at the onset of LHON. Therefore, speed of diagnosis is of the essence.

Thirdly, delays in diagnosis occur when blood samples are sent to the gene panel for analysis. The clinicians interviewed in my thesis all agreed that the lengthy delays in obtaining test results were in part due to the cuts to the NHS budget. In an age of austerity, the NHS is challenged by increased patient demand for healthcare with finite resources. A clinician in this study, Dr Penvenen suggested that budgetary controls within the NHS have resulted in genetic testing taking up to four months in some parts of the UK. Newdick (2005: 1) argues that the demand for health care continues to exceed the supply of resources made available by the NHS, with the

spending on healthcare ‘outpacing economic growth [...] forcing governments to find new funds or pass a larger share of costs onto individuals’. The age of austerity has thereby restricted the availability of specialist services including genetic counselling. A number of participants in this research explained that they did not receive a referral, experienced inordinate delays before receiving their appointment, or were only offered one genetic counselling appointment. As a consequence, some of them paid privately to see a genetic counsellor. Participants such as Beth who are well educated and, so equipped to negotiate the healthcare system, hold the appropriate capital for both articulating their views in interactions with doctors and paying privately for treatment. Not all people with LHON, however, will be so fortunate.

Normalcy, stigma and the everyday experience of illness

Throughout this thesis I borrow Bury’s (1982) concept of biographical disruption to explore the consequences of sight loss. Irrespective of age or gender, every person with LHON who participated in this study described experiencing disruption in the weeks and months following their diagnosis. Whilst acknowledging that Bury’s (1982, 1991) framework has been the subject of continuous debate (Lawton 2003; Pierret 2003; Williams 2000), I suggest that it has stood the test of time, especially in terms of making sense of the participants’ claims in this research. A number of participants expressed their frustration and at times irritation that they were no longer able to perform daily activities including ‘routine body maintenance’ (Barnes and Mercer 2010: 52), and required assistance from their family to accomplish these taken-for-granted tasks. It is in these moments that family relationships are re-negotiated, a key focus for the likes of Bury (1982) and Charmaz (1983). Whilst I have adopted Bury’s (1982) concept, I have also extended its application. Bury’s participants were relatively young when they experienced rheumatoid arthritis. Answering Williams’ (2000: 61) call for the extension of the ‘biographical focus’ of studies in the sociology of chronic illness ‘*to both ends of the life course*’ (original emphasis), I interviewed individuals, who were children, and individuals who were in their sixties, when they experienced sight loss.

Whilst Bury outlines how his young participants experienced biographical disruption at the *onset* of their illness. I argue that Bury’s concept is pertinent in

understanding how people with LHON negotiate the longer-term consequences of illness.

In locating this research within the wider scholarship, I acknowledge that different chronic conditions give rise to a range of varying experiences. However, as Kelly (1992: 48) puts it, ‘in many ways the problems posed by such illnesses share a quality which cuts across particular medical diagnoses and is common to them all’. In reviewing the classic chronic illness scholarship I have highlighted two points of departure. Firstly, there is an assumption that chronic illness is to be expected in older age (Pound 1998; Williams 2000; Sanders *et al.* 2002; Faircloth *et al.* 2004), and therefore, is not considered to be disruptive since it is a normal component of the life-course. Williams (2000: 51) suggests that in older age chronic illness may be perceived as a ‘biographically anticipated event’. Similarly, Faircloth *et al.* (2004: 245), also argue the onset of sudden illness such as stroke is not always perceived as a disruptive event, but instead may be viewed in terms of a ‘biographical flow’. This is not the case for older participants in this thesis, some of whom allude to being ‘healthy and feeling cheated (Sandra) by the loss of their sight. I argue, then, that rather than focus on the age of onset of sight loss we should instead adopt Lawton’s (2003: 29) suggestion that it is more appropriate to consider an ‘individual’s whole biography, not just the age at which they first contract a particular disease’.

Secondly, there is an assumption, predominantly within the psychological sight loss literature, that following sight loss people move in a unidirectional way through the five stages of grief, ultimately arriving at acceptance (Kübler-Ross 1963). Mirroring more recent research (Murray *et al.* 2010), the narratives of participants in this thesis suggest that the traditional grief model is too simplistic and fails to account for the diverse and highly individualised experiences of sudden sight loss. Many participants in this thesis continue to experience episodes of grief many years after the onset of sight loss—and thus, never reach acceptance. I have also captured the initial reactions of mothers to their child’s diagnosis. LHON is a sporadic condition, as in the majority of cases there is no family history. Mothers who were unaware of their carrier status experienced feelings not only of guilt and self-blame, but also shock and devastation after their child was diagnosed (Clarke 2016). Mirroring Thomas’ (2014) study with mothers of children diagnosed with Down’s syndrome,

they mourn the loss of the perfect idealised child. In the months following their child's diagnosis, mothers expressed the view that their children would never lead an independent life or get married and have children of their own. However, again mirroring Thomas's research, mothers later spoke of their pride in their children's achievements.

Following the early weeks and months, participants start to re-position themselves by recognising that LHON is a long-term condition. I have explored how participants negotiate the longer-term everyday realities of living with sudden sight loss by considering the significance (Bury 1991) for them of living with LHON. I again draw on Bury's (1982) conceptual framework of biographical disruption. Whilst acknowledging that Bury's framework refers to a single event occurring at the *onset* of illness, this is a point of departure for this thesis. Participants, some of whom had lived with sight loss for nearly 20 years, referred to experiencing *repeated* events of disruption throughout their lives—a kind of 'recurrent biographical disruption' (Saunders 2017: 726). As a chronic illness, LHON occupies an unusual space within the chronic illness literature. Many participants were in good health before the onset of LHON and (with the exception of the four women diagnosed with MS-like symptoms), remain so after their sight loss. However, although not terminal, LHON is a chronic condition as it is long-term and currently has no cure.

I identified that irrespective of age, a number of participants considered that after receiving their diagnosis a life was over. I have captured how some of the younger participants, have not yet come to terms with their sight loss. Charmaz (1983) has characterised living with long-term illness as leading a restricted life, experiencing social isolation, and negotiating discredited definitions of self, forcing individuals to reconceptualise their self-identity. The narratives of participants in my thesis resonates with Frank's (1995: 97) 'chaos narrative'. However, I also claim that participants experience emotions more severe than biographical disruption aligning more with Locock *et al.*'s (2009: 1043) concept of 'biographical abruption', that is when people consider a diagnosis to be a 'death sentence'.

I explored how participants overcome the challenges presented to them by sudden bi-lateral sight loss. One of the dominant themes that emerged from participant narratives was the desire to return to a 'normal' life. Normality was interpreted as

once again undertaking tasks that had previously been taken-for-granted or doing the same things as everyone else. My findings parallel current research that has considered how normalcy is negotiated in everyday life (Gregory 2005; Hanson and Philo 2007; Seidman 2013; Coleman-Fountain 2017; Buse and Twigg 2018; McLaughlin and Coleman-Fountain 2018). Green (2009: 4) argues that by promoting positive images of illness and disability, those who have previously been categorised as 'not normal' are challenging what it is to be 'normal'. There has also been a wealth of anti-discrimination legislation such as the Equality Act 2010, designed to protect the right of those living with a disability.

Such legislation is an example of how attitudes to those living with a disability are being challenged; (although the relative successes of such legislation in achieving this lofty aim has been debated, (Thomas 2012)). Disability activists continue to contest perceptions that disability is a personal tragedy and those living with disability are to be pitied. The old order, that positioned disability within a medical model preoccupied with the symptoms of disease, is being usurped by the introduction of a social model of disability, which acknowledges that it is not a problem located within the impaired body but is a consequence of an oppressive social environment (Oliver 1990, 1996; Barnes *et al.* 2002; Thomas 2007). As Green (2009: 9) comments:

'the social model shifts the problem away from people with disabilities and focuses upon society and societal attitudes that create a physical and social environment that socially excludes and oppresses disabled people.'

Notwithstanding these welcome changes in attitude, for many participants the social model of disability has failed to bring about changes to the physical environment which continues to exclude them and poses a threat to their desire to be perceived as normal. Whilst the built environment has proved challenging, the most pervasive challenge to normality identified by participants was that of experiencing stigma, both felt and enacted (Scambler and Hopkins 1986), which at times sabotaged their attempts to be perceived by others as normal. Participants did not believe they were stigmatised by their diagnosis of a genetic condition, but rather by the reaction of others to the physical manifestations of their sight loss. Echoing Watson's (2002) research, many of the participants in this thesis, felt that

they were 'normal'. Scambler and Hopkins (1986) claim that, for their participants felt stigma was more apparent than enacted stigma. However, although participants in my thesis referred to experiencing both felt and enacted stigma, it was enacted stigma that dominated their social interactions. Participants' narratives recounted numerous instances of (overt) stigma whilst using public transport, or whilst in the workplace, or out shopping. Scambler and Paoli (2008: 1851) have added a further dimension to understanding felt and enacted stigma by introducing the concept which they refer to as 'project', suggesting that people respond to discreditable or discredited illnesses, 'without either internalising cultural norms of shame and blame or becoming fearful, defensive or subdued by prospects of discrimination'. The authors argue that people living with disabling conditions 'formulate positive strategies and tactics that acknowledge the risks of enacted stigma and deviance whilst trying to avoid the pitfalls of felt stigma and deviance'. There was no evidence to suggest that participants in my thesis had formulated positive strategies when confronted with stigma (felt or enacted). For instance, whilst some participants identified the 'secondary gains' (Goffman 1968) of a white cane, they also conceded that they would avoid using it in certain situations.

Imagined futures

Over time, the majority of participants were able to adopt coping strategies to successfully live with their changed circumstances and imagine a future where they realise their aspirations, including finding a good job and buying their own home—where they could in short, live a 'good life' (Sakellariou 2015; Errington *et al.* 2018). In Chapter Seven I looked to the future and located my discussion in the sociology of hope (Petersen and Wilkinson 2014). Whilst a few participants claimed that they had accepted their sight loss and were not interested in finding a cure, the vast majority of participants invested emotionally and financially in the introduction of one new treatment—Idebenone—to restore their sight. I have alluded to how the effectiveness of Idebenone has divided the scientific community. I have also argued and that Santhera Pharmaceuticals provides an example of how a multinational company, seeking to recoup the costs of developing Idebenone, has attempted to mobilise stakeholders to have Idebenone routinely prescribed in the UK and in so

doing have participated in what Novas (2006: 289) refers to as the 'political economy of hope'.

I also explored the hope felt women who were interested in the development of mitochondrial replacement techniques (MRTs) which potentially will provide them with the opportunity to give birth to a genetically-related healthy child. I focused on the narratives of four women, in particular, two of whom at the time of interview were considering their reproductive options. Notwithstanding the extensive media debates surrounding the ethical, social and legal implications of allowing MRTs, the women were influenced not by the extensive media rhetoric but by their own experiential knowledge of being affected by LHON or living with a relative who is affected.

Chronic illness and disability

These three key themes, I argue, combine to suggest two key, unique contributions to the wider literature. First, the thesis extends Bury's (1982) conceptual framework of biographical disruption through exploring the lived experiences not only of men and women affected by LHON, but of their mothers who carry the mutation, a group of people rarely the focus of sociological interest. Using interview data and some observations, I also unpacked the work of the genetic ophthalmologists in diagnosing and treating LHON. In so doing, I would argue that this research contributes to the sociology of diagnosis scholarship (Blaxter 1978, 2009; Brown 1990, 1995; Jutel 2009; Jutel and Nettleton 2011).

I acknowledge throughout the thesis that Bury's conceptual framework has been the subject of much debate (Lawton 2003; Pierret 2003; Williams 2000); indeed Bury (2001) has revisited and extended his original work. I have also acknowledged that more recent research (for example, Locock *et al.* 2009; Saunders 2017) has provided a new perspective on Bury's concept. Nonetheless, the narratives of participants in this thesis chimes incredibly accurately with Bury's own insight – as well as those of scholars like Charmaz (1983) and Williams (1984). Such classical works, when used to frame the experiences of people with LHON in this research, show remarkable durability.

Finally, I suggest that by unpacking the complex interconnections between sight loss, disability, normalcy, and stigma, my thesis is located at the intersection of disability studies and medical sociology, thereby contributing to the ongoing debate surrounding disability and impairment. As Watson (2013: 78) has commented, the social model of disability was developed by disability activists and promoted by disabled scholars, who were 'disillusioned with the way that sociology in general, and medical sociology in particular, defined and researched disability'. In turn, Thomas (2007: 570) suggests that disability studies are informed by the idea that disability is 'culturally structured by social oppression, inequality and exclusion'. In contrast, medical sociology delineates disability as a consequence of illness and impairment and entails suffering and some social disadvantage.

The future

So what now for people with LHON? Against the background of austerity politics, people living with sight loss in the UK may be said to be facing a precarious future as financial cuts to the Health and Social Care budget impact on the provision of eye clinic services, the recruitment and training of ophthalmologists (Royal College of Ophthalmologists 2018), and support services provided by local authorities. The RNIB recently published a series of reports and consultation papers (2016) focusing on how cuts to the social care budget impact on the quality of life of the visually impaired. There are almost three quarters of a million blind or sight impaired people living in the UK and this number is predicted to increase by 12 per cent in the next five years (RNIB 2016). The RNIB claim that of the severely sight impaired and sight impaired people who are known to their local authority in England, 49 per cent do not receive assessment for vision rehabilitation support. Similarly, research undertaken by the Thomas Pocklington Trust (2018) suggests that reductions in the availability and amount of welfare benefits have caused exceptional financial hardship to those living with visual impairment.

There has been a lack of sociological scholarship exploring the lived experience of sudden sight loss. In what follows, I suggest some avenues for further exploration. For instance, whilst I have focused on affected men and women and their mothers, I believe there are a number of areas of further research including, firstly, collecting the narratives of unaffected siblings, who have to cope with 'survivor guilt' (Arribas-

Ayllon 2008: 1522) and who may themselves develop sight loss at some point in the future. Moreover, I did not have the opportunity to interview fathers to discuss their experience of living with a partner who carries or is affected by LHON and one or more children who are affected, this indicates a possible point of departure for future researchers. Finally, undertaking research with people currently enrolled into the gene therapy trial would provide the opportunity to consider in more depth the motivation for participating in an experimental treatment that may potentially restore sight but also may result in loss of visual acuity.

Conclusion

In drawing my thesis to a conclusion, I have alluded to the lack of sociological scholarship exploring LHON. Although this could be considered a disadvantage, I suggest that it has given me the opportunity to locate my thesis within the chronic illness literature whilst also providing the opportunity to unpack wider sociological debates exploring the complex interconnections between sight loss, disability and normalcy. From here, I have been able to both engage with and extend key themes (including biographical disruption (Bury 1982), loss of self (Charmaz 1983), uncertainty (Fox 1957; Atkinson 1984; Davis 1960), regaining a sense of normalcy (Davis 1995) and stigma (Goffman 1968; Scambler and Hopkins 1986, Scambler 1989, 2006, 2009, 2018)), and discuss their relevance to people living with LHON. I have suggested that LHON is a condition that is poorly understood, a condition that does not sit easily within the chronic illness literature with the majority of participants remaining fit and healthy after their sight loss—and yet having a long-term inherited condition for which there is currently no cure. In addressing the existing gaps in the literature, I argue that my thesis is located at the intersection of the chronic illness and disability studies.

I finish by reminding myself of Southwell's (2012: 109) comments that 'fear, pity and stigmatisation of visual impairment are not just somewhere out in the atmosphere, they are within all of us to a degree, until we take the trouble to learn that they are subconsciously assumed stereotypes that we can correct'.

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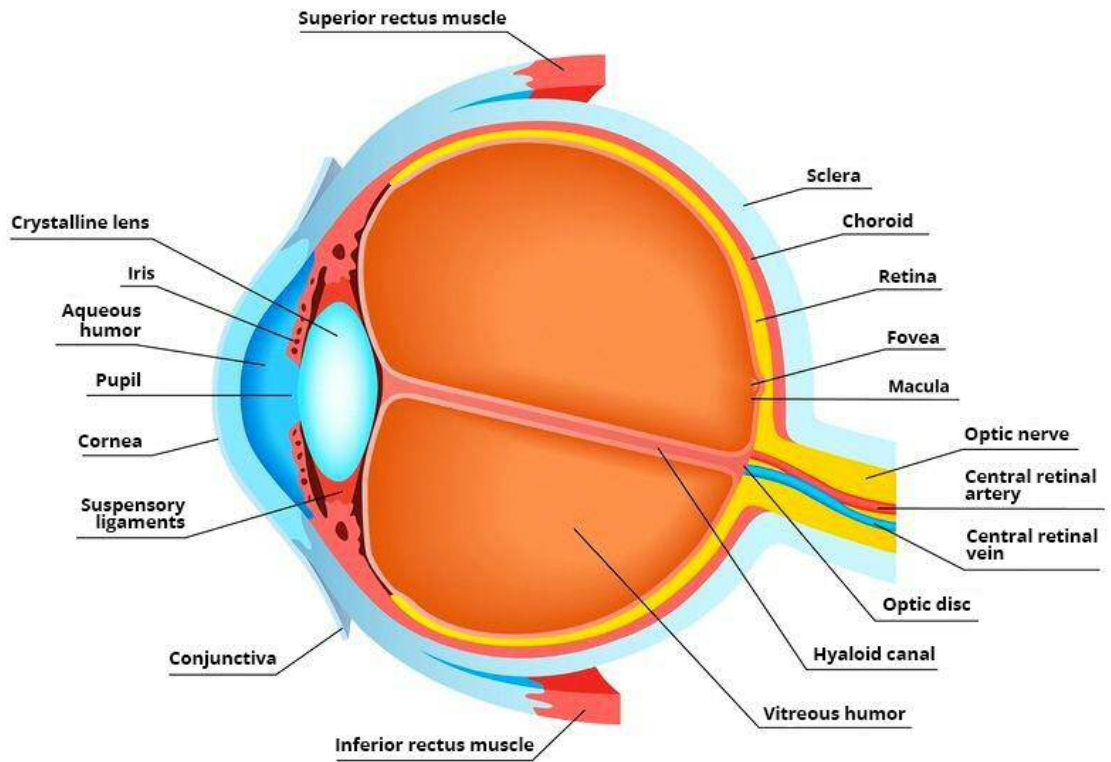
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Appendices

Appendix 1: Anatomy of the Eye



Source: The Royal College of Ophthalmologists (2018).

Appendix 2: Invitation letter adult participant interview and observation (example)

Study Title: Living with Leber hereditary optic neuropathy: exploring the experiences and perceptions of children, young adults and their families.

Date:

Dear

My name is Lydia Harper and I am a PhD researcher at Cardiff University. I am conducting a research study and would like to invite you to participate. My research is interested in finding out what it is like to have a genetic eye condition known as Leber hereditary optic neuropathy (LHON). I would like to be present when you attend the hospital for your eye clinic appointment and after the appointment, I would like to interview you.

I have included in this letter an information sheet and consent form. If you would like to take part in my research, please email me at HarperL@cardiff.ac.uk. I will then arrange to discuss my research in more detail with you and answer any questions you may have before you make a final decision.

Thank you for your help.

Yours Sincerely

Lydia Harper
PhD Researcher
Cardiff University School of Social Sciences

Appendix 3: Invitation letter child participant interview and observation (example)

Study Title: Living with Leber hereditary optic neuropathy: exploring the experiences and perceptions of children, young adults and their families.

Date:

Dear [*Insert Parent/ Guardian Name*]

My name is Lydia Harper and I am a PhD researcher at Cardiff University. I am conducting a research study and would like to invite your child to participate. My research is interested in finding out what it is like to have a genetic eye condition known as Leber hereditary optic neuropathy (LHON). I would like to be present when your child attends the hospital for their eye clinic appointment and after the appointment, I would like to interview you your child.

I have included in this letter an information sheet and parental consent form. If you are interested in my research, please contact me on the telephone number or email above. I will make arrange to discuss the information sheet with you in more detail and answer any questions you may have before you make a final decision.

Thank you for your help.

Yours Sincerely

Lydia Harper

PhD Researcher

Cardiff University School of Social Sciences

Appendix 4: Information sheet and consent form adult participant (example)

Participant Information Sheet and Consent Form for Adults attending the Eye Clinic

This Information Sheet and Consent Form is for research participants who are being invited to participate in a study that will explore what it is like to live with the genetic eye condition known as Leber hereditary optic neuropathy (LHON).

This form has two parts:

- Information Sheet (gives you information about the study)
- Consent Form (this is where you sign if you agree to participate).

You will be given a copy of this form.

Part 1: Information Sheet

Introduction

My name is Lydia Harper and I am a PhD Researcher at Cardiff University. As part of my degree I am inviting you to take part in my research project which aims to discover what it is like to live with the genetic eye condition known as Leber hereditary optic neuropathy (LHON). Your involvement in the research will be voluntary, your medical treatment will not change, and I will not have access to your medical records. Before deciding whether you would like to participate, I would like you to understand the reasons for my research and what it may involve for you. Reading the information sheet will take around 10-20 minutes. Part 1 of this formation sheet tells you the purpose of the study and provides more detailed information. Part 2 is the consent form which you sign if you are happy to participate. If you would like to participate, please contact me by telephone or email.

What is the purpose of the study?

Developments in genetic technologies to identify and treat rare genetic eye conditions are advancing at a rapid pace. This means that there is now far more information available than ever before. Some of the information may be very complex and difficult to understand. My research is concerned with the

experiences and perceptions of adults who have been diagnosed with LHON. If you previously attended the genetic eye clinic or genetic counselling clinic, I would like you to tell me about your experience. My research is also considering how adults and their families come to terms with a sudden loss of sight caused by a genetic eye condition.

Who has reviewed the study?

All research in the NHS is reviewed by an independent group of people, called a Research Ethics Committee, which is there to protect your safety, rights, wellbeing and dignity. This project has been approved by the Wales REC 1.

Why have I been invited to participate?

You have been invited to participate in the research because you have been identified by your ophthalmologist as having LHON.

What will happen if I take part?

If you agree to take part, I would like to be present during your eye clinic appointment. During the appointment I will be taking notes and may use a digital recorder to record the conversation between you and the ophthalmologist; this is to ensure that I type up accurately what is said during the appointment. I will only record the appointment if you agree to this. If you do not want me to record the appointment, then I will not do so. I will not be requesting any extra time on top of what is already allocated for the appointment.

After the appointment I would like to interview you about your experiences and perceptions of living with a genetic eye condition and if you have developed problems with your eyesight how this affects your ability to undertake daily activities. If you agree to the interview, this would take place immediately after your appointment or (if you prefer) at a later date. I will be taking notes during the interview and may also use a digital recording device to record the interview. This is to ensure I accurately record what you say during the interview. However, if you do not want me to record the interview then I will not do so. You can choose when and where you want the interview to take place. The interview will last approximately 1–1½ hours.

Is participation in the study voluntary?

Yes. You do not have to agree to take part in the research. If you do agree to participate, you will need to sign a consent form. If at a later date you change your mind, you can withdraw your consent to participate in the research. In the event that you do decide to withdraw consent, any information collected during the research will be destroyed securely.

What are the potential benefits of taking part?

The study will not help you directly, but the information you provide may help other people to understand what it is like to have a sudden loss of sight caused by a genetic eye condition.

What are the risks of taking part?

You will be asked sensitive questions about your eyesight and how it impacts on your ability to take part in daily activities and how your eyesight will potentially affect your prospects of continuing your education and obtaining employment. You may find talking about these things upsetting; however, this research has been designed to ensure that your welfare is the most important consideration. If at any time you find talking about your eyesight distressing, the interview will be stopped. You can also decide not to participate in the research if it is too upsetting for you to carry on. If you do decide to withdraw from the research any information I have collected will be destroyed securely.

What if I do not want to carry on with the study?

If you decide you want to withdraw from the study, please contact me immediately. Any information that has been collected up to this point will be deleted. However, if at a later date you change your mind and want to participate once again you can re-register an interest at any time during the study.

Will I find out anything about the outcome of the study?

Yes. At the end of the research, I will send you a summary of the results. I am also intending to use the results of the study as the basis for articles that may be printed in research journals, and for presentations at academic conferences. The data will be anonymised so that you cannot be identified.

Who is organising and funding the study?

This study is sponsored by Cardiff University and is funded by the Economic and Social Research Council (ESRC).

What consequences will this have for the data collected?

The ESRC requires that researchers send data to the UK Data Archive (UKDA). The data will include typed-up transcripts from the clinical appointments and the interview transcripts. The transcripts will be heavily edited to ensure that there are no identifiable individuals or locations. The data stored at the UKDA will be anonymised and made available to third parties. However, your data will only be sent to the UKDA and made available to third parties if you agree to this. The typed-up interview notes will be kept for a minimum period of five years from the end of the research. This is in accordance with Cardiff University's data retention period. After five years, the data will be destroyed consistent with Cardiff University's 'Complying with Data Protection and Freedom of Information' policy.

Will my participation in the study be kept confidential?

Yes. All the information about your involvement in this study will be confidential. This means that you will not be identified in any report or publication. The identities of everyone taking part in the study will be protected by being given false names or numerical codes. The data collected from you (audio-recordings and typed-up interview notes) will be stored securely either in a locked filing cabinet (which only I can access) on security-controlled Cardiff University premises or on a password-protected Cardiff University computer and a password-protected USB computer device. The data will have your name removed and replaced by a numerical code. The sheet containing these codes and your contact details will be kept as manual records in a locked filing cabinet on security-controlled Cardiff University premises. After the PhD thesis is completed, your data and details will be deleted and/or shredded in accordance with Cardiff University regulations. Please note you have the right to check the accuracy of the data that I hold.

What if I have a complaint about my participation in the study?

If you have concerns about the study, please do not hesitate to contact me, Lydia Harper at HarperL@cardiff.ac.uk. You may also contact my academic supervisors, Professor Adam Hedgecoe (02920 870027, HedgecoeAM@cardiff.ac.uk) or Professor Joanna Latimer (02920 876908, LatimerJE@cardiff.ac.uk). You may also wish to contact the Clinical Supervisor for this research: Dr Morgan, consultant ophthalmologist, St Tristan's Hospital. If you remain unhappy and wish to complain formally, you should contact the Complaints Department by email: Concerns@wales.nhs.uk.

What happens if I am harmed while participating in the study?

In the unlikely event that something goes wrong and you are harmed during the research, and this is due to someone else's negligence, then you may have grounds for a legal action for compensation against Cardiff University.

Further information and contact details

Thank you for taking time to read this Information Sheet and Consent Form. If you require further information about the study or wish to register an interest in participating, please contact me using the details below:

Lydia Harper
Cardiff University School of Social Sciences
PhD Office
1-3 Museum Place
Cardiff CF10 3BD
HarperL@cardiff.ac.uk

Part 2: Consent Form

Participant ID:

Please Initial Box

I have read and understood the information leaflet (version 2.0 dated 09/10/15).
I have had the opportunity to consider the information and ask questions and
have had these answered satisfactorily.

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

I agree/do not agree* to take part in an interview with the researcher which will
be anonymised. (*Delete as appropriate)

I agree/do not agree* to the interview being tape-recorded by the
Interviewer. (*Delete as appropriate)

I agree/ do not agree* to my data being sent to the UK Data Archaive
and made available to third parties. (*Delete as appropriate)

I acknowledge that my data may be used in the PhD thesis and academic or
other publications.

I acknowledge that I can request a summary of the study findings.

I agree to take part in the above study.

Research Participant

Date

Signature

Name of Person taking
Consent

Date

Signature

When completed: 1 for participant; 1 (original) to be kept by researcher.

Appendix 5: Parental Information sheet and consent form (example)

Parental Information Sheet and Parental Consent Form for Young People aged 13–15 with LHON attending the eye clinic

This Information Sheet and Consent Form is for the parents/guardians of young people who are being invited to participate in a study that will explore what it is like to live with the genetic eye condition known as Leber hereditary optic neuropathy (LHON).

This form has two parts:

- Information Sheet (gives you information about the study)
- Parental Consent Form (this is where you sign if you agree that your child can participate).

You will be given a copy of this form.

Part 1: Information Sheet

Introduction

My name is Lydia Harper and I am a PhD Researcher at Cardiff University. As part of my degree I am inviting your child to take part in my research project which aims to discover what it is like to live with the genetic eye condition known as Leber hereditary optic neuropathy (LHON). My research is also concerned with how doctors give complex information to patients about their eye condition and the questions patients ask during their appointment. I would like to be present during your child's appointment in the eye clinic. To do this this, I will require your permission. After you have heard more about the study, and if you agree, the next thing I will do is ask your child for their agreement as well. After the appointment in the eye clinic I would like to interview your child. Both of you have to agree independently before I can do this. You do not need to decide today whether or not you agree that your child can participate in this research. Before you decide, you can talk to anyone you feel comfortable with. If there is anything you do not understand, please ask me to stop as we go through the information and I will take time to explain. If you have any questions later, you can contact me by telephone, email or letter.

Your child's involvement in the research will be voluntary, their medical treatment will not change, and I will not have access to their medical records. Before deciding whether you would like to participate, I would like you to understand the reasons for my research and what it may involve for you. Reading the information sheet will take around 10-20 minutes. Part 1 of this information sheet tells you the purpose of the study and provides more detailed information. Part 2 is the consent form which you sign if you are happy for your child to participate. If you would like your child to participate, please contact me by telephone or email.

What is the purpose of the study?

In recent years doctors have been able to find out much more about how to recognise and treat sight loss caused by genetic eye conditions. This means that there is now far more information available than ever before. Some of the information is very complex and may be difficult to understand. My research is concerned with finding out how doctors diagnose genetic eye conditions, how doctors explain complex information to young people and how young people make sense of the information they are given.

Who has reviewed the study?

All research in the NHS is reviewed by an independent group of people, called a Research Ethics Committee, which is there to protect your child's safety, rights, wellbeing and dignity. This project has been approved by the Wales REC 1.

Why has my child been invited to participate?

Your child has been invited to participate in the research because they have been referred to the genetic eye clinic.

What will happen if my child takes part?

If you agree to your child taking part, I will be in attendance during the appointment in the clinic. During the appointment I will be taking notes and may also use a digital recording device to tape the conversation between your child and the ophthalmologist; this is to ensure that I accurately type up what was

said during the appointment. However, if you do not want me to record the appointment then I will not do so.

Is participation in the study voluntary?

Yes. You do not have to agree to your child taking part in the research. You can choose to say no and any services that you and your family receive at the hospital will not change. If you do agree to allow your child to participate, your child will also have to confirm that they would like to take part. If you change your mind later, you can withdraw your consent to your child's participation in the research. Your child may also decide to withdraw from the research at any time. In the event that either you or your child decides to withdraw consent/assent, any information collected during the research will be destroyed securely.

What are the potential benefits of my child taking part?

The study will not help your child directly, but the information your child provides may help to improve the way in which children are dealt with in the eye clinic and how complex information is given to them. The information obtained from your child may also help other people to understand what it is like at a young age to have a sudden loss of sight caused by LHON.

What are the risks to my child in taking part?

I will be present during your child's appointment with the doctor. The doctor may ask your child sensitive questions about their eyesight and how it impacts on their ability to take part in daily activities. Your child may find talking about these things in front of me upsetting. However, this research has been designed to ensure that your child's welfare is the most important consideration. If at any time your child finds talking about their eyesight in front of me distressing, the interview will be stopped. Your child can also decide not to participate in the research if it is too upsetting for them to carry on. If your child does decide to withdraw from the research any information I have collected will be destroyed.

What if I do not want my child to carry on with the study?

If you decide you want your child to withdraw from the study, please contact me immediately. Any information that has been collected up to this point will be deleted. However, if at a later date you change your mind and want your child to participate then you can re-register an interest at any time during the study. If this happens, your child will also be asked to confirm that they wish to be involved in the research.

Will I find out anything about the outcome of the study?

Yes. At the end of the research, I will send you and your child a summary of the results. I am also intending to use the results of the study as the basis for articles that may be printed in research journals, and for presentations at academic conferences. The data will be anonymised so that your child cannot be identified.

What if I have a complaint about my child's participation in the study?

If you have concerns about the study, please do not hesitate to contact me, Lydia Harper, on 07715254962 or HarperL@cardiff.ac.uk. You may also contact my academic supervisors, Professor Adam Hedgecoe (02920 870027, HedgecoeAM@cardiff.ac.uk) or Professor Joanna Latimer (02920 876908, LatimerJE@cardiff.ac.uk). You may also wish to contact the Clinical Supervisor for this research: Dr Morgan, consultant ophthalmologist, St Tristan's Hospital. If you remain unhappy and wish to complain formally, you should contact the Complaints Department by email: Concerns@wales.nhs.uk.

What happens if my child is harmed while participating in the study?

In the unlikely event that something goes wrong and your child is harmed during the research, and this is due to someone else's negligence, then you may have grounds for a legal action for compensation against Cardiff University. The normal NHS complaints mechanism will still be available to you (if appropriate).

Will my child's participation in the study be kept confidential?

Yes. All the information about your child's involvement in this study will be confidential. This means that neither you nor your child will be identified in any report or publication. The location of Dr Morgan's clinic and the identities of everyone taking part in the study will be protected by being given false names or numerical codes. The data collected from your child (audio-recordings, typed-up notes from your appointment and typed-up interview notes) will be stored securely either in a locked filing cabinet (which only I can access) on security-controlled Cardiff University premises or on a password-protected Cardiff University computer and a password-protected USB computer device. The data will have your child's name removed and replaced by a numerical code. The sheet containing these codes and your child's contact details will be kept as manual records in a locked filing cabinet on security-controlled Cardiff University premises. After the PhD thesis is completed, your child's data and details will be deleted and/or shredded in accordance with Cardiff University regulations. Please note you have the right to check the accuracy of the data that I hold.

Who is organising and funding the study?

This study is sponsored by Cardiff University and is funded by the Economic and Social Research Council (ESRC).

What consequences will this have for the data collected?

The ESRC requires that researchers send data to the UK Data Archive (UKDA). The data will include typed-up transcripts from the eye clinic appointment and the interview transcripts. The transcripts will be heavily edited to ensure that there are no identifiable individuals or locations. The data stored at the UKDA will be anonymised and made available to third parties. However, the data will only be sent to the UKDA and made available to third parties if both you and your child agree. The notes taken during the eye clinic appointment and the typed-up interview notes will be kept for a minimum period of five years from the end of the research. This is in accordance with Cardiff University's data retention period. After five years, the data will be destroyed consistent with

Cardiff University's 'Complying with Data Protection and Freedom of Information' policy.

Further information and contact details

Thank you for taking time to read this parental consent form. If you require further information about the study or wish to register an interest in participating, please contact me using the details below:

Lydia Harper
Cardiff University School of Social Sciences
PhD Office
1-3 Museum Place,
Cardiff CF10 3BD
HarperL@cardiff.ac.uk

Participant ID:
Part 2: Parental Consent Form

I have been asked to give consent for my child to participate in the above research study.

Please Initial Box

I confirm that I have read and understood the information sheet (version 2.0. 09/10/15).

I have had the opportunity to consider the information and ask questions.

I know I can ask further questions later if I want to.

I understand my child's participation is voluntary and my child may withdraw at any time without giving a reason and this will not affect my child's medical treatment.

I agree/do not agree* to the researcher being present during my child's appointment in the eye clinic. (*Delete as appropriate)

I agree/do not agree* to the appointment being tape-recorded. (*Delete as appropriate)

I agree/do not agree* to my child taking part in an interview with the researcher. (*Delete as appropriate)

I agree/do not agree* to my child's data being sent to the UK Data Archive and made available to third parties. (*Delete as appropriate)

I acknowledge that data collected during the study may be used in the PhD thesis and academic publications.

I acknowledge that I can request a summary of the study and findings.

I consent voluntarily for my child to participate in this research study.

Name of parent
/guardian

Date

Signature

Name of researcher

Date

Signature

When completed: 1 for participant; 1 (original) to be kept by researcher.

Appendix 6: Child participant information sheet and assent form (example)

Participant Information Sheet and Assent Form for Young People aged 13–15 with LHON attending the eye clinic

This Information Sheet and Assent Form is for young people who have been referred to the genetic eye clinic, and who are being invited to participate in a study that will explore what it is like to live with the genetic eye condition known as Leber hereditary optic neuropathy.

This Information Sheet and Assent Form has two parts:

- Information Sheet (gives you information about the study)
- Assent Form (this is where you sign if you agree to participate).

You will be given a copy of this form.

Part 1: Information Sheet

Introduction

My name is Lydia Harper and I am a PhD Researcher at Cardiff University. As part of my degree I am inviting you to take part in my research project which aims to discover what it is like to live with the genetic eye condition known as Leber hereditary optic neuropathy (LHON). My research is also concerned with how doctors give complex information to patients about their eye condition and the questions patients ask during their appointment. I would like to be present during your appointment in the eye clinic. To do this, you will have to agree. This study only talks about your experience of going to the clinics—you do not need to attend any extra appointments in the clinics. I am going to give you information and invite you to take part in my research study. Your involvement will be voluntary, and your medical treatment will not change. I have discussed this research with your Mum, Dad or carer and they know that I am asking for your agreement. If you are going to participate in the research, they will also have to agree. If you do not wish to take part in the research, you do not have to.

You may discuss anything in this form with anyone you feel comfortable talking to. You can decide whether or not to take part after you have talked it over; you do not have to decide immediately. If there are words you do not understand or things you want me to tell you more about, please ask me to stop and I will take time to explain.

Before deciding whether you wish to take part, I would like you to understand why this research is being done and what it involves for you. Reading this form will take about 20 minutes. **Part 1** tells you all about the study and why it is being done. **Part 2** is the assent form that you sign if you agree to being involved in the study.

After you have heard more about the study, and if you agree, after your appointment in the eye clinic I would like to interview you. You do not need to decide today whether or not to participate in this research. Before you decide, you can talk to anyone you feel comfortable with. If there is anything you do not understand, please ask me to stop as we go through the information and I will take time to explain. If you have any questions later, you can contact me by telephone, email or letter.

What is the purpose of the study?

In recent years doctors have been able to find out much more about how to recognise and treat sight loss caused by genetic eye conditions. This means that there is now far more information available than ever before. Some of the information may be very complex and difficult to understand. My research is interested in how the work of the genetic eye clinic is undertaken. My research is also concerned with how doctors explain complex information to young people and the questions young people ask during their appointment to make sense of the information they are given.

Who has reviewed the study?

Before my research can go ahead it has to be considered by a Research Ethics Committee. The committee makes sure that your interests are protected and that the research is carried out fairly. This project has been approved by the Wales REC 1.

Why are you inviting me to participate in the study?

You have been invited to take part in the study because you attend the eye clinic.

Do I have to do this?

No. You do not have to be in this research if you do not want to. It is up to you. If you decide that you do not want to be involved in the research, that's okay, it will not affect your medical treatment. If you decide to say "yes" now, you can change your mind later and that's okay as well.

What is going to happen to me?

Firstly, I have already spoken to your parents or carer and discussed my research in more detail with them. Your parents or carer have indicated they are happy for me to speak to you. I will answer any questions you would like to ask me. If you agree to take part, I will be present when you have your appointment in the eye clinic. I will be taking notes and I may also use a digital recorder to record your conversation. However, if you do not want me to record the appointment then I will not do so. I am interested in how information is given to you about your eye condition and the questions you ask during the appointment. I will not be getting involved in your treatment. Your appointment will not take any longer than normal. I would also like to interview you after your appointment. Again, you do not have to agree to be interviewed. The choice is yours.

What are the benefits of taking part?

The study will not help you directly, but the information you provide may help to improve the way in which young people are dealt with in the eye clinic and how complex information is given to them.

Are there any disadvantages or risks of taking part?

I will be present during your appointment with the doctor. You may find it distressing to have me present when you are being asked sensitive questions about your eyesight. However, this research has been designed to ensure that your welfare is the most important consideration. If you do find talking about your eyesight in front of the researcher upsetting, then I will leave the

appointment. You can also decide not to be involved in the research anymore. If you do withdraw from the research any information collected about you will be destroyed.

What if I do not want to carry on with the study?

If you decide you do not want to carry on with the study, just tell your Mum, Dad or carer at any time. They will not be cross with you. Information that has already been collected up to this point will be deleted. However, if you later change your mind and wish to take part in the study you may re-register an interest. Should this happen, I will need to speak to your Mum, Dad or carer to confirm that they are still happy for you to participate.

Will I find out anything about the outcome of the study?

At the end of the research, I will send you and your Mum, Dad or carer a summary of the results. After this I will be telling more people about the research. I will be doing this by writing research articles that may be printed in educational journals, and by speaking at academic meetings.

Who is organising and funding the study?

The study is sponsored by Cardiff University and is funded by the Economic and Social Research Council (ESRC).

What consequences will this have for the data collected?

The ESRC requires that researchers send data to the UK Data Archive (UKDA). The data will include typed-up transcripts from the clinical appointments. The transcripts will be heavily edited to ensure that there are no identifiable individuals or locations. The data stored at the UKDA will be anonymised and made available to third parties. However, your data will only be sent to the UKDA or disclosed to third parties if you agree. The notes taken during the eye clinic appointment and will be kept for a minimum period of five years from the end of the research. This is in accordance with Cardiff University's data retention period. After five years, the data will be destroyed consistent with Cardiff University's 'Complying with Data Protection and Freedom of Information' policy.

What can I do if I have complaint?

If you have concerns about the study, you should firstly tell your Mum, Dad or carer. You can also contact me, Lydia Harper (07715254962, HarperL@cardiff.ac.uk). You may also contact my supervisors, Adam Hedgecoe (02920 870027, HedgecoeAM@cardiff.ac.uk) or Joanna Latimer (02920 876908, LatimerJE@cardiff.ac.uk). You may also wish to contact Dr Morgan who is the doctor seeing you at in the eye clinic.

What happens if I am harmed while participating in the study?

In the unlikely event that something goes wrong and you are harmed during the research and this is due to someone else's fault, you and your parents/guardian may have grounds to take legal action against Cardiff University.

Will my participation in the study be kept confidential?

Yes. All the information about your involvement in this study will be confidential. This means you will not be identified in any report or publication. Fake names or numerical codes will be given to the location of Dr Morgan's clinic and the identities of everyone taking part in the study. The data collected from you (audio-recordings, typed-up notes from your appointment) will be stored securely either in a locked filing cabinet (which only I can access) on security-controlled Cardiff University premises, or on a password-protected Cardiff University computer and a password-protected USB computer device.

Further information and contact details

Thank you for taking time to read this Information Sheet and Assent Form. If you require further information about the study or wish to register an interest in participating, please contact me using the details below:

Lydia Harper
Cardiff University School of Social Sciences
PhD Office
1–3 Museum Place,
Cardiff CF10 3BD
HarperL@cardiff.ac.uk

Part 2: Assent Form

Young person (or, if unable, parent/guardian on their behalf) to circle all they agree with:

Has somebody explained the research to you? Yes/No

Do you understand what the project is about? Yes/No

Have you asked all the questions you want? Yes/No

Have you had your questions answered in a way you understand ? Yes/No

Do you understand it's OK to stop taking part at any time? Yes/No

Do you agree to the researcher being present during your appointment in the eye clinic? Yes/No

Do you agree to the researcher using a digital recorder during your appointment at the eye clinic? Yes/No

Do you agree to being interviewed by the researcher? Yes/No

Do you agree to your data being deposited at the UK Data Archive and made available to third parties? Yes/No

If you **do** want to take part, you can write your name below

Your name _____ Date _____

The person who explained this project to you needs to sign too:

Print Name _____

Signature _____

Date _____

Thank you for your help.

When completed: 1 for participant; 1 (original) to be kept by researcher.

Appendix 7: Participant interview questions (example)

Introductory question

Tell me about your experience of living with LHON, starting with when you first noticed problems with your eyes.

Conversational prompts

What information were you given by the doctors about your eye condition?

Do you think the doctors could have done more to help you with the emotional aspects of sight loss?

Tell me a little more about your feelings when you were given your diagnosis.

Did the relationship with your mother change after your diagnosis? If so, how did it change?

Did the relationship with your siblings' change? If so, how did it change?

Do you feel your friends and work colleagues treated you differently after you lost your sight? If so, give me some examples of how they treated you differently.

Were you offered sight loss/genetic counselling after you lost your sight?

When do you think the most appropriate time to have counselling would be?

What was your lifestyle like before your diagnosis?

Are there things you can no longer do? If so, what things do you miss doing?

Do you use low vision aids?

Has your attitude towards disability altered?

Concluding question

Is there anything else you would like to tell me about your experience of living with LHON?

