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**The Role of Experiential
Knowledge in the Reproductive
Decision Making of Families
Genetically At Risk: The Case of
Spinal Muscular Atrophy**

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**A thesis submitted in fulfilment of
the requirements for the degree of

Doctor of Philosophy in Sociology**

**University of Warwick
Department of Sociology**

July 2010

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Autosomal Dominant Spinal Muscular Atrophy- ADSMA	
Continuous Positive Airway Pressure Machine- Cpap Machine	
Cystic Fibrosis- CF	
Duchenne Muscular Dystrophy- DMD	
Huntingdon's Disease- HD	
Independent Living Fund- ILF	
Jennifer Trust for SMA- JTSMA	
Personal Assistants- PAs	
Polycystic Kidney Disease- PKD	
Pre-Implantation Genetic Diagnosis- PGD	
Spinal Muscular Atrophy- SMA	
Spinal Bulbar Muscular Atrophy- SBMA	
Spinal Muscular Atrophy with Respiratory Distress- SMARD	

This thesis is dedicated to my mother,
Jane Elizabeth Boardman
my biggest inspiration and supporter.

Acknowledgements

I would like to gratefully acknowledge the Jennifer Trust for SMA and their support of this research project, as well as the families and individuals living with SMA who shared their time, stories and lives with me. In particular, I would like to thank Georgia Tallis, for many years of friendship and inspiration (as well as tea drinking!) together with Jackie Doubtfire at the JTSMA, whose support of the study made it possible. I would also like to thank my supervisors, Hannah Bradby, Karen Throsby and Nickie Charles who have offered me the support, guidance and encouragement to keep going on this study- I am extremely grateful to you all. I would also like to pay special thanks to Sarah Mulcahy Lidster, Joannah Arias and Helen Starkey for their help with transcription, friendship and support, as well as Nigel Neilson, Samantha Lyle, Juliet Rayment and Joy Russell who were all endlessly supportive and made me smile, even through the difficult parts. Special thanks also go to my family whose support has been unfaltering, and in particular to Stephanie Boardman for her help with proof reading, as well as all my fellow PhD students at the University of Warwick Sociology department (you know who you are!) for sharing the trials and triumphs of completing our various studies. Lastly, I would like to gratefully acknowledge the support of the ESRC who funded this study (ESRC Award Number- PTA-031-2005-00137) and made it possible.

Declaration

This thesis constitutes my own original work and has been submitted to no other institute of higher education for the award of a degree other than the University of Warwick.

Abstract

This study reports on the analysis of 59 in-depth interviews conducted with people diagnosed with, or from families affected by, Spinal Muscular Atrophy (SMA). It focuses on attitudes towards, and actual uses of, prenatal testing and selective termination for SMA in reproductive decision making for this group of people, in order to focus on the role of experiential knowledge of SMA and its relationship to expert medical knowledge, within these highly complex decisions.

Experiential knowledge has been described in the literature as knowledge derived from experience, whether ‘embodied’ (i.e. sensory) or ‘empathetic’ (i.e. based on the experiences of others). Experiential knowledge has frequently been positioned as being in opposition to, or even conflicting with, medical knowledge, particularly by feminists and disability rights supporters, for whom the tensions between experiential knowledge and medical knowledge have political significance. However, this research found the relationship between expert and experiential knowledge to be both fluid and dynamic, which had important implications for the way in which SMA was conceptualised, understood and responded to by families living with it. Whilst participants’ accounts of SMA were thoroughly grounded in their day-to-day realities with the condition, this knowledge always existed in and through a relationship with expert medical knowledge of SMA.

The inherent uncertainties within and between experiential and expert knowledge, and the ways of conceptualising SMA that emerged from them, however, rather than alleviating, instead contributed to, and heightened, some of the social, ethical and moral dilemmas these families experienced around reproductive decision making. Indeed, many participants became trapped within these ways of knowing SMA and the internal contradictions they contained, whilst for others, the strategic privileging of one form of knowledge as ‘authentic’ over the other became the only way to escape some of these dilemmas, and clarify where their reproductive responsibilities lay.

Introduction

Developments in genetic and reproductive medicine throughout the latter half of the twentieth century have had a significant influence on the reproductive choices available to prospective parents. Advancements in screening and testing technologies and the development of procedures such as pre-implantation genetic diagnosis (PGD) have altered the number and nature of reproductive decisions presented to prospective parents, and these options appear set to increase over time as rising numbers of conditions are identified as being of genetic aetiology (Lawson, 2001; Shakespeare, 2005, 2008a). In line with these developments, there has been much research into how, and to what end, these decisions are approached by would-be parents, uncovering a range of responses and experiences. A key theme within this body of research is related to the issue of 'choice', and how far, given the rise in sophistication and social acceptability of such technologies, prospective parents are now free to exercise choice over the reproductive options available to them. It has been suggested that given the status and authority attributed to medical knowledge, as well as the negative value attributed to physical impairment and disability, prospective parents experience the use of prenatal screening and/or testing technologies as an obligation rather than a free choice (Lippman, 1989; Press and Browner, 1997). Indeed, there have been suggestions that the use of such technologies can be regarded as an extension of parental obligations (The Times, 1999; Harris, 1998, 2000; Purdy, 1996). Kenen (1994) has referred to a sense of 'genetic responsibility' which has emerged alongside the expansion

of genetic knowledge; as we come to learn more about the socially undesirable traits and propensities within our genetic make up, so our responsibility to prevent their transmission increases. This obligation, however, is not experienced equally. As women are more heavily implicated in reproduction than men, they often assume reproductive responsibility as well as the brunt of any negative consequences associated with having a disabled child (Steinberg, 1996: 267; Dragonas, 2001).

Whilst most of the research around the use of prenatal screening/testing technologies has focused on the standard screening/testing practices for conditions which are routinely tested for in most western countries i.e. Down's Syndrome, Spina Bifida etc., less attention has been paid to the reproductive choices made within families affected by a known inheritable condition (Kelly, 2009). For most families affected by inheritable conditions, awareness of the genetic trait arises at the point of the birth of an affected family member, or following the development of symptoms in an existing member. The subsequent reproductive decision making of family members in light of this knowledge has most widely been discussed in the literature in relation to the practice of genetic counselling together with the acceptability and uptake of testing technologies (e.g. Bryant et al., 2005; Wertz et al., 1992). Research within the social sciences, however, has focused on the way in which genetic risk is constructed by such family members in contrast to medicalised perspectives (Parsons and Atkinson, 1992; D'Agincourt-Canning, 2005), as well as how genetic information is managed

and negotiated within social relationships (e.g. Downing, 2005; Hallowell, 1999; Arribas-Ayllon et al., 2008a).

Increasingly, however, the importance of ‘experiential knowledge’, that is, knowledge derived from experience with a given phenomenon, of both pregnancy and of disability is being acknowledged as an important factor in the making of these prenatal testing decisions (Etchegary et al., 2008; Lippman, 1999; Abel and Browner, 1998) particularly for families affected by inheritable conditions (D’Agincourt, 2003; Kelly, 2009; Downing, 2005; Hallowell, 2006; Cox and McKellin, 2001). However, thus far, there have been relatively few studies that have examined how individuals with experience of a genetic condition within their family respond to the reproductive genetic technologies available to them, and the social, moral and ethical dilemmas that accompany them (Kelly, 2009: 82; Ferguson et al., 2000: 74). As Kelly (2009) points out, there has been little cross-referencing of the literature around the experiences of childhood disability within families and the growing literature on prenatal testing decisions, despite the relevance such familial experiences have to the decisions individuals make about the use of prenatal testing (Asch, 1999). Moreover, there has been even less attention paid to the way in which the concerns highlighted by disability rights supporters in relation to prenatal testing and the possibilities of selective termination arise or are experienced by families affected by genetic conditions (Asch, 2000; Shakespeare, 1999), nor how they are conceptualised and experienced by adults with genetic impairments approaching reproductive

decision making, although there is anecdotal evidence to suggest that these are significant issues (Gow, 2000; Kent, 2000).

The purpose of this study, therefore, is to bridge some of these gaps in the literature through an analysis of the influence and importance of experiential knowledge of a particular inheritable condition, Spinal Muscular Atrophy (SMA), on the reproductive decision making of individuals from families affected by SMA, and those diagnosed with SMA themselves. Previous studies have primarily focused on the experiences and reproductive views of specific family members within families affected by inheritable conditions, e.g. individuals diagnosed with the condition themselves (Gow, 2000), the parents of children with genetic impairments (Kelly, 2009; Wertz et al., 1992) or their siblings (Bryant et al., 2005). However, the inclusion of different family members (e.g. brothers, sisters, parents, grand parents) within this study as well as those diagnosed with SMA themselves allows an analysis of different forms of experiential knowledge and degrees of intimacy with the experience of SMA. As well as different types of experience with SMA, I further anticipate that this inclusive approach will allow an exploration of the views of people at different points of reproductive decision making (i.e. those who have not (yet) had children, those who have had children, those who have chosen not to have children etc.), which will potentially illuminate different perspectives and types of experience. The views elicited by this study will be set out, throughout this thesis, in the context of broader debates on prenatal screening and testing for genetic and other disabilities, to highlight the nature and influence of experiential knowledge.

Spinal Muscular Atrophy (SMA)

After Cystic Fibrosis, SMA is the most common (potentially fatal) autosomal recessively inherited condition (i.e. a single gene disorder requiring both parents to carry one copy of the gene each to transmit SMA), affecting approximately 1 in every 6,000 newborns in the North West European population (Dreesen et al., 1998; Spiegler et al., 1990). Werdnig-Hoffman Disease, or SMA type I, moreover, is currently the most common genetically inherited condition causing infant mortality in the UK and America (SMA Foundation, 2009), and it is estimated that there are between 5,500-6,000 people diagnosed with SMA currently living in the UK (NHS choices, 2009). Whilst some of the specific biological mechanisms by which SMA occurs remain poorly understood, SMA is a condition which is nevertheless defined by the medical profession primarily in terms of the presence of anterior horn cell degeneration in the spinal cord. These anterior horn cells are responsible for relaying nerve 'messages' from the brain to the muscles, and their deterioration and/or death results in disruption to neural pathways. This breakdown in the communication between brain and muscle means that the muscles cannot be activated, and, as a consequence of lack of use/stimulation, they gradually atrophy, or 'waste', leading to permanent weakness or even total paralysis of the muscle. Whilst SMA is generally described as a 'neurodegenerative' disease, that is, one that progresses over time, De Groot and De Witte (2005) have queried whether the disease progresses, or whether the increase in symptoms over time reported by those with SMA can be

attributed to the endurance of their symptoms causing further complications, rather than progression of the disease per se.

SMA has been categorised by the medical profession into different ‘types’ according to the degree of muscle weakness experienced. In its most severe form, SMA causes severe atrophy of the inter-costal muscles (those used to support breathing) and can lead to respiratory failure and premature death, often in the first two years of life. SMA presenting in this way in infants has been termed ‘type I SMA’ or ‘Werdnig-Hoffman Disease’ after the neurologists who first described the condition in the late 19th century (Werdnig, 1891; Hoffman, 1893). Less severe forms of the condition have been described as ‘SMA type II’ and ‘SMA type III’, although considerable confusion exists within the medical profession as to how the types of SMA should be differentiated from one another (Dubowitz, 1991, 2008). The age of onset, genetic profile of the individual, together with their achievement of certain developmental milestones (such as the ability to sit or walk unaided) are some of the commonly used determinants of the type of SMA diagnosed, with those able to sit but not walk being described as having type II SMA and those able to both sit and walk being described as having type III SMA (Dubowitz, 1995a).

The implications of SMA for those who live with it have been under researched within the medical literature, with the notable exception of Lamb and Peden’s (2008) qualitative study on the perspectives of adults with SMA. This situation is mirrored in the social sciences, where the perspectives of those living with SMA have been subsumed within broader research projects

addressing a range of topics, including the impact of genetic disease more broadly (Macaulay, 1996), experiences of disablement (Cardol et al., 2002), the ethics of medical decision making (Simonds, 2005) as well as the uses of genetic technologies (Franklin and Roberts, 2006). The relatively low profile of SMA relative to other genetic conditions such as Duchenne Muscular Dystrophy, Huntington's Disease and Cystic Fibrosis may have contributed to the lack of research into the perspectives of families and individuals living with SMA. Where research from a medical perspective has explored the experience of life with SMA, this has primarily focused on quality of life issues (Bach et al., 2003) family and personal adaptation to stress, disability and bereavement (Von Gontard et al., 2002b; Boyer et al., 2006; Lamb and Peden, 2008), medical complications associated with SMA (Von Gontard et al., 2001; Riddick et al., 1982; Carter et al., 1995; De Groot and De Witte, 2005) as well as appropriate interventions and care (Wang et al., 2007; Parker et al., 1999). No studies thus far have explored the way in which those with SMA in their family experience and conceptualise the condition, nor the way in which family members and those diagnosed with SMA approach reproductive decision making in the context of this medically defined genetic risk.

Using the example of the perspectives of families and individuals living with SMA, therefore, within this thesis I present an analysis of the way in which intimate experiential knowledge of an inheritable condition, SMA, is managed and negotiated alongside expert medical knowledge in the context of reproductive decision making. More specifically, I explore the way in which

these (often contradictory) bodies of knowledge are used to negotiate the complex, and frequently incompatible, social, ethical and moral dilemmas associated with reproduction in the context of genetic risk.

Chapter 1 presents the literature surrounding expert and experiential knowledge more broadly, situating the prominence of expert knowledge within 'risk society' theorising. Within this chapter I also consider the way in which the validation of experiential knowledge as an 'authentic' form of knowledge has become a political project for both feminists and disability rights supporters, highlighting the epistemological assumptions of these perspectives. Chapter 2 details the research methods that were used to carry out the study, my approach to analysis of the data as well as a reflexive consideration of my own values and assumptions as a researcher, considering the various implications these factors had for the study. Chapters 3 and 4 lay much of the groundwork for the later consideration of reproductive decision making by presenting the fluidity of the relationship between expert and experiential knowledge in understanding what SMA is (Chapter 3), as well as how it is experienced in day-to-day life (Chapter 4), which form a backdrop for reproductive decisions. Whilst genetic and clinical classifications have suggested particular ways of ordering the experience of SMA, these chapters demonstrate the messiness of such classifications as they are played out through the accounts of those living with SMA; experiential and expert knowledge both informed, but also contradicted each other in participants' understandings of SMA, contributing to the instability of knowledge of SMA. In Chapters 5 and 6, the way in which this knowledge is brought to bear in

reproductive decision making is presented. Participants strategically mobilised particular versions of knowledge about SMA, and privileged it as 'authentic' in similar ways, even when they arrived at very different reproductive decisions. For other participants, however, the contradictions within this knowledge of SMA trapped them in a state of indecision, unable to navigate the incompatible social, ethical and moral dilemmas surrounding reproduction. Chapter 7 draws together the analysis of the previous 6 chapters, highlighting the way in which experiential knowledge, whilst sometimes privileged as an 'authentic' resource, could never be fully disentangled from expert knowledge of SMA. Experiential knowledge emerged as similarly inflected with the ambiguities and instabilities that characterises expert medical knowledge of SMA, highlighting the deeply precarious position from which families affected by SMA approach reproduction.

Chapter 1

The Politics of Knowledge and Risk

The emergence of SMA as a genetic disease, and the subsequent development of testing technologies to monitor and predict its recurrence, have, according to writers within the social sciences, occurred in a context in which the way we think about health and illness, our identities and the way in which we orient ourselves in our daily lives, has drastically shifted. Technological developments, arising alongside processes of modernisation and globalisation, have fuelled a widespread sense of insecurity and risk, which have in turn altered the way in which we relate to one another and manage our daily lives. Beck (1992) and Giddens (1990) have argued that we now live in a 'risk society', a particular type of society in which the management of risk has become an integral part of daily life. Expert bodies of knowledge, such as genetic and medical knowledge, play a significant role in the context of the risk society, as it is by recourse to these professional bodies of knowledge that we both conceptualise, and adopt, strategies to manage risk. Whilst Beck (1992) and Giddens (1990) have pointed to the significance of expert knowledge in the risk society, however, they have simultaneously highlighted the paralleled development of distrust in these forms of knowledge. Contradictions within and between bodies of expert knowledge, together with the acknowledgement that risks can emerge out of, and through, these expert bodies of knowledge have resulted in widespread uncertainty amongst lay people as to what knowledge can be trusted. In more recent years, particularly within medical sociology, there has been an increased interest in 'lay' or

‘experiential’ bodies of knowledge, or knowledge grounded in everyday lived experiences, as an alternative, or supplement, to medical knowledge (Caron-Finterman et al., 2005; Abel and Browner, 1998; Etchegary et al., 2008; Popay and Williams, 1996). For writers and researchers exploring these forms of knowledge, experiential knowledge is an invaluable resource, particularly in relation to the management of risk, as it has the potential to challenge or displace expert accounts. For feminist writers and disability rights supporters, the validation of experiential knowledge as a valuable alternative to such expert knowledge has political significance, particularly in the context of reproductive risk, where the dominance of expert medical knowledge may have especially negative consequences for women and people with disabilities.

This chapter contextualises my research on SMA and the management of genetic risk through the presentation of the literature surrounding the apparent divide between ‘expert’ and ‘experiential’ forms of knowledge. Firstly, the literature surrounding the development of the ‘risk society’ is presented, before moving on to a discussion of the consequences the risk society has for the conceptualisation of different forms of knowledge, particularly medical and genetic accounts of health and illness. Finally, the growing literature on experiential knowledge, and particularly its relevance to feminist and disability rights political projects, is presented.

The Risk Society

Lupton (1999) has argued that the notion of 'risk' can be understood as the attempt by societies to deal with danger, frightening events and misfortune. In pre-modern societies, such events were explained by reference to supernatural forces, metaphysical powers or divine intervention, powers generally deemed to be beyond personal control. However, this form of reasoning has now largely been surpassed. Lupton (1999) has highlighted the period of enlightenment, in the 17th and 18th centuries, as signalling the ascendance of the notion of controllability and measurability in relation to the natural and social worlds; rather than behaving randomly, these worlds came to be seen as following particular laws that were amenable to calculation, measurement and prediction through rational thinking. It is through observation of these developments that Beck (1992), as well as Giddens (1991), developed their theorising around the emergence of a 'risk society'. According to Beck (1995) and Giddens (1998), society has now moved from pre-modern times into an era of 'reflexive modernity'. Beck (1995) has likened this epochal change to a second enlightenment; the emergence of a society characterised by particular conceptualisations of, and expected reactions to, risk. However, the risks of reflexive modernity to which Beck (1995) and Giddens (1998) refer are unlike the risks from the natural world which troubled pre-modern societies. Instead, risk in reflexive modernity has evolved out of the combined processes of modernisation, industrialisation, globalisation and the associated expansion in human knowledge. More specifically, the development of nuclear, chemical and genetic technologies have brought with them new insecurities,

uncertainties and manufactured risks; they are the unintended by-product of humanity's accumulated knowledge and increasing attempts to control the natural world (Giddens, 1998). As well as the prevalence of risk, a further key feature of the risk society as set out by Beck (1992) is the emergence, and predominance of, expert bodies of knowledge. As the risks confronting us in the risk society are no longer natural dangers, but instead emerge out of technological and scientific developments, expert knowledge is required to identify, and manage risk.

Genetic risk is a key example of a risk that has emerged out of, and is managed through, expert techno-scientific knowledge. Whilst notions of hereditary and biological relatedness have long shaped conceptualisations of kinship (Featherstone et al., 2006), expansions in genetic knowledge and associated technologies have introduced the notion of genetic risk to the sphere of reproduction, and impacted on forms of identification between individuals (Rabinow, 1996). For Novas and Rose (2002) this dominance of expert knowledge about risk has led to new forms of identity and personhood; the genetically 'at risk' individual is a form of personhood which has arisen out of these social conditions, and is associated with particular obligations and socially acceptable behaviours, more specifically, the obligation to manage genetic threats (Novas and Rose, 2000). Recourse to expert knowledge is a key means through which this management is undertaken. Through the identification of mutations on specific genes, geneticists and researchers not only have the technological capacities to diagnose genetic diseases, but they may also ascertain the probabilities an individual has of

developing a specific condition/impairment, or of being a carrier for it, and thus the likelihood of its transmission to future offspring. Thus, the emergence of genetic counselling services, where individuals may obtain information regarding their genetic risks and strategies to manage them, can be regarded as an example of such expert knowledge with reference to which individuals can calculate and manage their genetic risks (Polzer, 2002).

Whilst expert knowledge is set out as an important resource by risk society theorists, and as a means by which risks can be managed, these bodies of knowledge, are not always accepted as infallible by those who use and are subject to them. The paradox of the risk society, Giddens (1990) argues, is that whilst expert knowledge may offer us the means by which to manage the risks which impinge on our daily lives, these bodies of knowledge have simultaneously *generated* the very risks we seek to manage. As our capacities to identify and manage risk have increased, so, with that knowledge, have the number of risks, and potential risks, in our lives increased: risk has now become an overwhelming feature in our lives. The number of risks to our health, for example, as Flynn (2006) has argued, may be experienced as ‘all-embracing and ever-extending’ (Flynn, 2006: 79) so that any behaviour or activity may come to be defined as a health risk at any given point. For example, in a context in which people are now less vulnerable than in previous eras to certain types of infectious diseases, and are consequently enjoying better long term health and longer life expectancies, there is increasing scepticism amongst the lay population as to how ‘real’ health threats actually are, and which should be accepted as so (Flynn, 2006). Whilst

risk society theorists such as Beck (1992) have approached an analysis of risk from the ontological standpoint that risks are pre-social and pre-existing phenomena, and in this sense have an objective existence outside our descriptions of them, he also acknowledged the importance of social and cultural factors in contributing to risk perceptions; what society considers to be a risk and in need of managing at any given point reveals social and political judgements, what Beck refers to as 'cultural disposition' (Beck, 1995: 47).

In terms of how people respond to risk, this contextual and socially constructed character of risk has facilitated a weakening of trust in the capacities of experts to define risks, and studies have demonstrated the way in which lay people, rather than unquestioningly accepting, make judgements about the trustworthiness of expert knowledge in the context of competing knowledge claims (Wynne, 1989; William and Popay, 1994). Knowledge may indeed be derived from a variety of sources, including one's own experiential knowledge, which is then factored into, and weighed against, expert knowledge, in determining risk perceptions. Whilst risk society theorists may thus prioritise the role of expert knowledge in the joint processes of generating and managing risks, studies have revealed that this dominance is nevertheless highly contested, and trust in this knowledge is incomplete.

Expert Knowledge: Geneticization

The dominance of expert knowledge in the risk society takes a variety of forms, and much has been written about the particular character this dominance has taken in relation to genetic knowledge. Indeed, since the completion of the human genome project in 2000, the status and implications of expert genetic knowledge has frequently been the centre of media attention, controversy and speculation. The appeal of the new genetics to the public imagination is reflected in the wealth of articles, books, public debates and films that have been produced on this topic which consider the implications of developments within this field for our freedoms, identities, health as well as society, both present and future. In line with, and fuelling, this burgeoning interest in genetics has been a growing tendency to attribute a wider range of behaviours and health states to genetic status. Claims that scientists had found the ‘gay gene’ (Conrad and Markens, 2001; BBC News, 2004) or the ‘obesity gene’ (BBC News, 2007) all reached the headlines in the past decade and point to the range of human experiences that have been framed in terms of expert genetic knowledge. The completion of the first draft of the human genome project was presented in the media and to the general population as a working draft of the ‘book of life’ (Nerlich et al., 2002). Replete with religious references, the conceptualisation of this expert knowledge as an information system containing definitive explanations for particular forms of illness and health, and even our very ‘humanness’ (Nerlich et al., 2002), exemplifies the power attributed to genetic knowledge in prescribing and dictating life itself (Kay, 2000; Nelkin and Lindee, 1995). Thus, upon

completion of the human genome project, the benefits this expert knowledge offered in terms of improving the human condition were extolled, and whilst some concerns were raised in the public domain about the dangers of genetic determinism, the completion of the project was nevertheless overwhelmingly presented as a breakthrough to be celebrated (Nerlich et al., 2000; Juengst, 2000). The gene emerged not only as a vessel of information passed between individuals, but as a 'cultural icon' (Duden and Samerski, 2007), containing within it the means by which to understand our relationships, behaviours and social problems, illnesses and diseases (Ten Have, 2001).

Abby Lippman (1991), through an analysis of the evolving status of genetic knowledge, has coined the term 'geneticization' (p. 64) to describe this vast expansion in genetic knowledge as it occurred towards the end of the 20th century, with the use of genetic explanations to account for a broad range of experiences and physical states. Building on the previously defined notion of 'medicalisation', developed by Zola (1975, 1977) and Illich (1990) in the 1970s to theorise the extension of the dominance of expert medical knowledge into different facets of life together with the attendant negative implications of this, the concept of 'geneticization' refers to a similar process of colonisation, but in this case of the idea that genetic knowledge can explain a broad range of physical and behavioural phenomena. As Lippman notes, 'geneticization' refers to:

The ever growing tendency to distinguish people one from another on the basis of genetics; to define most disorders, behaviours and physiological variations as

wholly or in part genetic in origin. It is both a way of thinking, and a way of doing, with genetic technologies applied to diagnose, treat and categorize conditions previously identified in other ways.

(Lippman, 1991: 64)

The concept of geneticization has been taken up by writers in different fields since the 1990s to explore the various ways in which expert genetic knowledge has become the dominant means through which to tell stories of health, illness and behaviour, and the implications this has for our identities, interpersonal relationships as well as the way in which we perceive, and respond to, risk.

Related to, and emerging out of the concept of 'geneticization' is that which Holtzman (1999) and Fleising (2001) have referred to as 'genohype', a term that has been used to describe the over-inflation of expectations and promises of genetic knowledge that have accompanied geneticization. As Nightingale and Martin (2004) have argued, expectations of, and investments in, the promises of genetic technologies (both financial and emotional), have far outstripped the reality of progress in this area. In spite of media and researcher claims in the late 1980s and early 1990s for example, that the discovery of the gene responsible for Cystic Fibrosis (CF) would bring about the possibility of developing gene therapies to cure the condition within twenty years, such a cure remains elusive (Stockdale, 1999; Wailoo and Pemberton, 2006). Indeed, the very development of the concept of genes as a form of therapy and cure can be viewed as part of the process of

geneticization; treatments and cures become conceptualised in terms of the altering of our DNA rather than previously accepted treatments. As Kakuk (2006) and Raab (1993) have argued, this notion supports the promise that, through the manipulation of our genetic make up, many of the major diseases afflicting humanity will be alleviated or, better yet, cured. Despite these hopes, however, many of the gene therapy trials over the past two decades have failed to produce viable treatments for debilitating and life-shortening conditions such as CF, which has been accompanied by an increased scepticism towards such scientific claims by the lay population (Kakuk, 2006; Nightingale and Martin, 2004).

A further key implication of the dominance of expert genetic knowledge relates to the ontological status of diseases. Yoxen (1982) has argued that it is in a particular social milieu wherein genetic explanations for disease comply with the 'institutional, professional and conceptual structural constraints of the modern health-care system' that they become acceptable, or even desirable (Yoxen, 1982: 148). Hedgecoe (2003) and Kerr (2000) have highlighted this process through their respective analyses of the construction of CF as a genetic disease. Kerr (2000) has noted that the acceptance of genetic explanations for CF, and the use of language to support this definition (e.g. the transformation of genetic codes into pathogens by use of the language of 'mutation') have, rather than tightening the boundaries of CF, instead facilitated the inclusion of associated syndromes into a CF diagnosis (Kerr, 2000; Wailoo and Pemberton, 2006). Through the identification of 'mutant' genes present both in men experiencing infertility and those

experiencing CF, certain forms of male infertility came to be re-classified as a form of CF, and subsumed within this diagnostic category, highlighting the power of genetic explanations for a given disease in defining the very boundaries of it. Kerr (2000) notes however, that whilst genetic explanations of disease may be appealing as a means by which to more clearly mark out disease boundaries and offer definitive diagnostic testing, the apparent fixity of genetic accounts can mask the 'dynamism' of genetic disease categories and the uncertainties inherent therein (Kerr, 2000: 870). As Kerr (2000) has argued, the inclusion of male infertility into the diagnostic criteria of CF is of questionable value to the men so-diagnosed and their families, particularly when treatment options are limited (Kerr, 2000: 871). Thus, whilst scientific pursuits to pinpoint genes deemed responsible for particular conditions are widely regarded as progressive and paving the way for future treatments or even cures, the work of Kerr (2000) and Hedgecoe (2002, 2003) has pointed to some of the complexities associated with this process of geneticization. In the case of CF, the introduction of genetic explanations has not brought with it the anticipated clarity and confirmation of medical classifications, but rather a far messier picture of the interplay between genotype and phenotype (Stempsey, 2006). Such studies point to the importance of geneticization in accounting for the ways in which genetic explanations for disease come to be singled out as the single underlying causal factor *in spite of* these uncertainties, and the complications that arise from this (Hedgecoe, 2002; Stempsey, 20026).

As well as the shifting ontological status of diseases, the changing role of expert genetic knowledge in the diagnosis and management of disease has, furthermore, had implications for understandings of identity, personhood together with interpersonal relationships. At a very basic level, the study of genetics is a study of groups of similar individuals, defined in biological terms (Yoxen, 1982), and thus the practices associated with genetic technologies mean that the biological basis of family and kinship has the potential to be attributed greater importance (Featherstone et al., 2006; Finkler, 2001; Richards, 1996a). Not only may the boundaries of family groupings be altered and redefined by genetic information, both in legal terms (e.g. the importance attributed to biology in defining what constitutes a parent) as well as social ones, but new social roles, expectations and obligations may be forged by such genetic knowledge. As Novas and Rose (2000) have argued, the increasing reliance on genetics to explain health and illness occurs in a context in which identity practices have shifted. They argue that increasingly, individuals are encouraged to adopt 'life strategies' that maximise their life chances (Novas and Rose, 2000: 487), but which are simultaneously imbued with ethical responsibilities to others. An example of this new form of identity has emerged alongside genetic knowledge; the person 'genetically at risk' is associated with particular subjectivities, more specifically, the duty to act in accordance with one's 'genetic responsibility' (Kenen, 1994). 'Genetic responsibility' is a term that refers to the impetus to act in the present in relation to genetic information in order to manipulate potential futures (Novas and Rose, 2000: 486), and highlights the way in which the effects of

geneticization may be felt beyond the conceptualisation of diseases, informing the way in which we understand our own identities and our relationships to others.

Various studies have explored experiences of genetic risk and responsibility and its management within families and communities (Hallowell, 1999; Kerr, 2003; Schaffer, 2008; Polzer et al., 2002; Novas and Rose, 2000; Richards, 1996a; Merz, 1987), and further, the gendered ways in which this responsibility is experienced. It has long been acknowledged that women take primary responsibility for 'kinship work' in family groups (Di Leonardo, 1987), and, moreover, assume responsibility for the family's health (Graham, 1979). However, Lippman (1994), Reed (2009) and Hallowell (1999) have argued that these forms of responsibility are both heightened and reinforced by genetic knowledge, and further, they can operate to constrain the autonomy of women. The assumption of 'genetic responsibility' (Kenan, 1994) may dictate not only the management of women's *own* genetic status (for example overriding their right *not* to know about their genetic status in order to facilitate the genetic knowledge acquisition of other family members (Hallowell, 1999; Downing, 2005)), but also the management of the genetic status of their offspring, through a sense of obligation to undergo invasive prenatal testing (Lippman, 1994). Moreover, the management of the genetic status of other family members, for example through surveillance or the recruitment of family members into screening or testing practices may be felt and experienced as an obligation rather than a choice (Downing, 2005; Hallowell, 1999; Featherstone et al., 2006). Thus, as Beck (1992) has

acknowledged in relation to the risk society, expert information on risk, and in these instances, genetic risk, is experienced differently across social groups, and such groups may have unequal access to resources with which to manage such risk. For women, expert genetic information confers on them particular forms of responsibility and obligation that, in some instances, prove to limit, rather than expand the choices they are able to make.

Whilst Lippman's (1991) study of geneticization, and the work of those who have taken up her concept, point to the range of implications associated with the encroachment of genetics on an ever increasing number of facets of life, both actual and imagined, the notion has nevertheless been critiqued and adapted (Ten Have, 2001). Some of these critiques have focused on the methodological difficulties associated with researching the concept (Hedgecoe, 2001; Ten Have, 2001), whilst others offer more substantive insights, concerned with the usefulness of the concept. In light of evidence of extensive public debate on the new genetics, Ten Have (2001) has argued that, at least to some degree, expert knowledge is not automatically accepted but is instead reflexively considered by those who may use such information. Further, Novas and Rose (2000) have argued that the undertones of colonization inherent in the concept overlook the active and creative responses of individuals to geneticization, and the possibilities within this process for resisting the dominance of expert knowledge. Rather than passive recipients of genetic discourse who accept the fatalism of their inheritance and the self-identities it suggests, Novas and Rose (2000) argue that individuals engage actively with the new 'molecular optics' (Novas and Rose, 2000: 487)

to intervene in, manage and manipulate their potential futures (e.g. Abel and Browner, 1998). Moreover, those who have, are at risk for, or who may be carriers of, a range of genetic disorders are now questioning the relevance of genetic information to their lives, and are bypassing testing when it is offered (e.g. Cox and McKellin, 1999; Qian et al., 2001; Tibben et al., 1992). This rejection or avoidance of genetic information suggests that there may be more possibilities for resistance to expert knowledge than the geneticization thesis suggests; that people do not always accept genetic practices and information to be useful, relevant and valuable in their lives in an unproblematic way. Rather, this information is interpreted in different ways in the context of their own lives.

Cox and Starzomski (2004), in a study of Autosomal Dominant Polycystic Kidney Disease (PKD), a kidney condition generally understood to be linked to specific genes and for which genetic testing is available, for example, have argued that this refutation of genetic knowledge by both lay people and health care professionals mitigates the process of geneticization. They argue that health care providers and those living with PKD see little need or use for genetic testing for PKD, a finding that they link to the availability of treatment options for the condition. The clinical management of those affected by PKD generally focuses on the monitoring, prevention and treatment of renal failure rather than the inheritance of PKD per se. Cox and Starzomski (2004) suggest that the hope and sense of control offered by the availability of treatments for those affected by PKD contrasts sharply with the fatalism associated with genetic explanations of the disease, and works to

prevent its acceptance as a genetic condition. This irrelevance of the genetic aetiology of PKD is further reflected in the lack of a disease specific support group for PKD (Cox and Starzomski, 2004: 162). The families who took part in Cox and Starzomski's (2004) study, for example, instead participated in groups that offered support to those affected by generic kidney disease such as the US based 'Kidney Foundation'. Thus, unlike the support groups explored by Rapp et al. (2001), such as Little People of America (LPA), where a strongly geneticized identity existed, those affected by PKD were not a group self-defined by their genetic status. Indeed, it can be argued that those diseases which appear most highly geneticized are amongst those for which treatments are primarily palliative rather than curative, or are largely ineffective, such as those offered for Tay-Sachs, Huntingdon's Disease (HD) and Duchenne Muscular Dystrophy (DMD). The degree to which genetic information is accepted and used by those experiencing different conditions therefore varies according to the status of the disease and whether effective treatments or cures are available.

Meiser and Dunn (2000) have further noted that a sense of fatalism, a factor that Cox and Starzomski (2004) suggest accompanies geneticization, can also compel those at risk for non-treatable progressive conditions such as HD to *avoid* pre-symptomatic testing and information, on account of the distress such genetic information could generate (Meiser and Dunn, 2000; Wright, 1996; Madigan, 1996). The possibilities for psychological distress and 'ethical problematisations' (Novas and Rose, 2000: 488) that emerge from the application of genetic knowledge may thus also contribute to the

lack of acceptance of the information. The health care providers in Cox and Starzomski's (2004) study, for example, reported reluctance in discussing the genetic/hereditary aspects of PKD with their patients on account of the potential they have to generate fear or distress, and indeed the association of genetic conditions with shame, guilt and stigma has been widely documented in the literature (Dragonas, 2001; Ettore, 2002; Chapple et al., 1995; Arribas-Ayllon et al., 2008a; Hallowell et al., 2006; Markel, 1992). These findings highlight that the degree to which genetic information about a condition is accepted is highly contingent; the availability of effective treatments for the given condition as well as the likely psychosocial impact of knowledge genetic status are key factors which mitigate against the acceptance of genetic explanations for a condition (Cox and Starzomski, 2004), but which, however, may not indicate a direct subversion of geneticization. For conditions such as HD, where acceptance of genetic testing is low, for example, there is still widespread acceptance of the genetic aetiology of the condition, suggesting that whilst personal genetic information may not be used, genetic explanations still largely shape the way in which particular diseases are conceptualised. A rejection of genetic information, therefore, does not necessarily imply a resistance to the genetic status of the disease, nor the incomplete domination of expert genetic knowledge, but rather points to the varying ways in which this knowledge is responded to in the everyday reality of people's lives.

Thus far, geneticization, or the prominence given to genetic explanations for disease over and above others, has been presented as an example of the significance attributed to expert knowledge in what Beck

(1992) and Giddens (1990) refer to as the 'risk society'. The implications this expert knowledge has, in terms of the hopes and expectations it perpetuates for the treatment of debilitating conditions, and the shifts it produces in terms of identities, personhood and disease boundaries have all been presented in order to contextualise my own research on SMA. Indeed, SMA is a condition that has, over the past 50 years, come to be understood through this lens of expert genetic knowledge. However, as both Beck (1992) and Giddens (1990) have acknowledged, whilst expert knowledge may be given precedence in the context of the risk society, this knowledge can nevertheless be resisted, challenged or discounted in various ways, as acceptance of the infallibility of such knowledge has decreased. The rejection of genetic information on account of the emotional consequences or 'ethical problematisations' (Novas and Rose, 2000) that it suggests for those who encounter it, as well as its relationship to treatment options have all been suggested as factors which may mitigate the dominance of such expert knowledge. One area of literature where the relationship of lay people to expert knowledge has been extensively explored is in the area of risk perception. As risks are both produced by, and managed through, expert knowledge in the risk society, the way in which risks are conceptualised and responded to by individuals is an arena in which the contested status of expert knowledge may be played out.

Expert Knowledge and Risk Perception

A body of literature has emerged documenting, and accounting for, the reactions of individuals to expert knowledge and their conceptualisations of this knowledge, in the context of risk across a variety of disciplines (Slovic, 2000). Health risks, in particular have received much attention; as Lupton (2006) and Flynn (2006) have highlighted, health risks are frequently the subject of media and public concern, and the way in which they are conceptualised by individuals is of particular interest to the medical profession in order to account for adherence (or otherwise) to health promotion strategies and treatments.

Within the psychological and scientific literature, perceptions of, and responses to, genetic risk have received considerable attention with a view to documenting the disparities between lay and professional accounts of genetic risk and uncovering how and why lay people understand and reproduce genetic risk statistics (Evers-Kiebooms, 1992; Edwards et al., 2002; Evers-Kiebooms and Van den Berghe, 1979). Studies within this field have also sought to document the factors which influence decision making and subsequent behaviours in the context of genetic risk, in particular, to account for the uptake or avoidance of health services and information (Shiloh, 1996; Wertz et al., 1992; Vleck, 1987). Such studies have taken as a starting point a definition of risk which situates it as a pre-determined objective fact; risks are 'real' in the sense that they exist independently of our interpretation of them, and expert knowledge is viewed as a resource which may be used to circumvent and manage such risks. Studies utilising this approach to an

analysis of risk have pointed to a variety of factors which affect the way in which genetic information is retained and used by the lay population (Hallowell and Richards, 1997); these factors include reproductive intentions (Ekwo et al., 1985), the timing and presentation of the genetic risk information (Sorenson et al., 1979), familiarity with genetic disease as well as subjective interpretations of risk (Pearn, 1973).

One of the critiques of this approach to analysing genetic risk, however, is its focus on assessing the way in which lay people produce 'faulty' accounts of expert knowledge. As Polzer et al. (2002) have argued, this approach can be seen as pathologising the choices lay people make about the risks which confront them and privileging the knowledge claims of experts in defining and managing risks. Social theories of risk, rather than defining risks as pre-existing phenomena to be communicated to lay populations by those with expert knowledge, have instead sought to demonstrate the way which risks are collectively constructed and experienced within a broader socio-cultural context. Instead of examining the 'faulty' nature of lay accounts of risk, those empirical studies that have taken up Beck (1992) and Giddens' (1990) approach to risk instead seek to demonstrate the way in which lay people make judgements about the trustworthiness of professional knowledge in the context of other competing knowledge claims, for example knowledge derived from their own everyday lives (e.g. Wynne, 1989; William and Popay, 1994). Studies of risk perception utilising this approach have therefore moved beyond an analysis of the match between lay and professional accounts of risk, to explore the way in which individuals

make sense of risk through recourse to different bodies of knowledge. However, whilst Beck (1995) and Giddens' (1990) approach to an analysis of risk and its relationship to expert knowledge has highlighted the way in which risks are, at least in part, socially constructed, it has nevertheless been argued that such macro theories of risk and risk perception overlook important aspects of the way in which risk is *experienced* by individuals and groups, and further, how widely accepted definitions and understandings of risk may even be rejected or reconstructed (Wynne, 1996).

Beck (1992) and Giddens (1990) made reference to 'reflexive practices' to document the way in which individuals reflexively evaluate which interpretations of risk they will accept. An *interpretive approach* to an analysis of risk perceptions, however, moves beyond these strategies to account for the various ways in which individuals actively and reflexively construct and respond to risk in relation to their situated knowledge and circumstances. Expert knowledge is evaluated by social actors, not in a vacuum, but instead in the context of their own complex biographies, life experiences, relationships and sense of embodiment, and it is by reference to these cumulative stocks of knowledge that professional definitions of risk are processed, and responded to. Experiential knowledge is thus key to this interpretative approach to an analysis of risk perceptions.

Experiential Knowledge

The term 'experiential knowledge' has been used by various authors to account for the 'experiential and particularistic' (Abel and Browner, 1998:

310) forms of knowledge which inform perceptions of risk and relationships to expert knowledge in a variety of contexts (Etchegary et al., 2008; D'Agincourt-Canning, 2005; Lippman, 1999). More recently, its role in informing decision making in prenatal care, testing and screening decisions has been explored (Etchegary et al., 2008; Lippman, 1999; Abel and Browner, 1998). Experience with a condition, furthermore, has been suggested as a factor in shaping how people arrive at genetic testing decisions and conceptualise genetic risk in relation to cancer (Hallowell, 2006; Werner-Lin, 2007; Kenen et al., 2003; Babb et al., 2002; D'Agincourt-Canning, 2005; Burgess and D'Agincourt-Canning, 2001), Huntingdon's Disease (Downing, 2005; Cox, 2003; Burgess and D'Agincourt-Canning, 2001; Cox and McKellin, 1999), X-linked conditions (Parsons and Atkinson, 1992; Kay and Kingston, 2002) and CF (Wertz et al., 1992; Evers-Kiebooms et al., 1988), demonstrating the relevance, and uses of, experiential bodies of knowledge across a wide range of conditions in the evaluation of expert knowledge.

The term 'experiential knowledge' has been used differently by researchers. Caron-Finterman et al. (2005) who have used the concept do not substantively distinguish it from 'lay knowledge', a form of knowledge already much researched within medical sociology (Wynne, 1996; Popay, 1996). Indeed, Caron-Finterman et al. (2005) simply use the term 'experiential knowledge' as a means by which to rebuff any suggestions of inferiority imbued in the term 'lay knowledge', which is defined primarily through its relationship to medical or scientific knowledge. Borkman (1976; 1990), however, suggests that experiential knowledge has two defining

characteristics: firstly, it is knowledge based upon the experiences of an individual, and secondly it is highly valued by the individual and deemed to be 'authentic', as it has been acquired through this individual's direct interaction with the physical, social and intellectual world. In addition to these defining characteristics, Borkman (1976) also emphasises the pragmatic uses of experiential knowledge: it is translated into strategies and methods for living with a particular problem, which is then pooled with the experiences of others, typically in the context of a self-help organisation (Borkman, 1976: 450).

More recent research, however, has moved away from a definition of experiential knowledge as a template for action, instead emphasising its contextual, subjective and emotional properties, as well as the different forms of experiential knowledge which exist. When defining experiential knowledge, Abel and Browner (1998) differentiate between two distinct types of knowledge: embodied knowledge and empathetic knowledge. Embodied knowledge refers to personal perceptions of bodily experiences and sensations (e.g. pregnancy), whereas empathetic knowledge is derived from close association with others living through a particular experience (e.g. caregiving). Thus, 'one derives from direct sensory experience, the other from close emotional ties between individuals' (Abel and Browner, 1998: 315). Abel and Browner's (1998) definition of experiential knowledge differs from that of Borkman (1976) in that they conceptualise such knowledge as a *process* rather than strategy, and one that is in an endless state of flux; continually accumulated and subject to revision across different social

contexts. Whilst Borkman's (1976) definition furthermore sets out experiential knowledge as an abstract end product or 'personal insight', which is of strategic use to people living with long term health conditions, Abel and Browner's (1998) definition incorporates those aspects of experience which may not have been subject to rational thought and processed into a prescription for action. Rather, experiential knowledge, according to this definition, includes unconscious awareness of bodily experiences, or even experiences acquired indirectly, for example, through the body and subjectivity of another person. D'Agincourt-Canning (2003), taking up this distinction of experiential knowledge set out by Abel and Browner (1998), has used it to separate the experiences of those family members who live alongside a relative who has cancer, from those who have cancer themselves. Whilst D'Agincourt-Canning (2003) recognises the possibilities of these two forms of knowledge becoming 'intertwined' as individuals may experience caring for a relative with cancer whilst simultaneously having the condition themselves, this distinction is nevertheless used to mark out two different ways in which people come to know cancer, which, she suggests, should be regarded as equal in status. Indeed, whilst it might be assumed that those diagnosed with cancer themselves are 'closest' to the experience of cancer, D'Agincourt-Canning (2003) is keen to emphasise that empathetic knowledge can be just as 'poignant' or 'real' as embodied knowledge of cancer (p. 151).

In their respective analyses, both Etchegary et al. (2008) and D'Agincourt-Canning (2003) have further expanded on this primary distinction of experiential knowledge, developing sub-categories of

experiential knowledge within 'embodied' and 'empathetic' knowledge. Etchegary et al. (2008), for example, have employed a quasi-visual metaphor to explore 'vivid' (personal), and 'vague' (more distant) forms of empathetic knowledge that pregnant women mobilise in their decisions about the uses (or otherwise) of prenatal screening and testing technologies. 'Vivid' forms of empathetic knowledge include having experience of caring for a child who has the disability which is being tested for, whereas 'vague' knowledge could refer to knowledge about the condition or the screening process gleaned from the media or the stories of more distant acquaintances (Etchegary et al., 2008).

For D'Agincourt-Canning (2003), 'empathetic knowledge' could be further sub-categorised into what she refers to as 'tangible knowing' (or the knowledge derived from physically living with someone affected by cancer), but also 'recent' and 'accidental' knowing, which account for the more distant ways in which people come to know about cancer in their family, e.g. through stories about unknown relatives. The concepts of 'distance' and 'closeness' in empathetic knowledge, moreover, have been outlined in the work of Kay and Kingston (2002) in their exploration of the reproductive decision making of female carriers of X-linked conditions. Through conducting interviews with women who had 'close' relatives (e.g. first degree relatives) with an X-linked condition and comparing the accounts of their reproductive decisions with women who had more 'distant' relatives affected by an X-linked condition (e.g. cousins, uncles), they suggest that proximity to

the experience of disability may be associated with higher levels of guilt and anxiety around reproductive decision making.

The different forms of experiential knowledge identified by various writers, determined in part by 'closeness' and 'distance' from a particular phenomenon, suggest that, in a similar way to expert knowledge, experiential knowledge may be subjectively appraised and accepted to varying degrees according to its perceived authenticity. The studies that have used the concept have asserted that experiential knowledge is an important resource through which expert knowledge is assessed and framed as well as challenged or resisted. However, the privileging of experiential knowledge as an alternative to, or site of negotiation, of expert knowledge, has also been critiqued. In the first instance, Abel and Browner (1998) have argued that a focus on everyday stocks of knowledge as an alternative to, or challenge to, expert biomedical knowledge, can be seen as romanticising the uses of experiential knowledge. Indeed, by emphasising the legitimacy of experiential knowledge as an alternative to expert knowledge, as Prior (2003) has argued, there is a danger of overstating its value. Through an analysis of the emergence of the 'lay expert' in relation to medical knowledge, Prior (2003) has argued that knowledge based on an individual's experience of a given condition is *necessarily* limited and idiosyncratic; restricted to the specifics of that person's situation, and can simply be 'plain wrong about the causes, course and management of common forms of disease and illness' (Prior, 2003: 45) . Whilst we may refer to experiential and lay knowledge as separate from expert knowledge, moreover, the two forms of knowledge may not

necessarily be clearly distinguishable (Markens et al., 2010). Through their study of pregnant women's decision making processes around prenatal testing for conditions that are routinely screened for such as Down's Syndrome (DS), Markens et al. (2010) have argued that a refusal of testing does not necessarily imply a rejection of expert biomedical knowledge and an acceptance of experiential knowledge, as many studies have suggested. Whilst some women in their study cited an embodied sense of their pregnancy being fine or 'safe' as a justification for by-passing testing, Markens et al. (2010) have argued that these women's very conceptualisations of a 'safe' pregnancy were imbued with expert medical knowledge about the nature of pregnancy. Experiential knowledge may thus be a means of appraising medical knowledge even as this expert knowledge frames and contributes to it in a 'synergistic' process, suggesting that it may never be possible to fully disentangle experiential and expert forms of knowledge in a meaningful way (Markens et al., 2010).

Issues around the status of different sorts of knowledge have further been debated extensively within the feminist epistemological literature (Hartstock, 1983; Hekman, 1997; Ramazanoglu and Holland, 1999; Code, 1991). In particular, for feminist standpoint theorists, the epistemological basis for, and status of, experiential knowledge, has political implications. Experiential knowledge, within the feminist literature, has been strongly correlated with women's ways of knowing and being in the world, in contrast with the 'abstract and universalistic' masculine ways of knowing associated with expert knowledge (Abel and Browner, 1998: 310; Code, 1991). The

status accorded to this knowledge is thus of political significance for feminist writers. Feminist standpoint theorists, following the work of Hartstock in the early 1980s, have argued that women, through their subjugated position in society, can offer unique insights into the experiences of oppression inaccessible to those not in this position as they offer a 'true' account of the internal workings of patriarchy. Consequently, the accounts of women should be prioritised in feminist research as authentic and valuable (Hartstock, 1983).

Whilst feminist standpoint theory has been criticised both from within and without feminism on the basis of the privilege it attributes to certain perspectives over others instead of acknowledging knowledge from different standpoints as simply *different* (Haraway, 1988; Tuana, 1993), the value and status of women's experiential knowledge, particularly in the negotiation of expert knowledge and risk, has political implications. These implications are presented below in relation to feminist debates around prenatal testing and screening.

The Politics of Experiential Knowledge

Feminism and Experiential Knowledge

As Beck (1992) and Giddens (1990) have argued, a defining feature of the risk society is the prominence of technology and expert knowledge, which have both created, but also provided us with the means by which to manage risk in various ways. In terms of reproduction, the development of maternal serum screening and ultrasound screening technologies amongst others, have

now become a routine part of prenatal care in modern western societies (Bankier and Cram, 2008). In line with these developments, new groups of women have come to be considered 'at risk' of having babies affected by particular conditions; women over the age of 35, those with a family history of an inheritable condition and those who have a positive (i.e. abnormal) screening result are now all considered 'at risk' of having a child with a condition or disability. Such women are thus encouraged to obtain, and make use of, expert advice and technologies to manage this risk, primarily through means of prenatal testing and selective termination.

For feminist writers, this move towards technological and expert intervention in child bearing has been responded to in different ways. Whilst some have argued that the availability of technology and expert knowledge extends the reproductive control and freedom of women, particularly in a context in which the responsibility for raising a disabled child falls primarily to women (Sharp and Earle, 2002; Brookes, 2001; Wertz and Fletcher, 1996), for others, the dominance of expert knowledge over, and intervention in, pregnancy has been viewed as a site of disempowerment for women.

Much of the feminist literature offering a critical evaluation of reproductive technologies, particularly as their use became more widespread from the 1970s onwards, has focused attention on the way in which the gradual encroachment of technological medical interventions into the sphere of reproduction has altered the way in which power and control over pregnancy are negotiated (Squier, 1994; Rowland, 1984). In particular, feminist writers have called into question the rhetoric of 'choice' which often

surrounds prenatal testing (Lippman, 1993), by highlighting the various ways in which the use of reproductive technologies, and compliance with expert medical knowledge, may be experienced as an obligation for pregnant women, rather than a choice. Indeed, feminist writers have argued that in a society in which women have historically been held primarily accountable for pregnancy outcomes and conceptually positioned as the ‘gene transmitters’ (Steinberg, 1996: 267; Dragonas, 2001; Rapp, 1999), the argument that prenatal testing is a free ‘choice’ is difficult to sustain. Responsible behaviour in pregnancy is primarily defined by the submission of pregnant women to medical surveillance, together with adherence to dietary and other restrictions (Ivry, 2007; Charo and Rothenberg, 1994), and for writers such as Rapp (1999) and Lippman (1991), it is in this context that prenatal testing comes to be experienced as an obligation (Franklin and Ragone, 1997; Farrant, 1985; Bailey, 1996). The ‘technological imperative’ of prenatal testing, created by virtue of its existence and availability, together with its offer by experts who may be considered to possess authoritative knowledge (Markens et al., 2010), may impose on women a ‘burden of not doing enough’ (Lippman, 1991: 28) to ensure the wellbeing of their pregnancy, as Clarke (1993) notes:

...an offer of prenatal diagnosis implies a recommendation to accept that offer, which in turn entails a recommendation to terminate a pregnancy if it is found to show any abnormality.

(Clarke, 1993: 1000)

Indeed, despite aspirations of non-directive counselling, there is evidence that the attitudes of health care professionals themselves may further reduce the choices of women being offered prenatal testing (McLaughlin, 2003; Marteau et al., 1994). Brookes' (2001) research discovered, for example, that whilst obstetricians aimed to be non-directive in their recommendations, many held firm beliefs that women should only have an amniocentesis test (see Appendix V for a description of amniocentesis) if they intended to terminate in the event of a 'positive' (i.e. abnormal) diagnosis, a belief that was communicated in subtle ways to participants in her study.

Abel and Browner (1998), in their study of women's experiences of pregnancy, have argued that this dominance of medical knowledge and technology, and the framework it provides for women through which to interpret their experience of pregnancy, directly contributes to the shape and nature of that experience (Squier, 1994; Corea, 1985; Katz Rothman, 1986; Arditti et al., 1984). Women adapt to the testing schedule of doctors, and moreover, come to distrust their own experiences of their bodies, in favour of medicalised reassurances (Lippman, 1991, 1999; Abel and Browner, 1998). For Katz Rothman (1986; 1985; 1984), this process fundamentally alters the way in which women experience their pregnancies, as they are encouraged to orient themselves around medical readings of their bodies, and thus may relate to their pregnancies only as 'tentative', and suppress their embodied experiences of the pregnancy, until medical validation of the quality and safety of their foetus is secured.

For some feminist writers, therefore, the encroachment of medical technologies and expert medical surveillance into the sphere of reproduction is regarded as having particularly negative consequences for the autonomy and reproductive freedoms of women. The reproductive choices available to women are seen as being constrained by the perceived obligation to undergo various forms of testing and intervention to manage reproductive risk, which can, in turn, have negative health implications for women, and have been regarded by some feminist writers as contributing to a relegation of women's bodies to the role of 'foetal containers' (Squier, 1996; Jordanova, 1985; Petchesky, 1987; Martin, 1989, 1998).

It is within this context that women's experiential knowledge of their bodies and their pregnancies has political value, and an assertion of women's rights to define such experiences on their own terms rather than through the lens of expert medical knowledge can be seen regarded as a political project. Studies such as that by Markens et al. (2010), Abel and Browner (1998), Dragonas (2001) Lippman (1999) and Etchegary et al. (2008) have paid particular attention to experiential accounts of pregnancy, and more specifically, the way in which knowledge accumulated from women's everyday stocks of knowledge, and more specifically, embodied experiences of their own pregnancies, and those of other women, are used to inform prenatal testing decisions. Typically, as Markens et al. (2010) have argued, studies have emphasised the way in which such experiential knowledge has become an *alternative* source of knowledge to biomedical knowledge, and a means of resisting authoritative expert knowledge of pregnancy. Through

trusting their own interpretations of their bodies and the experiences of those around them, reliance on experiential knowledge has come to be interpreted as a form of resistance to biomedicine's domination of pregnancy and an assertion of women's autonomy. Whilst, as has been stated previously, concerns have been raised within the feminist epistemological literature as to whether feminist writers should be concerned with which forms of knowledge are more 'accurate' than others, Hartstock (1997) has argued that there nevertheless may be ethical, social or political justifications for the privileging of particular vantage points over others, in certain contexts (Hartstock, 1997). By this, Hartstock (1997) had in mind the possibility of setting aside such objections to standpoint epistemology if it is possible that more equal social relations could be envisaged. As the encroachment of medical expert knowledge and technologies in reproduction may have particularly negative consequences for women, it is within the feminist literature that experiential knowledge, as a means to contest and challenge expert authority, has political dimensions. Whilst Beck (1992) has acknowledged within his theorising on the risk society that not all social groups experience risk equally, or have the same access to resources by which to manage risk, the gendered consequences of the elevation of expert knowledge and technological dominance within the sphere of reproduction highlights the ways in which the everyday realities of life within the 'risk society' may reproduce traditional ideas about gender. However, feminist writers have drawn attention to the way in which the intersection of risk with the sphere of reproduction has created an arena in which negotiation of these concepts may simultaneously take place.

Disability Rights and Experiential Knowledge

Feminist writers, however, are not the only group concerned with the way in which ideas of risk have entered the sphere of reproduction. Disability rights supporters have also raised concerns about the implications of the introduction and routinisation of prenatal testing and screening practices for the lives of disabled people. In particular, they have highlighted the problematic context in which this testing occurs by pointing to the (predominantly negative) way in which the lives of disabled people are valued and represented in wider society (Parens and Asch, 2000). The ever-increasing number of conditions and impairments that are being tested for, and thus the expanding number of pregnant women deemed to be 'at risk' of having a child with an impairment, moreover, has meant that medical professionals are frequently being called upon as a source of expert knowledge. This knowledge concerns not only medically defined risks to future offspring, but also the nature and severity of the condition in question, in order to support prospective parents' informed decision making. The responses of disability rights supporters to this situation have coalesced around two central concerns: firstly, whether the practices surrounding prenatal testing and selective termination can be understood as 'eugenic', and secondly, whether prenatal testing communicates a negative valuation of what it means to be a disabled person. The experiential knowledge of disabled people has been validated by some disability rights supporters as a particularly valuable political tool within the context of these debates (Fletcher, 2002) as it is this source of knowledge which poses the most

fundamental challenge to the perceived restrictive medical model representations of disability which are deemed to be perpetuated by expert medical knowledge. Indeed, a resistance to such ‘medical model’ thinking about disability has formed the backbone of much of the theorising of disability rights supporters and writers as well as the grassroots political activity of the disability rights movement, in the form of the ‘social model of disability’, which is presented below.

Within the disability rights literature, there is a strong division between what has been termed ‘social model’ thinking about disability, and ‘medical model’ thinking (Oliver, 1990). Whilst writers such as Shakespeare (2008a) have suggested that these binary divisions may be overly simplistic as there is more overlap between the two models of disability than the terms imply (and indeed, many internal contradictions within the models themselves), these two models of disability nevertheless remain largely distinct in much of the literature on disability, and at the very least serve a symbolic or heuristic function to differentiate between two polarised conceptions of disability. On the one hand, supporters of the medical model of disability (which is regarded as the traditional and dominant model of disability, and one espoused and supported by expert medical knowledge) define disability as the malfunctioning of an individual’s body and deviation from perceived normal functioning. Oliver (1996a) has aligned the medical model of disability with a narrow and inherently negative interpretation of disability; disability is perceived as a ‘tragedy’ that has befallen a person, and an experience which is largely negative. In line with this reasoning, supporters

of this approach to disability have viewed medical interventions as the most appropriate means by which to manage disability, more specifically, curative and therapeutic medicine (Oliver, 1990; Shakespeare, 1996). Social conceptions of disability, however, emerging out of grassroots disability activism during the 1970s and theoretically articulated through the work of Mike Oliver (1990), transformed many of the medical model notions of disability, instead defining disability as the social product of a society which fails to meet the needs and rights of disabled people. Disability came to be viewed as a social product that occurs when inaccessible environments and prejudicial social attitudes prevent disabled people from full participation in society. Through a separation of the 'impaired body' from the socially created 'disability', social model of disability theorists asserted that disability, and not impairment, is inherently problematic and should be remedied through social transformation (Shakespeare, 2006). For many people with disabilities, this move of the locus of the problem of disability from the impaired body onto society was experienced as liberating, as the onus was shifted from disabled people onto society to remove experiences of disablement (Morris, 1991).

The sharp contrast between social and medical models of disability, however, has contributed to the suspicion and hostility that many staunch supporters of the social model of disability feel towards medical approaches to disability (Shakespeare, 2008). Expert medical knowledge is deemed to belong to an oppressive and outdated conceptualisation of disability, which should be resisted and challenged in order to envision a society free of disablism. The responses of some disability rights supporters to the

introduction of prenatal testing and screening technologies which detect particular types of disability have thus mirrored this broader distrust of medicalised approaches to disability. Rock (1996) for example, has argued that such practices can be understood as a form of modern eugenics (p. 121), designed with the goal of eliminating disabled people as a social group. As the impaired body is deemed to be the problem, as opposed to social conditions, the removal of disabled people through screening and testing has opened what Duster (1990) has referred to as a 'back door' to eugenics, whereby the gradual elimination of people with specific impairments is being normalised through technological interventions.

Objections to prenatal testing and screening on the basis of their eugenic origins are not arguments, however, widely accepted by disability rights supporters. Writers such as Shakespeare (2008a) for example, have favoured a more graduated interpretation, suggesting that eugenic outcomes may be an 'emergent' or unintended consequence of screening practices, rather than regarding them as part of a straightforward 'hunt and destroy' mission of modern medicine (Shakespeare, 2006: 88; Sparrow, 2008;). The arguments put forward by Shakespeare (2006, 2008a) and Sparrow (2008) highlight that the issues surrounding screening and testing practices are complex, and there are suggestions within the literature that these practices are responded to very differently by different groups of disabled people (e.g. Gow, 2000; Guillemin and Gilman, 2006; Asch, 1999, 2000). Indeed, there has been extensive debate within the disability rights literature as to the meaning and consequences prenatal testing and selective termination have for

disabled people (Parens and Asch, 2000). The term the ‘expressivist objection’ (Asch, 1999, 2000) for example, has been developed to refer to the objection some disabled people hold to the negative valuation of disability deemed to be communicated by prenatal screening, testing and selective termination (Asch, 2000). By focusing on disability as the singularly most important characteristic of an unborn foetus, Asch (2000) has argued, and to make judgements about the potential quality of the child’s life based solely on this characteristic is to reaffirm medical approaches to disability. Within such medical approaches, disability is considered to be a wholly negative trait, leading to lives governed by ‘pain, burden and personal tragedy’ (McLaughlin, 2003: 300; Hubbard, 1996; Morris, 1991; Wendell, 1996; Kaplan, 1993). For Asch (2000) moreover, prenatal testing practices attribute not only a negative valuation of the particular foetus being tested, but of *all* disabled people, as it communicates the idea that it would have been preferable had they not been born at all. For Asch (1999), such an evaluation of disability is based on ignorance and overlooks the predominately positive experiences of childhood disability that are reported in the literature (e.g. Ferguson et al., 2000).

The expressivist objection to prenatal testing, however, has been critiqued both from within the disability rights movement and without (Sparrow, 2008; Shakespeare, 2008; Edwards, 2004). In particular, as Shakespeare (2008a) has argued, a distinction should be drawn between overtly prejudicial or ignorant attitudes towards disability and the, often constrained, social contexts in which prospective parents make decisions

about having a disabled child. Indeed, he argues, it is feasible that an individual may positively value the lives of disabled people whilst simultaneously acknowledging that they do not have the personal and/or financial resources required to adequately care for a disabled child who may remain permanently dependent on them (Shakespeare, 2008:97). Sparrow (2008), furthermore, has suggested that the arguments championed by supporters of the expressivist objection focus too narrowly on the decisions and decision-outcomes of would-be parents facing prenatal testing, as opposed to the policies from which these decisions emerge. Rather than framing the critique of prenatal testing in terms of the relationship of prospective parents to their future children, Sparrow (2008) proposes a shift in critique to the relationship between disabled people and the (largely) non-disabled people who make decisions about research findings and testing policies; in short, he has argued that the debate about prenatal testing and disability rights should be recast as a critique of relations between social groups rather than an issue arising in and through the relationship between prospective parent and child (Sparrow, 2008).

In spite of these critiques of the expressivist objection to prenatal testing and selective termination, however, there is evidence that this response to the practices can nevertheless cause particular emotional, ethical and social dilemmas for disabled people in various forms (e.g. Gow, 2000; Kent, 2000; Saxton, 1984; Atkinson, 2008; Bowler, 2006). Whilst theoretical inconsistencies within the expressivist objection to prenatal testing argument have been highlighted by different authors, the accounts of disabled people

who have encountered prenatal testing in different ways within their own lives, suggest that emotional responses to it are rarely formed through the lens of theoretical abstraction, but are rather grounded in everyday experience and subjective interpretation (Kent, 2000; Saxton, 1984; Atkinson, 2008; Bowler, 2006).

The political concerns raised by disability rights supporters around prenatal testing, including expressivist and eugenicist objections, have given leverage to the calls by some disability rights supporters for the greater inclusion of the perspectives of disabled people in the practices surrounding testing and counselling (Fletcher, 2002; Asch, 1999; Alta Charo and Rothenberg, 1998; Saxton, 1999). Given that the value of life with disability is one of the factors prospective parents are encouraged to assess when making decisions about the uses of prenatal testing and selective termination, disabled people, it is argued, are in a unique position to contribute their experiential knowledge of life with that disability, to counter-pose or supplement medical expert knowledge (Shakespeare, 1998: 673). Indeed, Williams et al. (2002) have argued that there has been a tendency within the medical profession to focus primarily on the medical complications associated with particular disabilities, rather than other aspects of life with the condition when presenting information to prospective parents. Similarly, Kelly (2009: 82) and Asch (1999) note, there has been little crossover of the literature on prenatal testing and the growing literature which challenges many of the commonly held assumptions about the experiences of disability within families (e.g. Ferguson et al., 2000; Ali et al., 2001). Whilst there have

been some moves to incorporate experiential accounts of life with disability into the information provided during prenatal testing and screening through website resources such as www.antenataltesting.info (a web resource set up so that prospective parents may access interviews with, and accompanying photographs of people living with the conditions for which prenatal testing is currently available in the UK), there remains little evidence that these accounts are being included in medical advice and information, and medical models of disability continue to dominate prenatal testing consultations (Williams et al., 2002).

Whilst the validation of experiential knowledge as an alternative source of knowledge has political significance for disability rights supporters in a society in which expert professional knowledge dominates, however, the status of experiential knowledge within the broader political project of the social model of disability has been contested. Indeed, as Paterson and Hughes (1991) Morris (1991) and Crow (1996) note, social model of disability theorists have traditionally excluded from their analysis experiential aspects of impairment (i.e. what it feels like to live in an impaired body) in favour of an analysis of the social and economic constitution of disability. To acknowledge impairment effects, such as pain and fatigue, it has been suggested, is to risk confirming medicalised conceptions of disability; that the problem of disability had been impairment all along, rather than inaccessible social and physical environments. Oliver (1996a) has tentatively suggested the development of a 'sociology of impairment' as an arena in which to interrogate these experiential aspects of impairment, including pain,

discomfort and malaise whilst retaining a critical distance from the medical model of disability which has traditionally been concerned with them (Oliver, 1996a: 49). Paterson and Hughes (1999), taking up this work, have attempted a politicization of the experiential aspects of disability by developing a sociology of impairment through a phenomenological lens, demonstrating that oppression is not only located in the fabric of society, but also in the 'flesh and bones' of disabled people; oppression is an embodied experience (Hughes and Paterson, 1999: 606).

Whilst the value of experiential knowledge as a means by which to challenge dominant forms of knowledge has thus been a highly contested topic within disability politics, and to a higher degree than within the feminist literature presented previously, over the past two decades, there has been an increased acceptance of the political significance of such knowledge (Morris, 1991; Paterson and Hughes, 1999). Indeed, the disability rights response to prenatal testing is a key example of an arena in which such knowledge has political significance, and, despite its exclusion from mainstream social model theorising, has been suggested as a means by which to challenge the same medical model conceptions of disability that social model theorists seek to overturn.

Conclusions

In conclusion, I have set out within this chapter some of the bodies of literature that surround and inform this study, in order to contextualise and locate my own research. I have presented Beck (1992) and Giddens' (1990) theorising around the development of a 'risk society', which has suggested that we now live in a society in which the management of risk has become imperative. The combined processes of globalisation, industrialisation and increased use of technologies have not only brought risk into our everyday awareness, but the technologies themselves have also been suggested as a means by which to manage these risks, in conjunction with specialised expert knowledge. The increased importance attributed to genetic codes in explaining a broad range of behaviours, traits and impairments may be regarded as a key example of the way in which such expert knowledge, in this instance, genetic knowledge, and associated technologies, has come to dominate the way in which we account for a plethora of states and conditions, particularly those which are deemed to be problematic by society. Lippman (1991) has coined the term 'geneticization' to refer to this domination of expert knowledge, which has impacted not only on the way in which diseases are defined and responded to, but has also given rise to new forms of personhood and 'genetic responsibility', as we are called upon to manage genetic risks to our person as well as those to our (future) kin (Novas and Rose, 2002; Kenen, 1994). Whilst new responsibilities and ways of understanding, and responding to, health states have been suggested by the domination of genetic knowledge, however, this is not to say that such expert

knowledge has not been contested. Indeed, Giddens (1990) has argued that in the context of reflexive modernity, individuals are in a position not only to think reflexively about the forms of knowledge they accept, and those they do not, but also to critically evaluate different knowledge claims. Whilst expert knowledge claims may dominate the way in which particular phenomena are defined and approached, risk society theorists have also highlighted that this knowledge is not infallible, and may be open to challenge and contestation from different sources.

Experiential knowledge, or knowledge derived from the everyday stocks of knowledge, subjective interpretations and meanings accumulated by individuals through their daily realities has been highlighted in the literature as one site in which expert knowledge has been challenged and even subverted (Wynne, 1996; Abel and Browner, 1998). Using an interpretive perspective, which has been acknowledged as providing the theoretical structure by which to interrogate subjective experiential accounts, a broad range of literature has emerged, particularly within medical sociology, wherein the possibilities for experiential accounts of health and illness to provide a critique of medical knowledge has been emphasised, and lay accounts validated as an alternative source of knowledge (Gabe, 1995). Reproductive decision making, particularly in the context of medically defined genetic risk, is an arena in which many of these contestations and negotiations between expert and experiential knowledge take place. Indeed, the feminist literature has highlighted the way in which the domination of expert medical knowledge can have a negative impact on the way in which

women experience and relate to their pregnancies (Katz Rothman, 1986; Lippman, 1993), and that in this context, experiential stocks of knowledge may be called upon by which to process, challenge or subvert such knowledge (Etchegary et al., 2008; Abel and Browner, 1998; Lippman, 1999; Markens et al., 2010). Similarly, experiential accounts of disability have been posited by disability rights supporters as offering an alternative reading of disability within the context of reproductive decision making and genetic risk, and a means by which to challenge medical model thinking about disability, which currently dominates (Fletcher, 2002; Asch, 1999). There has, however, been little crossover of the feminist and disability rights literature on prenatal testing, and indeed, some have argued that there may be ‘irreconcilable differences’ (Sharp and Earle, 2002) between these two perspectives, as a woman’s rights to determine her reproductive future has been posited as incompatible with disability rights supporters’ assertions that termination on the grounds of disability represents a devaluing of that particular impairment, and of disabled people as a whole.

The internal tensions, within, and contradictions between these two perspectives, as well as the way in which they are negotiated alongside expert genetic knowledge in the context of reproductive decision making will provide the grounding and backdrop to my own research. By focusing on the experiential knowledge of families affected by an inheritable condition, and the way in which this knowledge conflicts, and intersects, with medical knowledge, my study will explore some of the internal tensions highlighted by feminist and disability rights supporters. As Kelly (2009) and Gow (2000)

have argued, the experiential accounts of families and individuals living with conditions which can now be prenatally tested and screened for, and the way in which these accounts inform approaches to reproductive decision making, have been relatively under-researched. Where such experiences have been explored, there has been a tendency to focus on those conditions where there is a traceable family 'legacy' or recurrence of the condition within the family, such as cancer or HD, from which individuals accumulate their perceptions of the condition and perceptions of risk (Hallowell, 2006; Werner-Lin, 2007; Kenen et al., 2003; Babb et al., 2002; Kelly, 2009). Far less attention, however, has been paid to the value of experiential knowledge in families where there is not a long family history with a particular condition, such as SMA, where most parents did not know they were carriers for SMA until they had an affected child, and thus must negotiate the 'epistemic shock' (Kelly, 2009: 94) of having an (unanticipated) seriously impaired child, or developing a serious condition themselves in adult life.

Where experiential knowledge has been the focus of studies on reproductive decision making, the emphasis has furthermore either been placed on those who have 'empathetic' experiential knowledge of the condition or impairment being tested for (e.g. Etchegary et al., 2008; Parsons and Atkinson, 1993; Kay and Kingston, 2002), or those who have been diagnosed with it themselves (e.g. Gow, 2000). Very few studies have explored the intersection between these perspectives (D'Agincourt-Canning, 2005), or the reproductive decision making of those who have an inheritable

condition themselves (Gow, 2000), in order to explore the evaluation of knowledge claims from different standpoints.

This study will therefore draw together these different strands of the literature at the point at which experiential and expert knowledge, in their various forms, intersect, conflict and challenge one another through use of the example of reproductive decision making for families affected by an inheritable condition, SMA.

Chapter 2

The Research Methods

This chapter will outline the main aims of the study as well as detailing the research process through its various stages, from the initial research design to the practicalities of fieldwork, the analysis and finally, the reporting of the data. Particular attention will be paid to the theoretical, practical and ethical issues which arose during the course of the research and how these various concerns were addressed and managed. Not only are the claims of research important, but also the processes through which these knowledge claims are acquired and interpreted (Altheide and Johnson, 1998). The chapter will therefore also include a reflexive account of my own role as a researcher and the way in which my identity, biography and values additionally informed the development and direction of the research process, thus ‘writing myself’ into the research process in an attempt to render these structuring influences transparent (Seymour, 2001a).

Aims

The main aim of the study is to examine the influence and importance of experiential knowledge of SMA in the reproductive decision making of those from families affected by SMA. Through an analysis and comparison of family members’ accounts of the impact of SMA on their lives, the study considers the role of personal involvement with SMA as well as the level of severity of the SMA in shaping experiential knowledge of the condition to consider how this confirms or conflicts with medical knowledge of SMA.

Such an analysis will be undertaken in order to trace the way in which experiential knowledge affects family members' interpretations of and responses to the widely documented social and ethical dilemmas associated with selective reproduction practices that are available to families affected by inheritable conditions.

Research Questions

Whilst addressing the broad question of:

In what way does experiential knowledge of SMA inform conceptualisations of genetic risk and reproductive decision making in families medically defined as 'at risk' of transmitting SMA to future generations?

The study also examines the following secondary research questions:

1. What are the main concerns and challenges faced by families and individuals affected by a diagnosis of SMA?
2. Is SMA perceived differently by those with a diagnosis of SMA, in contrast to the perceptions of their family members?
3. How do families and individuals affected by SMA relate to medical definitions of SMA?
4. To what extent are the concerns of disability rights activists about prenatal testing and selective termination reflected in the views and concerns of families affected by SMA?
5. How do families and individuals interpret the value of medical knowledge vis-à-vis their experiential knowledge in reproductive decision making?
6. How are notions of reproductive and relational responsibility negotiated in the context of experiential and medical knowledge of SMA in reproductive decision making?

Methodological Considerations

I decided to adopt a qualitative approach to the research in order to capture, in depth, the viewpoints of individuals living with SMA in terms meaningful to them. Qualitative research has been recognised as being particularly well suited to these ends in terms of allowing an analysis of detail and depth with an emphasis on meaning and understanding (Bryman, 1988). As May (2001) has pointed out, however, it is important to note that the approach taken to research necessarily reflects underpinning epistemological and ontological assumptions as to the nature of the reality of the phenomenon under study. As the aim of the study was to capture the way in which SMA is experienced and understood by those who live with it, a theoretical approach to research was required that regards individuals as active agents who ‘construct the meaning and significance of their realities’ (Jones, 2004: 257), rather than assuming shared understandings. An interpretative approach, and one closely associated with qualitative methodology, is particularly well suited to these ends. This approach, following the theorising of Max Weber (1949) takes the subjective constructions of reality espoused by social actors as the starting point for research. Rather than exploring pre-determined theoretical constructs, those working within this paradigm derive their understandings of the social world directly from the everyday life worlds of individuals (Schutz, 1979).

‘Common-sense’ understandings of the social world are viewed as worthy of analytic attention from an interpretive perspective as they are deemed to reveal much about the way the social world is co-created in an inter-subjective process between individuals, rather than existing as an objective

and pre-determined phenomenon. Everyday understandings and interpretations are thus deemed to *constitute* the social world, and an interpretive perspective is thoroughly grounded in these subjectivities (Garfinkel, 1967).

The use of an interpretive approach has implications for the choice of methods, particularly for analysis. As social actors are deemed to actively and creatively construct the social worlds around them, a grounded theory approach to data collection and analysis is often employed (Charmaz, 2003a). Based on the theorising of Glaser and Strauss (1967), a grounded theory approach emphasises the emergent, constructed nature of reality, thus data analysis strategies using this approach are inductive, allowing concepts to arise out of the data themselves, and theoretical frameworks to be constructed from them.

Whilst an inductive approach to data collection and analysis has been critiqued for its atheoretical nature (Scambler, 1987), for an exploratory study with an aim of understanding the way in which families affected by SMA experience their lives and the meanings they attach to reproductive genetic technologies, an approach which allowed such concepts to emerge from the data rather than through the researcher's preconceived ideas of what it means to live with SMA was deemed most appropriate. Whilst Thomas and James (2006) point out that it may never be entirely possible to 'bracket off' one's preconceived ideas about what the data contain, the adoption of a grounded theory interpretive approach and a close analysis of the data enabled me to

think reflexively about what I brought to the analysis as a researcher and to be open to the challenging of these preconceptions.

Practical Considerations

SMA was chosen as a condition to study as it is the most common recessively inherited condition in the UK after CF, and a condition for which carrier testing, diagnostic testing, prenatal testing and PGD (Pre-Implantation Genetic Diagnosis) is currently available. SMA, furthermore, has a wide variety of presentations, which means that comparative analysis could be undertaken, particularly of the different diagnostic subcategories of SMA (types I-III). Unlike other common genetic conditions, such as CF and DMD, however, SMA has been under-researched in the social science literature. Indeed, I was only able to identify one previous qualitative study from the field of rehabilitation medicine which specifically focuses on the experiences of those living with SMA (Lamb and Peden, 2008). As Gow's (2000) attempts to contact women with CF were hampered by her discovery that young people with CF had already been 'much researched' (Gow, 2000: 111), I speculated that an absence of previous research might positively impact on participants' willingness to participate in the study.

Through preliminary research into the condition, together with prior knowledge of SMA through having a close friend with the condition, I identified the possible routes of sampling families and individuals affected by SMA as being through dedicated neuromuscular clinics, or through the national charity for SMA in the UK, the Jennifer Trust for SMA (JT SMA).

However, after making some initial enquiries with the head of the Dubowitz Neuromuscular Centre, London, (the largest neuromuscular clinic in the UK with a specialist interest in SMA), I was advised that the clinics run for SMA are primarily paediatric, and that adults with SMA have far less contact with clinicians. As I wanted to include both families with young children, but also adults living with SMA in the study, the JTSMA (a national charity which currently supports approximately 2,000 families and individuals affected by SMA) was contacted, informed about the proposed research, and readily agreed to offer support (JTSMA, 2010).

As well as methodological and theoretical considerations when designing the study, the practical considerations associated with having a neuromuscular condition myself, as well as researching others affected by neuromuscular disease had to be accommodated. As the JTSMA is a charity based in a town near to my institution, with its annual conference held within easy driving distance, this was particularly well suited to my own mobility restrictions, as well as the management of other impairment effects such as fatigue and pain which are exacerbated by travel. As members of the JTSMA are geographically dispersed, many with young children or managing a condition resulting in complex needs, the means of interviewing needed to be as flexible as possible. For this reason, participants were offered a choice of a face to face, e-mail or telephone interview depending on their needs, preference and abilities. There was also flexibility over when the interviews took place, to allow for care needs and family commitments, and consequently many took place late in the evening or at weekends.

The Interviews

In-depth interviews were chosen as a means of producing data. In-depth interviews have been documented as offering researchers the means of obtaining 'deep understanding' (Johnson, 2001), particularly when researching lived experiences. As Johnson (2001) has noted, in-depth interviews allow the researcher to:

...explore the contextual boundaries of that experience or perception, to uncover what is usually hidden from ordinary view...or to penetrate to more reflective understandings about the nature of that experience.

(Johnson, 2001: 106)

Whilst in-depth interviews are typically unstructured in nature, I aimed to generate both 'thick description' (Geertz, 1973, 1988) of life with SMA, but also participants' perspectives on very specific issues, such as their views on the availability of genetic testing for SMA and their thoughts and feelings about having children. Four pilot interviews were conducted with three personal contacts (one participant was interviewed twice) who had experience of SMA to elicit feedback and discussion around the type of interview questions that would generate the sort of data I hoped to analyse. This proved to be a productive exercise as it highlighted relevant issues to those living with SMA, and helped me develop a more informed and sensitive interview guide.

Following the pilot study, the interview questions were refined so that after demographic information (age, gender, ethnicity, disability, geographical

location, occupation) was noted and introductory questions, or ‘ice breakers’, were asked (e.g. ‘can you tell me a bit about yourself?’), all participants were asked the question, ‘can you tell me the story of how your life has been affected by SMA?’. This open-ended question allowed participants to direct the interview to the issues they felt were significant, in terms meaningful to them, which has been documented as one of the main advantages of using unstructured interviewing techniques (Bryman, 1988). Participants were encouraged to explore topics that they felt were particularly important to their experience of life with SMA, which elicited broad discussions, however, during the second half of the interview, an interview guide (see appendix III) was used to prompt for answers to specific questions. In many instances, the stories told of life with SMA led into the topics set out in the interview guide without need for prompts. However, where this was not the case, participants were asked specific questions on their views on the severity of SMA, their feelings about the availability of genetic testing technologies (both for SMA and other conditions) and their ideas about reproductive decision making in the context of prenatal testing for SMA. Whilst an interview guide was used, the interviews were flexible enough to accommodate participants’ diversions into different topic areas, and further, the use of hypothetical scenarios to broach sensitive topic areas proved to be a useful strategy, particularly in situations where participants were not comfortable discussing their own experiences directly.

In total, 59 interviews were completed with 61 participants which lasted on average 1 hour and 10 minutes each, with the shortest being 45

minutes and the longest 2 hours and 57 minutes. The interviews via e-mail took place over periods lasting from three weeks to eight months. All interviews were tape recorded and transcribed verbatim and for e-mail participants, e-mail communications were gathered and compiled into single documents. All names and specific place names (e.g. names of towns or hospitals) were removed or changed during transcription to avoid identification. However, names of countries, where referenced, were left in to highlight the different national contexts in which SMA was experienced. Care was taken to ensure that all interviews were transcribed within two weeks of completion of the interview and the transcripts were annotated with my observations and thoughts from the interview. Most of these supplementary reflections had been recorded in my research diary, but some were noted during the process of transcription itself. Notes were also taken during the process of transcription, recording the themes and patterns that I felt were emerging from the data as the interviews evolved.

Consent to undertake the interview and a reiteration of their rights to withdraw and withhold information during the interview was agreed with all participants prior to the interview taking place. A consent form (see appendix II) was signed by all participants who took part in a face to face interview, whereas for e-mail participants this was done by a typed 'signature', and a verbal acknowledgement by those who took part in telephone interviews (this will be returned to later). Once the interviews had been carried out, the audio files and e-mails were removed from my computer and transferred to discs which were stored, together with completed transcripts and the names and

addresses of participants, in a lockable cabinet to which only I had access, to ensure as far as possible, the confidentiality of participants.

Table 1: The Interviews

Type of Interview	Number of Participants
Telephone Interview	44
E-mail Interview	10
Face to Face Interview	5
Total	59**

*** Four interviews were joint interviews, and one participant was interviewed twice: first alone and then with their sibling.*

Telephone interviewing has been shown to offer many advantages over face to face interviewing; including logistical and economic benefits (Rhode et al., 1997; Sturges and Hanrahan, 2004) and the potential to include those who may have been hesitant to participate in a face to face interview, an issue which may be particularly relevant for sensitive research projects (Fenig and Levav, 1993). As Sturges and Hanrahan (2004) have argued, it is equally as possible to develop rapport during in-depth interviews conducted over the telephone as it is in face to face interviews. However, this method of interviewing, in practice, presented me with particular technological difficulties. Firstly, there were problems in ascertaining the quality of the sound recording prior to the interview, and despite stopping and re-starting the interview to check the recording (in itself a disruptive exercise), five interviews had to be repeated due to unintelligible recordings. Secondly, as Creswell (1998) has argued, in the case of telephone (as well as e-mail interviewing), the loss of nonverbal communication can pose difficulties in

interpretation. It was difficult to know, for example, how to respond appropriately when participants fell silent during a telephone interview and to accurately detect emotional distress.

Difficulties with reading emotional responses, and in particular being attentive to signs of emotional distress, can also pose problems for e-mail interviewing as auditory cues (e.g. sighs, hesitation) are further absent. McCord and Schwaber Kerson (2006) however, through their interviews with women who had undergone termination of pregnancy for foetal anomaly, have argued that the disadvantages of e-mail interviewing, such as the invisibility of visual cues, are outweighed by the advantages they offer, including the obviation of transcription and travel costs. For example, e-mail interviews can be conducted over several months, introducing a longitudinal element to studies and asynchronous communication allows participants to respond in 'instalments', at times and dates convenient for them, which works particularly well for research on topic areas that are potentially emotionally demanding, or for participants with time constraints. In the case of my study, the flexibility this format permitted was useful as the majority of participants were managing complex disabilities and/or had young children. Rather than obtaining a 'snapshot' of their lives, furthermore, the e-mail interviews allowed me to gain more insight into participants' perspectives over several months. In three instances, the interview period covered a time of major upheaval and distress in their lives. Whilst these participants were offered the opportunity to withdraw from the study, all three opted to continue the

interview and reflected on the changes in their lives in their responses to my questions.

As Kivits (2005) has noted, however, the timescales involved in e-mail interviewing can make it harder to draw a close to the interview upon completion, a situation which has been described as commonplace across sensitive research projects (Dickson-Swift et al., 2008; Lee, 1993; Hubbard et al., 2001; Warren, 2002; Burr, 1995). Unlike the telephone and face to face interviews, the protracted length of my relationship with the ten e-mail respondents, and our continued communication (in some cases, through traumatic life events) led to difficulties in breaking off the e-mails. These difficulties have been noted in the literature, particularly in relation to sensitive research, and researchers respond differently to these situations (Cannon, 1989; Stebbins, 1991; Burr, 1995). In all instances, I still received e-mail communication from participants (to which I responded) for up to four months after the completion of the interview.

Once all of the transcripts were written up, participants were sent a copy to verify the accuracy of the transcription and to make amendments to the text where they felt appropriate (Gershick and Miller, 1995). As Pahl (1995) discovered, the practice of returning transcripts is useful in terms of offering participants the opportunity to make amendments to the text, but also for further comments. In practice, most transcripts were approved following a brief scan or were not read. However, seven participants used this opportunity to remove sections or revise words they felt, on reflection, to be too personal to include in the research, and four participants added further comments and

insights to their transcript. Returning transcripts to participants a short time after their interview further provided me with an opportunity to ‘check in’ with the participants in the study and to conclude my contact with them by offering them the opportunity to discuss any concerns or reflections they had on the interview process itself.

Access and Sampling

The JTSMA were the primary gatekeepers and facilitators of my research. Once ethical approval of my research project had been obtained from them (which will be returned to in the ‘ethics’ section), access to and sampling of participants occurred through a variety of different channels, a strategy commonly recommended for sensitive research (Lee, 1995).

Firstly, individuals were approached at the JTSMA’s annual conference, held every summer in the Midlands and attended by many families and individuals living with SMA, as well as health care professionals. The annual conference provides the means by which families and individuals can meet to discuss SMA, and thus the conference was a useful opportunity for me to introduce myself and the research to those in attendance. A leaflet outlining the research aims and the implications of participating were distributed (see Appendix I), and the details of those who agreed to participate were taken down to be contacted at a later date. The response to this approach was overwhelmingly positive, and all 16 individuals who were approached to participate agreed to do so. Whilst it has been argued that sampling for research should be theoretically informed (Glaser and Strauss, 1967; Denzin,

1970), due to the small population size and the exploratory nature of the study, strategies for sampling were kept broad initially with a view to recruiting as many people as possible with a later refinement of the sampling strategy. One of the difficulties with approaching individuals to request participation in a research study is the possibility of researcher bias. Arber (2001) has highlighted the concern that researchers may consciously or unconsciously only approach those individuals who they perceive to be friendly and accommodating, and indeed, as I was introduced to some families by JTSMA staff, it is likely that they may have selected families who they anticipated would be likely to respond positively. Whilst the possibility for such bias in sampling strategy is largely unavoidable, the use of further sampling strategies accommodated for some of this bias.

Further sampling strategies that were employed included the placement of an advertisement about the research in the JTSMA's newsletter, together with snowball sampling. The advertisement, which briefly stated the purposes of the research and what it would include was published in the JTSMA's quarterly newsletter as well as the electronic newsletter which is e-mailed to members. A small advert was also placed on the JTSMA's website and remained there for several months. A dedicated research telephone number was available for participants to contact me if they wished, and four participants made use of this option. Whilst there are issues associated with sampling through electronic means, as computer use is both a classed and gendered phenomenon (Hewson et al., 2003), this bias was addressed through the use of alternative sampling methods.

In total, 17 individuals responded to the advertisements placed (5 to the printed newsletter and 13 to the electronic one) and agreed to be interviewed. Two participants were excluded from the study, one on account of the fact that her child was affected by a different condition to SMA and the other due to inappropriate communications. All participants recruited from the conference and newsletters who took part in these first 32 interviews were asked if they could recommend a family member or another person living with SMA who might be interested in taking part. Biernacki and Waldorf (1981) have argued that snowball sampling, or 'chain referral' sampling is particularly well suited to studies where populations are small and insider knowledge is advantageous, and this technique expanded the group of participants by a further 22. Those who agreed to participate out of this snowball sample were primarily the family members of the original participants, although some were their friends or acquaintances. To my knowledge, the majority of those who were asked by friends or relatives to take part agreed to do so, and I was only informed of two refusals. The first was due to lack of interest in the project, and the second due to feelings that the nature of the study was too personal to discuss with a researcher. However, nothing can be known about the participants who may have asked their friends or relatives to participate, but subsequently lost contact with me after the interview.

Biernacki and Waldorf (1981) have referenced some of the difficulties of enlisting participants as what they term 'de facto research assistants' (Biernacki and Waldorf, 2001: 153) in the practice of snowball sampling,

which include a loss of control over the communication of the research's goals and procedures to would-be participants. Little can be known about the way in which the research was presented between participants, and further, I had no control over which individuals the participants chose to approach about the study. Whilst they were asked to recommend those who they thought might be interested, there was evidence that some participants interpreted this to mean those people who had a particular story to tell, as one participant commented *'I'll ask my friend, she's been through an awful lot with it so she'll have a good story for you'* (Abi, mother of child diagnosed with type II SMA). These forms of bias that are introduced through the use of snowball sampling are difficult to avoid and necessarily impacted on the composition of the sample. Whilst 'understanding' rather than representativeness is regarded as one of the main goals of qualitative enquiry (Mays and Pope, 1995), it has been argued that researchers should attempt to achieve a sample that is as unbiased and representative of the population under study as possible. For this reason, different sampling strategies were also attempted.

Whilst my initial sampling efforts yielded an overwhelmingly positive result, I acknowledged that people may not identify with one another on the basis of a diagnostic category such as SMA (Anderson and Bury, 1988), and that charities (such as the JTSMA), are sometimes avoided by people with disabilities on account of the medical model of disability often espoused by such groups (Drake, 1996). For these reasons, I felt that it was important to sample outside of the JTSMA, to give voice to those individuals living with

SMA who might not have been a member of the JTSMA, for various reasons. Sampling outside of the JTSMA was attempted through the following routes; firstly, personal websites set up for those affected by SMA (e.g. for the purposes of blogging, fund-raising, awareness) were located through search engines. Specifically, personal web pages which were not linked to the JTSMA and those based in the UK were identified. Only two such pages that did not reference the JTSMA were identified, and the page creators for these sites were contacted. One page creator did not respond, while the second, a mother of two young children diagnosed with SMA, responded and agreed to participate. Further to this, opportunistic recruitment of individuals was achieved through two disability-orientated organisations. The first, Motability, a national charity for disabled people of which I am a member, ran a feature article in their newsletter on a person living with SMA. Following contact with Motability, my details were passed on to this individual, who responded stating their agreement to participate in the study. Two further individuals were identified through their participation on an online forum on a website for an organisation supporting disabled parents. Recruitment through online resources, however, raises particular ethical issues (Berry, 2004). More specifically, it has been argued that the taken-for-granted boundaries between 'public' and 'private' life are blurred in the context of internet research (Bakardjieva and Feenberg, 2001), and it cannot be unproblematically assumed that websites and forums are considered public domains by those who post their information on them. Such issues of privacy raise particular

concerns for researchers wishing to recruit participants, and the ethical issues related to this sampling strategy will be returned to in the 'ethics' section.

As analysis of the interviews progressed, further sampling was informed by the analysis, a process Glaser and Strauss (1967) have referred to as 'theoretical sampling'. I became aware of particular perspectives that were missing from the study, such as experiences of parenthood of those diagnosed with severer forms of SMA. One participant was therefore purposively included in the study on account of the fact that she had received a diagnosis of type II SMA and was the mother of a young child. She was contacted through an existing participant who was aware of her experiences. Efforts were also made to include those who had known, through prenatal testing, about their child's SMA before birth, and decided to continue with their pregnancy. A call for participants was placed on the JTSMA's website online forum where participants were actively discussing this issue, and the JTSMA also forwarded letters to members they believed to fulfil this criterion, inviting them to participate. However, there was no response to these strategies. Herring (1996) has argued that online forums and chat rooms are attractive research spaces due to their accessibility and ease of use. However, it has been noted that ethical guidelines for internet research are not as well developed as for other forms of research (BSA, 2002), and thus particular care needs to be taken when using online methods of research. The purpose of online forums for JTSMA members, for example, is for participants to offer informal support and advice to one another as well as swapping stories and experiences. Whilst these forums were readily accessible to me, therefore, I

also acknowledged that individuals who posted on them may well have been doing so with the expectation that they were addressing an exclusive audience (Berry, 2004). There has been much debate about the extent to which researchers can use data posted on the internet for research purposes. Herring (1996) has argued that if a user chooses a public forum to post comments, then this makes them available for research purposes on account of the fact that more private forms of communication are available. However, as Bassett and O’Riordan (2002) have argued, it simply may not be possible to know a user’s intentions when posting in forums and chat rooms, or even to know how they conceptualise the virtual space they inhabit, which raises ethical concerns for researchers. Thus, rather than approaching directly the individual forum members who appeared to meet the recruitment criterion on the basis of their posts, an advertisement was placed on the forum with the prior approval of the JTSMA, announcing the research and inviting participation. As this advert attracted no responses, none of the postings, whilst relevant to the research, were treated as data as consent could not be obtained (AoIR, 2002). Grinyer (2007) however, has argued that whilst such data may not be treated as primary data, it may be regarded as contextual data, framing the researcher’s understanding of a particular topic. In this instance, despite the lack of response to the advertisement and letters sent, the existence of the forum posts confirmed that several JTSMA members continued with their pregnancies following a prenatal diagnosis of SMA, even though their perspectives and experiences are missing from the study. The absence of this particular group from the study highlights the way in which those who

participated in the research can be understood as a self-selecting population in the sense that participation may have been more appealing to those individuals who felt able to talk about their experiences, or whose stories conformed to prevailing norms and expectations (Landsman, 1998). As the issues with which my research is concerned may be considered sensitive with the potential to evoke strong feelings, the research may only have appealed to participants who were clear in their views or who felt comfortable with the issues it raised.

Table 2: Sampling Strategy and Number of Participants

Sampling Strategy	Number of Participants
Recruited Through Attendance at JTSMA Conference	16
Responded to Electronic or Postal Newsletter Advert.	16
Snowball Sampling	22
Disability Organisations	3
Personal Contacts	3
Websites	1
Total	61

The Participants

At the start of each interview, each participant was asked to offer a brief description of themselves including their age, occupation (if any) and a description of their ethnicity in order to provide some demographic data. Out of the 61 participants who were interviewed, 13 were male and 48 were female. The over-representation of women in the sample may be attributed to a variety of factors. In the first instance, it was apparent that participants in the

study perceived the subject matter of the interviews to be the concern of women, who bear the brunt of the responsibility for childbearing and raising. Ideas about gender which position women as both the 'gene transmitters' (Steinberg, 1996: 267) of genetic conditions, but also as responsible for the family's health more generally reinforce the notion that reproductive decision making in the context of SMA is primarily a female domain (Graham, 1979; Ruddick, 1989). Further, all but three of the mothers of children with SMA who participated in the study had left work or reduced their hours following their child's diagnosis in order to care for them. This assumption of care work by women is a finding supported by the literature (Finch and Groves, 1983; McLaughlin, 2006) and reinforces the notion that both childbearing and child rearing, around which the study is based, is primarily the domain of women.

The method of sampling may also have influenced the number of women who participated in the study. As participants were asked to recommend friends or family who might be eligible and interested in taking part, women tended to suggest their female friends and relatives. There was evidence that there existed networks (primarily) of mothers of children diagnosed with SMA who were in regular contact with one another and who knew each other's experiences in great detail. Schaffer et al. (2008), through their study of mothers of children with genetic conditions using the internet, have argued that women take responsibility for networking and participating in 'genetic communities' (p.156) in order to learn from the experiences of others in the management of their child's condition, and indeed, it appeared

that in this study women were the primary networkers and information gatherers on SMA.

All participants were asked to describe their ethnicity at the start of the study. Fifty-two (82%) described their ethnicity as white British and 8 (13%) described themselves as belonging to an ethnic minority group; one as Iranian, two described themselves as having mixed heritage, one as Indian, two Irish, one Romanian and one as white European. Whilst the JTSMA is a UK charity, not all participants were living in the UK at the time of interview; one participant was interviewed living in France, one in the Republic of Ireland, one in Nepal and one in Switzerland. The remaining participants were geographically dispersed throughout the UK- in Scotland, Wales, Northern Ireland and across 22 different counties in England. Seven British participants had had the SMA diagnosed in their family during a time when they were living outside of the UK (in Australia, Italy, Spain and Holland), but had returned to the UK upon diagnosis.

The ages of participants ranged from 9 to 63 with an average age of 37. Whilst the study set out to interview participants exclusively over the age of 18, one child, (aged nine and the able-bodied sibling to a six year old with SMA) requested to be interviewed following her mother's participation in an interview. A separate guide was prepared for this interview which focussed on the child's experiences of having a sister with a disability and how it impacted on her family's life. This interview was arranged through the child's mother and upon completion of the transcript, it was returned to her mother who read it through with her. Whilst interviewing a child posed particular ethical

dilemmas, more specifically the risk of emotional harm, or of unsettling the relationship between the siblings, Eder and Fingerson (2001) have nevertheless argued that interviewing children can give voice to their unique interpretations of reality ordinarily inaccessible to adults. Indeed, as the initial interviews highlighted the importance of childhood experiences of disability in informing participants' (adult) perspective on SMA, it seemed important to include a child's perspective that had not been reinterpreted retrospectively but was grounded in the present- the everyday reality of growing up with disability in the family, which was so central to the research.

The participants varied in terms of the diagnosis within their family, but also in their relationship to the person diagnosed with SMA. Such diversity enabled a broad range of perspectives to be explored within the study, and is reflected in Tables 3 and 4.

Table 3: The Participants

Participants	Numbers	Gender	
		Female	Male
Diagnosed with SMA	25	21	4
Sibling of person with SMA, without SMA themselves	7	4	3
Parent of person diagnosed with SMA	24	21	3
Grandparent of person diagnosed with SMA	1	1	0
Son or daughter of person diagnosed with SMA	2	1	1
Partner or spouse of person diagnosed with SMA	2	1	1
Total	61	49	12

Table 4: The Diagnoses of SMA

Diagnosis	Number of Participants with Diagnosis in their Family	Gender	
		Female	Male
Spinal Muscular Atrophy Type I	12	11	1
Spinal Muscular Atrophy Type II	32	25	7
Spinal Muscular Atrophy Type III	11	10	1
Spinal Muscular Atrophy with Respiratory Distress	3	2	1
Spinal Bulbar Muscular Atrophy	2	1	1
Autosomal Dominant Spinal Muscular Atrophy	1	0	1
Total	61	49	12

Whilst there is little published information on the prevalence of different types of SMA, estimates suggest that anywhere between 50 and 70% of diagnosed cases of SMA are type I (Brichta et al., 2003:2481; Swoboda et al., 2007). However, within my study, those diagnosed with SMA type I in their family comprised only 20% of the sample, with a much higher representation of those diagnosed with type II (52%). As severer forms of SMA are associated with a greatly shortened life expectancy (around 18 months), it is unsurprising that the majority of adults with SMA who were interviewed and whose families also took part were diagnosed with intermediate or milder forms of SMA; only one adult with a diagnosis of SMA type I was interviewed.

Ethical Considerations

The Risk of Emotional Harm

It was decided early on in the project that the subject matter of the research warrants its definition as a 'sensitive' research topic. Lee (1993) defines sensitive research as covering topics that pose either intrusive threat, political threat, or the threat of sanction to participants (Lee, 1993: 4-9). The term 'intrusively threatening' research refers to research which involves deep invasion into participants' personal or emotional lives. My research indeed invoked stories covering a range of emotive and personal issues, including participants' experiences of living with a life-limiting chronic illness or disability, experiences of bereavement, experiences of, and attitudes towards, selective abortion and/or involuntary childlessness, as well as ideas about the prevention of lives of people with disabilities. These topic areas are highly personal and potentially emotionally charged, and it was acknowledged that discussing them with a researcher could induce a range of emotional responses including stress, grief, guilt, shame and anxiety. A key concern in the study of sensitive topics is the possibility of psychological harm to participants through the very process of addressing difficult issues.

Shakespeare (2008b), for example, has highlighted the way in which discussions around the existence of genetic technologies may raise difficult issues specifically for people with genetic impairments, as it may invite them consider whether or not their parents would have used these technologies if they had had knowledge of them or access to them. Such thoughts may be deeply unsettling or threatening as they suggest the possibility of one's own

non-existence, and indeed, several participants reported such thoughts and ideas. Similarly, various studies have highlighted difficult emotional responses to the knowledge of a genetic condition within a family. Guilt, for example, has been a well documented response of parents to children with inherited conditions (Kessler, 1989) as is the experience of ‘survivor guilt’ in siblings to people with inherited conditions (Madigan, 1996). The risk of emotional harm was therefore one of the most troubling ethical considerations of my research project. Although participants generally articulated their experiences of emotional distress in matter-of-fact ways and appeared to have anticipated the emotional implications of the research, some participants nevertheless became distressed during the interview, and one person refused to participate on account of the emotional difficulties the research could raise for them. Measures were therefore put in place, to minimise, as far as was possible, the risk of emotional harm.

One way of managing emotionally sensitive interviews has been described by Brannen (1988) who argues that topics likely to provoke emotional responses should be allowed to develop gradually over the course of the interview, rather than being approached directly early on. This technique proved useful in practice as it allowed me to gauge the emotional response of the participants and to end a line of questioning if it was apparent that the participant was experiencing distress. It further allowed the participant an opportunity to set the boundaries of which topic areas they were happy to talk about and which they were not, and four participants steered the

interview in this way and closed down topics of conversation that they were not happy to discuss.

A further means of minimising emotional distress in research that has been noted in the literature is the practice of allowing the participant to 'debrief' after the interview, or talk about the aspects they found particularly difficult or challenging (Rodham, 1998). Whilst this technique has been criticised for further blurring the (often) hazy line between interviewing and counselling (Etherington, 1996), and also for assuming that this form of support 'works' or is desirable (Hubbard et al., 2001), it was felt that it would be equally unethical to deny research participants the opportunity to discuss the effects the interview had on them after it had taken place. In practice, this debriefing often occurred spontaneously with participants offering me their immediate reflections once the interview had ended. However, for others, reflections on the interview did not occur immediately after the interview, but rather emerged after the participant had time to reflect on the event. As contact was made with all participants within two weeks of the completion of their interview to provide them with their transcript, this offered an opportunity for participants to discuss any effects the interview had for them. Only five participants took up this opportunity. However, all of them indicated that the interview had been an illuminating or even cathartic experience, which has been noted in other research on sensitive topics (Pillow, 2003; Hutchinson et al., 1994; Lee, 1981). In addition to the opportunity to express reflections on the interview, participants who expressed distress during the interview were further advised of the availability

of other forms of professional support, such as counsellors and outreach workers based at the JTSMA whom they could contact for further assistance.

A further ethical concern regarding emotional harm to participants is the potential effects the research could have on family dynamics and family relationships. Genetic conditions necessarily implicate other family members, and as my interviews often involved interviewing more than one family member, I frequently found myself in a position similar to that experienced by D'Agincourt-Canning (2003) when she completed her doctoral thesis on breast cancer: one of holding more information on the decisions and perspectives of their family members than participants appeared to have themselves, and there was evidence that the research did raise some issues around familial relationships. Two participants from different family groups stated that the interview had sparked family discussions about SMA and genetic testing (which they stated had been productive) whereas three participants expressed concern about how their family members had answered, as one commented:

I would recommend you interview my brother...but then there's a danger with that that I might not like what he says...I don't think he would, but he could have a very different perspective on SMA to me and that might be hard for me...to [pause] um... accept.

(Gill, diagnosed with type II SMA)

The potential for tension between family members, brought to the surface by the interviews was therefore a very real concern. In order to circumvent or, at

the very least, minimise damaging effects to familial relationships, great sensitivity was required to protect the interviewee, but also any of their family members who had participated. All participants were reminded prior to the interview that their family members or partners need not be recruited into the study if they felt this would be problematic or would raise difficult issues for them. Further, it was emphasised that interviewees' anonymity could not be guaranteed in situations where members of the same family were interviewed. Even if names were removed, the personal and unique nature of individuals' stories could render them recognisable to other members of the same family (LaRossa et al., 1981). Further, as I recruited through the JTSMA, I was aware that it was possible that participants could recognise other JTSMA members' stories. As there are many close knit networks within the JTSMA, and the ethos of the JTSMA is to encourage its members to share their stories with one another as a form of mutual support, during the course of the interviews, I became aware that many of the participants knew each other, or knew *of* each other, including their stories with SMA. Indeed, some recounted these stories during their interview in order to highlight and reinforce the point they were making. This familiarity between JTSMA members, whilst facilitating snowball sampling and introducing the possibility for comparative analysis of the accounts (i.e. comparing the stories participants told with the way in which they were told by other participants), also had the potential to render participants' stories recognisable to one another. These limits to the anonymity of participants meant that relationships could be disrupted both within family groups and also friendship circles. Whilst it was recognised that

it may be difficult, if not impossible, to anticipate the existence or even the nature of such potential relational difficulties, obtaining informed consent could not absolve me of responsibility for these unintended consequences of the research (Lee, 1993). Whilst no evidence of relational difficulties were brought to my attention however, nothing can be known about the experiences of participants with whom contact was lost after completion of the interview.

Risk to Researcher

It has been argued that an area often neglected in a discussion of ethics is the risks the research may pose to researchers themselves, not only in terms of physical risk (Jones, 1991), but also in terms of the psychological and emotional consequences of the research (Hubbard et al., 2000; Dickson-Swift et al., 2008). If ethically responsible research is characterised by its avoidance of harm to those involved in it, then it follows that this should apply to the researcher who is deeply embedded within the research for a much longer time frame than the participants themselves.

It has been argued that personal safety in the research field is often treated like a ‘non-issue’ for social researchers, with relatively few studies addressing this concern directly. As Keynon and Hawker (1999) have argued, it is often assumed that researchers will use common sense and intuition to avoid potentially risky situations, and restrict their research to ‘safe’ areas such as public spaces and conduct their interviews at ‘safe’ times such as during office/daylight hours. Assumptions about which temporal and spatial situations are ‘safe’ and which are not, however, are highly cultural (Furedi,

1998), and indeed it may be impossible for researchers to predict where and when dangerous or threatening situations may emerge. My own research involved one-to-one interviews with strangers, in two instances in their homes, although the remainder of the interviews were conducted over the telephone or via e-mail. Whilst Kenyon and Hawker (1999) recommend the planning of 'quick and easy' escape routes when conducting interviews, my physical impairment (which necessitates the use of a wheelchair), combined with the inaccessibility of participants' homes meant that such routes were not available to me. In order to minimise the safety risks to me, I was therefore careful to ensure a procedure of 'checking in' either with a friend, relative or colleague before and after the interview.

An additional issue relating to the possibility of harm to researchers concerns emotional wellbeing. As Lee (1993) has argued, interviews have an effect not only on respondents, but also on researchers themselves who may react emotionally to their data, particularly when the issues under discussion relate to their own lives and experiences. As someone living with a neuromuscular condition myself, the issues addressed by my research sometimes resonated with my own experiences. Furthermore, I frequently encountered harrowing stories, particularly of grief and loss, which had a great emotional impact on me and required what Dickson-Swift et al. (2008) have referred to as 'emotion work', or the active management of feelings. This 'emotion work' was done primarily through accessing the support of friends and colleagues which facilitated the more emotionally demanding aspects of the research (Brannen, 1998).

Reflexivity

Reflexivity, or reflection on the way in which our selves and our identities impact on the knowledge our research produces, has been considered a crucial part of the research process (Seymour, 2007). Whilst Patai (1994) has argued that discussion of the researcher's identity can be considered egocentric or even self-indulgent, Seymour (2001a) and Ellingson have countered this argument by suggesting that to write oneself out of the research project is to overlook a critical part of the research process and leads to 'deceptively tidy' accounts of research (Ellingson, 2006: 299). Whilst reflexivity has been acknowledged as an (often) uncomfortable part of writing up process (Ryan-Flood and Gill, 2009), it may nevertheless present opportunities to explore what initially may have been invisible to us: that is, the influence of our identities and selves on the research (Finlay, 2002). Acknowledgement needs to be made not only of the way in which participants respond to the interview questions that are asked, but also of the way in which they react to the researcher, and how the researcher themselves impacts upon the interview situation. As the values, insights and preconceptions of the researcher invariably feed into the research process at different points of the research process and in subtle ways, there have been calls for careful reflective thinking on the part of researchers about how this situation informs the overall shape of the research, or even the advantages it may offer (Goodley et al., 2004).

As well as their interpretations of the research, the participants' personal reactions to, and assumptions about, the researcher may additionally

inform the shape of the study and influence the version of the stories that are told, and hence the data that are collected. Much of the literature exploring these so-called 'interviewer effects' have focused on the possible benefits of 'matching' researcher/researched identities in terms of gender (Oakley, 1980; Oakley, 1981; Finch, 1984), ethnicity (Kohler Riessman 1987; Edwards 1990;), culture (Song and Parker, 2005) or sexuality (Platzer and James 1997), as a means of facilitating rapport and trust building in the context of the interview. However, far fewer studies have explored the influence of a disabled identity (Barnes 1992; Oliver 1996b; Vernon 1997) or the influence of the impaired body on the interview or the research process more broadly (Ellingson, 2006). In the case of my own research, my identity as a female in my mid twenties with a visible disability invariably shaped the research process in a myriad of ways.

As Dickson-Swift et al. (2008) have noted, researchers frequently gravitate towards research projects that explore aspects of their own experiences and subjectivities (p. 91). This closeness to the research project, or even shared experiences with research participants, can facilitate rapport building in the interview context through mutual sharing and self-disclosure (Dickson-Swift et al., 2008). However, it may also make researchers more sensitive to stories that resonate with their own experiences and perceptions, and to overlook, or attribute less importance to, those which do not; the stories we are able to hear may thus be informed by our subjective experiences and values. My own experiences of living with a disability have inevitably fuelled my academic interest in disability, as well as in the social implications of

reproductive genetic technologies. In this way, my personal experiences both influenced my interest in the topic of the research, but also called for me to pay greater attention to the way in which my pre-existing beliefs, values and potential prejudices stemming from these experiences could shape the research process (Ribbens and Edwards, 1998). Reflexive thinking, undertaken at all stages of the project, aided by the maintenance of a research diary in which personal reactions and thoughts were recorded and reflected upon, facilitated sensitivity to these more subtle influences on the research.

As well as informing the design of the research, my visible disability informed participants' responses to me. Four participants who were themselves wheelchair users, reported feeling more comfortable talking to me as disabled person:

...I'm so glad that it's a disabled person doing this research, it makes it a lot easier to talk about things. I thought you were going to be able-bodied.

(Megan, diagnosed with type II SMA)

Seymour (2007) and Andrews (2005) have noted the way in which the shared experience of disability can signal the 'right' to research disability, as well as facilitating rapport and trust building in the interview context (Seymour, 2007: 1193). As my research addressed issues around the way in which disability is perceived and valued in the context of prenatal testing, it may be that participants felt more able to discuss these issues openly with a disabled researcher, and indeed many readdressed the interview questions back to me, suggesting that they felt these issues were relevant to my own life as well as

theirs. Whilst there may be benefits to *not* sharing a disabled identity with participants, indeed Gow (2000) notes the way in which the women in her study positioned themselves as the ‘experts’ on their respective conditions and thus may have gone into more detail in explaining their experiences to her as an ‘outsider’ (p. 118). In my study, having a shared disabled identity often provided the opportunity for reciprocity which has been acknowledged as a means of addressing power differentials between researchers and the researched (Harding, 1987; Cotterill, 1992; Tregaskis and Goodley, 2005). For the interviews conducted with the able-bodied parents of children with SMA, however, my disability appeared to have a different influence. I was concerned that such parents might feel unable to discuss their feelings about having a disabled child with me on account of the fact that I am a disabled person, and there was some evidence that parents felt uncomfortable with this, as one woman commented:

J: Um it’s very hard to um comment on the test

[prenatal test for SMA]...because I mean [pause]

you’ve got a disability yourself, haven’t you?

F: Yes

J: Yeah well you see...I wouldn’t want to offend

anyone...you know it’s not like saying disabled

people are bad or anything, but I think that it could

still be considered offensive to disabled people really,

and I don’t want to offend anyone.

(Jessica, mother of child diagnosed with type I SMA)

The potential offense that disabled people can take to prenatal testing has been documented in the literature (Asch, 2000), and it appeared that through reading my identity as a disabled person, Jessica was reluctant to discuss her feelings about testing and selective termination. Due to the methods of sampling used in the study, however, the extent to which participants had access to knowledge about different aspects of my identity varied and thus the impact was hard to ascertain.

Data Analysis and Reporting

As the aim of this study was to better understand the experiences of those living with SMA and the relationship of these experiences to perceptions of genetic risk and reproductive decision making, an inductive approach was taken to data analysis. Rather than testing pre-existing hypotheses, a grounded theory approach was taken to the data, which allows theoretical concepts to emerge from the data itself (Glaser and Strauss, 1967). Charmaz (2003b) has noted that there are two central approaches to grounded theory data analysis, constructivist and objectivist approaches, each with different epistemological assumptions. Objectivist approaches to grounded theory analysis treat data as pre-existing facts that represent a reality readily accessible to the researcher through participants' accounts. Constructivist accounts, however, acknowledge that the data are accounts co-produced by researchers and participants, located in 'time, place, culture and context' (Charmaz, 2003b: 313) and constructed through the interpretive lens of both the researcher and the researched. As discussed in Chapter 1 in relation to risk, an interpretative

approach to the research has been adopted; writers supporting this theoretical perspective acknowledge that risk perceptions are constructions that emerge out of, and through, the everyday meanings and experiences of those who encounter them. Thus, a constructivist grounded theory approach to data analysis, which accommodates this role of interpretation, was the most appropriate for data analysis.

Analysis of the transcripts was a long process, begun early on in the research (during data collection), and one which did not follow clearly defined stages. Upon completion and transcription of the first seven interviews, manual coding was undertaken of the transcripts, so that every line of text was attributed a code. Memos were written during the process of coding to document thoughts and reflections on the data, including those recorded in my research diary. In this way, the analytic process of interpreting and making sense of the data began at an early stage. Gibbs (2007) has argued that this process of ‘open coding’ of every line of transcript text is a particularly important aspect of grounded theory analysis as it keeps researchers close to the data. Whilst the suspension of all preconceived ideas about the data, with the researcher acting as a ‘tabula rasa’ during analysis, has been acknowledged as an unrealistic expectation of those undertaking grounded theory analysis (Bulmer, 1979), close reading and re-reading of the texts ensured that as far as possible, concepts that emerged from the data were ‘data driven’ rather than ‘concept driven’ (Gibbs, 2007). Initially, the codes attributed to the first seven transcripts were largely descriptive and derived from participants’ own phrases, referencing the topic areas under discussion

e.g. 'getting to a diagnosis of SMA', 'managing SMA in daily life'. As Ritchie et al. (2003) have noted, this initial 'sorting and sifting' of data into descriptive codes is particularly useful to aid later finer analysis as the data are organised into manageable sections.

As one of the aims of the research was to examine the importance, and uses of, experiential knowledge of SMA in the context of reproductive decision making, participants' descriptions of their experiences with the condition were not interpreted literally during analysis, but rather as accounts presented and interpreted in continuous relationship to these decisions. Stories of life with SMA were thus not treated as objective facts but rather as contextually bound accounts subject to different presentations and forms of justification at various points.

As the number of interviews increased, coding was transferred to Nvivo 7 and the codes were organised hierarchically into 'coding trees' to categorise and sub-categorise the emergent concepts according to their relationship to one another. The use of coding trees added a further dimension to the analysis (Bazeley, 2007); by the appearance of the same sub-code under different 'parent codes', I was able to explore how particular experiences or instances were talked about in the context of different topics, which facilitated the identification of conceptual relationships not only within codes, but also between them. Proponents of grounded theory analysis support the use of what has been termed the 'constant comparison' method of data analysis (Pidgeon, 1996) as a means of developing theory from the data. The comparison of codes, and texts within each code, specifically enabled me to

identify contradictions and inconsistencies both within, and between, participants' accounts. These contradictions were treated as significant as they highlighted ways in which different versions of life with SMA were presented at different moments, which in turn led to the refinement of emergent concepts. Once this process of analysis had been undertaken on half of the interviews, written summaries of the 'parent codes' were completed which highlighted missing perspectives in the study and pointed to areas where more data were needed to extend and modify the emerging picture. This process of coding, the refinement of concepts through data interpretation followed by re-coding and further sampling, which are central features of grounded theory analysis, were carried out over a period of eight months until 'theoretical saturation' (Glaser and Strauss, 1967), or the point at which no new concepts are emerging from the data, had occurred.

The writing up of the research, as has been described, started during the analysis of the coded transcripts, and these written summaries of the emergent themes went on to become the basis of the drafts for Chapters 3-6. As it was discovered that the boundaries between diagnostic categories of SMA are far more blurry than medical classifications suggest (as will be discussed in Chapters 3 and 4), I decided to present participants' experiences thematically rather than stratified according to type of diagnosis, reproductive status or relationship to a person with SMA. However, where quotations from participants are used, the type of SMA diagnosed within their family is included with the quotation. This decision was taken because whilst many participants were generally critical of the medical classifications of SMA,

these classifications nevertheless provided a framework through which they evaluated their experiences of SMA. Through knowledge of the prognosis associated with diagnostic categories as well as the experiences of those with the same diagnosis, participants assessed their own abilities and those of their family members. Furthermore, the accounts of some participants, who articulated specific concepts particularly clearly or had certain types of experience with SMA, reappear in several chapters, e.g. Fraser.

Limitations of the Study

One of the main limitations of this study is the possibility of the sample of participants being unrepresentative. By recruiting through the JTSMA, my sample was limited to those individuals who identified themselves with this organisation, its aims and its ethos, and thus may have excluded the perspectives of those who do not identify in such a way. The potential exclusions this recruitment strategy may have brought about are set out below.

Whilst the JTSMA was cited as an invaluable resource by the vast majority of participants, there were nevertheless some participants who saw the objectives and strategies of the JTSMA as having less relevance to their lives than others. As Epstein (1996) as well as Stockdale and Terry (2002) have noted, patient organisations are often formed by white, middle class individuals, and, disproportionately, by wives and mothers (Rapp, 1999; Rapp et al., 2001). It is these individuals who have the financial, cultural and practical means at their disposal to become educated about 'their' condition

as well as to mobilise resources in the formulation of self-help groups. A consequence of this is that people from ethnic minority or working class backgrounds are under-represented in organisations such as the JTSMA, as well as those individuals who prefer to access other forms of support (Rapp et al., 2001).

In addition, the JTSMA's focus on cure and medicalised approaches to SMA may serve to alienate some people living with the condition and preclude their attendance at JTSMA events. As Stockdale (1999) has noted in relation to the Cystic Fibrosis Association, a focus on 'cure at all costs' by medical charities can actually lead to an oversight of the 'many ways people live with and experience the disease' and lesser importance being attributed to areas that make a real difference to the everyday lives of people living with CF (Stockdale, 1999: 594). Shakespeare (1999) has posited these contrasting approaches in terms of medical and social discourses around disability and genetics. Medical discourses around genetics and disability which position the use of genetic technologies as a means of avoiding the 'tragedy' of the birth of a person with an impairment may be more closely aligned with the approach of medical charities such as the JTSMA and in stark opposition to disability rights approaches (Shakespeare, 1999: 673). Whilst the 'tragedy' approach may be used primarily to generate funds for charities (Stockdale, 1999), for those who favour social approaches to disability, the agenda of the JTSMA may be experienced as alienating or patronising. Andrew, in his late 40s and diagnosed with SMA commented that he did not have extensive

involvement with the JTSMA as their priorities did not match what he felt was important to his own life as an adult with SMA:

I had some [contact with JTSMA] in the past, I just haven't had much currently and I think it's an interesting....I mean I've been to their conference a couple of times and it's an interesting space for a lot of people, particularly kids with SMA coming together. I think that the absence of a space for people with SMA, particularly adults, to get together and share different bits of our lives...that would be useful and the Jennifer Trust don't really do that...we have different concerns, particularly around access to...I know that people with SMA, a lot of people, don't very often come into contact with disability politics themselves and sometimes that interaction and that understanding of what might be possible sometimes isn't there at the Jennifer Trust.

(Andrew, diagnosed with type II SMA)

Despite these criticisms levelled at the JTSMA by some of the adults living with SMA, the JTSMA emerged not as an organisation designed to affront disability politics, but rather one founded by the able-bodied parents of young children with SMA and has retained its focus as a 'family' charity; its annual conferences are primarily attended by able-bodied parents and children with SMA leaving adults with SMA largely outnumbered. The consequence of this focus on the needs and concerns of able-bodied parents and children with SMA has been the under-representation of adults with SMA, many of whom

drift out of the JTSMA as they ‘age out’ of its target age group, a finding also recorded by Gow (2000) in her attempts to access women aged between 18 and 25 through the Cystic Fibrosis Trust. Indeed, eight of the adults interviewed with SMA stated that after frequent contact with the JTSMA as children they now rarely get involved with its activities. Whilst three of these adults made specific reference to a lack of engagement with disability politics by the JTSMA as a reason for their lack of identification with them, the remaining five simply stated that the JTSMA became less important as they grew up with SMA. This may relate to the under-representation of issues of importance to them within the JTSMA’s activities or it could be that the JTSMA is particularly important for families and individuals at ‘critical points’ (Bury, 1982) such as diagnosis, but has less relevance as people with SMA gain experience and knowledge of how to live successfully with the condition.

Despite these potential limitations to the study, however, a broad range of individuals living with SMA agreed to participate, and actively and thoughtfully engaged with the research, as their accounts in the following analysis chapters reveal.

Chapter 3

What is SMA?

SMA is a condition which is associated with a broad spectrum of presentations and experiences, and thus is not easily described or defined. Despite all being attributed the same diagnostic label, the families and individuals who took part in this study all had very different experiences of SMA; some had lost children in early infancy, some had been unable to walk since birth, whilst others had experienced being able-bodied well into adult life at which point they began to experience increasing muscle weakness. This diversity in presentation and severity of muscle weakness has been identified as a major contributing factor to the documented confusion within the medical literature on SMA, particularly around its definition and the categorization of variant forms (Dubowitz, 1991). Indeed, Dubowitz (1995a) has described SMA as having the widest variety in presentation out of all of the neuromuscular disorders of childhood. Despite the identification of the 5q11.2-13.3 chromosome in 1990, which has been described as being involved in the causation of most forms of ‘classical’ SMA (i.e. SMA displaying all of the features traditionally associated with it) (Melkin et al., 1990), uncertainty remains as to where the diagnostic boundaries of SMA lie, and how the different ‘types’ of SMA may be differentiated from one another. This concern has further been exacerbated by the apparent discontinuity between genotype and phenotype expression of the condition (Wirth et al., 2006). Nevertheless, the ability to identify different ‘types’ within a diagnosis of SMA, has been considered a particularly important exercise from a medical

perspective, not only to understand the biological mechanisms by which SMA occurs, but also in terms of offering a useful diagnosis and prognosis to families and individuals experiencing SMA. Indeed, the division of SMA into 'types' is commonly regarded by the medical profession and those living with SMA as a shorthand to describe the severity of muscle weakness and anticipated life expectancy of those with different forms of the condition. For families and individuals living with SMA, therefore, the development of specialist medical knowledge around SMA over the course of the 20th century has been highly significant. Its emergence as a genetic condition and the uncertainties and inconsistencies inherent within the medical knowledge around it, have inevitably shaped the way in which it is experienced, conceptualised and responded to as a condition. As this chapter presents, participants' accounts revealed that there was not always an easy 'fit' between expert knowledge of SMA and the way in which it was experienced, and lived with, in daily life; families negotiated different forms of knowledge from contrasting sources to construct their definitions and understandings of SMA and to formulate their responses to it.

The focus of this chapter, therefore, is on some of these negotiations of expert and experiential knowledge as they occur through the definition, diagnosis and geneticization of SMA. The contradictory, yet also interconnected, relationship between these two forms of knowledge will be emphasised, with a consideration of the consequences this has had for the way in which families and individuals relate to the condition. This setting out of how SMA is understood, as well as how it is lived with (in Chapter 4), will set

the scene for a later presentation, in Chapters 5 and 6, of the meaning these conceptualisations took on in the context of reproductive decision making.

Defining and Describing SMA

Defining SMA

SMA is a condition which has been attributed within the medical literature to a genetic mutation of the SMN1 (survival motor neuron) gene on chromosome five. The genetic mutation is widely understood to have occurred during the evolution of the human genome from that of apes (Fortna et al., 2004). However, the condition was not systematically described by the medical profession until the 19th century. Guido Werdnig (1891), Johan Hoffman (1893, 1897) as well as others around this time (e.g. Beevor, 1902; Thomson and Bruce, 1893) described patients who experienced severe muscle weakness in the first months of their lives and died in early infancy, for which they coined the term ‘Spinal Muscular Atrophy’, also known as ‘Werdnig-Hoffman Disease’. It was not until later in the 20th century however that debates began to emerge as to where the diagnostic boundaries of SMA should lie, given its similarities with other conditions, such as Muscular Dystrophy and Motor Neurone Disease. The SMA described by Werdnig (1891) and others in the 19th century was an early onset form of SMA that resulted in premature death. However, over the course of the 20th century, clinicians and researchers came across individuals whose symptoms mirrored those of the early cases reported by Werdnig, but who nevertheless achieved developmental milestones (such as the ability to sit or walk unaided) or

experienced prolonged survival, beyond infancy. A doctoral thesis describing SMA produced by Brandt in 1950, for example, described both a ‘severe’ type of SMA, but also an ‘intermediate’ type, with those so-affected capable of sitting and surviving infancy (Brandt, 1950). Further, the work of Dubowitz (1964) and Byers and Bankers (1961) in the 1960s pointed to the various different forms SMA could take, even within sibling groups.

From the 1960s, therefore, clinicians and researchers began to speculate about the possibility of an SMA ‘continuum’ (Dubowitz, 1967) with the most severe infantile forms of the condition at one end (associated with poor life expectancy), and the milder juvenile, or adult onset, forms at the other end, with various other presentations in between. However, where the dividing lines between these types should be positioned continued to be debated within the SMA medical community through the 1970s and 1980s. Pearn (1980), in an English study of 240 people affected by SMA, for example, suggested that seven different SMA syndromes could be identified, caused by 13 different genetic mutations, whereas Fried and Emery (1971) argued for the existence of three clinically differentiated forms of SMA, defined by the age at which symptoms first appear, and associated with different genetic mutations.

The range of biological explanations offered by the medical profession to make sense of the experience of SMA, and the movement of its boundaries over time has had a considerable influence on the prognostic and diagnostic information given to individuals experiencing SMA over the past fifty years. Indeed, all participants in this study who were diagnosed with SMA in the

1960s and 1970s reported being offered conflicting biological explanations for their condition as a consequence of these shifts in definitions of SMA. Five participants, for example, were initially diagnosed as having Duchenne Muscular Dystrophy (DMD) at a time when SMA was poorly understood, and described the devastating emotional consequences this had.

Matilda is 63 years of age and was initially suspected of having Polio due to an epidemic of the disease in the late 1940s. However, she was later reclassified as a Muscular Dystrophy patient before finally having her experiences reclassified by a doctor who went on to write a definitive paper on type III SMA based on her experiences in the late 1960s (Hausmanova-Petrusewicz et al., 1968). Matilda recalls the mental anguish this initial uncertainty and diagnosis caused herself and her family, as a consequence of being at the centre of shifting disease boundaries:

...I think it was in 1947 there was a Polio epidemic and I was sort of lumped together with the other children who had Polio. One of my earliest memories is my mother arguing with the consultant... that she'd talked to the other mums in the waiting room and their children who had Polio had been ill and I'd never been ill...but you know they thought at the time that doctors know best and mums don't have a clue. Then I think when I was 18 they used a situation to come up with a condition, I had deteriorated considerably and then they diagnosed me with Muscular Dystrophy and they said I'd be dead within a couple of years-that I'd die before I was

21...and it was completely... devastating...for everyone.

[pause] It was at a time when they were beginning to

separate out the atrophies and the dystrophies but it

[SMA]was a completely unknown quantity in the early

1960s...I mean they knew about SMA type I, but not II or

III. And one of the doctors who wrote one of the definitive

papers on SMA talked um about a 19 year old with these

symptoms um and that was me. So that's when I found out

what I had.

The shifting nature of disease categories in line with changes in scientific understanding over time is of crucial importance to people who are the subjects of such classificatory systems (Hedgecoe, 2002; Bowker and Star, 1999). Medical diagnostic categories are significant not only in terms of their impact on individuals' identities, but also their subjective experiences of their reality, their imaginings of their future as well as their entitlement to services and support. However, as Hedgecoe, through his analyses of the emergence of Cystic Fibrosis (2003) and Diabetes Mellitus (2002) as disease categories has noted, despite their appearance of fixity, disease boundaries are socially constituted in that they represent 'compromises' within and between medical communities and are never devoid of inconsistencies, uncertainties and ambiguities (Hedgecoe, 2003: 55). These uncertainties, as Matilda's account highlights, can have catastrophic consequences for those who are the subjects of such medical classifications. The emotional impact of being diagnosed with a terminal condition, only to have this prognosis displaced a year or two later

can have serious implications for psychosocial wellbeing, as well as feelings of trust in the stability of medical knowledge. Indeed, a similar account to that of Matilda is explored in the autobiographical film '39 pounds of love' (Menkin, 2005), which tells the story of an American born Israeli man whose ambition it becomes to confront the doctor who diagnosed him with SMA and predicted a certain death before the age of two, which later proved to be inaccurate, highlighting the distress associated with these medical uncertainties.

As well as the impact shifts in medical knowledge can have, Matilda's account further highlights the way in which the experiential knowledge of individuals living with SMA can, in itself, play a role in challenging medical knowledge. Recollections of the various processes around getting to, and receiving, a diagnosis of SMA for many participants in this study, for example, were accounts of fighting, perseverance and the challenging of health care professionals for their symptoms of SMA to be taken seriously. The diagnostic process frequently took participants on a 'medical merry go round' (Peterson, 2006: 35) of undergoing invasive tests and procedures before what was often described as a 'lucky' encounter with a health care professional who had some knowledge of SMA.

As early signs of SMA may be experienced as 'floppiness' and difficulties with feeding in babies, delays in walking and crawling in infants and the onset of muscle weakness and fatigue in adults, symptoms can be confused with late development (in children), or the onset of age-related fatigue (in adults), which can contribute to some of the obstacles to diagnosis

experienced by those living with SMA. Mothers of young children who are experiencing symptoms of SMA appeared to be at the forefront of many of these battles; in total, 17 mothers of children with SMA were interviewed and 10 specifically mentioned having their concerns about SMA dismissed, particularly by health care professionals, before a diagnosis of SMA was established. For such mothers, there was a fine line to be trodden between being regarded as a 'good mother', who assumes responsibility for promoting her child's health, and being a 'neurotic' mother (Graham, 1979), the suggestion of which being the basis on which many mothers had their concerns about SMA disregarded (e.g. Macaulay, 1996: 41).

Liz is the mother of a (now 28 year old) daughter, Cara, who was diagnosed with SMA type II at the age of 4 (considered medically as a 'late' diagnosis for SMA type II). She described her experiences of having her concerns invalidated in the following way:

Cara was minded whilst in school by a very close friend who has many other young children of her own and through family friends, so Cara grew up amongst many other cousins and small children. And we were aware from about a year that perhaps she didn't move as fast as others, she didn't crawl, she bottom shuffled, and the GP always said she was fine, I asked about it several times. At two, we were still worried so we went to...initiated by us...to a specialist who said that 'she was short and fat like her mother' and that was why she couldn't turn her head

enough and that I was a 'fussy' mum and nothing else was picked up until she was four. It was hard to keep pushing for information when I got responses like that from them, but it's your child's health and as a mother you've got to put it first...you know, I *knew* there was something not quite right... call it mother's instinct [laughs].

Avdi et al. (2000) have explored the role of parents' knowledge in the diagnosis of children with autistic spectrum disorders. They have suggested that parents' input is central in ascertaining an accurate diagnosis on account of the fact that parents can be regarded as the true 'experts' on their children. For Liz, the responsibility that mothers, in particular, assume for this expertise is naturalised through the notion of a 'mother's instinct'. By virtue of being in a close and intimate relationship with Cara, Liz could both detect a problem and contribute expert knowledge to the diagnostic process that was inaccessible to health care professionals. As Avdi et al. (2000) argue, medical knowledge may be deemed fallible by such parents as it is rooted in seemingly abstract concepts and language rather than 'infallible' everyday sensory experiences by which they come to know their child's condition (Peters et al., 1998). For Avdi et al. (2000) therefore, the process of diagnosis is a 'meeting of experts' where medical and experiential knowledge combine, intersect and interrogate one another in order to produce an explanation for the child's behaviour or symptoms which is acceptable to both parent and doctor. Whilst experiential knowledge was thus a valued resource to parents, and could be used to reject the diagnoses or dismissals of health care

professionals, the very perseverance of parents to obtain what they considered to be the ‘correct’ diagnosis for their child nevertheless points to the significance medical knowledge retained in spite of these issues. Obtaining a medical diagnosis of SMA was indeed an important point of validation for such parents and offered them a socially sanctioned framework through which to interpret their experiences, as well as hope that something could be done for their child. Experiential knowledge may therefore be understood as a key driver in the processes of arriving at a diagnosis of SMA, but as knowledge that was also interpreted in continual relation to medical knowledge. A sense of there being ‘something wrong’ for parents was indeed grounded not only in intimate knowledge of their child, but also in (largely) medically defined notions of child development and normality.

Describing SMA

Whilst many participants were certain in their knowledge that ‘something was wrong’ prior to a diagnosis of SMA in their family, when asked how they would define or describe SMA, there was a diversity of responses.

Participants used a combination of medical, subjective and functional descriptions of the effects of the condition. The key biomedical feature which was present in all of the participants’ descriptions of the condition was muscle weakness, sometimes referred to as ‘muscle atrophy’ of varying degrees.

Understandings of the biological mechanisms through which this muscle weakness occurred, however, were far less clear. Participants explained the aetiology of SMA in very different ways and had contrasting ideas about the

biology underpinning their experiences. SMA was described as a miscommunication between nerve and muscle, as the failure of muscles to grow properly, as a wastage of the nerves supplying the muscle, as a failure of the brain to produce certain proteins required for healthy muscle function, or in terms of genetics, either as a 'deletion' of a particular gene or the 'breaking down' of the gene responsible for muscle growth. Whilst medical terminology and explanations were thus used to describe SMA, this particular conceptualisation of the condition was interpreted alongside, and through, experiential knowledge of living daily with the condition. Trisha is in her thirties has a seven year old daughter, Joanna, who was diagnosed with type I SMA in infancy, and feels that the medical descriptions of muscle atrophy to explain her daughter's decrease in ability over time do not tie in with her observations of her daughter's development, nor discussions with others who experience SMA:

When I describe SMA what I normally say to people is...when you're born the neurons in the spine usually die off...but in SMA, too many die, therefore you lose the ability to walk and you lose the start to the nerves that causes the innervation to the muscle. And um as people get older the muscles kind of get weaker as they get more pressure on them, as the body gets bigger, there is more pressure. I don't actually believe that the muscles waste as such. I don't think I can believe that, from seeing Joanna grow up and watching her and just from listening to

different people's discussions... it's not progressive; it's just that the more weight on the muscles the less they're able to do because there is more weight.

For Trisha, medical descriptions of muscle atrophy were negotiated alongside her personal interpretations of her daughter's disability; medical knowledge became re-interpreted in the face of her experiential knowledge. Whilst Trisha privileged her experiential knowledge in forming her view of the nature of the condition, this knowledge was nevertheless imbued with medical language and descriptions. In challenging dominant medical conceptualisations, Trisha both mobilised medical knowledge but also recast it in terms of her subjective interpretations of SMA. Markens et al. (2010), in their study of women's uses of expert and lay knowledge in decisions about the use of prenatal testing decisions, have similarly argued that the relationship between experiential and medical knowledge in this context may not be oppositional, as it has frequently been described in the literature, but rather *dynamic* and synergistic. Whilst some of the women they interviewed used their experiential knowledge to challenge medical definitions of their reproductive risk, this same medical knowledge was also used to validate and interpret their experiential knowledge. Similarly, for participants living with SMA, experiential and medical knowledge could not always be separated in a clear way when defining SMA, but rather were interpreted, challenged and revised through one another. A challenging of medical descriptions of SMA, as Trisha demonstrated therefore, did not necessarily imply a rejection of medical definitions of SMA more broadly, but rather the selective acceptance of

knowledge that tallied with her own subjective interpretations of her daughter's experiences.

Aside from discussions around what are considered to be the 'classic' features of SMA, some participants further noted additional characteristics which they assigned to the diagnostic category of SMA, but which are not ordinarily offered as part of medical descriptions of SMA. These were derived both from medical sources but also from lived experiences with the condition. The possibilities of those diagnosed with SMA having heightened intelligence, together with an outgoing, sociable personality were among such features. Whilst some medical researchers have attempted to demonstrate the association of SMA with enhanced cognitive ability through the use of Binet and Wechsler IQ scales (Ogasawara, 1989) and more recently through multidimensional tests such as the Kaufman Assessment Battery for Children (von Gontard et al., 2002a), these have failed to demonstrate that intelligence is related to SMA. However, they have suggested that children with SMA may have some heightened cognitive abilities when conducting certain tasks due to environmental factors. It is argued that children with SMA develop cognitive skills to 'compensate' for their lack of physical abilities, as a positive resource for coping with their 'adversity' (Von Gontard et al., 2002a: 134). Whilst these studies are problematic in that they are based on the concept of 'IQ' (intelligence quotient) as an objective and measurable phenomenon (Lezak, 1988) as well as an assumption that SMA is experienced by those who live with it as an 'adversity' to be overcome, for twenty participants in the study, heightened IQ and an outgoing, confident and

sociable personality were as much a feature of SMA as low muscle tone. Dave and Fiona are the foster parents of an 8 year old boy diagnosed with SMA type II and took part in an interview together. They described their son's strong personality and intelligence, which have been attributed by the medical profession to his diagnosis of SMA:

You know he's such a happy boy, he always comes in with a smile, he's always pleased to see you and he learns things every day, he's learning and you know he comes home and he delights in showing us that he's learnt something, and to us, that's fantastic...and he's a comedian, you know, ever since a young age, he's always wanted to make people laugh...and this is not just us, this is to everybody...anybody who's around, he wants to see them smiling. He has this positive energy, much more than anyone else I've ever known. And that's part of SMA children, because I know a couple of other children with SMA who are exactly the same, and it's linked to the SMA, we've been told this, we asked Dr [name] about it and he said 'yes that's part of the SMA condition'. Intelligent, outgoing, lovely children really, yeah.

These personality and intelligence features of SMA, however, were not universally described by all participants. Some participants described feeling very negative and withdrawn as a consequence of their experiences with SMA. This is not to say that these participants did not have outgoing or positive personalities, but rather that the perception supported by some

participants- that people with SMA *necessarily* have positive responses to their situation by virtue of their condition- did not tally with the realities described by some participants. Four participants with SMA, all with different forms of the condition and different ages of onset, for example, reported experiencing depression, suicidal thoughts or had even attempted suicide, in response to the implications SMA had for their lives. The experiences that were described as contributing to this were not only embodied experiences of SMA, but were also social and environmental; two of these participants had lost their jobs as a result of their decreasing physical abilities and one had experienced institutionalisation. These different forms of experience with SMA will be returned to in Chapter 4.

The often contradictory accounts about what SMA is, including the key features of the condition, suggest that SMA was experienced and defined by those who live with it in markedly different ways in line with different knowledge sources, and within a particular social context. The personal and community investment, for example, in medical speculations about personality and intelligence traits associated with SMA, point to the need to affirm the value of the lives of those affected by SMA in the eyes of others, in turn highlighting the devalued status of the lives of people with disabilities in society more broadly. Different conceptualisations of SMA, moreover, were derived as much from living intimately with the condition as they were from medical knowledge of SMA, highlighting the way in which medical definitions and descriptions of SMA were not straightforwardly accepted, but

instead came to be challenged, revised or reinterpreted through experiential accounts.

One area of medical knowledge of SMA which has been particularly contested, both within and without the medical profession, is the way in which SMA's diagnostic sub-categories are ordered, and prognoses offered to families. As has previously been stated, the diagnostic boundaries of SMA have been chronically contested within the medical profession (Munsat and Davies, 1992), and current classification systems do not offer enough information for prognosis (Zerres et al., 1997). The degree of muscle weakness to be expected over the life course, the susceptibility to chest infections, the likelihood of premature death and the extent of anticipated disability are all highly uncertain following a diagnosis of SMA. The medical profession's response to this situation has been the sub-categorisation of SMA into 'types' to differentiate between different clinical trajectories. The resulting clinical classification system (Munsat, 1992) has had a big impact on those diagnosed with SMA, as it is on this basis that diagnosis and prognosis are ascertained. However, as will now be discussed, participants' lived experiences with SMA often contradicted or transcended this typology, leading them to revise their conceptualisations of SMA in line with this knowledge.

The Categorisation of SMA: Diagnosis and Typing

In 1991, an SMA Consortium meeting convened in New York to address, amongst other issues, the ‘diagnostic quandary’ around SMA (Munsat and Davies, 1992). There were debates between those who preferred descriptive classifications of SMA (i.e. dividing those affected by SMA into those who cannot sit unsupported, those who can sit but not walk, and those who can walk), and those who preferred a numerical system for marking out the boundaries of the different types of SMA. However, by the end of the consortium, a consensus was reached for the classification of the childhood SMAs, together with inclusion and exclusion criteria for a diagnosis of SMA (Munsat and Davies, 1992). The classification system drawn up is represented in table 5 below, as well as the ages at which participants in this study received their diagnostic classification, in table 6.

Table 5: Spinal Muscular Atrophy: Clinical Classifications

Type	Onset	Course	Age at Death
1 (Severe)	Birth to 6 months	Never sit	Usually <2 years
2 (Intermediate)	<18 months	Never stand	>2 years
3 (Mild)	>18 months	Stand alone	Adult

From: International Consortium on SMA (Munsat, 1991)

Table 6: Timing of Diagnosis for Participants Diagnosed with SMA

Age at Diagnosis	Number of Participants
< 5	21
5-10	1
10-20	1
20-30	0
30+	2
Total	25

The classificatory system in table 5, together with molecular analysis, continue to be the most common means by which diagnosis and prognosis are reached for SMA today (JTSMA, 2010). The inclusion and exclusion criteria associated with them have further contributed to the differentiation of ‘classical’ SMA from its variant forms, such as Autosomal Dominant Spinal Muscular Atrophy (ADSMA), Spinal Muscular Atrophy and Respiratory Distress (SMARD) and Spinal Bulbar Muscular Atrophy (SBMA) (La Spada et al., 1991) (see Appendix VI for a description of these variant forms of SMA). Whilst sharing similarities in clinical presentation, these conditions have nevertheless been demonstrated to have contrasting genetic aetiology and patterns of inheritance to classical SMA and thus are considered to be separate forms of the same condition.

As well as distinguishing SMA from its variants, research following the establishment of the 1992 classificatory system has attempted to show that the different severities of SMA are linked to different genotypes. As Lefebvre et al. (1995) and Roy et al. (1995) have argued, the extent of the deletion in the 5q-region of the gene deemed responsible for SMA, and the dose of the compensatory SMN2 gene (the so-called ‘rescuer’ gene as it is understood to

compensate for deletions on SMN1- the ‘SMA gene’) appear to influence the clinical course of SMA, so that SMA type I is associated with the largest deletions in the 5q-region and with fewer copies of the rescuer SMN2 gene, and the milder SMA type III with smaller deletions and higher copy numbers of the SMN2 gene. Thus, this form of genetic diagnosis of the type of SMA became incorporated into diagnostic procedures from the 1990s (Dubowitz, 1995a).

Despite this classificatory system and molecular analysis being widely accepted and used to diagnose and classify different forms of SMA, the shortfalls of these methods have been documented in the medical literature on SMA. The considerable variability in presentation and abilities within each classificatory type of SMA (Dubowitz, 1991; Hausmanowa-Petrusewicz et al., 1992), together with examples of incongruence between genetic and clinical findings (Cuscó et al., 2006), have raised questions about whether more genes might be involved with SMA causation than originally thought (Zerres et al., 1997). Indeed, one participant in this study found herself classified under two diagnostic brackets on the basis of contradictory genetic and clinical findings: *‘They say I’m type II, because I’ve got all the signs of type II, but then genetically I’m type III, so it’s quite confusing’* (Beth).

The range of muscle weakness experienced by those within one diagnostic category of SMA as well as the variable age of onset, and also age at death, within these groupings have led to some further attempts within the medical SMA community to expand on the 1991 classificatory system. Zerres and Rudnik-Schöneborn (1995), for example, have suggested an expansion to

include a type IV SMA, with an onset after age 35 and involving relatively mild weakness, whereas Dubowitz (1999) pointed out the possibility of SMA occurring in utero and has thus supported the creation of another diagnostic category, 'type 0 SMA', defined primarily by a prenatal onset. In 1995, Dubowitz (1995b) also tentatively suggested the introduction of a decimal point system, so that each type of SMA could be further sub-categorised on a scale between .1 and .9, depending on severity in order to more accurately capture some of this diversity in abilities and muscle weakness whereas Zerres et al. (1997) suggested a less discriminating sub-category system, between a type I SMA 'a' and 'b'. However there remains little evidence that these systems have been implemented diagnostically, and no one who took part in this study reported being given a diagnosis whereby the type was sub-categorised.

Attempts to produce an accurate categorisation system for SMA within the medical profession have been further hampered by contextual factors. Given that the only fatal component of SMA is reduced respiratory function (due to weakening of the muscles needed to support breathing), improvements in ventilatory support technology and the widespread availability of antibiotics to treat chest infections have meant that many more children diagnosed with severe SMA are surviving infancy, beyond their anticipated age of death, as set out by the 1991 classificatory system; 32% of infants diagnosed with Type I SMA are now surviving past age 2 (Zerres et al., 1997) and type II SMA, whilst once understood to be a life-limiting condition has now been re-classified as many people so-diagnosed are now

experiencing far longer than anticipated life spans. This ‘prolonged survival’ has been attributed in the medical literature to increased knowledge about maintaining appropriate nutrition, posture and respiratory function in this group of individuals (Zerres et al., 1997; Willig et al., 1995; Gilgoff et al., 1989). These compounding factors further highlight the difficulties of predicting the disease trajectory on the basis of genetic and clinical evaluations alone.

For families and individuals keen to obtain an accurate diagnosis and prognosis for SMA, the diagnostic sub-types of SMA may therefore offer parents and individuals an *estimate* of the likely degree of muscle weakness or respiratory difficulties to be experienced by the individual. However, as Nicole et al. (2002) have argued, the typing system that emerged out of the 1991 SMA consortium is best understood as a loose guide for both clinicians and families diagnosed with SMA (p. 4), and there remains no thorough and reliable indicator of the disease trajectory to offer those diagnosed with SMA.

Experiencing the Diagnosis

The arrival at a diagnosis of SMA for the participants in this study typically marked a particularly important point in participants’ accounts of living with SMA, and was the point around which many of their descriptions of life with SMA began. As Jutel (2009) has argued, the power of diagnosis rests in its ability to ‘sort out the real from the imagined, the valid from the feigned, the significant from the insignificant’ (Jutel, 2009: 279), and for participants who had experienced the dismissal of their concerns by other family members or

health care professionals, the diagnosis of a recognisable medical condition provided a validation of their experiences; it was the confirmation that something was 'really' wrong. For parents of children who were diagnosed with type I SMA, however, the diagnosis of SMA typically did not follow a protracted search for answers, but rather was the result of a fraught period of rapid tests following a dramatic decline in health of their child, sometimes only over a few hours. Shannon is 37 years old and was living in Australia at the time she received a diagnosis of SMA type I for her daughter, Millie, following the sudden onset of feeding difficulties at just 6 weeks of age. After spinal x rays and a brain scan failed to identify an explanation for Millie's feeding difficulties, a consultant ordered blood tests:

[Dr's name] looked at her lack of movement in her legs and said that he wanted to take blood and he was testing for Spinal Muscular Atrophy. It was then that it started to become a reality. [Dr's name] said that he wanted the blood tests to rule out SMA, but the best case scenario for Millie was that she would have some kind of Muscle Dystrophy and require a wheelchair. Aidan [husband] and I took Millie home and once she was asleep we spent the whole night crying. The following day [Dr's name] phoned us at home to see how we were and obviously we were distraught. He asked us did we want a second opinion to which we said yes. Unfortunately this day was a Friday and he couldn't get anyone to see us until Monday at 5.15pm.

We had to wait the whole weekend which seemed like a lifetime. Eventually the Monday came around and we saw a neurologist who examined Millie and very bluntly told us that he agreed with [Dr's name] that Millie had SMA. I was stunned but because he was so cold and blunt I didn't cry until we were out of his room. On leaving the hospital I called [Dr's name] on his mobile and said what the neurologist told us and the likelihood of the blood test coming back negative ...he said very unlikely. I felt like my heart had been pulled out of my body and squashed on the floor. And that's how it was diagnosed because the test came back positive.

The importance of biomedical information in shaping the experience of Millie's SMA is evident in Shannon's comment that it was at the point that testing began that SMA 'started to become a reality', despite the feeding difficulties beginning prior to this. The diagnosis of a serious disability in the family has been described as 'family crisis' in the literature and one frequently surrounded by reactions of horror, dismay, guilt, grief and disbelief (Ellis, 1989; Fortier and Wanlass, 1984; Buchanan et al., 1979; Ferguson, 2002). In such 'critical situations' (Giddens, 1979: 127), taken-for-granted assumptions about reality are disturbed and individuals are thrown into a state of confusion and displacement (Bury, 1982). Such reactions were reflected in the accounts of other participants in the study; the diagnosis of SMA was described as a situation in which the 'world was turned upside down' (Trisha,

mother of girl diagnosed with SMA type I). Young et al. (2002) in their studies of mothers of children with cancer and borrowing from the work of Bury (1982), have argued that the point of diagnosis for parents of children with serious conditions represents a point of 'biographical disruption', 'signalling their transition from mothers of a 'healthy' child to mothers of a child in crisis' (Young et al., 2002: 1837). Bury (1982) coined the term 'biographical disruption' to refer to a multi-faceted effect of, and response to, chronic illness. The onset of a chronic and debilitating illness can pose fundamental challenges to individuals' life worlds, exposing them to experiences and realities that were hitherto incomprehensible (Bury, 1997). Whilst Bury (1992) had in mind the onset of a long term illness or disability in adult life when considering biographical disruption, Young et al. (2002) have extended the concept to mothers of children who are diagnosed with cancer. For such mothers, Young et al. argue, the diagnosis of cancer marked a period of major transition where roles, identities and meanings were fundamentally challenged and renegotiated (Young et al., 2002: 1825). Indeed, such mothers had to redefine their caring responsibilities and roles as mothers to incorporate their child's (often intensive) treatment regime and to adjust to a changing relationship with their child, which involved new forms of 'emotion work' and altered conceptions of childhood (Young et al., 2002: 1841). A similar sense of 'biographical disruption' was reported by the three participants who received their diagnosis past the age of ten. For two of them, this occurred well into their adult lives, at a point when they had full time jobs and dependent children. For Brian who was diagnosed with SBMA in his

late forties, the diagnosis marked a point in his life where he had to re-evaluate his life, values and sense of self: *'I had to really work to put my life back together again after that point [diagnosis] because it was just shattered...absolutely shattered. I wasn't who I used to be, and I never could be again'*. The diagnosis of SMA, therefore, whilst on the one hand, marking a point at which participants could establish a socially legitimated reading of their experiences, was simultaneously a point at which previously taken-for-granted roles, identities and relationships were fundamentally challenged. After this, participants often invested large quantities of energy and resources to 'normalise in the face of disruption' (Bury, 1982: 177) and come to terms with the changes the diagnosis meant for their lives and those around them.

Type and Prognosis

The type of SMA diagnosed was described as a particularly significant aspect of the diagnostic process for many families and individuals, as it provided an outline of the anticipated course of the disease. As Rachel, whose daughter was diagnosed with type II commented *'...it lets you know what you're up against'*.

All participants who took part in this study except four were aware of the type of SMA diagnosed within their family, and described it as belonging to one of the 'classic' three types (I-III) or its variants, SBMA, SMARD or ADSMA (see Appendix VI for descriptions). From the four participants who reported that they were unaware of the type of SMA within their family, three stated that they were not interested in knowing a great deal of medical

information about SMA as they felt it to be irrelevant, and the fourth could not recall what type of SMA their sibling had been diagnosed with in the interview.

Despite the apparent importance attributed to diagnostic categories at the point of diagnosis however, as many families reported, there was often an imperfect match between the prognosis offered for a particular diagnosis of SMA and the way in which the condition was actually experienced in day-to-day reality. The classification system and the inconsistencies within it, as highlighted by Matilda's account previously, had implications not only for families' expectations and hopes for the future, but also for their present identities. Trisha, whose daughter, Joanna, was diagnosed with type I SMA described how Joanna's survival beyond the life expectancy assigned to her diagnosis had implications for Trisha's sense of belonging amongst others with SMA:

T: When she was diagnosed, she was diagnosed with type I and we were told she wouldn't see her second birthday, as you are...And obviously, well she's 8 in April, so clearly for her that was the wrong clinical decision...and they won't re-classify her, as they say that clinically she is type I.

F: What sort of impact has that had on you?

T: Well...I'm a volunteer for the JTSMA and I really ummed and ahhed about it because I don't really feel like there's a place for me within it. You know we're not really type I or type II, we don't really fit their...either of those bands because Joanna's is

still here, and when I go to the conferences I get a mixed reaction from people, erm most don't believe she's been diagnosed with type I, because a lot of people there have lost their children to type I and they find it very hard to... understand and accept why Joanna's still here...and their child isn't.

Hedgecoe (2002) has argued that diagnostic categories are important in terms of the development of patient identities, as well as their imaginings of the future. Trisha and Joanna occupied a space between diagnostic categories which served to alienate them from other families affected by SMA. Rather than clarifying the prognosis, the diagnosis of type I SMA reinforced the haziness of the boundaries of the types of SMA, and the large degree of overlap between the types. Whilst Trisha's experiences with her daughter forced her to reconsider the diagnostic boundaries of SMA type I and to re-conceptualise it as a condition with which children can survive early infancy, for other participants, the uncertainty about the boundaries of the types of SMA occurred through the experiences of families other than their own.

The experiential knowledge gained through meeting other people affected by SMA and listening to their stories -what Etchegary et al. (2008) refer to 'vague' experiential knowledge- had an important role to play in clouding the waters of the classifications of SMA and introducing uncertainty to what had hitherto been taken for granted medical information. Lily was diagnosed with SMA in childhood but did not know this was a diagnosis of SMA type II until she was 15 years of age. Whilst not having much contact with anyone else with SMA throughout her life, Lily became more involved

with others with SMA in her twenties when she decided she wanted to have more information about the condition. Lily attended her first JTSMA annual conference when she was 28, an experience which she found particularly bewildering in terms of her understanding of SMA:

When I was 15, I found out that I had type II and you read about what type I, II and III entail, and you go on the Jennifer Trust [website] and you type in SMA type II and it comes up with all this information and you know...I just thought I was so lucky to be type II because you look at type I and you know the outcome and I just thought 'phew' you know, 'you're out of the woods' sort of thing, does that make sense? You sort of think 'Oh thank God, you know, I've had a lucky escape, I've not got type I'. And then you go to the Jennifer Trust [conference] and you see all of the type II people and you think there's *so* much variation and there are people who look nothing like what I thought type II would look like because I'd never had contact really, but I just presumed they'd be like me...So in hindsight, I hung on to the fact that I only had type II and I clung to that and I took that for granted, but having gone to the Jennifer Trust conference...I just don't think the typings are very informative anymore...because the spectrum is so massive and they can't even remotely try to whittle it down to like... descriptions, if that makes sense.

Lily's embodied understanding of what type II SMA was like was fundamentally challenged through attending the JTSMA conference and gaining indirect experiential knowledge of other people's type II SMA. Whereas prior to this insight, the categorization of SMA had been a source of security and certainty for Lily, enabling her to see type II SMA as 'not that bad' and 'taking it for granted', seeing others with type II forced her to reconceptualise the types of SMA in ways that were highly uncertain. For Lily, this blending of medical and experiential knowledge, and the emergent conceptualisations and ways of thinking about SMA, were not necessarily welcome, as she commented: "*I don't think it's that helpful to experience and see it really [different people affected by SMA at the JTSMA conference] I always say 'ignorance is bliss' and I still swear by that*" (Lily). Henwood et al. (2003) in their analysis of debates around the expert/lay knowledge divide use the example of the emergence of the 'informed' or 'reflexive' patient to argue that a fundamental oversight in these debates exists. They argue that there are particular constraints on patients' abilities and willingness to become informed about their own health condition. One such constraint is the reluctance of patients to take on board health information about their condition (p.604); in many instances, they argue, the preference of the patient is to defer to expert medical knowledge in order to remove difficult or challenging decisions or distress that may emerge from this knowledge. In a similar way, Lily wished to avoid coming into contact with extensive information about the condition affecting her. For her, medical information about SMA could potentially be distressing and unsettling rather than

transforming her into an ‘informed patient’, and thus in order to minimise the influence the condition had on her life, she actively avoided the acquisition of such knowledge.

For other individuals, the classification system for SMA, rather than blurred and unreliable as Lily’s account suggested, was instead presented as being definite and clear cut. This was especially so for parents who had children who died of SMA type I. For some of these participants, the apparent certainty around the classifications of SMA (and thus the associated prognoses) enabled these parents to accept their child’s prospects. Charlie is in her 50s and experienced the deaths of two children (Jack and Alexis) from SMA type I, both within the first year of their lives. Presenting the classificatory system and associated prognoses as definite played a pivotal role in aiding her acceptance of her first baby (Jack)’s diagnosis and subsequent prognosis:

...when the doctors got the tests back and they told us they were 100% sure and that he would *definitely* die and there was nothing I could do about it, I felt quite relieved at that because I knew that there was nothing I could physically do for Jack, you know, he would die, and that created a sort of comfort for me because I knew that no matter what I did, you know, he *would* die. It wasn’t like cancer you know where there’s a chance of them living but there’s a chance of them dying, I don’t think I could have coped with that. I couldn’t have coped

with that kind of uncertainty. And so we could accept that, and that was a fact.

Charlie's interpretation of the medical classifications, and thus the prognosis associated with type I SMA as an undisputable and inevitable 'fact,' once again points to the way in which experiential and medical knowledge are mobilised together in forming conceptualisations of, and responses to, SMA. Whilst Charlie's experiential knowledge of SMA was, at this point, limited to that of Jack who had just been diagnosed with the condition, her experiential knowledge of how to make sense of, and attribute authority to, genetic test results, and, further, her conceptualisation of a comparable condition (cancer), were used to reaffirm medical classifications of SMA and the certainty of the fatality of Jack's condition. The perception of certainty around diagnosis and prognosis offered Charlie a sense of reassurance and acceptance, and consequently relieved her of the difficulties which she associated, through her experiential knowledge, with an uncertain or changeable prognosis.

For other participants, however, the presentation of *uncertainty* surrounding the different types of SMA had functional benefits. Rhona is 27 years old and was diagnosed with SMA type III in early childhood. However, at this time, she was not given any indication as to the type or prognosis associated with this diagnosis. She was able to remain ambulant into her teens and now in her twenties uses a wheelchair, but is able to walk short distances with the assistance of elbow crutches. For Rhona, the uncertainty surrounding her prognosis and the lack of expectation was central to the maintenance of her mobility:

...There wasn't much information available at all [at the time of her diagnosis], we were told that the outcome was uncertain really and I think in the long run that served me well...the one doctor I was seeing when I was going through a rough patch when I was 13... I was putting weight on and I was finding mobility really really difficult, he strongly believed that from then I could quite easily have made the decision to go into a powered wheelchair and then I would have lost the ability to walk, but because we just weren't expecting that to come, we expected me to be mobile... forever sort of thing, and hoped that I would be, it was very much, you know, I was really encouraged to walk as much as possible, by my family. And I kind of got through that phase through doing a lot of exercise and I do feel that it would have been a very logical point to say, 'right, stop the fight, kind of thing, start using a powered wheelchair' and then I think that that obviously would have had a knock on effect on...I would have deteriorated then, but it was *because* we weren't sure and we didn't know to expect that I kept going.

For Rhona and her family, uncertainty had a large role to play in her experience of SMA; the optimism from her family derived from the lack of a clear prognosis and publicly available information on SMA. Davis (1960) in a study of parents and children affected by Paralytic Poliomyelitis, refers to

‘functional uncertainty’ (p.45) as being a mechanism utilized in doctor-patient relationships to divert the emotional distress associated with a poor prognosis and a means by which families and patients can remain optimistically hopeful about the future. The lack of information concerning Rhona’s prognosis and the associated uncertainty opened up a space for Rhona and her parents to remain optimistic about the future and led to the avoidance of interventions that, Rhona feels, would ultimately have been counter-productive in her development. Through not knowing what to expect from life with SMA, Rhona was able to adjust in a way that she felt she might not have been able to had she known from the outset that she would eventually lose the ability to walk. In this way, uncertainty within medical knowledge both shaped Rhona’s experiences with SMA, but also became the basis from which she challenged medical categorisations:

...I’m just not sure that the types [of SMA] can tell you very much, as you can tell from my experience [laughs] and there can be so much variation...I mean, they still don’t really know what type I am, one doctor’s said ‘you’re definitely a type III because you’ve got your mobility’ and another one has said ‘you’re definitely a type II because you’re too poor to be a type III’ ...[...]... So really, it’s not all set in stone and there’s such a wide spectrum...we don’t all fit into the boxes!

Experiential knowledge and medical knowledge of SMA, therefore, both fed into and informed one another in the construction of Rhona’s perception of

SMA, and her responses to it. Whilst Rhona was aware that the medical typology of SMA had limited value in terms of offering a prognosis, and her experiences defied medical classification, the centrality of this system as a way of conceptualising SMA nevertheless was apparent in Rhona's response, and was reflected in many participants' responses. Whilst medical knowledge of SMA, therefore, was contested, it nevertheless was central to the way in which participants constructed their perceptions of SMA.

As has been discussed within this section, families and individuals living with SMA had to negotiate and reconcile different forms of knowledge in arriving at understandings of what SMA is, and how it should be classified. Contradictions and tensions often existed between expert medical knowledge of SMA and the way in which it was experienced by those living with it in their daily lives. For some participants, this lack of clarity had functional benefits, whereas for others, the presentation of medical and experiential knowledge as certain enabled them to make sense of, and manage the lived realities of SMA.

Experiential knowledge emerged as a particularly important resource within participants' accounts; it could be drawn upon for practical advice and reassurance, or to challenge medical knowledge. As one mother, Natasha, who has identical twin boys both diagnosed with SMA type II, commented about the importance of information gained from JTSMA members:

...The prognosis we got from the hospital for the boys was 'may not survive childhood' but the prognosis from the Jennifer Trust and the families we meet there is '*can*

survive into adulthood' and we prefer to look at the positives. It is important for us to be a part of the Trust and see all the children and young people because you realize that SMA is so broad and that the doctors can only tell you their perspective, but there is a lot to be gained from speaking to the people who live with it...they are the ones who *really* know what it's like.

The contrast Natasha draws between the medical prognosis she received for her sons, and the 'prognosis' gained from experiential knowledge, from 'the ones who *really* know', is revealing in terms of the way in which experiential knowledge can be mobilised not only to support and validate certain experiences, but also to displace and contest medical knowledge in different contexts. Whilst Natasha may still value medical knowledge of her sons' condition, her assertion highlights the perception, held by participants in this study, that the knowledge of SMA possessed by those living intimately with it is a multi-faceted form of knowledge, inaccessible to the medical profession, and one based in lived realities, rather than abstract notions, as Natasha went on to comment:

...doctors obviously have their views on SMA, and you take that on board... what they recommend, but you also keep in mind that they're coming at it from the point of view of muscles and cells and nerves and all that, and...really, the way they see it isn't necessarily how we live it.

As Natasha highlights, not only are experiential and medical knowledge of SMA derived from different sources, but their content is also substantially different. Indeed, medical knowledge of SMA could often be experienced as alienating given its highly technical and seemingly abstract nature, which did not necessarily tally with everyday experiences of SMA.

Genetic knowledge of SMA was an aspect of medical knowledge which further had a substantial impact on the way in which SMA was understood and responded to, both by the medical profession, but also by those living with SMA, as presented below.

The Geneticization of SMA

The location of the gene deemed responsible for causing SMA in 1990 by Gilliam et al.'s group in New York, and shortly after by another team in Paris (Melki et al., 1990), was heralded as a landmark in the history of SMA (Dubowitz, 2008). It enabled the use of prenatal diagnosis of SMA through the use of DNA markers, and was widely regarded as a starting point for the development of effective treatments or even a cure for SMA. SMA became classified as an 'allelic disorder', meaning that it involves a pair of genes on a chromosome, and linkage analyses mapped this chromosome as 5q11.2-13.3 (Melki et al., 1990). SMA came to be understood as inherited in an autosomal (i.e. relating to a chromosome that is not a sex chromosome) recessive pattern. However, as the region of the aforementioned chromosome is regarded as unstable (Nichole et al., 2002), there are also a high number of so-called 'de novo' mutations deemed to cause SMA. 'De novo mutations' refer to genetic

mutations that are not inherited, but occur during conception. These de novo mutations can subsequently be passed on to future generations. Melki et al. (1994) estimate that approximately 2% of those diagnosed with SMA have such de novo mutations. However, the vast majority of those diagnosed with SMA are found to have mutations in the SMN1 (Survival Motor Neuron) gene, on chromosome 5q. In a study of 500 patients with SMA by Zerres et al. (1997), 96% of those with SMA type I were found to have deletions on this chromosome, 94% of those with type II and 82% of those with type III SMA (Zerres et al., 1997: 202). Whilst the identification of the gene deemed responsible for most cases of SMA did little to resolve debates about how to classify the various presentations of SMA, genetic understandings of SMA have altered the way in which it is diagnosed, and also the way in which cures and treatments are conceptualised. Research on cures for SMA, for example, has concentrated efforts on different ways of maximising the function of SMN2, the so-called 'rescuer' gene, to compensate for the deleted SMN1. This process is known as 'transcriptional activation' and correction of the 'splicing' of a copy gene (Wirth et al., 2006). Activation of this process has been attempted through different chemicals, such as sodium butyrate (Chang et al., 2001) and valproic acid (Brichta et al., 2003), as well as through stem cell and gene therapies (Kerr et al., 2000; DiDonato, 2003). More recently, the restorative possibilities of embryonic stem cells administered following muscle atrophy have been suggested (Corti et al., 2008). These studies highlight the potential to reverse the effects of SMA after they have started,

although the direct benefits of this research to those living with SMA through a definitive cure remain elusive.

The Genetic Diagnosis

Whilst the diagnosis of a serious and incurable physical condition in the family was experienced as a major disruptive event for participants in this study, the genetic aetiology of the condition carried with it its own specific implications for the families and individuals who took part. The medical and psychological literature points to the possibility of parents and families of children with serious conditions experiencing a sense of responsibility for the condition afflicting their offspring (Chapple et al., 1995; O'Dougherty and Brown, 1990; Kerr and McIntosh, 2000; Young et al., 2002). However, for families diagnosed as being affected by an *inheritable* condition, the experience of guilt may be particularly pronounced (Kessler, 1998; Kay and Kingston, 2002) and has been documented as being a common parental reaction in relation to the diagnosis of other genetic conditions of childhood such as DMD (Buchanan et al., 1979), Myotonic Dystrophy (Faulkner and Kingston, 1998), Fragile X Syndrome (James et al., 2006), Sickle Cell Disease (Evans et al., 1988) and CF (Fanos and Johnson, 2005) as well as being observed in anecdotal accounts of parents of children with genetic conditions (Gore Olsen, 2006). Indeed, by their very nature, genetic conditions implicate more than one individual, and the consequences genetic information has for family relationships as well as individual subjectivities has been widely explored, particularly in relation to the notion of

responsibility (Burgess and D'Agincourt-Canning, 2001; Dragonas, 2001; Kay and Kingston, 2002; Hallowell et al., 2006; Hallowell, 1999; Hallowell, 2003; Hallowell et al., 2005; Downing, 2005; D'Agincourt-Canning, 2001; Reed, 2007; Rhodes, 1998; Rose and Novas, 2004). For Polzer et al. (2002) and Novas and Rose (2000), the increasing availability of genetic knowledge has led to new forms of personhood; our notion of self has become inherently relational, and imbued with responsibility to actively manage health information, not only for ourselves, but for our biologically related kin. Polzer et al. (2002) have related this shift to 'neoliberal programmes of governance' (Polzer et al., 2002: 156), whereby individuals are encouraged to regulate their actions and selves in line with political objectives. Thus, the assumption of responsibility for one's own health is a characteristic of neoliberal societies, and the practices of genetic testing and the procurement of genetic risk information, not only for ourselves, but for our biological kin, has been regarded as an extension of this obligation (Lupton, 1995; Peterson, 1998). As Hallowell notes:

Biomedical discourses construct genetic risks as internally imposed involuntary health risks. However, the fact that these risks are involuntary does not absolve gene carriers of the responsibility to act to protect their health. Indeed, it could be argued that because genetic risks are portrayed as part of the individual's make up, their responsibility to act to protect their health, or the health

of future generations, is emphasised, for inherited risk cannot be blamed on external sources.

(Hallowell, 1999: 599)

For families affected by SMA, particularly for parents, there was a strong sense of responsibility attached to the genetic diagnosis of SMA and many parents, and grandparents, blamed themselves (or each other) for having passed on the condition to their offspring. Guilt is strongly associated with responsibility, in that guilt can be understood as an emotional response to feeling responsible for some (perceived) offence. For five parents of children with SMA, the knowledge that SMA had been inherited was described as the hardest part of the diagnosis, whereas for others, this experience of guilt was transient and recurred in particular contexts. Paula has a 13 year old daughter, Tamara, who was diagnosed with SMA type II, and a 9 year old son, Ethan, who is able-bodied. Paula described her reaction to the inheritable nature of SMA in the following way:

When we first found out it was genetic...um... I suppose you just accept it really because I don't think there's anything you can really do...if you've got that gene... and I suppose we just accepted it really. We knew it came from one of the parents, you know, of ours [Paula and her husband's]. But then just sometimes... you know we look at Ethan and then you know look at Tamara and sometimes you know she will say 'why can't I do this?' or 'It's not fair' or 'how do you think I feel?' and when

you do hear her saying these things sometimes you do think well 'it is our fault that you're like it', you know me and my husband's, you know, we gave it her.

For Paula, her initial acceptance of the genetic diagnosis as being beyond her personal control became displaced by her everyday experiences of her daughter's condition. Watching her daughter struggle or comparing her son's abilities to her daughter's triggered feelings of responsibility or guilt; they were contextually dependent and not a constant factor in her life. For other participants in the study, reactions to the genetic diagnosis and the implications for the experience of guilt were inextricably tied to the nature of the SMA experience; where the effects of SMA were perceived to be particularly severe or debilitating, guilt reactions appeared to be stronger, or surfaced at 'critical points' (Bury, 1982) e.g. at diagnosis, when witnessing suffering, when undergoing major surgery, or at the death of someone from SMA (which will be returned to in Chapter 4). Genetic information was thus received and responded to in the context of the experience of SMA and was understood both in relation to, and through, this lived reality.

The intersection of experiential and genetic knowledge of SMA, as well as prompting guilt reactions, has also meant that the 'geneticization' (Lippman, 1991) of SMA has not been universally accepted by those living with SMA as relevant or significant in their lives. Isabella is 21 years old and was diagnosed with SMA type II in early childhood. She has used a wheelchair all of her life and is currently attending university, completing an

undergraduate degree. For Isabella, genetic information about SMA has very little meaning for her:

Genetics is something I've never really thought about a great deal to be honest. I mean I know it's [SMA] genetic, and I saw a lot of posters when I was little for 'Jeans for Genes' day and I'd see SMA and I'd be like 'oh that's what I've got' but genetics...it just doesn't really mean anything to me. I don't really understand how it relates to my life...I mean, I know what genes are, I did Biology A level, I know all about the science or whatever but I just don't really like to put a label on it.

For Isabella, the genetic language medical professionals use to describe the aetiology of SMA did not speak to her own life; identifying her condition in this way was, for Isabella, part of a medicalised way of thinking about, and understanding her experiences, in which labelling and categorisation are of fundamental importance. However, Isabella's subjective experiences of her self, identity and life allowed little room for such clearly defined categories:

Do you know though, I don't even think of myself as a disabled person either, I just don't like categorising it in that way because yeah I'm 100% reliant on people physically, if I was left on my own for three days I would die because I can't get to the tap, but then mentally I'm not reliant at all. But then you know some people who may be able to look after themselves physically might not be able to sort out their

money, or they may not be very strong, you know, psychologically, in their thinking...so there are loads of things and thousands of different ways of looking at abilities and disabilities. So I just don't really see the need to label it at all.

The medicalisation of Isabella's life through the application of a diagnostic label, and the social categorisation to which she felt subject, did not sit comfortably with her perception of her own life (or the lives of others), in which identities and subjectivities were far more fluid and unstable than such compartmentalised thinking would permit.

Genetics and the Notion of Cure

Whilst some experienced the geneticization of SMA as a way of viewing SMA abstracted from their daily realities, for others, the genetic status of SMA was more intimately bound to the notion of cure. The construction of SMA as a genetic condition which carries with it the suggestion that it may one day be cured through advanced genetic technologies, is a representation of the condition frequently harnessed by the JTSMA and other 'genetic advocacy groups' (Novas, 2007) as a means by which to secure public funding for research activities and to support fundraising activities. The priority given to genetic explanations for SMA by organisations such as the JTSMA has meant that the experiential knowledge of SMA accumulated within and between members of the JTSMA has thus, inevitably, been shaped by this genetic knowledge. For some members, this genetic knowledge became a means by which to deal with everyday experiences of SMA and

frames their experiential knowledge of the condition. Rakesh is 51 years old and was diagnosed with ADSMA in his 40s after experiencing increasing muscle weakness over a period of several years. For Rakesh, the concept of a cure through the use of stem cells was particularly important:

...I have to believe that they will find a cure for SMA. I just have to. You see I follow the developments in the research, they are developing it every day and they can use stem cells now to treat it, don't they? In China...there was an article, the other week, or the week before.... in the Observer, a woman from Manchester and they found the gene that causes her condition, and they replaced it with stem cells in the brain. One month after the treatment, the necessity for her to use the wheelchair is not there anymore so she is moving about without help of wheelchair...so I am hopeful for that. I remember that when I am experiencing my problems, a cure will be on its way.

Holtzman (1999) and Fleising (2001) have used the term 'genohype' to describe the way in which advances in genetic medicine have encouraged the development of unrealistically high hopes and expectations for treatments and cures amongst those living with genetic conditions (as discussed in Chapter 1). Indeed, whilst Rakesh appeared to acknowledge that there was a possibility that such a cure may not be forthcoming, the anticipation of it nevertheless shaped his daily life with SMA, enabling him to stay hopeful that his symptoms would one day be alleviated. Shakespeare (2008a) has

pointed to the way in which such genotype, or the rhetoric of cure being ‘just around the corner’ (p. 101) is one that is frequently mobilised by medical researchers in order to secure funding for, and to validate, their own work. However, the reality of progress in this area has not kept pace with the raised hopes of many families and individuals living with SMA, as I witnessed myself at the 2009 JTSMA conference, in the fraught exchanges between the guest speaker, a leading geneticist researching SMA, and the parents of young children living with SMA.

However, the geneticization of SMA and subsequent suggestion of cure or intervention has not been accepted as relevant or significant by all of those living with SMA. Kristen is in her late twenties, was diagnosed with SMA type II in childhood and has never been able to walk. She has an older sister, Ellie, who was diagnosed with type III SMA at the same time that Kristen received her diagnosis:

I don't really know and I've always um not really been that bothered about a cure. I know it's [SMA] genetic, but in my day to day life that doesn't really mean that much...and I've never really, um, been, um, focused on treatment and 'there *has* to be some sort of cure'...some people are like that and...that's not really been something I've been that bothered about. I'd rather focus on what I can do now. Often these things [cure] don't end up happening anyway, do they? I'm sure if you spoke to my sister, she'd have a different opinion, even

though people expect me to feel more strongly about it because it [SMA] affects me in a more severe way than her.

For Kristen, maintaining her focus on her experiences of SMA in her daily life was a means by which to manage the hype surrounding genetic technologies and consequently her expectations of a cure that may, or may not, materialise. She further relates this disinterest in the concept of cure to the very *nature* of her impairment, and her experiential knowledge of it. Shakespeare (2008a) in his writing on the response of disability rights supporters to the notion of cure, has highlighted the nature of impairment experiences as a significant influence on disabled people's attitudes towards, and acceptance of, the notion of cure. Those with permanent and unchanging impairments, particularly those present since birth or early childhood, he has argued, have typically been less interested in the notion of cure than those whose impairments may be described as degenerative or painful. Within this latter impairment group, the prospect of cure often develops into a quest to return to a (previously known) state of higher functioning or able-bodiedness (Shakespeare, 2008: 106), articulated through the language of genetic medicine. Indeed, for Rakesh who, unlike Kristen, had witnessed a decline in his abilities over time and wanted to regain the life he had previously experienced, approached genetic research into SMA from a different perspective. The framing of his experiences of SMA as amenable to both change and recovery, moreover, not only enabled him to remain hopeful about the future, but also to manage the uncertainty of his present with SMA.

The nature of impairment experiences associated with SMA will be returned to in Chapter 5.

Conclusions

In conclusion, using the examples of the emergence of SMA as a disease entity, the development of its medical typology and finally the implications of its geneticization, this chapter has set out some of the tensions between expert knowledge of SMA and the experiential knowledge of those who live with it, and how they are played out in these contexts. The uncertainties inherent within this expert knowledge- not only as to where the boundaries of SMA lie, but also with regards to how it can be understood, predicted and treated- have, in various ways, impacted on the way in which SMA is lived through in the daily lives of those experiencing the condition, and, consequently, the experiential knowledge accumulated from these experiences. Whilst in some instances medical knowledge of SMA was challenged by reference to experiential knowledge, expert medical knowledge nevertheless framed and contributed to that experiential knowledge even as it was resisted. As Markens et al. (2010) have suggested, the relationship between expert and experiential knowledge may be more synergistic and dynamic than previous research has suggested. Researchers, for example, have tended to emphasise the uses of experiential knowledge as an alternative to medical knowledge and have interpreted its validation as a form of resistance to medical knowledge. However, even though many participants challenged medical descriptions of SMA and questioned the relevance of

genetic explanations to their lives, they nevertheless framed their conceptualisations of, and experiences of, SMA in medical frameworks. Indeed, an understanding of oneself as a '*good type II*' as one participant, Georgia, commented, is grounded in a notion of how SMA type II *should* be experienced, according to this medical knowledge.

As Abel and Browner (1998) have argued, experiential knowledge was, moreover attributed a very particular status vis-à-vis medical knowledge. For families and individuals living with SMA, this knowledge was highly valued; it offered them a form of security and point of reference at times when they felt the most vulnerable, particularly in instances where expert knowledge was felt to be especially uncertain and fallible. As Natasha highlighted, experiential knowledge was often felt to be more secure and '*real*' (Natasha), being grounded in the everyday realities of life with SMA, onto which medical experts have only a limited window. The experientially based ways of understanding SMA, as they emerged through participants' accounts, will now be presented in Chapter 4.

Chapter 4

Living with SMA

In Chapter 3 I presented conceptualisations of SMA and the impact of the diagnosis of SMA on families and individuals. This analysis has revealed the divergent experiences and conceptions of SMA amongst those who participated in this study, as well as reactions to clinical and genetic understandings of the condition. This chapter will present participants' accounts of the impact of SMA beyond the diagnosis, in their day to day lives. Whilst the diagnosis was a crucial point in participants' accounts of their lives with SMA and offered a socially legitimated reading of their experiences, it was the meaning SMA took on and the consequences it had in their everyday lives that formed the bulk of their stock of experiential knowledge of the condition. It was through living with the condition that participants came to reassess their initial reactions to diagnosis and prognosis, and also redefine the condition's significance over time. It was, furthermore, through these experiences that participants accumulated knowledge that became both a resource for the management of day to day living but also a point of reference in the context of familial reproductive decision making (as presented in Chapter 6). It was the sorts of experiences they felt they had with SMA, and perceived others to have, that were important in imagining future lives with, or without, the condition.

These experiences will be presented under three thematic sub-headings- 'experiences of disability', 'embodied experiences of impairment and disability' and 'experiences of illness, death and bereavement', rather

than using the medical typology to categorise and order the divergent range of experiences associated with a diagnosis of SMA, which were discussed in Chapter 3. Indeed, this chapter will address the question of whether such medical categories can accurately account for the range of experiences within each 'type' of SMA, and the associated assumptions about the correlating level of severity of the condition. Participants' experiences often extended beyond the boundaries of each medically defined 'type' of SMA in a non-linear fashion with a constant movement back and forth between the different types of experience. Finally, I will present the ways in which participants made sense of these experiences, and the strategies they used to overcome the problems associated with them will also be presented. Despite commonly held assumptions about the lives of individuals with SMA as necessarily difficult and constrained, participants reported a range of creative and innovative coping strategies and philosophies to counterbalance any negative implications of living with SMA. By presenting these accounts of SMA, this chapter will problematise medicalised and popular presentations of the experience of SMA.

Experiential Accounts of Living with SMA

Despite there being an extensive literature documenting the medical complications associated with severe SMA, less is known about how it is experienced by affected families and individuals. A study by Bach et al. (2003) attempted to measure clinicians' perceptions of 'quality of life' for children diagnosed with type I SMA as compared to care providers'

(primarily the parents) perceptions, and discovered a wide discrepancy between the two. The care providers, in contrast to the clinicians' perceptions, reported many positive and fulfilling experiences of living with SMA and rated their children as being happy, despite physical difficulties. A similar study examining the impact of SMA on familial stress levels reported that families with children diagnosed with SMA often develop good coping strategies, compared with families with children diagnosed with Fragile X Syndrome, which the authors attribute to the physical nature of SMA in contrast to Fragile X Syndrome which can involve physical and mental disabilities (von Gontard et al., 2002b: 955). Such findings are also supported by the social science literature on the impact of a disabled child on the family (Asch, 1999; Ferguson et al., 2000), which suggests that the experience is not always negative (as is so often assumed), and that parents of disabled children can and do have rewarding experiences of parenthood. This is not to say that SMA cannot be challenging for families, indeed, studies have pointed to the 'burden' of care involved with caring for a child with high support needs (Boyer et al., 2006), and the difficulties experienced by able-bodied siblings who may receive less parental attention than their disabled sibling (Laufersweiler-Plass et al., 2003), but rather, that research suggests that families affected by SMA may nevertheless thrive in spite of the financial, practical and emotional demands associated with care for a child diagnosed with severe SMA.

As discussed in Chapter 3, SMA can be experienced in lots of different ways, and as such has been categorised into 'types' by the medical

profession to reflect these differences. Each type is associated with a particular level of anticipated ability, life expectancy and 'quality of life', and the type of SMA diagnosed is often used as a proxy to refer to the sorts of experience that the individual or family can expect. However, these medically defined types do not necessarily correlate with the experiences of those living with a particular 'type' of SMA; many spoke of a lack of identification with the experiences of others who have been diagnosed with the same type of SMA as themselves:

When my family met another girl with the same type of SMA as me, they just could not believe we had the same condition, we're so different.

(Lily, diagnosed with type II SMA)

This could also be experienced as a lack of identification with the diagnosis of SMA itself :

I just sometimes think it would make more sense to have different names for it, the kind of concerns we have as parents of a type I baby are so different to those of parents who have children with type II or III...we're worrying about CPAP machines [positive airway pressure] and suction and how long we'll have our babies for, whereas type II parents are worrying about wheelchairs, access to schools, equipment for cars, those sorts of concerns. I guess it's just because it's caused by

the same gene, that's what connects us, but it presents very differently.

(Mark, father of baby with SMARD)

For Mark, as for many other participants, it was their experiences of SMA that came to have a significant impact on their perception of the condition and the similarities and differences between others with the condition. This was not only informed by medical definitions of SMA, but also a sense of shared experience; shared challenges, shared losses and shared grieving. The next sections present some of these shared experiences of SMA, and have been categorised according to experiential theme rather than diagnostic category. Whilst the medical typology of SMA has many uses, and proved to be a helpful reference point for participants, informing their overarching conceptualisation of the condition, the typology cannot fully account for the range of experiences within each category. This chapter thus problematises some of the assumptions inherent in medical definitions of the SMA experience, particularly the assumption that SMA can be understood as being measurable on a scale of severity. In particular, the suggestion that those types of SMA medically defined as more severe *necessarily* involve higher degrees of suffering, or are experienced more negatively than those that are defined as less severe, is explored. This analysis is organised under thematic headings, 'experiences of disability', 'experiences of embodied impairment and disability' and 'experiences of illness, death and bereavement'. It is important to note that these themes are not designed to describe the experience of any particular diagnostic type of SMA, but rather they serve to demarcate

different forms of experiential knowledge, between which individuals with SMA and their families may pass through and between at various points during their experience with the condition.

Experiences of Disability

One of the central emergent themes when participants described their experiences of living with SMA was the prevalence of the experience of disability, and also participants' strategies for overcoming or managing it. Definitions of disability have altered over time, ranging from a physically based description of 'abnormality', bodily 'deficit' or 'incapacity' (Thomas, 2002: 38) to contextual definitions, as supported by social model of disability theorists (e.g. Oliver, 1996b; Barnes, 1992). The writers and activists who developed this latter definition of disability drew attention to the social and physical restriction experienced by disabled people, which they saw as arising from society's failure to accommodate their needs and rights as opposed to any bodily difference or deficit (Oliver, 1996a). Thus, definitions of disability which follow social model of disability theorising have removed the association of impairment (physical difference) with disability (its social product), and emphasised the role of the social and spatial environment in creating disabling barriers (Barton and Oliver, 1996). In order to support an emphasis on the social origin of disability, social model of disability theorists have simultaneously removed the body from an analysis of disability, the site at which traditional (i.e. medical model) explanations have focused (Paterson and Hughes, 1999). This contextually based definition of disability supported

by social model of disability theorists will be used to explore the experiences of individuals with SMA, as for many participants, experiences of restriction, whether social or physical, were often seen as arising as a consequence of environmental, rather than physical factors.

Despite Lamb and Peden's (2008) exploratory study of peoples' experiences with SMA focuses on SMA as an 'illness' with reference to strategies of 'symptom management', individuals who took part in my study rarely spoke of their experience in these terms. Instead, for many participants, the emphasis was on the way in which their social and physical environment shaped their experience of SMA, and mobility was a key element of this experience.

Powered wheelchairs are the most commonly used mobility aids for those living with SMA as they require minimal muscle strength to operate and can be used by children as young as 20 months. These chairs allow for independent movement and current models allow the user to be raised to standing height or lowered to the floor to retrieve objects, which has altered the autonomy and capacities of those affected by SMA to control their own environment (Jones et al., 2003). Whilst these chairs offer users increased autonomy, their weight and size (which is considerably greater than that of manual wheelchairs) together with their use in a society in which physical access for wheelchair users is limited, means that many families and individuals living with SMA continue to face environmental obstacles in their daily lives. Kate is in her 30s and is the mother of a 7 year old boy, Jamie, who has been diagnosed with SMA type II. Jamie now uses a powered

wheelchair full time which prompted the relocation of the family to a house which can accommodate the extra space required to manoeuvre the chair as well as the spending of considerable resources to adapt the house to make it fully accessible for him. For Kate, her experiences of SMA were defined primarily by her experiences of social and environmental problems:

I think a lot of the things I go through with Jamie on a day to day basis, I think it's not his condition that's the problem, it's everything else that goes with it, you know, the lack of help, the lack of adapted places, all those sorts of things which are the hardest... I do think that could be easier... it's like 3 years ago I wanted him to go to a school which wasn't adapted, and they couldn't do it, I fought them, but it was too much money so...It's difficult getting transport to the school he does go to now as well, so we walked it. We can get transport, but he can't use it because of the wheelchair he needs. So really you need a hell of a lot of money, you know they out-grow their wheelchairs, and you have to keep replacing them, you can never go on holiday where you want to....You know it's all this kind of thing all the time, things never turn up at the school or things never get ordered, that sort of thing, it's that stuff that makes it so bloody hard, not really the SMA itself.

Families affected by SMA frequently spoke of the need to 'fight' to get appropriate support (the right social security benefits, equipment, adaptations

and access), constructing 'battling' as an important strategy by which to overcome difficulties associated with a society poorly equipped to cater for people with disabilities. Whilst experiences of disability were therefore presented as amenable to social and environment manipulation, the emotional and physical demands of this 'fighting' and need to be assertive was also evident in participants' responses. As one foster mother of a young boy, Marcus, diagnosed with SMA type II commented:

When everything's in the right place, all the equipment's sorted, that's when you can forget about SMA because it's not so much of a struggle. But you do have to be incredibly assertive. Since having Marcus in my life, I've had to learn to really fight for things and stand up for his rights, things that I previously took for granted.

The familial stress potentially involved with managing the high care needs of a disabled member have been well documented in the literature (Farber, 1960; Baxter, Cummins and Pollack, 1993; Floyd and Gallagher, 1997), and the availability of appropriate support, interventions and adaptations is acknowledged as crucial to the adjustment of such families to disability. Children with SMA in particular may require regular physiotherapy, hospital check ups, surgery and the regular replacement of their equipment and aids (such as wheelchairs) as their bodies grow and change, on top of their day to day care needs. The medial literature points particularly to the orthopaedic complications associated with prolonged sitting, which may pose specific challenges to those affected by SMA such as joint contractures and scoliosis

(curvature of the spine) (Evans et al., 1981). Whilst the suppleness of joints may be maintained to some extent through the use of physiotherapy exercises and orthopaedic interventions (such as standing frames and serial plaster casting), many people affected by SMA experience permanent joint contractures in their legs, which can interfere with personal care and daily activities, necessitating the use of aids and adaptations (Wang et al., 2007). Scoliosis is ordinarily treated with bracing and/or a spinal fusion operation, which involves fusing the spine in a straight position surgically. This operation is a major procedure with a long recovery period (Aprin et al., 1982), but one which may improve respiratory function and sitting balance (Wang et al., 2007), in spite of the potential side effect of further loss of mobility and flexibility post surgery (Furumasu, 1989).

The management of children living with SMA in their day to day lives therefore involves the input of a range of professionals from both health and social care, as well as considerable amounts of the family's resources which may constrain the family's social and other activities (Wang et al., 2007). Seven siblings of people with SMA reported that they felt that they had received less parental attention than they otherwise might have done due to their parents' distraction with caring for their child with SMA; a finding reflected in many studies on sibling relationships within families with disabled children (Laufesweiler-Plass et al., 2003; Labato, et al., 1988; Fleitas, 2000).

For adults with SMA as well, access to resources appeared crucial to how SMA was experienced. Two participants with SMA who reported

particularly negative experiences with the condition had both lost their full time jobs as a result of their deteriorating abilities and many others emphasised the importance of access to resources in overcoming the restrictions in terms of housing, jobs and education. For Hannah, who is 42 years old and was diagnosed with SMA type I in childhood, having the right resources at her disposal is crucial to the way in which she experiences her life:

...having a quality to my life, for me, means having somewhere to live that's an ok environment, in ok surroundings, transport, enough finances to be able to pay for the people that I want assisting me, e.g. not from an agency but privately employed [this makes the difference of disabled people being able to hire employees of their own choosing rather than allocated workers]. I mean you know it's a whole package. It means being able to get up when I want to get up, being able to go to bed when I want to go to bed, go out in my vehicle when I want to go out in my vehicle, and if I want to drive around at two in the morning then that's....you know, up to me, that is quality of life. My life is not determined by the condition I've got, to be honest it's more about the quality of the care I receive, that is what makes the difference.

Community care policies such as the introduction of the Independent Living Fund (ILF) and direct payments from statutory bodies have been introduced to enable people with disabilities to direct the personal assistance they receive

(through their ability to hire and fire their own workforce and negotiate working hours and pay with their employees) (Morris, 1994), and nearly all of the adults living with SMA who were interviewed in this study reported being in receipt of this monetary support. Morris (1994) has highlighted some of the conceptual difficulties of these schemes, including the underpinning assumptions about disabled people's need for 'care' rather than autonomy, together with some of the practical issues associated with their implementation, including complexities in the negotiation of the relationship with personal assistants (PAs) (more specifically, enforcing professional boundaries), and the potential inflexibility of the services when erratic or unsociable hours of work are required. However, all but three of the participants in this study diagnosed with SMA were able to live independently as a consequence of their receipt of ILF and direct payments which they felt positively about, in spite of reported difficulties with identifying and retaining appropriate PAs and the personal intrusion posed by the need for continuous support.

The importance of the social and physical environment in determining the life experiences of people with disabilities is crucial to many disability rights supporters' perspectives on prenatal testing and selective termination in relation to disability. Writers such as Asch (1999) have argued that the availability of prenatal testing draws attention away from social arrangements that create disability and instead focuses on the impaired foetus as 'the problem'. As Shakespeare (2006) reminds us, environments can be equally as disabling as bodies, and thus the way in which participants in this study

constructed the causes of the problems and restrictions associated with life with SMA is important in relation to reproductive decision making. Indeed, for many, the problems associated with disability (i.e. restriction arising from social arrangements) were seen as potentially open to interpretation or even change; and thus experiencing disability was not *necessarily* a negative phenomenon.

Lamb and Peddon (2008) have described the way in which participants in their study developed innovative strategies to manage their daily lives with SMA. Such strategies included maintaining an optimistic life view, establishing networks of strong relationships, maintaining a 'normal' life through independent living and adopting creative approaches to dealing with symptoms. Similarly, participants in this research described the way in which experiences of disability could be mediated by various factors. In particular, the importance of optimism, perseverance and 'thinking of ways around things' emerged as significant coping strategies for individuals living with or alongside SMA. Geraldine is in her 40s and has been diagnosed with SMA type II. She has used a wheelchair all of her life, is currently unemployed and lives in her own bungalow with the support of 24 hour personal assistants:

Well I've got a very positive outlook on life I mean obviously it is quite debilitating and it is quite limiting but I mean obviously it is all down to your view on life and your outlook. I'm not going to just sit in and let it get to me, but you know there's a lot of inspiration belonging to the Jennifer Trust, you know I can get inspiration from other

people affected by the condition and at the end of the day, you can go about your daily life and be as normal as you want. If you're willing to fight for the things you need and you keep positive, there's no reason why your life can't be as good as anyone else's.

For many participants, dealing with disability included dealing with others' assumptions about what life as a disabled person meant, and indeed, many were keen to dispel this assumption by *presenting* their experiences as overwhelmingly positive. By maintaining a positive outlook and developing strategies to circumvent obstacles, participants presented their experiences of disability surmountable, as mediated by personal attitudes and actions, for which individuals must take responsibility.

For many, this personal attitude was attributed to positive experiences in childhood and a supportive family. Isabella is 22 years old, has been diagnosed with SMA type II and has been a wheelchair user all of her life. Isabella was born in Australia, but moved to the UK with her mother following the death of her father when she was three. Isabella's mother has been her full time carer since this age:

I've never really thought about me having SMA because...I've just grown up the same as everybody else and I've never really thought about it. I think I was about 13 in school when people started doing things that were a bit difficult or whatever...and I'd still take part but I was actually like 'ah...I can't do some of these things' but if that was the

case then I'd just find something else to do, you know we'd find a way round it ...and when I was growing up I did everything that my friends did...I even had a skateboard and my mum made sure I had whatever anybody else had I had, so my SMA didn't really matter...my mum's attitude has always been 'never say never' and so that's been my attitude too. I was always the disabled person who's never been disabled because of that.

Despite the wealth of studies documenting the difficulties and challenges faced by families affected by disability, participants overwhelmingly reported positive familial experiences and adaption to life with SMA. Moreover, recent research has reflected these more positive accounts of families thriving in spite of disability (e.g. Ferguson et al., 2000). However, as the participants in this study can be considered a self-selecting population, it may be that families with positive experiences of SMA were more likely to volunteer to participate than those with more difficult experiences, which may have biased the reporting of family life with SMA.

Disability could also be circumvented by more practical rather than simply attitudinal strategies. As Lamb and Peddon (2008) have suggested through their interview study with individuals affected by SMA, living with a disability requires the use of 'innovative and creative strategies' in order to overcome obstacles. There was evidence that participants in this study engaged in similar 'thinking outside the box' (Lamb and Peddon, 2008:255) to bypass these difficulties arising in day to day life. These strategies often

involved research, experimentation, the development of particular ways of undertaking tasks or the use of specialist pieces of equipment. Lily, for example, a mother with SMA in her twenties with a three year old daughter, designed and had various pieces of assistive equipment custom made to manage some of the physical and practical difficulties she encountered when caring for her daughter, including the adaptation of her wheelchair to accommodate her transportation.

Disability therefore, was experienced as an aspect of SMA that was mediated by various factors. Whilst the implementation of legislation to eliminate the problems associated with disablement was emphasised, participants also cited access to resources, personal attitudes and philosophies, the use of creative strategies and the availability of strong support networks as means by which the experience of disability could be transformed or manipulated. However, in spite of the possibilities of transcending traditional understandings of disability as necessarily negative and restrictive, there were also elements of the experience of SMA which were conceptualised as being beyond the scope of individual, social or environmental change. These aspects of the experience of living with SMA will be referred to as ‘embodied experiences of impairment and disability’ and ‘experiences of illness, death and bereavement’ and will be analysed below.

Embodied Experiences of Impairment and Disability

Whilst individuals who had been diagnosed with SMA and their families experienced disablement in various different ways, there were further

dimensions of their experiences which transcended the social and environmental. Indeed, participants also spoke of their bodily experiences of their condition, what it feels like, physically and emotionally, to live in a body affected by muscle weakness. Family members also described this bodily impact of living alongside SMA, not only in terms of the physical consequences of caring for an individual affected by SMA, but also the emotional implications of such work. Indeed, whilst family members did not have access to knowledge about what it feels like to experience SMA in their own bodies, they nevertheless experienced SMA through their bodies as a consequence of living intimately with the condition.

D'Agincourt-Canning (2005), in her study of genetic risk perceptions in families affected by hereditary breast/ovarian cancer has suggested that individuals who are in close association with individuals affected by cancer 'participate directly in the cancer experience' (p. 56). By offering care, as well as emotional and practical support, these individuals come to know cancer through their relative's experience of the condition, and these experiences in turn influence their perception of their own genetic risk. For D'Agincourt-Canning (2005), however, as well as Abel and Browner (1998), this form of experiential knowledge can be distinguished from that possessed by individuals who have received a diagnosis of a particular condition. Abel and Browner (1998) for example, refer to the experiential knowledge of relatives who care for individuals with dementia as 'empathetic knowledge' as it is grounded primarily in emotional ties (p. 315), whereas D'Agincourt-Canning (2005) defines the knowledge of care-givers as 'tangible knowledge', or

subjective knowledge of the condition derived from close association with those living with it and observing their realities. Both D'Agincourt-Canning (2005) and Abel and Browner (1998) reserve the term 'embodied knowledge', as a component of experiential knowledge, to describe the experiences of those individuals who have received a diagnosis of the condition affecting the family. However, upon analysis of the accounts of the family members of individuals with SMA, these participants' experiences of SMA were thoroughly embodied; the care work they offered their relatives was often both physically and emotionally demanding, and they came to know and understand the impact of SMA through their own bodies. Rachel is the mother of a 12 year old girl diagnosed with SMA type II, Anna, and is her main carer at home. For Rachel, caring for Anna is a thoroughly embodied activity:

SMA is extremely tiring for the families, really, because you basically have to take over what their bodies can't do and that's a hell of a lot of stuff, you know...Anna's not strong enough to reach out and pick a drink up off the table, so I do it...you know, she can't lift herself up er she can't support her weight, so I lift her. You become their muscles and their strength, in effect, for them, if that makes any sense, you do what they can't...so it is very tiring and I end up thoroughly exhausted at the end of each day if I'm honest.

For Rachel, caring for her daughter meant blurring bodily boundaries, a finding which has been documented in the literature in relation to caring

practices (Meyer et al., 2007). Through becoming the muscles and strength for another body, Rachel became two bodies in one, posing a fundamental challenge to the notion of the autonomous, discrete individual, and highlighting the inseparability of embodied and empathetic experiential knowledge. Stress and physical exhaustion featured strongly in family members' accounts of living alongside SMA, the consequences of which were felt in the bodies of individuals caring for someone with SMA. Needing to lift their relative, assisting with physiotherapy exercises, turning their family member in bed several times a night alongside other daily care activities such as assistance with washing dressing and eating, all contributed to an increased workload for the family, and, often, physical fatigue and stress, particularly for women, to whom much of this workload fell. Thus, whilst differentiating between 'empathic/tangible' knowing and embodied ways of knowing SMA may be useful in distinguishing the different ways in which individuals come to know SMA, these distinctions draw on notions of Cartesian dualism. Cartesian dualism refers to a reading of the body whereby a conceptual gulf is positioned between mind and body, between 'empathy' and 'embodiment', and reflecting an underlying assumption that these are distinguishable and independent of one another (Grosz, 1994). Grosz (1994), drawing on the works of Lacan, however, has developed the notion of a 'Möbius strip', an inverted three dimensional figure of eight (8), to present an alternative reading of the body, and one which demonstrates the

...inflection of mind into body and body into mind, the ways in which, through a kind of twisting or inversion, one side becomes the other.

(Grosz, 1994: xii)

Grosz's conceptualisation of the fluidity of body and mind such that mind and body flow into one another, inside and outside the body in a continuous loop may be more easily mapped onto the accounts of the families living with SMA, whereby the boundaries between emotion and body, and between bodies themselves, became blurred.

The way in which emotion and embodied experiences of SMA fed into one another occurred via various routes; two participants who were diagnosed with SMA themselves (and thus had experience of the condition directly) also performed assistance work for other relatives affected by SMA, and thus had 'empathetic' knowledge of the condition through observing their relative, but also embodied knowledge of the condition. Whereas for other participants, the inseparability of bodies and emotion was apparent in the way in which they recounted particularly difficult experiences of SMA. Miriam is 40 years old and experienced the death of her daughter, Skyla, to SMARD when she was just 10 months old. Miriam and her husband were firstly told that Skyla had Peripheral Neuropathy and had to be permanently ventilated due to breathing difficulties. However, when the family were relocated to the UK, Skyla was re-diagnosed with SMARD:

We were sent to [hospital] in the UK where sadly they could only confirm what we had been told in Holland

and that there was nothing that could be done for Sklya. That was the hardest part for us, and our families, was that we just could not accept that no one could do anything, she looked perfect, it was very hard to accept that she was dying and I couldn't help her. She remained on a ventilator until [date] when we made the heartbreaking decision to withdraw the ventilator and she slipped away in my arms. As devastated as we were, we also felt an element of relief for our precious girl that she did not have to suffer anymore. I remember waking up the day after she had died and thinking that at least she did not have to be suctioned anymore which was something that she hated having done. The suffering was so much for little Skyla to bear, and the whole family suffered with it and continues to do so. It's a devastating disease.

Through describing SMA as a 'devastating disease', Miriam highlights both the bodily aspects of devastation, caused by the disease to Skyla's body, but also the devastation and suffering experienced by herself and her family; the embodied and emotional aspects of the condition are merged into, and experienced, as one and the same, reaffirming the familial nature of genetic disease.

It is important to recognize these various forms of experiential knowledge as participants came to 'know' SMA through a variety of means.

Whilst carers have been conceptualized as having empathetic knowledge of the condition affecting the person for whom they care, an analysis of the families of individuals affected by SMA revealed that the way in which these individuals experienced SMA was thoroughly embodied as well as empathetic, with a fluid interchange between the two. This finding is significant in terms of an analysis of the accounts of families living with SMA. As D'Agincourt (2005) has highlighted, it is often taken for granted that 'embodied' experiences, by virtue of their emergence from sensory perception, are to be regarded as more being more 'authentic' or reliable forms of experiential knowledge than those gleaned from empathy or the experiences of others. This blurring of boundaries between emotion, embodiment and experience, however, suggests a more complex picture of experiential knowledge and the way in which the different forms of it can be conceptualised.

For participants who have been diagnosed with SMA themselves, embodied experiential knowledge again took on a different form to that of family members. Despite the fact that social model of disability theorists have largely shied away from an analysis of the experiential aspects of impairment (or what it feels like to live in an impaired body) in favour of an analysis of the social and economic constitution of disability (Paterson and Hughes, 1999), participants' lived sense of their own bodies was so central to how they perceived SMA as a condition, this dimension of experiential knowledge cannot be discounted.

All participants who took part in the research were asked to explain how they describe SMA to others in order to elicit their experiences and perceptions of the condition. For individuals diagnosed with SMA, this question evoked a range of what can be termed ‘embodied descriptions’; descriptions of what it feels like to live in a body which cannot always easily be independently manoeuvred or controlled in the desired manner; a body which is subject to muscle tremors, fatigue and difficulties with breathing (Wendell, 1996; Öhman et al., 2003). Whilst social model of disability theorists have dismissed such experiences of bodily hindrance as belonging to medical models of disability, these experiences were often intermeshed with experiences of disability, as will be discussed later. Analogies were used as a way of capturing these experiences in a way that could be easily understood:

When people ask me what it’s like to live with SMA, I always say it’s like living in a strait jacket, but when your mind is fine so you are incredibly constrained. You live your life through other people.

(Fae, in her 30s, diagnosed with type II SMA)

...I always say to people that having SMA is a lot like having the flu but without the fuzzy head, you know you feel like you’ve got a lead blanket over you, your body’s too heavy for you to lift. Just lifting your arm is so much effort so you’re completely exhausted.

(Kristen, in her late 20s, diagnosed with SMA type II)

I always describe it in terms of a remote controlled car with the batteries dying. The muscles are too weak to do what they want so it's like the car trying to get over a tiny bump and it just stalls. And when I say that people say 'o yeah I know what you mean'.

(Paul, in his 30s, diagnosed with SMA type II)

For Hughes and Paterson (1999), as for the participants with SMA, these embodied experiences of impairment are important, and thus cannot be written out of thinking about, and theorising disability. Indeed, our perception of the world is created through and within our bodies (Bendelow and Williams, 1995), and thus embodied experiential knowledge is crucial in framing the reality of life with SMA.

For Paterson and Hughes (1999) the means by which to realign these experiences of the body with the social and political domain in which disability is produced and reproduced lies in the theoretical contribution of phenomenology. Following Leder's (1990) use of the concept of 'dys-appearance', Paterson and Hughes (1999) have argued that the impaired body emerges to awareness through its encounter with the social world. Unlike work in the field of medical sociology, which has used the concept to explore the way in which the chronically ill body rises up into conscious awareness through its deviation from ordinary functioning (Williams, 1996), for Paterson and Hughes (1999), the impaired body is brought into the disabled person's consciousness only at critical points; the points at which it is not adequately

catered for by society. Indeed, like the respondents in Watson's (2002) study of identity and disability, embodied experiences of impairment were not experienced as a 'hindrance', but as simply their way of being in the world. Embodied experiences of impairment thus emerged through their interaction with a disablist society. Rhona is in her late 20s, diagnosed with SMA type III, and described her sense of SMA in the following way:

I really don't think about myself as having SMA at all, I don't notice it on an everyday basis I think it bothered me more...I mean when I think back to times when I was down and I was a child and the condition really affected me, it was because I couldn't do something everybody else could do and I wasn't being treated the same, and that's the only time I remember thinking the whole 'why me?' and...um because I literally wasn't able to do...not necessarily not able to do, I probably could have done, people *assumed* I couldn't, or there wasn't the facilities to enable me to get involved like everybody else, those were the only times that I thought that I'm physically different to everyone else because when you're able to just get on with things you don't notice it at all and I never think about having SMA.

For Rhona, her embodied sense of being 'different' emerged at the points in her life in which her social and physical environment prevented her from participating in activities she would have otherwise liked to; her impairment

‘dys-appeared’ (Leder, 1990), whereas ordinarily her embodiment was not experienced as disruptive. Nick Watson (2002) has used Somer’s concept of the ‘ontological self’ to argue that impairment often becomes incorporated into disabled people’s realities so that it may be a phenomenological impossibility for them to imagine life without it. Indeed, many of the participants with SMA had never been able to walk and had always lived with reduced strength, and thus their awareness of their embodied impairment came through their social interactions with the world, others’ judgements of their bodies as intolerable to live in or inadequate, or the failure of society to enable their bodies to function in ways in which they desired.

Rosie is 28 and has been diagnosed with SMA type II, uses a powered wheelchair full time and requires the use of night time ventilation on occasion. As SMA involves weakening to the intercostal muscles (those used to support breathing), people diagnosed with SMA often experience insufficient oxygenation during sleep and thus many make use of overnight ventilation technologies (Lamb and Peden, 2008). Rosie relies on a 24 hour PA system to live independently and works full time as a solicitor in London. She contrasted her own sense of her body and her life to how she felt others perceived it:

And I mean I get, I get very strange reactions from people when I meet them. I always say that actually the reaction I get, people react to me *as if I am feeling as they are*, because they react in a way, you know they are completely new to it and they think ‘well how would I feel if I were in

this position?' er and then they assume that I feel like that forgetting that I've had 28 years of it, um and that I've got a very different perspective...so there are um, there are people who um have that attitude, who just, their experience of their lives and their bodies is so far away from mine that they are scared – it's a fear thing, because they are worrying about how they'd live with it. I do know some people who would be devastated to live the way I do.

For Rosie, her sense of embodiment was entirely different than she perceived others to be who had not lived her life. Her body was as she had always experienced it and thus her point of orientation in the world differed from that of non-disabled people who could not contemplate these experiences. Thus, a heightened awareness of the differentness of her impaired body emerged not only through being excluded, as in Rhona's experiences, but also through the attitudes, and perceived attitudes, of others.

However, for a minority of participants, SMA was not a condition which they had experienced since birth, and for others, their embodied experiences of impairment were unstable, ever changing and unpredictable. Shakespeare (2006) has made distinctions between those impairment groups for whom experiences of disability remain stable over time, and those who experience deterioration, suggesting that they may be conceptualised as two different groups with different viewpoints and concerns (Shakespeare, 2006: 106). Whilst the progressive nature of SMA has been debated within the medical literature, it was a generally accepted fact amongst those interviewed that the

symptoms of SMA can be expected to worsen with age. In particular, an increase in fatigue, respiratory problems and issues with swallowing and speech are reported as being amongst the most common emergent difficulties experienced by ageing individuals with SMA (De Groot and De Witte, 2005). Moreover, individuals with SMA type III and rarer variants of SMA such as SBMA and ADSMA (see Appendix V for descriptions of these variants) are all medically defined as being conditions with late onset (i.e. in the second or third decade of life) and marked by gradual decline with age. Whilst the explanations for this worsening of symptoms vary, for the participants who noticed such changes, their embodied experiences of impairment emerged in a different way to those individuals whose SMA had remained stable, and it was many of these participants who experienced their bodies as a ‘hindrance’. Indeed, their sense of impairment was reported as emerging specifically at points at which they experienced such deterioration *as well as* through their interaction with a disablist society. Hayley is 34 years old and was diagnosed with SMA type II at 18 months of age. She described her changing sense of her own body in the following way:

But I mean everyday I notice things that are harder and that are more difficult but people that are around me don't necessarily notice, but I'm much more aware of my condition now than I was when I was growing up...like my skin's breaking down now, and I'm noticing that, but I'm still fighting and I'm still trying but things become harder and harder as you get older and like at the moment I have pain constantly in my right arm...I'm

now having to lift my arm to the control box [on wheelchair] by biting my fingers with my teeth [...] And I've always fought it and never let it get the better of me, but it *is* beginning to get the better of me now, because I can see it getting worse, whereas before it was just how I was, you know. Now, I just get used to one thing, and then something else packs up, and that's hard.

For Hayley, her awareness of her body and her condition arose from the deterioration she experienced. The 'breaking down' of her skin, and pain in her right arm represented a form of 'dys-embodiment' (Williams, 1996) or a fracturing of her otherwise taken for granted sense of embodiment; 'that's just how I was' (Hayley). Unlike Rhona, whose sense of impairment and physical difference arose from her interactions with a disablist society, for Hayley, it was her own sense of 'dys-embodiment' that constituted this aspect of her sense of self. Williams has suggested that at critical junctures in the chronic illness trajectory individuals experience dys-embodiment and what frequently follows is the development of coping styles and strategies to realign body, self and society, in an attempt at 're-embodiment' (Williams, 1996: 34). For Hayley, 'fighting it' and 'not letting it get to her' represented these strategic attempts at re-embodiment, and her struggles with maintaining this 'fight' highlight some of the pressures on those living with SMA to appear positive in spite of the challenges facing them. The relentless nature of this process of re-embodiment, for Hayley, was driven by the deterioration of her condition; SMA was at the forefront of her consciousness because she was living in a

continual ‘oscillation’ between dys-embodiment and attempts at re-embodiment (Williams, 1996: 39).

Whilst there were a range of embodied experiences associated with SMA, including a sense of ‘dys-appearance’, of ‘dys-embodiment’, ‘re-embodiment’, or a movement between all three of these states at various points, these embodied states did not necessarily correlate to medical classifications of SMA in any clear way. Whilst neurologists and geneticists have attempted to classify SMA on a numerical scale of severity to mark out those experiences most likely to be characterised by suffering and intolerability, this scale did not always match participants’ accounts. In fact, for some participants, the experience of ‘dys-embodiment’, and particularly that which emerged from the deterioration of SMA symptoms, was perceived to involve suffering of a higher degree than that of ‘dys-appearance’, whereby an embodied sense of impairment was an ‘absent presence’ (Leder, 1990), emerging through an interaction with a disablist society rather than through bodily decline. Kristen and Ellie are two sisters in their twenties, and have been diagnosed with SMA type II and SMA type III respectively. Whilst medical definitions of SMA position Kristen’s experiences of SMA as more severe and involving a higher degree of physical restriction than Ellie’s, Kristen reported that these medical categories did not fit her own, and her sister’s experiences of the condition:

I don’t really think you can know much about the sort of life someone will have from the type of SMA they’ve got because, I think SMA is much less of a big deal to me

than it is to Ellie, you know, because I've always had it, it's always been there. I could never walk whereas she could walk and had a normal life up until she was a teenager, and she has found that so hard as she's got weaker and she really hates her disability, I know she does, even though people expect that to be *my* attitude, not hers, because it affects me in a more severe way I suppose, but I'm not really bothered by it. I guess the other thing is that I'm a constant reminder to her as well, like about how she could get, physically, and I think that scares her. It's got to be much harder if you've got to keep adjusting and looking at things you can't do anymore whereas...you know I've always been this way.

I never lost anything.

Shakespeare (2006) has highlighted the potential differences in perspective and attitude towards their impairments of different groups of disabled people. In particular, Shakespeare notes there can often be differences in perspective between those with 'congenital impairments which are largely static in nature' (p. 106) and those individuals with 'acute degenerative conditions' which may have been present at birth but which only become problematic in childhood or midlife, as described previously in relation to Hayley's experiences (Shakespeare, 2006: 106). As Shakespeare (2006) has noted, the concerns and viewpoints of these groups of individuals may be very different and there is much potential for vastly differing experiences of impairment

between them. Whilst Shakespeare (2006) appeared to be referring to different impairment groups when describing the possible associated experiences and perspectives, SMA could be experienced in both of the ways Shakespeare has outlined. For some individuals their SMA had been present from birth or within the first year of life, whereas for others it had a gradual onset, for others still, deterioration occurred alongside ageing. De Groot and De Witte (2005) in a questionnaire survey of adults living with SMA found that some of the most common medical complications associated with ageing with SMA included difficulty with sleeping, speaking, coughing and swallowing and fatigue. In defining those forms of SMA present from birth as the most severe, however, medical typologies fail to accommodate these fluctuating embodied experiences. Indeed, such a system does not acknowledge the potentially different perspectives embedded within these embodied experiences of SMA. Being diagnosed with type III SMA does not necessarily guarantee an experience of SMA that is fundamentally 'less severe' than those of individuals with type II. As Kristen's account highlights, it appeared to be the *process of deterioration* itself, the gradual loss of abilities or the onset of ill health, that was associated with the greatest level of (perceived) suffering and distress as opposed to the resultant level of impairment per se. Disability activists have worked hard to disentangle the automatic amalgamation of 'suffering' with 'impairment' and the associated assumption that the degree of suffering experienced by an individual can be read off from their level of impairment. As these interviews revealed, it is not necessarily the degree of disability and impairment that leads to SMA being

experienced negatively (although this can be the case), but that particular embodied experiences were more closely aligned with suffering than others.

These findings have implications for an analysis of experiential knowledge. The literature around experiential knowledge has tended to conflate 'empathetic' and 'embodied' forms of experiential knowledge as being similar in constitution, but having been arrived at with differing degrees of intensity or closeness to the experience (D'Agincourt, 2005; Etchegary et al., 2008). However, interviews with people diagnosed with SMA and their families have revealed that 'empathetic' and 'embodied' experiential knowledge can produce inconsistent, or entirely contradictory, accounts of the same life with SMA. Many people diagnosed with SMA felt that those who would empathise with their experiences brought with them assumptions about which experiences involve suffering, and which do not- assumptions that could be entirely inconsistent with the embodied experiences of people diagnosed with SMA themselves. Whilst the literature on experiential knowledge has thus drawn attention to the way in which accounts of experiential knowledge can contrast with medical knowledge, little attention has been paid to the way in which experiential knowledge accounts can similarly contain internal tensions and inconsistencies which render them as unstable as the forms of medical knowledge that they are frequently presented as an alternative to.

A further dimension of experience associated with SMA is that of illness, death and bereavement. These experiences were presented by participants as instances where SMA involved unequivocal suffering. Unlike experiences of

disability or embodied experiences of impairment, these experiences were presented as involving levels of distress which could not be remedied or reduced by personal, social or environmental intervention, and thus were of a fundamentally different character to other experiences of SMA.

Experiences of Illness, Death and Bereavement

The conceptual differences between 'illness' and 'impairment' have been the subject of much debate both within and between the fields of medical sociology and disability studies. However, theoretically robust distinctions which account for the differing contours of both impairment and illness are still lacking (Oliver, 1996a; Bury, 1996; Mulvany, 2000; De Wolfe, 2002; Corker, 1999). Much work in the field of medical sociology has been concerned with the social significance of illness as well as strategies of adaptation used by those who experience it (Barnes and Mercer, 1996). Social model of disability theorists, however, have instead bracketed off the experiential aspects of illness or impairment in favour of an analysis of the structural origin of disability. As dialogue between these two fields has developed, the similarities, particularly between what medical sociologists term 'chronic illness' and disability theorists term 'impairment' have been emphasised (Mulvany, 2000: 592) or the terms have been treated as synonymous (Oliver, 1996a: 40). However, tensions remain, as evidenced in the difficulties reported by those with chronic illness in identifying their experiences as an 'impairment' or 'disability', as suggested in the previous sections (De Wolfe, 2002: 257). The social model of disability in particular

has been criticised for sidelining the experience of illness and suffering, which can be an integral part of impairment experiences (Morris, 1991). Social model of disability theorists such as Oliver (1996a) have tentatively suggested a 'sociology of impairment' as an arena in which to interrogate these experiential aspects of 'impairment', including pain, discomfort, fatigue and malaise whilst retaining a critical distance from those theoretical models which have traditionally been concerned with them, including individual and medical models of disability (Oliver, 1996a: 49). Paterson and Hughes (1999) have attempted to develop this 'sociology of impairment' through a phenomenological lens, demonstrating that oppression is not only located in the social fabric of society, but also in the 'flesh and bones' of disabled people; oppression is an *embodied experience* (Paterson and Hughes, 1999: 606).

For writers such as De Wolfe (2002) herself living with Myalgic Encephalomyelitis (ME), any attempts to subsume those with chronic illness into social model thinking will necessarily be inadequate as the proposed adaptation of society is 'contingent on the absence of suffering' (De Wolfe, 2002: 262). Indeed, both De Wolfe (2002) and Shakespeare (2006) have argued that there are certain aspects of impairment that are experienced negatively and which defy accommodation within society. Impairments can be problematic in and of themselves (Shakespeare, 2006: 43), and it is these aspects of impairment that more easily render themselves to the term 'illness'. De Wolfe (2002) equates illness specifically with 'suffering', which, despite the possibilities for emergent meanings and narratives (e.g. Frank, 1995), is

largely experienced negatively; as a state of embodiment most people would seek to avoid:

It would be perverse to prefer illness and misery to health
and happiness...virtually anybody...would rather be well.
Illness should not be romanticised. It ruins lives.

(De Wolfe, 2002: 263)

Similarly, when talking about living with SMA, participants drew conceptual distinctions between those experiences of impairment, disability and embodiment which were mediated or counter-balanced by contextual factors (for example having a positive attitude or a supportive social and physical environment), and those aspects of SMA which allowed little room for interpretation; the experiential aspects of SMA which were deemed to be imbued with suffering.

Paula is the mother of a 13 year old girl who was diagnosed with SMA type II. For Paula the possibilities of chest infections and illness were seen as so separate from the rest of the experience of SMA that she spoke about them as being separate conditions:

...well with Spinal Muscular Atrophy, I was always told,
and it's always stayed in the back of my mind,
that...about 3 years ago we had a cluster group, we had
an outing [through JTSMA] and you get to talk to
different people there and a lady said to me, another
parent, and she said 'it's never actually SMA that kills
the person or the child, it's more the respiratory side of it'

you know, with them being unwell etc. You know it's not actually the spinal muscular atrophy, it's if they become really really unwell, and then they get the respiratory and then the chest infection and then the pneumonia, that's what normally, you know, they pass away with that. It's not actually the condition, Spinal Muscular Atrophy, that's the problem. I always say to Rob [husband], I could live with that [SMA] if it was *just* that, but it's the illness side of it that's the problem, that's what they suffer with and that's what makes it hard I think.

Individuals diagnosed with SMA are deemed medically to be at a continuous risk of (potentially fatal) chest infections and pneumonia due to the weakening of the chest muscles. Paula's daughter passed away from such a sudden onset chest infection a few months following participation in this research. Due to the risks posed by potential chest infections, many people with SMA attend regular respiration clinics and keep a supply of dried antibiotics at home to treat the sudden onset of an infection, which is regarded as a medical emergency (Loos et al., 2004; Wang et al., 2007). For Paula, these respiratory difficulties associated with SMA were conceptualised as entirely separate from her everyday experiences of SMA that she regarded as separate entities; as entirely distinct experiences which could be ranked according to their tolerability.

The 'suffering' associated with SMA, furthermore, transcended the medical typology of SMA in the way that it was presented by participants.

Rather than being confined by and to particular ‘types’ of SMA, suffering was more closely correlated to specific embodied experiences. The lived reality of chest infections, discomfort, going through, and recovering from, spinal fusion surgery as well as the deterioration of abilities were all embodied experiences that participants associated with suffering. However, the most undisputed form of suffering associated with SMA by participants was represented by the individuals who developed severe breathing difficulties and subsequently died. This form of SMA was most commonly associated with type I SMA. However, seven individuals with type II SMA in their families had experienced the death of a relative from SMA due to respiratory difficulties (and one person’s daughter with type II SMA died shortly after being interviewed) and furthermore, three individuals had lost children to SMARD. Thus, these experiences were not solely associated with a diagnosis of type I SMA. For families who experienced the death of a relative with SMA, the suffering was both innate and intractable for the individual concerned as well as for their relatives; unlike those experiencing embodied impairment or disability, this suffering could not be displaced by social or environmental changes. Indeed, despite the ‘genohype’ (Fleising, 2001) surrounding the treatment of SMA, including the more recent ‘recreation’ of the early stages of the condition using stem cells which has been heralded as heightening understanding of the biological mechanisms of condition and consequently ‘paving the way’ for future treatments or cures (BBC News, Dec. 2008; Yu et al., 2008; Sendtner, 2009; Nicole et al., 2002), medical science is still unable to offer these options for those diagnosed with SMA.

For many parents and relatives then, experiencing severe SMA involved a sense of helplessness; the only release from the suffering their children endured occurred when they passed away.

Charlie and Fraser are in their 50s and experienced the deaths of two of their children from SMA type I. For Charlie and Fraser, the deaths of their children marked the end of a relentless and futile battle with the condition which was conceptualized as a form of release:

When I think back to when our children were really ill...

You knew that the time had come when you were actually sitting beside them and wishing that they would just take their last breath, you know? You know and they were struggling to breathe, their breathing was so restricted, they couldn't take a breath, they couldn't cry, their cries were weak...and I just knew when the time had come when I was wanting my children's lives to end...because I thought they were suffering.

For Charlie and Fraser, the experience of losing their two children involved suffering for the whole family which many individuals found simply too much to bear. Charlie further recalled being actively avoided by friends who could not cope with the reality of her situation after she had given birth to her second child with SMA type I:

...I remember when I'd had Alexis and people used to cross the road, you know, good friends of mine and they'd cross the road and only speak to me from across the road when I was

with the pram...and I'd think 'I've got a lovely wee baby in here, come over and look at her' but it was because they couldn't cope...but, you know, I forgave them all. And I thought 'it's you that can't cope with my situation' and I didn't lose friends through that, they all came back to me after it was all over. You know and they say now, 'I didn't know what to say and I didn't know what to do' and they feel terrible about it now.

Experiences of illness, death and bereavement therefore were presented as experiences of suffering which extended beyond the individual diagnosed with SMA to the family, friends and more distant relatives. Whilst experiences of disability and embodied experiences of impairment primarily related to the individual diagnosed with SMA, experiences of illness, death and bereavement were presented as involving collective suffering; embodied and empathetic forms of experiential knowledge became intermeshed in the familial experience of suffering, which, as will be discussed in Chapters 6 and 7, had consequences for the way in which this form of suffering was conceptualised in the context of reproductive decision making.

Conclusions

In conclusion, in addressing the question of how families and individuals experience SMA and related to medical knowledge, this chapter has explored the content of participants' experiential knowledge of SMA and made conceptual distinctions between the different forms of this knowledge. Whilst

medical descriptions of SMA have distinguished between different ‘types’ of SMA to account for the range of experiences associated with SMA, this chapter has suggested alternate ways of understanding these differences. Distinguishing between ‘disability’ ‘embodied impairment’ and ‘illness, death and bereavement’ as different forms of experiential knowledge associated with SMA reveals the limitations of the medical typology in accounting for the lived reality of those who live alongside SMA. Indeed, despite the suggestion by the medical definitions presented in Chapter 3 that there is a linear progression of severity running from the mildest type of SMA (type III) up to types I or even 0, the narrative accounts of those individuals living intimately with SMA suggests a far more complex picture. The marrying of SMA type with assumptions about quality of life, moreover, is problematic when viewed in the context of the different forms of experience associated across each type. By distinguishing between ‘disability’, ‘embodied impairment’ and ‘illness, death and bereavement’, which transcend medical classifications, it became clear that everyday experiences with SMA did not map clearly onto the medical typology, and moreover, that there was little correlation between these everyday experiences and the genotype of individuals diagnosed with SMA. Despite the primacy attributed to genetic knowledge in accounting for SMA, and its role in facilitating the ‘hype’ around the possibility of future treatments, genetic information emerged as having extremely limited value in predicting or mediating the various ways in which SMA was experienced in daily life -effective treatments for SMA

remain elusive, and as has been described in Chapter 3, there is a documented incongruence between pheno- and genotype expression of SMA.

By focusing instead on different forms of experience rather than medically defined types of SMA, important conceptual distinctions have been made between those experiential aspects of SMA which were considered to be mediated by social and environmental factors, and those aspects which were deemed to be pre-social -beyond medical, environmental and social resolution. Such aspects of SMA were presented by participants as fundamentally negative and involving physical and psychological distress not amenable to any form of intervention. Whilst medical definitions have located the presence of such suffering in what are regarded as the more 'severe' types of SMA (e.g. type 0-I), analysis of the accounts of participants have revealed that such experiences were not actually contained within any particular type of SMA. Rather, SMA was experienced often as a complex interplay between these different types of experience; a movement that took place over time and in a range of different contexts.

As well as highlighting the difficulties in medical accounts of SMA, in analysing the accounts of different family members' experiences, this chapter has further highlighted some of the difficulties associated with an analysis of experiential knowledge. Whilst 'empathetic' and 'embodied' forms of experiential knowledge have been studied in the literature as knowledge derived from the same experience but from differing levels of abstraction, this chapter has highlighted some of the internal tensions and inconsistencies with this conception of experiential knowledge. Firstly, the lines between

embodied and empathetic forms of experiential knowledge were blurred and intertwined in families affected by SMA, and secondly the accounts of life with SMA emerging from experiential knowledge could be entirely contradictory; empathetic accounts of SMA did not necessarily tally with the way in which individuals diagnosed with SMA reported their embodied experiences, leading to internal tensions within experiential knowledge itself.

An understanding of these different forms of experience of SMA, their mapping onto medical classifications and the experiential knowledge of SMA emerging from them are of crucial importance to an understanding of how families affected by SMA approach reproductive decision making. The negotiation of these different factors led to contrasting accounts of reproductive decision making, as Chapters 5 and 6 present.

Chapter 5
Reproductive Decision Making: Experiential Knowledge as Valued
Resource

In the previous chapters, I have analysed the nature of experiential knowledge for families affected by SMA with an emphasis on the range of experiences reported, and the problematic nature of the medical taxonomy for classifying these experiences. I have argued in Chapter 4 that in spite of medical definitions, the way in which SMA is experienced cannot be neatly contained within diagnostic categories; families affected by SMA instead report a far more complex picture of living with the condition, with intersecting experiences of impairment, illness and disability across medically defined ‘types’ which are, moreover, rooted in ever-changing social, political and economic circumstances across the life-course. The following two chapters will take further the analysis of these experiential aspects of SMA to explore the ways in which they not only create a backdrop for, but are also strategically mobilised within, reproductive decision making.

Medical descriptions of the inheritance of SMA assert that in order for a person to inherit SMA when it is not caused by a de novo mutation (which account for 2% of diagnoses), both parents must be ‘carriers’ of SMA. This means that each parent has one copy of the faulty SMN1 gene each. As it requires two copies of this gene to result in a case of SMA, the carrier parents do not display symptoms of SMA, but can have a child who does (See Appendix IV for diagram). It is estimated that approximately 1 in every 40-50 people in the general population have this deletion on the SMN1 gene and

thus are carriers (Wirth, 2000). When two carrier parents reproduce, they can therefore have children who display symptoms of SMA, are asymptomatic carriers or non-carriers. The widely accepted medical estimates of *each* pregnancy are as follows:

- 25% chance of producing a child who would be affected by SMA.
- 50% chance of producing a child who would be an SMA carrier
- 25% chance of producing a child who would not have SMA and would also not be an SMA carrier.

(Simard, 2007)

(see Appendix IV for diagram)

These genetic risk estimates are different for some of the variant forms of SMA such as ADSMA and SBMA which have dominant (i.e. only one copy of the gene is needed to result in an affected child) and x-linked (i.e. transmitted on the 'x' chromosome) inheritance respectively. SMARD involves a different gene to classical SMA. It is, however, recessively inherited in the same way as SMA (see Appendix V for an explanation of the types of genetic testing available to families affected by SMA and Appendix VI for a description of ADSMA, SBMA and SMARD).

For families affected by SMA, this medical knowledge of the inheritance of SMA must be negotiated alongside experiential knowledge of SMA. In order to present this complex relationship between experiential knowledge and medical knowledge in reproductive decision making, the analysis will specifically focus on the interaction between experiential knowledge and notions of 'reproductive responsibility'. Responsibility is a key theme that has been analysed by different writers in relation to

reproduction and disclosure, particularly in the context of genetic conditions within families (e.g. Downing, 2005; Hallowell, 1999). Despite the fact that responsible attitudes and behaviours are emphasised for any couple approaching reproduction, and particularly for pregnant women (Ivry, 2007), for families with a known history of a genetic condition, the imperative to make ‘responsible’ reproductive decisions is even more pronounced (Reed, 2009). This chapter will therefore analyse the negotiations that occur for families affected by SMA around ‘responsible reproduction’ and experiential knowledge, paying particular attention to the strategies used to ameliorate the tensions and contradictions emerging from their intersection. More specifically, in this chapter I will argue that experiential knowledge is presented by participants as clarifying the perceptions of their reproductive responsibilities, either through its interpretation as a ‘warning’, or as ‘reassurance’ as to the possible outcomes of future reproductive decisions, before moving on to the more problematising interpretations of experiential knowledge and reproductive decision making in Chapter 7.

Responsibility and Reproduction

The concept of responsibility has been identified as a central area of inquiry in the literature, both in relation to genetic knowledge more generally, as well as in relation and parenthood. Studies around the management of genetic knowledge within families have specifically addressed the existence and operation of notions of responsibility through an analysis of the dynamics of disclosure and the ownership of genetic information within families (Arribas-Ayllon, M. et al., 2008b; Burgess and D’Agincourt-Canning, 2001;

Chadwick, 1999; Hallowell, 1999; Downing, 2005; Polzer et al., 2002; D'Agincourt-Canning, 2001; Hallowell et al., 2003), whereas the specific forms of responsibility attached to parenthood, or more accurately, motherhood, have also been highlighted in the literature in order to account for the meanings, roles and socially accepted behaviours assigned to it (Ivry, 2007; Green, 1997; Wilson, 2007; Charo and Rothenberg, 1994; Dragonas, 2001; Ettore, 2002; Reed, 2009). For families affected by SMA considering reproduction therefore, both 'genetic responsibility' (or a responsibility to negotiate the genetic risks to one's own health and the health of others) (Kenen, 1994), as well as 'parental responsibility' (or the responsibility to prioritise the welfare and well-being of a (potential) child) may need to be negotiated, as well as other forms of responsibility to wider family and kin. This section will present participants' experiences and conceptualisations of responsibility in relation to reproductive decision making under the headings of 'relational responsibility' and 'accountability'. These thematic headings will be used to distinguish between the experiences of responsibility grounded in everyday experiences and relationships (Burgess and D'Agincourt-Canning, 2001) and the more abstract notions of responsibility to distant, unknown others or society, and will include issues around responsibility for disclosure of genetic information, as well as within participants' own reproductive decisions. It will be argued that the presentation of particular accounts of experiential knowledge of SMA is an important means by which participants negotiate these various, and sometimes competing, responsibilities.

Relational Responsibility

Despite the development of the new genetics being based in rhetoric of individual choice and freedom, promising more information and suggesting enhanced possibilities for the management of health risks (Kenen, 1994), the discovery of a genetic condition within one's family, as has been discussed in Chapter 2, is often experienced as a familial discovery, necessarily involving others and carrying implicit, and explicit, responsibilities. By its very nature, genetic knowledge is situated within the context of biological and social relationships, and for individuals who took part in this research, knowledge about their genetic risk implied perceived responsibilities not only in relation to their own reproductive decision making, but also the feelings, judgements, freedoms and reproductive decisions of others within and outside their family. Burgess and D'Agincourt-Canning (2001) have described this sense of responsibility as 'relational responsibility' in the context of their research on HD and hereditary breast/ovarian cancer. For Burgess and D'Agincourt-Canning, relational responsibility captures the moral dimensions of responsibility that are born out of '...specific life stories, shared understandings and mutual and self-expectations' of individuals (p. 363). For Finch and Mason (1993), these moral dimensions refer to the impetus to present one's actions as falling within the boundaries of the pre-determined conceptions of responsibility that mark out one's identity as a responsible brother, sister, daughter or father etc.

In the interviews that I conducted, participants expressed a strong sense of what their relational responsibilities were, who they were accountable to,

and who should enact them. Louise is 35 and has a seven year old son, Will, diagnosed with SMA type II. For Louise, the discovery of a genetic condition in her family led her to a series of difficult decisions in which her perceived moral obligation to disclose had to be balanced against the possibilities of an emotionally challenging situation for herself:

Um I think basically when he [Will] was diagnosed, and they said it was a genetic thing and I said 'well where the hell's that come from?' you know I don't know anyone who's got it in my family or anyone else's family, because I'd never heard of it before. But obviously we know it's genetic and since then I've done a family tree and I think there were 3 or 4 people in the family with it. But it's all linked off different ways with my, my husband's dad, his half cousin, really really far away in America, she's got type II and she's really bad, she can't sit up unsupported, she has a ventilation thing at night time, and then I've got my mum's second cousin who lives in [town] where I live, and she can't sit up unsupported which is quite bad, so these people I knew nothing about...but the biggest shock for me was that it brought up other things in the family. I found out that I actually have a half brother which had been kept from me before. So I thought he should know about the SMA but I was confused about how I felt about meeting him, you know, if I can handle all the information about my family. In the

end I decided to contact him through [website] because I felt he needed to know, I believe we're a similar age and he has a right to that information, you know he might want kids himself, he might already have them, so no matter how I feel about doing everything, I've got to lay down the facts because it's only fair. And he is my brother.

As Hallowell (1999) has argued, the construction of genetic risk management as a moral issue has the possibility of constraining women's choices. Despite her reservations, and the risks to her own emotional well-being, Louise felt an obligation to inform her half brother of his potential genetic risk, and this responsibility overrode the potential emotional risks to Louise involved in uncovering her family's long kept secret. Despite not knowing her half-brother, Louise prioritised her blood ties with him when counting him as family, and thus was clear in what her 'sisterly' duties involved, including her responsibility for his reproductive decisions in light of the information she possessed (Rhodes, 1998). For writers such as Reed (2009) and Hallowell (1999), the operation of these forms of responsibility are, however, necessarily gendered; that is, the responsibility to enact these relational responsibilities fall primarily to women.

Chloe is 36 years old with a two year old son, Tommy, who was diagnosed with SMA type II a year prior to the interview taking place. Upon Tommy's diagnosis, Chloe felt a similar responsibility to disclose this information to her female relatives and her husband's female relatives in order to alert them to the possibility of them being carriers with a view that

this information might be of use to them in their own reproductive decision making:

It was important for me to know it was genetic because I have 4 nieces and Ryan [husband] has nieces on his side as well, you know all at an age that they might be thinking...My feeling all along was if they have the knowledge of SMA, then they have the options. What they do with it is entirely their decision, but I had to let them know the facts and make the information available so I printed off information for all of them to take to their GPs if they wanted to test. But I don't know how many of them went, I think part of it is a 'it'll never happen to me' attitude, but also not many GPs are knowledgeable about SMA here in Northern Ireland.

Despite the fact that SMA is a recessive condition and thus transmitted by both parents equally (as Chloe demonstrated an understanding of when later talking about her own reproductive decisions), she nevertheless felt that it was her responsibility to inform her *female* relatives about their potential genetic risk (despite the fact that her husband's brother would medically be defined as being at higher 'risk' of being a carrier of SMA than any of her husband's nieces), perhaps reflecting her assumption that it is women who take primary responsibility for reproductive decisions. Reed (2009) has argued that women's assumption of responsibility for reproduction can be related to the fact that pregnancy takes place in women's bodies; the embodied connection with the foetus and the association of certain behaviours with negative

pregnancy outcomes (e.g. smoking, alcohol consumption) positions women as the guardians of the foetus whose actions have direct implications for its well-being. Other writers such as Hallowell (1999) have further suggested the notion that women tend to ‘...position themselves as self-in-relation, to see their lives as interconnected to others, or to define themselves in terms of their social relationships with, and obligations to, others’ (Hallowell, 1999: 616). This can be used to understand women’s felt sense of genetic responsibility to their family members. The sense of responsibility, as has been demonstrated, not only included the assumption of responsibility for reproductive decision making, but also the dissemination of genetic risk information to family members. Hallowell (1999) has noted that this gendered assumption of relational responsibility could be experienced as both burdensome and constraining, not only for women, but also those for whom they enacted their responsibilities. Nevertheless, by enacting such genetic disclosure responsibilities, however emotionally challenging, women could define their actions as justified and maintain their identities as responsible family members.

As well as a responsibility to disclose genetic information, there was also evidence that participants took emotional responsibility for their other family members as an integral part of their ‘relational responsibility’. Reproductive and disclosure decisions were never made in isolation, but instead in a social context, and there was evidence in participants’ accounts that the emotional responses of their family members had to be factored into any decisions they made about how to use genetic information. Not only were

responsibilities to existing children and the relationship between partners considered, but also the feelings and viewpoints of extended family members and kin. Charlie and Fraser, having experienced the birth of a healthy son, Toby, and then the deaths of their second and third child (Jack and Alexis) to SMA type I, described the way in which their responsibilities to their existing child and extended family members were implicated in their decision to undergo prenatal testing for their fourth pregnancy:

...well just when Alexis was born they [medical profession] had made the break through with prenatal diagnosis. So they took blood off Alexis so they could do the tests, but I don't know where we'd have gone with that one [future reproductive choices] if the test hadn't come about, because it wasn't just, you know, us dealing with the situation, it was grandparents and Toby himself, all of our families and our friends, you know. And they were all wishing that we would chuck this, you know. They were all wishing that we would just get a grip and give it up, stop, you know? You were just aware of people wishing you'd stop, they didn't tell you, but you know we've got friends and they knew it was really hard for us, you know. If people care about you, it affects them as well. So you know that whatever decision you make you have to take into account what effect it's going to have on them, you have to take that into account because you have to live with the consequences of that.

(Fraser)

Roberts and Franklin (2004) as well as Rapp et al. (2001) have emphasised the way in which reproductive decisions in the context of genetic information are *necessarily* social decisions, and not simply personal ones. Despite the emphasis on patient autonomy in relation to genetic testing more generally (Skene, 1998; Chadwick, 1999; Arribas-Ayllon et al., 2008b) individuals approach reproductive decision making in the context of ‘a range of overlapping contexts, including personal, familial, and wider social contexts’ (Roberts and Franklin, 2004: 289), and thus this broader social context needs to be accommodated in a consideration of how individuals approach their own reproductive decisions, and the additional dimensions this may bring (Gilbar, 2007).

That participants felt a responsibility or accountability to their family members for the reproductive decisions they made was also reflected in the number of couples who reported that their reproductive decisions were kept a secret from their families. Indeed, unlike the disclosure of a genetic condition within the family, the responsibilities around reproductive decision making were experienced as particularly fraught as they directly implicated the lives of future generations of the family, and participants saw themselves as being particularly vulnerable to the judgements and criticisms of others. Keeping secrets served to protect both the emotional well-being of families, but also to deflect this criticism, judgement or interference from other members concerning the decisions made. Becky is 40 years old, lives in Switzerland with her two children and partner and experienced the death of her first baby,

Lucy, to SMA type I. At the time of Lucy's death, Becky was pregnant again and underwent a CVS which revealed that the foetus also had the genetic deletion associated with SMA. Becky and her partner, Rico, decided to keep their decision to terminate this pregnancy a secret from their families:

When we got the results of the CVS we didn't tell any of the family members because we didn't want to be touched by their views. It hadn't occurred to them that I was pregnant when Lucy died. We were aware of how guilty our parents had felt after the diagnosis and death of Lucy, and it was enough for us to deal with our own grief without them knowing about our decision to terminate. I think they would have taken it very hard so close to Lucy's death. Our families are close knit, but are located in Northern Italy and Belgium, so it was not difficult for us to keep it a secret. We didn't tell them anything until after I'd had the termination, I was pregnant again with my son and I'd had the all-clear test results.

For Becky, managing her relational responsibility meant not only managing her responsibility to her prospective child, but also to her own, and Rico's, parents. By referencing the 'guilt' her parents experienced at the death of her first child, Becky's account highlights the way in which responsibility for reproductive decisions can be experienced not only by the parents in question, but can operate both vertically (i.e. grandparents-parents-children) as well as horizontally (siblings-cousins), as has been suggested previously. Moreover,

the guilt associated with a genetic diagnosis can be heightened by, and intermeshed with, the (often) traumatic experience associated with terminating a wanted pregnancy, amplifying the grave sense of responsibility surrounding reproductive decision making for such families and the need to safeguard against the potential emotional harm of, and from, different members.

For some individuals, enacting relational responsibility not only meant taking into account the emotional, social and practical well being of existing family members, but also unknown future generations of the family. Rakesh is 51 years old, married with two children, and was diagnosed with a rare adult onset form of SMA, Autosomal Dominant Spinal Muscular Atrophy (ADSMA), in his 40s. Despite reporting a happy and supportive marriage and positive relationships with his two adolescent children, Rakesh's diagnosis of ADSMA in his adult life led him to re-evaluate his pre-diagnosis decisions about marrying and having children, and also to fear for the future well-being of his own children and any subsequent generations of his family:

SMA can be active, it can affect you [symptoms can first appear] from the age of 2 up until about 40 or 50 or even 70, so if you know about it, it could help to prevent it... I wouldn't have got married, if I'd known earlier. Knowing that the gene would affect future generations then I wouldn't no...because, the thing is, right, that you're talking about future generations and four maybe five generations down the road could be affected, you know?

And you know the condition will spread...and ten years time, you don't know what the future will hold...and I think if you've got the condition, you've got that responsibility to stop it in its tracks.

For Rakesh, SMA was conceptualised as a relentless condition which he had a responsibility to stop. Two other participants, one with an adult onset form of SMA (Ian, diagnosed with SBMA), and the other with an earlier onset in infancy (Cara, diagnosed with SMA type II), described a similar concern about future generations and the possibility of having children who are carriers of SMA, regardless of whether or not they displayed symptoms of SMA. Whilst primarily presented as a concern for the prevention of disease in future generations, there was also a social component to the responsibility experienced by these participants. As Cara, commented, *'I suppose it's about preserving the integrity of your family lineage, you don't want any nasties to come from your side of the family'*. Rakesh additionally perceived a stigma associated with disability and genetic disease more generally within the Asian community in which he lived and worked, which could potentially extend to his children, and one which Rakesh saw himself as having primary responsibility for:

I'm very cautious, you know about telling them [people in the community] what I'm suffering from. Medically, they don't understand it so they think it's contagious and stay away from me, and I'm noticing that. You know I say it's a muscle condition, you know, but that's all I tell them, I

can't tell them anything else and if they ask me a specific question, I can't lie to them so I tell them. But the thing with that, right, is that it could affect the children because if they [community members] know that it is genetic, if they know that it's hereditary, my children will be affected if and when they do want to get married... the prospective family might look at it differently, won't they? I mean they might not develop the condition, but they might be carrying the gene, it might be dormant and four generations down it might come out again, and that would affect my children's prospects, so that's my responsibility, right?

Whilst the testing is not yet available for Rakesh's family to know for certain whether his children are likely to develop ADSMA later in their adult lives, for Rakesh, the possibility of his children experiencing the stigma attached to his condition existed regardless of their genotype. The existence of a stigma around carriers of genetic conditions has been considered in the medical and social sciences literature (Kay and Kingston, 2002; Parsons and Atkinson, 1993; Kenen and Schmidt, 1978; Evers-Kiebooms et al., 1994), with the suggestion that carriers of genetic conditions may experience stigma particularly when approaching reproduction, or self-stigmatisation (including experiencing guilt and shame) as a result of the diagnosis of a genetic condition. For some participants, such as Rakesh, through prioritising his relational responsibility to his children and future generations of his family,

the issue of stigma became particularly central to the negotiation of responsibility and led to practices of ‘perception management’ (Goffman, 1963) when interacting with other members of the community.

A final area of relational responsibility that has not been extensively acknowledged in the medical and social sciences literature is a sense of responsibility stemming from the existence of a family member living with SMA. For some participants, having a family member affected by SMA produced a particular form of emotional responsibility which was referenced in their reproductive decision making. For the able-bodied adult siblings of individuals with SMA, the possibility of being perceived as under-estimating the quality and value of their siblings life by undertaking carrier testing or prenatal testing with a view to prevent the birth of someone with SMA produced a particular form of responsibility. Disability rights supporters have referred this particular notion as the ‘expressivist objection’ to prenatal testing (Parens and Asch, 2000), as it draws attention to the way in which the existence of such tests express a negative valuation of disability. There was evidence in this study that this expressivist objection was an important factor that could both justify and constrain the reproductive decisions of siblings or parents of individuals with SMA and inform how participants understood what acting responsibly meant. Zoe is in her late twenties and is the older sister to Rhona, who was diagnosed with SMA type III at two years old. Zoe has a three year old daughter, Freya, and made the decision to avoid carrier testing of herself or her husband before, or during her pregnancy. Zoe cited her responsibility to protect her Rhona’s feelings as an important factor in her

decision to avoid testing, despite Rhona's encouragement for her to use the tests:

The possibility of me being tested to see if I was a carrier was openly discussed in the family. Rhona had mentioned that I could be tested if I wanted to and that it would be quite interesting to know who in the family is a carrier, but I always said the only reason I would ever do it would be just for medical research purposes, you know if they wanted to know to further the research then I would happily help out, but I wouldn't know otherwise because it wouldn't affect me having children or not and my husband was in agreement with that. Then when I became pregnant with Freya the midwife asked me again if I wanted to be tested...but I said absolutely not, because my husband and I both agreed, you know, who wouldn't want another Rhona? We just thought if it happened it happened, and I wouldn't have had a termination anyway. It turns out Freya was fine, but I know that I would have also have felt...very guilty, maybe guilt's not the right word...but it would have been horrible for Rhona if we had undergone the testing. I know she told us she was happy about it, but it would be like us saying that it mattered to us, you know if we had a baby that was like her, it mattered enough to go through the testing, and it absolutely wouldn't...and I wouldn't want her to

think that or feel that way about it. It could be like saying that we actually didn't want her either. Which is rubbish.

Despite the fact that a considerable amount has been written about the expressivist objection, particularly in relation to social policy (Fletcher, 2002; Shakespeare, 1998; Parens and Asch, 2000) and attitudes of, and towards, disabled people (Chen and Schiffman, 2000; Middleton et al., 1998; Press et al., 1998; Shakespeare, 2008), very little attention has been paid to the way in which it is experienced *within* families in the context of relational responsibility. This may be on account of the fact that the expressivist objection to prenatal testing intersects with socially based approaches of disability, which, for political purposes has tended to shy away from psychological responses to disablement. Whilst the concept has been used to describe reactions to antenatal screening practices, anecdotal evidence has pointed to its existence within families affected by genetic conditions (Kent, 2000 pp.61-63; Atkinson, 2008; Bowler, 2006), and for some participants, avoiding this form of emotional harm to family members affected by SMA was an integral part of their relational responsibility to be considered in the context of reproductive decision making.

The different forms of relational responsibility that were experienced within reproductive decisions are important to highlight as they had a profound impact on the way in which reproductive decisions were approached and accounted for to others. As Downing (2005) has highlighted, *which* responsibilities are prioritised in the context of reproductive decision making plays an important role in the negotiation and maintenance of moral identities

and also had serious consequences for the way in which experiential knowledge was (re) conceptualised and expressed in the interviews. Aside from relational responsibility, however, participants also referenced a broader sense of accountability for their reproductive decisions, an accountability to unknown others as well as a moral accountability to society. There is a broad literature documenting the development of new medical technologies and casting speculation over the future of society in the context of these developments (Gurnham, 2005; Brownsword et al., 1999; Fukuyama, 1992; Gosden, 2000). In particular, concerns have been raised about the associations of reproduction with consumerism as prospective parents are able to deselect traits in their future offspring, and a loss of the spontaneity and chance ordinarily associated with reproductive outcomes (Gurnham, 2005). For some writers, this is part of a growing shift towards a society in which children have come to be valued as ‘commodities’ designed to parental specifications, the fears around which have been articulated into the notion of the ‘designer baby’ (Lee, 2002). The sense of ‘accountability’ reported by parents in this study incorporated these wider societal concerns and judgements about ‘designer babies’, as well as responsibilities to more distant or imagined others.

Accountability

Whilst relational responsibility, in its many forms, played a significant role in the reproductive decisions taken by families affected by SMA, a less personal and more abstract notion of responsibility was also expressed which I will

refer to as ‘accountability’. The term ‘accountability’ captures the responsibilities felt towards unknown others, communities and society. These forms of responsibility are captured in the work of Rapp (1999), who has referred to women as ‘the philosophers and gatekeepers of the limits of who may join our current communities’ (p. 318), and are thus accountable for their reproductive decisions at not only a personal level, but also a social one.

There was evidence to suggest that participants were acutely aware of the way in which their decisions were judged and perceived by others and their responsibility to maintain their moral identities in this sphere. Press coverage of the issues around reproductive decision making, as well as comments from the general public served as reminders to families affected by SMA that their accountability for their reproductive choices extended beyond their own family; that these were social issues in which other individuals had a stake.

One mother of a child with SMA recalled an incident of being asked by an acquaintance if she planned to get her subsequent pregnancy ‘properly tested’ so as to avoid tax payers taking the brunt of the financial consequences of her having a second disabled child (Tara, mother of child diagnosed with SMA type II). Kate is 37, has a seven year old child diagnosed with SMA type II, and at the time of interview was about to embark on her third cycle of Pre-implantation Genetic Diagnosis (see Appendix V for a description of PGD).

She described her encounters with the attitudes of others in the following way:

K: Most people I tell about the PGD are fine with it,
but I did have one comment off one person saying that

I'm playing God, I'm messing in things that I shouldn't be, and don't I think about the direction this is heading in, but that's the only negative comment I've had.

F: and how did you respond to that?

K: Well she went off to do IVF herself anyway so, what's the difference? But, she wasn't a close enough friend for me to challenge her on that one or to worry about it. If she had've been closer to me then I would have done but she wasn't and the ones who have an answer for everything...the people who aren't in that situation are the ones who tend to not agree. And I just think just wait and see if you're in this situation then see how you feel then.

The way in which PGD is accounted for in the narratives of those undergoing or moving on from the process has been described by Franklin and Roberts (2006) and Roberts and Franklin (2004). One participant in Roberts and Franklin's (2004) paper, Anne, who lost her daughter to SMA before undergoing PGD, experienced a similar sense of wider interest in her reproductive decisions, this time in relation to the 'designer baby' debate (Roberts and Franklin, 2004: 289). The idea that the use of PGD (and indeed other reproductive technologies) are usurping God's role in deciding 'who should and should not inhabit the world' (Hubbard, 2006:93), or paving the way for future generations of genetically enhanced children (Franklin and Roberts, 2006) have been discussed in the literature, particularly in relation to

popular representations and public opinions (Scully et al., 2006; Bloomfield and Vurdubakis, 1995; Lee, 2002; Gosden, 2000; Nerlich et al., 1999). For Kate, (as well as for Anne, in Roberts and Franklin's (2004) study), however, these forms of public accountability were not always experienced as a form of responsibility. The closeness of Kate's relationship to her friend was an important factor in determining the boundaries of her accountability in reproductive decision making. Finch and Mason's (1993) study of the negotiation of familial obligation has suggested that closeness and involvement are important mediating factors in the experience of familial responsibility, in that those individuals with close emotional ties may experience their familial responsibilities in a different way to those with more distant relationships. Beyond the family, the emotional and psychological connectedness of individuals appears to affect their sense of accountability to them. As the woman to whom Kate refers was not a 'close' friend, and was not in the same situation as her, Kate was able to take her accusation less seriously, but nevertheless still felt that she had to defend her actions. By referring to her own personal knowledge of her standpoint, the experiential knowledge she has obtained through both being a mother to a seven year old child with SMA, and having already gone through two cycles of PGD, Kate could defend her position and discredit her friend's opinion and standpoint as a 'non-knower'; her lack of intimate knowledge of her life was used as a means by which to deflect her judgement and surpass any responsibility to wider society, the future of reproduction, or God.

Thus far, I have presented the different forms of responsibility that emerged in the accounts of, and reflections about, reproductive decision making in families affected by SMA. These have included, but are not confined to, forms of 'relational responsibility' and 'accountability'. Whilst these forms of responsibility have been discussed as discrete categories for illustrative purposes, there is a degree of fluidity and inter-connectedness in the way in which they are experienced. Individuals may maintain their broader accountability by prioritising and upholding their relational responsibilities, and a sense of accountability may correspondingly shape the nature and ideals of relational responsibility as they change and transform over time. A presentation of the different ways in which responsibility is experienced in reproductive decision making is important as it is through attendance on these responsibilities that social and moral identities are sustained (Walker, 1998; Finch and Mason, 1993). Reproduction is an arena in which powerful norms and values exist; values which sustain the boundaries of these obligations. It is through these that would-be parents must negotiate their own reproductive choices. I will now present these strategies used to negotiate responsibility in further detail in the following sections, specifically focusing on the different meanings attributed to experiential knowledge in these contexts, as both 'warning' and 'reassurance'.

The Negotiation of Experiential Knowledge and Responsibility

In the previous section, I have considered the various definitions and types of responsibility that were considered in the imagined and actual reproductive decisions made by those with SMA in their families. This section will examine the way in which these responsibilities were negotiated in reproductive decision making in the context of experiential knowledge of SMA. The literature has suggested that the experience of a condition, whether first-hand or through a family member has the potential to affect feelings about having a child oneself with that condition (Gow, 2000; Wertz, 1992; Asch, 1992; Parsons and Atkinson, 1993; Kelly, 2009) and, more recently, how these forms of experiential knowledge may influence screening decisions through more distant forms of knowing has been highlighted (Etchegary et al., 2008). However, no study thus far has specifically examined the intersection of these different forms of experiential knowledge with the specific responsibilities associated with reproduction, and the tensions and (dis)continuities associated with them that are negotiated by families affected by an inheritable condition.

Shakespeare (1998) has suggested that in spite of the implications of genetic technologies for people with disabilities, the voices of disabled people have been largely absent from discussions around the implications of their uses (Shakespeare, 1998: 673), with researchers instead focusing on the needs and concerns of prospective parents. This research seeks to address this gap in the literature by simultaneously presenting both the concerns of people with disabilities and their family members. It is within the relationship between those diagnosed with SMA and their family members that one of the central

issues raised by disability rights supporters about prenatal testing, the expressivist objection, is experienced and negotiated.

Experience as Warning/Reassurance

The different ways in which SMA was experienced and described by participants has been discussed in Chapter 5. This chapter emphasised the way in which participants distinguished between different forms of SMA experience; ‘disability’, ‘embodied experiences of impairment’ and ‘illness, death and bereavement’. It is in the context of reproductive decision making, however, that these experiences and understandings of SMA took on new significance and meaning. Indeed, participants frequently reported contrasting views about the possibilities of having a child with different types of SMA, on the basis that they are associated with different experiences, different abilities and different levels of suffering. Whilst the prenatal test available for SMA is not able to determine which type of SMA a foetus is likely to develop, it is generally accepted within the medical profession that future generations, if found to have two copies of the deleted SMN1 gene, will be affected by SMA in a similar way to existing or previous relatives. Whilst the experiences of some sibling groups within this study suggested that this was not always the case (e.g. Kristen and Ellie, sisters who were diagnosed with different types of SMA), participants nevertheless reported that they anticipated any future generations diagnosed with SMA to be affected in a similar way to existing family members. Within this context, the meaning attributed to that particular

relative's experiences with SMA came to be of crucial importance in imagining future lives with SMA.

Experience as Warning

In speaking about, and attributing meaning to, this experiential knowledge in the context of reproductive decision making, participants presented their conceptualisations of SMA in more or less certain ways. For parents of children who experienced severe SMA and died in infancy or experienced ongoing illnesses with a shortened life expectancy, the suffering associated with SMA was an unquestionable fact. Fraser is in his 50s, has two children without SMA, but experienced the deterioration, suffering and eventual deaths of his second son and first daughter due to SMA type I at 10 months and 8 months of age respectively. Fraser had witnessed his children gradually decline over their short lives, requiring tube feeding, and finally being unable to breathe unassisted, spending much of their short lives in a paediatric intensive care unit. For Fraser, the suffering his children had gone through before their early deaths left no room for interpretation, and left him feeling secure in the meaning he attributed to his experiential knowledge- that it was a warning of the suffering involved with SMA and an impetus to prevent the recurrence of SMA in future generations:

....the ability to under go prenatal testing [after the deaths of 2 babies, and before going on to having child without SMA] was a God send for us, because no one would want that if they could avoid it and I think everybody would say the same who

is affected by it. I don't see any dilemma at all with type I testing, the test is there and I think every parent should take advantage of that. I can't understand where they're coming from if they don't...I know some parents face a big dilemma around type II testing, but if type I is the biggest genetic killer of children under the age of 1 in the UK, which I'm told it is, then I'm sitting in the prettiest spot in the argument in a way because I don't see any dilemma with using the test. You know, and very few people could argue against my position...I know some people talk about type Is living past their first birthday, but that wasn't going to be the case for our children, so I think I'm sitting in the securest spot in the whole argument, I think.

For Fraser, the suffering and the shortened life of any future children affected by SMA were presented as a certainty. In the same way that parents of type I babies in Roberts and Franklin's study (2004) emphasised the certainty of illness and premature death of babies affected by type I SMA, so Fraser attributed this meaning to his witnessing of his children's deaths. By presenting this outcome as inevitable, Fraser saw his responsibility as lying primarily as preventing the recurrence of SMA in future generations, which in turn enabled him to see his position regarding prenatal testing as secure; by undergoing testing and selective termination of any future pregnancies found to be affected by SMA, Fraser and his wife, Charlie, were enacting their responsibilities as parents to prevent the future suffering of another child, to

divert the possibilities of their own grief and to safeguard their family's emotional well being. Their perceived sense of responsibility lay not in their attendance to a broader, ethically framed accountability around prenatal testing, but instead emerged from the realities of the pain, sadness and horror of watching their two young children succumb to SMA.

Hallowell's (2006) study of women's perceptions of the risk of developing ovarian cancer in the context of a familial history of the disease has emphasised the way in which analyses of negotiations around risk, and genetic risk in particular, have been abstracted from reality; the risks are not treated as immediate or 'real', and the management of these abstract risks are typically cast as an issue of identity (Giddens, 1991; Downing, 2005; Novas and Rose, 2000; Polzer et al., 2002), devoid of any emotional content. By drawing on interviews with women at risk, Hallowell (2006) has demonstrated the emotional dimension to genetic risk perception; the women she interviewed had nearly all witnessed the gradual decline and eventual death of a close relative with ovarian cancer. Their perception of their genetic risk was both grounded in their intimate encounters with suffering as well as their obligations to maintain their other social relationships (e.g. to partners, children, parents). Within this study, the emotional weight of participants' experiential knowledge of SMA was felt in their accounts of suffering; it was heard in the voices of parents as they recounted their experiences of witnessing their children dying; it was visible at the bereavement support meetings, the memorial service at the JTSMA annual conference and it was felt every time a sentence was left unfinished during an interview, the

emotional weight of experiential knowledge often too heavy and painful to translate into words. As one participant eloquently stated, recounting her experiences of witnessing the deaths of her three sons in infancy due to SMA, *'bereavement is such a...hollow word...I cannot name what it is that I have lost, can't identify... I have no language... there are no words'* (Denise).

Frank (2001) has questioned the ability of researchers to accurately depict the experience of suffering, as, by its very nature, suffering cannot be contained within language; to tie it to words is to fail to accurately represent it, reducing feelings and sensations to 'complaints and specific concerns' (Frank, 2001: 354). Hollowell (2006) and Bendelow (2006), however, have argued that whilst dimensions of suffering are experienced privately and may defy representation, suffering can nevertheless be communicated, recognised and understood in ourselves and others. Whilst representations of it may only ever be partial at best, Hollowell (2006) has argued that we nonetheless have an obligation to write personal experiences of suffering into research; a process that may challenge researchers emotionally, but to neglect this duty is to risk overlooking an integral aspect of human experience.

In terms of the perception of genetic risk and responsibility, emotional suffering incurred through witnessing the deaths/frequent illnesses of close family members with SMA fundamentally altered the way in which participants approached reproductive decision making. Experiential knowledge of the horrors of past suffering, moreover, compelled participants to formulate their relationship to the future in specific ways; enacting reproductive responsibilities became a means by which to circumvent future

suffering and alter the relationship to past hurts, as Fraser went on to comment:

Despite the two affected by SMA, we have got two children both free from SMA, and it's my belief that you get paid back, you know Alicia's doing great and Toby's just started at medical school. You know, for all that we went through, you get paid back. And I believe it's changed me as a person, what I've been through. I'm more comfortable talking about death with folk, I feel ok to do that. I'm probably a nicer person than I would have been [laughs].

The suffering experienced by Fraser, his wife and his two children affected by SMA, enabled him to regard the successes of his children without SMA as a form of compensation or reward for what they had gone through, reinforcing his identity as a responsible parent. Giddens (1991) has argued that self-identity is not fixed throughout the life span, but is managed in relation to lived realities and experiences. The 'reflexive project of self', or the rational decisions taken to manage self-identity may be triggered by significant experiences which compel individuals to redefine their identities. Experiential knowledge of suffering, and the steps taken to avoid its recurrence, not only had implications for the identities of individuals in their present, but was also a means of maintaining continuity of these identities for future selves. By defining his children's various successes as a form of payback, Fraser

continued to actively manage his identity as a responsible parent and one humbled and fundamentally changed by his experiences of trauma.

Whilst the narratives of those parents whose children died in early infancy from SMA invariably involved descriptions of experiences characterised by unequivocal suffering, of both the children and their families, descriptions of the lives of children who survived infancy but incurred frequent bouts of illness and high degrees of restriction were also talked about in these terms. It was in these circumstances that the experiences of SMA were interpreted as a warning of the possibilities of suffering in future generations, and the focus of reproductive responsibility was presented as the prevention of its recurrence. Downing's (2005) research on the perception and enactment of responsibility in families affected by Huntington's disease when approaching reproductive decision making has emphasised the way in which the nature of social relationships alters the perception and enactment of responsibility. The changing prioritisation of relationships (i.e. whether prospective parents prioritise their relationship with each other, themselves, their existing children or any other relationship which they see as having value and meaning), alters the way in which they relate to, and experience responsibility. For participants who defined their relatives' experiences of SMA as involving suffering, there was little room to accommodate potentially conflicting responsibilities. The obligation to protect the interests of a potential foetus or child was seen to override any parental objections or reservations about prenatal testing.

Trisha is in her 30s and has a seven year old daughter, Joanna, diagnosed with type I SMA. Whilst Trisha and her husband are not certain about whether or not they would want any more children, Trisha reported that she felt the use of prenatal testing and other antenatal tests is an integral part of accepting the responsibilities of parenthood as they protect the best interests of the child:

I suppose I think that now you have all these tests, you can test for other conditions and SMA as well, there's no need to have disabled children you know. If you don't need to, then don't, you know? At the hospital where we take Joanna there are children there who are in a lot of pain and who have no quality of life at all...and I think if that could have been avoided, then that probably should have been the case. Without wanting to sound callous...but I think that because I've got Jo I can say that, it gives me some legitimacy to say that. I see what a burden it is for Jo to be carrying on, she's so often ill and I watch her suffer with it everyday so...if you've got a pregnancy, you've got a responsibility to do what you can for that child, what's in their best interests even if it goes against your instincts as a mother.

The issues suggested in the balancing of parental autonomy with the responsibility to protect the best interests of future lives in the context of potential disability or impairment has been discussed widely in the literature

(Downing, 2005; Vehmas, 2001, 2002; Skene, 1998; Charo and Rothenberg, 1994; Rhodes, 1998; Shakespeare, 2008). Whilst for writers such as Green (1997) and Purdy (1996), there are moral limits to parental freedom in reproductive decision making in that parents must 'strive to give...children lives unimpaired by genetic (or congenital) disorders' (Green, 1997: 6), for other writers such as Vehmas (2001) and Asch (2001), the decision to undertake to parent *any* child, regardless of impairment or disease, can also be considered a hallmark of responsible parenthood. Indeed, the widespread abhorrence expressed by the general public to the use of genetic technologies such as PGD to select future children on the basis of social reasons, (e.g. preferred eye/hair colour) can be regarded as an expression of this conception of parental responsibility (Scully et al., 2006); 'responsible' parents, according to this perspective, are expected to love and value whatever child they produce, regardless of their own personal preferences and desires. Parents such as Trisha were acutely aware of the existence of this broader accountability through which reproductive decisions were scrutinized, and many reported feeling guilty or judged by others (whether imagined or enacted) for transgressing (for example through invasive testing or termination) what Trisha refers to as the 'natural instinct' as a mother to nurture and protect their growing foetus.

Importantly however, for Trisha, experiential knowledge of her daughter's condition offered her a means by which to escape this paradox in a way that maintained her identity as a responsible parent. Trisha's experiences of being a primary carer to her seven year old daughter with SMA who

requires 24 hour care, and a witness to her frequent illnesses with chest infections, led her to perceive SMA as a burdensome. For Trisha, having SMA meant ‘suffering’, not only for Joanna, but also for herself and husband who described physical and psychological exhaustion meeting her needs and that of her other daughter, aged 11. Trisha mobilised her experiences of her daughter’s impairment to validate her perspective on reproductive decision making; by emphasising the burden of her daughter’s condition for both of their lives and regarding their experiences as a ‘warning’ as to the quality of future lives with SMA, Trisha was able to ‘trump’ the discourse of parental responsibility which positioned her with a duty to care for *any* child she would have. Moreover, Trisha additionally positioned herself as being in a privileged position vis-à-vis other parents in that she had intimate experience of living with SMA which qualified her to lay claim to experiential knowledge and thus enabled her to make ‘insider’ judgements about the sorts of lives children with severe disabilities possess, implicitly suggesting that differently positioned parents may not be able to justify such a standpoint in the same way. By laying out the boundaries of experiential knowledge and maintaining the unacceptable standard of life experienced by children with SMA, Trisha was able to exonerate herself from one of the perceived central tenets of parental responsibility: to accept any child.

Experience as Reassurance

Whilst the existence of genetic testing technologies have set a new precedent in terms of extending the boundaries of responsible behaviour in pregnancy

(Lippman, 1991), for some participants in this research, the decision not to use genetic technologies could equally be presented as acting responsibly through the mobilisation of experiential knowledge and the transformation of reproductive responsibility. For individuals with SMA themselves, and thus with first-hand experiential knowledge, this standpoint was particularly relevant to the presentation of their reproductive decision making. Rhona is in her twenties, has been diagnosed with SMA type III, and hopes to have children in the near future. For Rhona and her husband, the process of undergoing genetic testing in preparation for childbearing was not a priority, despite it being strongly recommended to them by health care professionals. While Rhona recognized that prenatal testing can offer benefits to some people, she felt that selective termination or PGD would not be an option to her in the event of her husband being found to be a carrier of SMA. For Rhona, her experiences with SMA were central to this decision:

I suppose actually in hindsight now, SMA's been a positive thing really. I think that a lot of sort of what I've done and what I've achieved has been hugely down to sort of having the personality to overcome the problems that have come along with SMA. Um I think when you sort of know your own experiences and when you think about having children and whether they will be affected by SMA, the way I look at it is, 'well I've coped and I'm fine with everything' so, you know, it's not all bad. I mean there're always going to be a certain amount of people who feel it's irresponsible to bring in a child to the world knowing that they're going to be

disabled or because you're disabled yourself, but they tend to be the people who have no experience of disability themselves, they don't know what it's like and it's just their perceptions... and that's just part of the harshness of life and I don't take a lot of notice of them.

Saxton (1984) and Morris (1991) have drawn attention to the way in which women with disabilities in particular are often regarded as being incapable of caring for children, and are thus excluded from the sphere of parenthood. This situation may be particularly exacerbated for individuals with genetic impairments who are presumed to automatically transmit their impairment to their offspring, regardless of the nature of its inheritance. Thus, both parent and offspring are positioned as being potentially problematic (Kallianes and Rubenfeld, 1997). The prevalence of negative attitudes towards people with disabilities reproducing may introduce a degree of accountability to the decisions made by those with disabilities, which Rhona acknowledges. However, by distinguishing between others' perceptions and her own experiences, Rhona was able to resist the discourse of responsibility that she felt others applied to her as a disabled mother and potentially the mother of a disabled child. Defining her embodied knowledge of living with a genetic disability as being '*not so bad*' (or even positive retrospectively) enabled her to understand and represent her decision as responsible. SMA for her was not harmful or undesirable (as others may suggest), but a condition which instead instilled character and around which a happy and fulfilling life could be built.

As well as a positive image of what a person's life with SMA could be like, Rhona also challenged the idea that someone with SMA would not be able to adequately parent a child, particularly a child with SMA themselves, by drawing on her experiential knowledge:

I mean I think a child with SMA would be really lucky to have a parent with SMA really, because as someone with the same disability, you already know about the sorts of obstacles you're going to come across, getting into schools and the surgery...things like that, and you already know about how to get round them...you know where to look for help and support and you've already made those sacrifices in your life and live with the restrictions that can come with it. You don't get upset when you can't take your child on holiday because there's nowhere accessible, because you never had those holidays yourself, you know? So I just think I'd be a lot more confident and have a hell of a lot more insight than, say, an able-bodied parent who has a child with SMA, you know, who didn't even know what it was before the child was diagnosed.

Rhona was aware that others might regard the decision to risk having a child with SMA as irresponsible and of her own need to be accountable to this accusation. However, by drawing on her own life experiences and by defining them as positive, she was able to redefine the boundaries of parental responsibility and justify her own standpoint on parenting with SMA. Being in the privileged position of knowing SMA from the inside, Rhona felt that

she had the authority to rewrite notions of responsibility, over and above those who might define her as irresponsible.

Whilst Rhona drew on her experiential knowledge to disregard the responsibility that was so central to the decisions undertaken by Trisha and Fraser (to prevent the future suffering of children with SMA), being reassured that SMA was a condition that could be lived with (with minimal or no suffering) did not necessarily mean that prospective parents felt willing or able to undertake this parenting. Megan is 32 and was diagnosed with SMA type II at the age of 18 months. She has never been able to walk and has used an electric wheelchair since childhood. She lives independently with the assistance of PAs. Whilst currently not in a relationship or considering having children, and indeed in doubt about her ability to have children, Megan nevertheless described having thought carefully about what the possibilities of childbearing would mean for her:

I've thought about it quite a bit actually because...um If I was having a child with SMA, well any...if I was having a child with any disability, I would want to be in a very secure relationship with someone who was very supportive and I think particularly if that child had SMA. I don't think...I would have no worries about bringing a child into the world who has SMA because I know my life, although it's been difficult at times-I'm not going to say it's been easy- but I do know that it's been enriched by it as well. And you know I would have no feelings of 'oh I shouldn't put somebody else through that' at all, because I don't

think it's a bad thing, but I would think...I would have to seriously consider how my partner would feel about that. And I think that would be my major concern, you know the practical side of things and having a partner who could provide that support, to support me and a child with SMA. It'd be more, can I put all that on my partner's shoulders? Because I certainly wouldn't think 'oh the child will have a terrible life I couldn't put them through that', it'd be more 'can I ask that of a partner?' and I don't think I could.

For individuals with SMA themselves, the uncertainty, risk and responsibility associated with reproduction related not only to the risk of them having a child affected by SMA (which is medically defined as a 3 out of 4 chance if the partner is a carrier of SMA and a certainty if their partner had SMA as well), but also uncertainty about their physical ability to undertake parenting tasks associated with raising a child, with or without SMA. Downing (2005) has suggested a 'model of responsibility' to describe the way in which people approach genetically reproductive decisions (Downing, 2005: 220). By examining the way in which people evaluate their ideas about the future, personal values, prioritization of relationships and access to social support in relation to their conceptualisations of risk, Downing has argued that the negotiation of responsibility in genetically risky reproductive decision making is in a constant state of flux (Downing, 2005: 221). As each of these factors change and shift in terms of the importance attributed to them over time and context, so the approach to, and actual reproductive decisions made,

may alter. For Megan, her experiential knowledge of SMA was an important aspect of this negotiation, as it was her primary resource in her evaluation of a child's potential life with SMA. By defining a life with SMA as a worthwhile life, and instead prioritising the relationship with a potential partner, Megan was able to discount the responsibility that had so guided Fraser's and Trisha's decisions. Unlike Rhona, however, who felt secure in her abilities to undergo childbearing, Megan's prioritisation of her relationship with a potential partner enabled her to regard the possibility of foregoing reproduction (despite her positive evaluation of life with SMA) as responsible.

The Specificity of Experiential Knowledge

Whilst experiential knowledge of SMA, whether conceptualised as warning or reassurance, was spoken about primarily in terms of the suggested implications for future lives with SMA, for some participants, the knowledge acquired by living through a relative's SMA was not related to one particular diagnostic category, and thus could be used as a resource to guide reproductive decisions relating to other disabilities and conditions. For such participants, their experience of SMA, rather than being centred around a diagnostic label, was instead identified according to the *type* of experience they had had with SMA whether this was of 'disability', 'illness and death' or 'embodied experiences of impairment'. Becky, who lives in Switzerland and whose first baby, Emma, died of SMA type I at eight months of age described

how her experiences of SMA altered her attitudes to testing for her three subsequent pregnancies:

B: When I was pregnant with Emma I didn't have any tests at all, I just had the nuchal screening for Down's [syndrome], but that was all fine. I was told I was low risk. But after the sadness and grief of losing Emma we were a lot more careful about the other pregnancies, we had one termination for SMA. We also requested to be tested to see whether we were carriers of Cystic Fibrosis, which I don't think is available in the UK.

F: Um, I don't think so...why was that?

B: It was to do with the fact that there is no cure for Cystic Fibrosis. We didn't want to bring another ill baby into the world when there's nothing that can be done. I would have accepted a test for anything like that, we researched the various conditions and which we could be tested for.

Whilst Becky's experiences had been labelled as 'SMA', she described them in terms of the suffering, sadness and loss associated with having a terminally ill baby. For Becky, these experiences traversed medically upheld boundaries and compelled her to avoid, not just SMA specifically, but any condition deemed incurable and fatal. Her experiential knowledge instilled in her not simply a responsibility to prevent another life with SMA, but instead a responsibility to prevent the birth of a terminally ill baby. Her sense of responsibility was thus grounded in her experiences rather than being attached to a specific medically defined condition.

Whilst Becky's experiences of the loss of her daughter warned her of the difficulties she may face as the mother of another seriously ill baby, for participants whose experiences of SMA were reassuring, a similar projection of the experience of SMA onto other conditions took place. Matthew is in his twenties and has a sister, Cara, who was diagnosed with SMA type II at 18 months of age. Matthew and Cara currently live together in a shared flat. Whilst not considering reproduction in the immediate future, Matthew reported that, on reflection, his perspective on disability generally has been shaped by his experiences of his sister's SMA:

I think having the family history I have, living with my sister for 25 years, makes me far less inclined to be worried about having a disabled child myself, be it SMA or Down's Syndrome, or whatever, because, [pause], because I don't see...you know I went on to take a job with disabled people, I work with students who have Down's Syndrome and similar conditions and it certainly does not make them of less worth. Er, you start to see disability as simply a different way of doing things rather than a problem...And er, I suppose being close to a disability, being close to differences gives you that awareness. Disability is an example of a difference, within individuals...and that's what we're talking about really, aren't we? Attitudes to disability.

Despite the fact that Press et al.'s (1998) and Etchegary et al.'s (2008) respective studies suggest that women's personal experiences of disability,

whether within their own families or broader social circles do not necessarily lead to more accepting attitudes about the possibility of having a disabled child themselves, for Matthew, familiarity with SMA and his choice to work with disabled people allayed any fears about having a disabled child himself. For him, feelings about having a child with SMA were synonymous with feelings about disability more generally. This attitude was reflected by other participants who had grown up alongside SMA, and in particular, participants with SMA who defined themselves as ‘disabled’ as opposed to identifying themselves as ‘a person who has SMA’, as Kristen (who has been diagnosed with type II SMA) commented:

Being a disabled person, you come into contact with other people who are disabled just in life generally. I went to PHAB [Physically Handicapped and Able-Bodied] club and met people with all kinds of disabilities which taught me to be tolerant and have a greater awareness of others with disabilities- you face a lot of the same problems.

These different ways of understanding and identifying with SMA are all relevant to the way in which experiential knowledge was conceptualised. For both Matthew and Becky, their experiences of SMA had implications for their reproductive responsibilities, not just in relation to SMA, but for other conditions that are currently tested for. Their respective experiences of disability and illness, and the meaning they attached to them, were projected onto other conditions which they deemed to involve similar consequences. Some of these projections were based on direct experiences with others with

these conditions, whilst others were based on cultural representations and medicalised information. The research by Wertz (1992) has suggested that parents of children with a genetic condition, CF, express a range of attitudes towards selective termination for other conditions, with some fully supporting the practice and others less comfortable. This study suggests that it is the experiential knowledge accumulated through living intimately with a given condition that is important in shaping these attitudes towards other conditions, and consequently, in shaping reproductive responsibilities towards them.

Conclusions

This chapter has demonstrated, through the presentation of different participants' accounts, how the meaning attributed to experiences of SMA informs participants' conceptualisations of their genetic risk and, consequently their reproductive responsibilities. As Downing (2005) has argued, the strategies thus used to diminish genetic risk by individuals from families affected by SMA may offer more of an insight into the way in which genetic risk and responsibility are experienced than what is actually decided. Whilst Megan, Rona, Fraser and Trish arrived at different (anticipated) reproductive decisions, the strategies they employed to arrive at, and assert, these decisions were similar in that they all mobilised their various experiences of SMA to ascertain where their responsibilities lay. Their experiential knowledge was, however, not only a resource with which to imagine future lives and discern responsibilities, but it was also used to deflect the felt and anticipated criticisms of others. By constructing

themselves as the true ‘knowers’ about SMA, in that they live(d) with it, they positioned themselves as possessing a privileged standpoint from which to best assess the risks to any future offspring associated with being born with SMA; what they knew about SMA was, therefore, less important than the way in which they presented and positioned this knowledge. Indeed, by the strategic presentation of their experiential knowledge, participants could exonerate themselves from the demands of specific aspects of parental responsibility, by invalidating the (felt) criticisms of others, and also by transforming its meaning. For Trisha and Fraser, experiences of SMA instilled in them a responsibility to prevent another life with SMA, whereas Rhona and Megan’s experiences of SMA meant that they could transform what it meant to be a parent, or a child, in the context of having SMA and to regard these as positive, rather than the negative way they assumed them to be evaluated by others. However, for some participants, experiential knowledge, rather than bringing into focus the felt responsibilities associated with reproduction, instead heightened the some of the dilemmas around reproduction. Further, for some participants, experiential knowledge was presented as being an irrelevant or unreliable knowledge source. I will present these contrasting accounts in Chapter 6.

Chapter 6

The Limits of Experiential Knowledge in Reproductive Decision Making

In the previous chapter, I have presented the way in which experiential knowledge can be drawn upon as a constructive resource in reproductive decision making. Whilst these participants attached vastly different meanings to this knowledge of SMA, it was nevertheless presented by all as a reliable and valuable form of insight, around which future reproductive decisions could be oriented. However, for some participants, the usefulness of this knowledge was less clear. Indeed, experiential knowledge could be experienced as problematic when its meaning was less certain, less fixed, or conflicted with other deeply held values. It is these instances wherein experiential knowledge was felt to be burdensome, challenging or unstable that will now be examined.

The Burden of Experiential Knowledge

In the processes of thinking about, and recalling, reproductive decisions, participants spoke about the range of responsibilities for which they were accountable. Whilst for Fraser, Trisha and Rhona and Megan presented in Chapter 6, experiential knowledge was described as enabling them to navigate these competing demands and maintain their identities as responsible (potential) parents, for others, their experiential knowledge was burdensome and in conflict with their perceived responsibilities.

Kate is 37 and has a seven year old son, Jamie, diagnosed with SMA type II. At the time of interview Kate and her husband were waiting to undergo their third cycle of PGD. Kate described her arrival at the decision to undergo PGD as arising out of a long period of not conceiving despite trying to, and having been offered the procedure when seeking medical advice for infertility:

I think we took the PGD because somebody told me about it and then we got offered the funding so we just sort of went along with it...we wanted another child and it had just never happened for us. PGD seemed like it would be the easiest route, because I couldn't have another child like Jamie, it wouldn't be fair on him because he needs so much attention, and I couldn't cope with all the lifting. We didn't have the...but going down the route of testing and terminating... well it would be like getting rid of Jamie, that's how I'd look at it. And you know I love him to bits, he's so funny, and yes he's in a wheelchair what have you, but he's a lovely little boy and that's how I see him. And it would just be like saying, well wiping him away really, saying he wasn't really what we wanted...and that would be a very hard thing to explain to Jamie when he's older, but because of not being able to conceive naturally as well...I don't know if that makes it easier for me, because it's not like I just chose it, it

was because I couldn't conceive naturally it was the only option.

Kate's description of her arrival at PGD touched on some of the recurring themes across many of the interviews undertaken with parents of children with SMA who wanted to have further children. For Kate, her experiential knowledge of SMA presented her with a difficult dilemma; it enabled her to see not only the reality of the care work involved in looking after a child with SMA, but it also simultaneously enabled her to discount SMA when evaluating her son's life: *'when you have a child and live with a child, you see past the wheelchair and all the problems'* (Kate). This personal knowledge of what it's like to live with SMA, and also feeling that caring for a second child with similar support needs would stretch the family's resources beyond their capacity, left Kate trapped between incompatible responsibilities, to both protect the emotional and physical well-being of her existing child with SMA, but also to safeguard the future welfare of the family unit. Larson (1998), through an empirical study of the impact of disability on maternal-child relationships in Mexico, has referred to this situation as the 'paradox' of disability. Through her interviews with mothers, Larson discovered that a constant tension existed between the mothers' feelings about loving their child for who s/he was, but simultaneously wishing to erase the disability, and the problems that came along with it (Larson, 1998: 865). Larson's (1998) study highlights the oversimplifications of the expressivist objection to prenatal testing- pointing to the possibility of concurrently valuing the lives of those with disabilities, but also

wishing to avoid the challenges associated with life with a disability. For Kate, in spite of the fact that PGD is generally not positioned within the medical-scientific literature as an infertility treatment, presenting her use of PGD not as a 'choice', but instead as *inevitable* given her difficulties with conceiving, enabled her to negotiate these competing responsibilities that her knowledge of SMA presented.

Whilst Kate's experiences with SMA and PGD enabled her to navigate a pathway through the competing responsibilities and inherent tensions she perceived to be involved with loving and valuing a child with SMA whilst also preventing future affected lives, other participants became trapped by the dilemma, unable to reconcile their conflicting feelings. Claire is 30 and lives in a neighbouring town to her sister, Megan (32), who was diagnosed with SMA type II at the age of 18 months. Megan works as an artist, and Claire works as a part-time teacher and is also employed by Megan as one of her personal assistants for the remainder of the time, replicating some of the assistance work Claire undertook for Megan when they were growing up. Both sisters took part in the study and described their relationship with each other as close. Claire has a long term partner with whom she has had discussions about the possibility of having children. However, for Claire, her close relationship with her sister, as well as her knowledge about the possibility of genetic testing created a painful dilemma for her and her partner:

Yeah I think I'm in quite a difficult position with that really, with having children. Because my first thought was that I

would never bother with any sort of testing, if we were going down that route, because I know SMA quite well really and I see what a wonderful person Megan is, and you know we often say that her condition has given her strength, mental strength that she may never have developed, so I know it on that level, but... I also know the other layers of it...um [pause] I suppose it sort of sounds cold to say it but, it's human nature to sort of think that if it could be preventable for somebody...Is it going to be really difficult or is it something that you can take on? Of course you're going to want the child that doesn't have to struggle and things, no one wants to see their child go through that. So I've got that, but on the other hand I've got 'what would that say to Megan?' It would be like me saying that she wasn't important or that her life wasn't worthwhile with her condition, that there was something wrong with her. So where you go with that...um...Yeah I'm stuck between a rock and a hard place [laughs].

In the same way that Kate feared explaining her decision about avoiding the birth of a child with SMA to her son in the future, so Claire feared her sister's interpretation of a decision to avoid a similar situation. Whilst for Kate, her resolution to her dilemma lay in use of PGD, for Claire, there was no such means of reconciling her experiential knowledge of SMA with her responsibility to protect her sister's feelings and possible interpretations of her actions. Downing's (2005) research has suggested that social context is

important in the approach to reproductive decision making, and that these contexts may inform the values individuals bring to reproductive decisions, as well as for what, and to whom, they consider themselves accountable. What emerged as particularly important in the narratives of those who took part in this research was the point in their reproductive lives at which the interview took place. Interviews were conducted with those who had had children or were in the process of deciding, whilst for others, the possibilities of reproduction were hypothetical or imagined. Some participants narrated well-rehearsed accounts of the reproductive decisions they had already made and appeared familiar with telling these stories, highlighting the fact that negotiations of responsibility for reproductive decisions within families affected by SMA extend beyond the point at which decisions were enacted, and indeed the work of managing and justifying these decisions for such families was ongoing. For other participants, however, the possibility of reproduction had not been thought about in any depth before; whilst some had undergone carrier testing and therefore had been given statistics as to their degree of genetic risk, others were only aware of this risk by virtue of having an affected relative. For individuals such as Claire, reproduction, as a distant possibility rather than an immediately threatening risk, was considered from a level of abstraction that allowed more room for uncertainties and imaginings, than for those participants who were reflecting on decisions that had already been made. It may be unsurprising therefore, that reproductive dilemmas were more frequently expressed by those participants who had not yet made a reproductive decision that involved medically defined genetic risk. Indeed,

such accounts many not reflect the reproductive decisions participants may go on to make, as the context of reproduction changes with increasing life experience, age, competing responsibilities, financial status etc. Nevertheless for Claire, experiential knowledge of SMA and the context of her relationship with her sister, complicated her feelings about her own possible genetic risk. Whilst experiential knowledge, therefore, could in certain situations be used strategically to clarify and justify reproductive decisions, for other participants, it left them stuck ‘between a rock and a hard place’, with a risk of being constructed as ‘irresponsible’ whatever decision is taken.

As well as fears about the possible impact of an expressivist objection to genetic testing from family members affected by SMA, participants also spoke about their responsibility to protect their own emotional well being, and the ways in which their knowledge of SMA posed particular threats to this responsibility. Evelyn, for example, who is in her 20s and has been diagnosed with SMA type II, had never met another person diagnosed with SMA until the age of 25 when she first attended a JTSMA conference. Up until this point, Evelyn’s knowledge of SMA was primarily derived from medical sources (e.g. consultations with clinicians, leaflets on SMA distributed at the hospital), as well as seeing other people who she described as ‘*looking like me*’ at the specialist SMA clinic she attended throughout her childhood. Evelyn described her first experience at a JTSMA conference as a ‘*real eye opener*’ which had a substantial impact on her perception of SMA as a condition, and also on her feelings about having a child affected by SMA herself:

I'd always said that I knew what I'd do if I were to have a child of my own with SMA, that it was fine if it was a type II or III, but that I wouldn't have a type I baby as I think it's cruel really to have them when they suffer and die so young, you know, if you can stop it, if you have a choice about it. I know a lot of parents don't get a choice in that...[It is not possible to diagnose the type of SMA prenatally] But yeah, that was before I went to conference for the first time, and it threw all that out of the window [laughs] because I met an eight year old with type I, and they're doing really well and the parents love them to bits and...I don't know, it's made me reconsider it all really um because now that I know *that*, I would feel guilty for the rest of my life...if you got rid of a type I, and then you saw that...I wouldn't want to put that that weight on my shoulders, knowing that it could have been ok, they might have lived and I'd just got rid of them. So now...I just don't know what to think about it really, it's more complicated now isn't it [laughs]?

The work of Lyons (2000) and Etchegary et al. (2008) have highlighted the way in which experiences of health, illness and disability presented in the media, or the lives of unknown others can contribute to an individual's understandings and feelings of 'knowing' about a condition. Whilst Etchegary et al. (2008) have contrasted this form of knowing with more 'vivid' knowledge obtained through the lives of friends and family, Evelyn's

first meeting with a family affected by type I SMA nevertheless had a significant impact on her feelings about the types of SMA, and the experiences of suffering therein. Seeing that a person diagnosed with SMA type I could survive past infancy introduced a new form of responsibility for Evelyn; she now had to weigh the doubt this knowledge brought to her previously steadfast evaluation of life with SMA type I against her desire to protect herself from the possibilities of regret and guilt. Her indirect experiential knowledge of another family's experiences with SMA, combined with her direct knowledge of SMA in her own life led her to re-evaluate her responsibilities in reproduction rendering them both uncertain and contradictory; both terminating or continuing with a pregnancy affected by SMA type I could cause imaginings of what '*could have been*' in light of her indirect knowledge of SMA type I, with the attendant guilt and regret if these imaginings appeared more favourable than reality.

This section has demonstrated the way in which experiential knowledge, whether obtained through having SMA oneself, or through less direct means, for example, living with, or meeting someone with SMA, rather than clarifying reproductive decisions, could actually introduce uncertainties and tensions to reproductive decisions. For both Claire and Evelyn, experiential knowledge of SMA both enlightened them to the possibilities as to what life with SMA could be like, but also created confusion as to which responsibilities they should prioritise when making reproductive decisions. Indeed, whether SMA was perceived as entailing suffering and whether or not there was a close family member affected by SMA are examples of factors

that not only shaped participants' experiential knowledge, but also impacted on the way in which participants conceptualised their felt sense of obligation.

Whilst thus far, experiential knowledge has been described as a stable resource, created, re-created and passed between individuals living closely alongside SMA, offering possibilities for the creation, subversion and transformation of felt responsibilities in medically defined 'risky' reproductive decision making, not all participants conceptualised this knowledge in this way, or indeed, as being available to the majority of people living with SMA. Some participants provided tightly defined accounts of experiential knowledge, regarding it as the property of a small minority of those diagnosed with SMA and not transferrable within and between social groups, whereas other participants provided far less lucid accounts of their lives with SMA. For these participants, the experience of SMA could not be described or owned in any meaningful way due to its unstable and ever-changing character. In contrast to the *bounded* knowledge presented by participants so far, these participants regarded their experiences of SMA as far more fluid and uncertain and thus an unreliable basis for knowledge claims. These accounts of contestation around experiential knowledge, and their interaction with responsibilities in reproduction will now be examined.

Ownership and Privilege

Whilst experiential knowledge of SMA was treated by some participants as a taken-for-granted product of their experience of the world, for others, it was described as a privileged form of insight available to only a minority of individuals. This contestation around ownership of, and rights to claim,

experiential knowledge of SMA is important as it strikes at the very heart of what *constitutes* experiential knowledge of SMA. Indeed, as has been discussed previously, participants came to know SMA from a variety of different perspectives; as the parents, carers, spouses, siblings, partners or children of individuals with SMA, or as people who have been diagnosed with SMA themselves. The question as to which of these individuals had access to the appropriate experiences, identities and subjectivities to give them a validated standpoint from which to lay claims to ‘knowing’ SMA, however, was highly contested. Given that experiential knowledge of SMA was regarded as a crucial resource in reproductive decision making, and an index by which to organise responsibilities, the way in which individuals lay claim to, or invalidated claims to, experiential knowledge is another important centre of negotiation and tension in the reproductive decisions of families affected by SMA.

As the development of genetic and screening technologies has brought with it the possibility of the prenatal detection of a range of conditions, so has concern risen amongst disability rights supporters as to the sources of information by which prospective parents come to know about the conditions their foetus is being screened/tested for. In particular, concern has been raised about the communication of overly medicalised portrayals of conditions to prospective parents which simply emphasise the medical complications associated with it (as found by Williams et al. (2002) in relation to Down’s syndrome) as opposed to the possibilities for a fulfilling life (in spite of impairment) that are often reported by the families and individuals currently

living with that condition (Asch, 1999; Ferguson et al., 2000). By elevating the status of the knowledge of disabled people in defining the reality of life with impairment, and rendering them the 'best experts' (Peterson, 2006:32) in their own conditions, disability rights supporters have attempted to counter balance some of the medicalised assumptions deemed to inform many people's attitudes to disability, and subsequently, their approach to selective termination decisions (Asch, 2000).

For some individuals with SMA, their experiential knowledge of SMA was viewed in these political terms and perceived as a resource for would-be parents considering the possibility of having a child with SMA, as Hannah, diagnosed with SMA type I commented:

I definitely think there should be a space for us in this debate about selective termination, I mean I basically think it's genocide for the contemporary world. The medical view of disability is so narrow and they only see like the worst angle of it. Even in families with SMA...you know, how can these parents know what it's like *really*? Just because they've got a kid with SMA doesn't mean they know what it's like or put them in a position to make that sort of judgement on whether or not the kid should live. Only people with SMA really can say what it's like, so the decisions to terminate are based on ignorance rather than fact.

Hannah's concerns about the uses of selective termination have been echoed elsewhere in the literature (e.g. Rock, 1996) and present a view of experiential knowledge not simply as a valuable form of insight into life with SMA, but instead as *the* only authentic version of what living with SMA means. As presented in Chapter 1, issues around the status of different sorts of knowledge have been debated extensively within the feminist epistemological literature (Hartstock, 1983; Hekman, 1997; Ramazanoglu and Holland, 1999; Code, 1991). Feminist standpoint theorists, following the work of Hartstock in the early 1980s, have argued that the unique standpoint of women offers a justification for the knowledge claims of feminist research (Hartstock, 1983). From the starting point that all knowledge is 'situated', standpoint theorists have argued that women, through their subjugated position, can offer unique insights into the experiences of oppression, they offer a 'true' account of the internal workings of patriarchy (Hartstock, 1983). Whilst feminist standpoint theory has been heavily criticised, particularly for its inability to account for difference without attracting the criticism of relativism (Hekman, 1997; Ramazanoglu and Holland, 1999), as well as its inability to disentangle truth claims from their discursive construction, it nevertheless appears to be echoed in the accounts of disabled people who claim epistemic privilege. This claim can be understood in terms of feminist theorising as an attempt to create a counter-hegemonic discourse to the disablism inherent in western societies. Indeed, despite the criticisms levelled at the valuing of some voices over others, Hartstock (1997) maintains that there are ethical, political and social justifications for doing so; she argues

that the privileging of certain knowledges may offer ‘possibilities for envisioning more just social relations’ (p. 373) which should be taken seriously as a basis for discounting relativist accusations. The possibility of altering the value attributed to lives affected by impairment has certainly been suggested as a justification for privileging the voices of people with disabilities in the debates about prenatal testing and selective termination.

Tom Shakespeare, a prominent disability rights supporter and writer on the ethics of prenatal testing, has nevertheless occupied a more graduated position, regarding his experiences (as a person with a genetic impairment) as being *relevant* to, but not necessarily offering a *better* account of, life with disability as presented to would-be parents facing the possibility of selective termination (Shakespeare, 2005). In terms of families affected by SMA considering reproductive decisions, the ownership of, and privilege accorded to, experiential knowledge of SMA appeared to carry moral weight, and was a point of contestation for many participants. Whilst for Hannah, claiming a privileged perspective on SMA was part of her broader political commitment, such claims could also be used strategically within families to justify particular standpoints and to discredit others.

Luke is 40 years old and lives and works in London for a consultancy company. His sister Gill, with whom he lived whilst he was growing up, was diagnosed with SMA type II in childhood. Luke has two children, and underwent carrier testing before having them. When found to be a carrier, Luke’s wife was also tested and was found not to be a carrier. Luke described

his feelings about undergoing testing before having children in the following way:

I have to say that I wasn't overly concerned about getting the testing done, but my wife thought it might be a good idea to get it done. I wouldn't say I was worried about it though, because you know I've had real experience of what it's like, living with Gill and knowing what SMA is like. But I've certainly seen friends of mine and I guess I might have been in the same sort of state of mind...that unless you know what the impact is on your life, and what your child's life will be like, you know, you worry. There's a sense that your life quality changes, you know as a parent, and that it's going to be an incredibly difficult life for you and your child, and I don't agree because I've got first-hand experience of it, so I guess I've got that unique perspective of it.

The medico-scientific literature in particular has pointed to the significant influence having a relative affected by a particular condition has on feelings about one's own genetic risk, reproductive choices and sense of responsibility (Kay and Kingston, 2002; Wertz, 1992; Elkins et al., 1986; Beeson and Globus, 1985). Whilst for some conditions, this experiential knowledge meant that relatives were less afraid of the possibilities of having an affected child themselves (Wertz, 1992; Elkins et al., 1986), for others, knowing the 'intimate detail about the realities of care' (Beeson and Globus,

1985: 110) associated with the condition created more unease in relatives of affected individuals about such a possibility (Kay and Kingston, 2002; Beeson and Globus, 1985). Gow's (2000) study of women with conditions for which prenatal tests or screening are available (which included interviews with women affected CF), highlighted the women's feelings about how their siblings would perceive their condition and also their feelings about having a child affected by CF themselves (p. 262). As unaffected siblings do not have the same constraints on their reproductive decisions imposed by a chronic illness or disability as individuals diagnosed with SMA and parents of affected children, the experiential knowledge of siblings may offer particular insights into experiential knowledge and its role in reproductive decision making (Bryant et al., 2005). In total seven able-bodied siblings of individuals with SMA took part in the study, all of whom emphasised the positive view of SMA they had developed having grown up alongside their sibling, and also the resourcefulness of their parents in accommodating the family's needs. A further six individuals with SMA described their anticipation that their siblings (who had not taken part in the study) would share a similar view to this, whilst three commented that they were uncertain about their sibling's feelings. Whilst five of the able-bodied siblings who took part specifically mentioned experiences of guilt akin to 'survivor guilt', or feelings of being overshadowed by their sibling's high support needs whilst growing up, all were keen to state the potential of people diagnosed with SMA to achieve and thrive in spite of disability- a finding reflected in the literature in relation to other conditions (e.g. Scelles, 1997; Van Riper, 2003).

Luke's feelings about growing up alongside SMA and his confidence in the insight this perspective offered him, therefore, are representative of this group of siblings.

Despite the 'empathetic knowledge' (Abel and Browner, 1998) of SMA accrued by this group of siblings, however, the accounts of individuals diagnosed with SMA suggested different criteria by which a person can 'know' SMA, and thus who may be qualified to ground their reproductive decisions in this knowledge. Indeed, for many participants diagnosed with SMA, siblings' knowledge of what it's like to live with the condition was partial and incomplete, although it was acknowledged that they may be able to reflect on the experiences of caring and living in a disabled family. Gill, Luke's sister and diagnosed with type II SMA, for example, disqualified her brother's knowledge claims when she talked about her life with SMA:

I definitely feel that no one can ever know what SMA is really like until they live with it themselves, you know, they've got it. Because other people, outsiders, no matter how close they are to you, they could be in your family even, they can't put themselves in your position. And my brothers, they don't really know how much help I need, because while I was at home with them growing up I was quite capable of doing quite a bit for myself, it's only since I've moved out and lived on my own that now I need PAs [personal assistants] all the time... They [brothers] know I need help, but I don't think they know how much help I need. So it's

really just between me and my PAs. Not even my friends...it's not something you...talk about...everything that's physically hard is usually done in the house, you know all the personal help and looking after the house and everything, no body sees that apart from the person who's helping me. And even then, how do they know how it feels to you? So yes it's so individual in that respect, you can't know unless you've lived that life.

Whilst Abel and Browner (1998) distinguished between 'embodied' and 'empathic' knowledge in the way in which people gain experiential knowledge, for Gill, experiential knowledge was a bounded form of knowledge; it could only be accessed by those living with the condition themselves, and was not transferrable to 'outsiders' by virtue of them living with her. There is a strong contrast between Luke, who described his experiences with SMA as '*first-hand*', and Gill who referred to all people as '*outsiders*', outside of her body, perspective and standpoint and thus ineligible to evaluate her life. Indeed, as her care needs changed over time, and she moved out of the parental home, her experiences of SMA began to change; she took on PAs and experienced independent living, and thus her brothers' knowledge of what life with SMA was like became outdated. For Gill, being able to discredit Luke's perception of her life with SMA was particularly important in relation to his decisions about undergoing carrier testing, as being able to claim authoritative knowledge of SMA could serve as a bastion against the emotional harm associated with the thought that Luke

might wish to prevent a life affected by SMA. As Gill commented when recalling her brother's decision to undergo carrier testing:

I actually gave Luke all the information [about the genetics of SMA] before he and his wife started trying for a baby, so they could know about their options and choices and I was glad they had it, but once I'd given the information, I realised that information is really powerful and that then they had a choice and I started to think, if I had known that she [Luke's wife] was pregnant and she had a little baby with SMA, and they got rid of it, I mean that would have been shattering. Because that tells you a lot about what they think about my life. And actually my life, from where I'm seeing it, is probably a hell of a lot better than most people's...but they can't see it from where I'm seeing it, only from where they are, which is probably quite different, so their decision wouldn't really be based on reality, if you know what I mean.

Being able to police the boundaries of who could claim experiential knowledge was crucial, not just for individuals with SMA considering the reproductive decisions of others, but also prospective parents (with and without SMA), as it enabled participants to discredit the perspectives of others who may criticise their decisions or viewpoints or protect against the emotional harm inflicted by the reproductive decisions of others.

Importantly, however, claims to ownership of experiential knowledge additionally served to strengthen identification with others who are deemed legitimate holders of this knowledge. For those people who have been diagnosed with SMA themselves, a common identity and sense of empathic understanding with other people with SMA was emphasised, as Hannah, diagnosed with type I SMA commented:

I am actually very interested in other people that have SMA because there is a commonality between us, you know, when you meet someone else with SMA it's like meeting a long lost member of your family. We're all linked in that way and there's that...[pause]familiarity in how we do things, how we move, how we look, the things we can do and the things we can't and the things we face in life that are unique to us I guess, that those people who don't have SMA don't really understand.

The term 'biosociality' (Rabinow, 1992) is one that has emerged in the fields of sociology and anthropology to account for the ways in which groups form around common biological or genetic identities (Hacking, 2006); according to Rabinow (1992), the new genetics are no longer grounded in the natural world, but instead are imbued with cultural understandings of relatedness. For writers such as Schneider (1980), biology can be understood as a cultural system whereby kinship and biogenetic relatedness are conceptualised as one and the same thing, so that the uncovering of new 'biological facts' triggers the reconfiguration of kinship. Thus, kinship ties and bonds can be

scientifically ‘discovered’ where there were none before (Franklin, 2001: 306). Rapp et al. (2001) have argued that, in line with Schneider’s thinking, genomic practices have increasingly come to dominate the way in which we scientifically ‘discover’ kin and additionally account for ‘atypical’ members of kin groupings (Rapp et al., 2001: 384). For Rapp et al. (2001), the emergence of lay organisations akin to the JTSMA are founded on the basis of a shared genetic identity which serves to normalise, empower and construct ‘imagined kindred’ for affected families and individuals (Rapp et al., 2001: 393). Through a study of the meanings of kinship in relation to lay organisations, Little People of America (LPA), National Marfan Foundation (NMF) and a proliferation of Down’s Syndrome support groups, Rapp et al. (2001) have demonstrated the way in which collective identities are forged on ‘genealogies of affliction’ (p. 386), which may be claimed through readings of the body. Indeed, for Hannah, it is through reading the bodies of other people affected by SMA that she recognises herself; the familiarity of appearance and abilities that signal that she has met someone who belongs to her ‘family’, linked by a shared physical and genetic identity. Rapp et al. (2001) have suggested that the forming of these kinship groups are important in the undertaking of kinship work, and the specialised bodies of knowledge that emerge from them, namely the experiential and medical knowledge obtained by living intimately with a condition. Whilst the knowledge production and collective identities to which Rapp et al.’s (2001) research points, however, may be possessed by individuals diagnosed with a condition, as well as their families (indeed it is highlighted within Rapp et al.’s study

that a large portion of the aforementioned knowledge is accumulated and disseminated by and for the mothers of affected children), Hannah's description of her close identification exclusively with others with SMA suggests a more tightly defined version of 'biosociality', whereby identification may be based on very specific readings of both the body and assumptions about genetic make up. Whilst carriers of SMA may have certain genotype similarities with those with SMA, for Hannah, the specific features of those with SMA marked them out as a group and defined their experiences as inaccessible to others. The importance of Hannah's assertion in terms of the ownership of experiential knowledge lies in its support of a conceptualisation of experiential knowledge as a discreet form of knowledge, only accessible to, and shared exclusively among, those united by a common genetic and physical identity.

This section has suggested that issues around ownership and privilege are of central importance to an analysis of experiential knowledge. Etchegary et al. (2008) have employed a visual metaphor to describe the different intensities of experiential knowledge that are mobilised around prenatal screening decisions, contrasting the more 'vivid' accounts of women's experiences (those which are close to them) with the more 'distant' forms of knowledge that women use, such as those obtained through the media or the narratives or contact with unknown others. Whilst describing these more 'distant' forms of knowledge as 'less personal' (p. 122) than vivid accounts, Etchegary et al. (2008) do not extensively consider the relative value of these different forms of experiential knowledge. My research suggests that not only

can contrasting ways of knowing SMA be differentiated, but also that these bodies of knowledge have a *hierarchical value*; not all sources of knowledge are considered equal, and there was a tendency to view embodied experiences of SMA as more 'accurate' accounts of life with SMA, with various degrees of removal from this experience. Those who had lived with someone with SMA were considered to possess more expertise in what life with SMA is like than more distant family members and friends.

This hierarchy serves important functions in the negotiation of reproductive responsibilities for families affected by SMA. By offering participants a means by which to claim authority on a resource deemed so central to reproductive decisions by these families, individuals were able to strategically discredit or subvert the knowledge claims of others in order to justify their own reproductive choices, or to protect their own feelings of self-worth and value. These strategies additionally served to draw closer together those individuals at different points in the hierarchy; laying claim to universal experiences was a means by which to forge collective identities, which in turn reinforced the foundations of the hierarchy. The hierarchy of experiential knowledge was not necessarily experienced as oppressive, but instead can be viewed as a creative strategy by which participants could negotiate the complexities of reproductive decision making whilst simultaneously safeguarding their own emotional and social well-being.

Experiential knowledge has thus far been treated as a form of knowledge to which different meanings may be attached, and around which negotiations of status, responsibility and ownership circulate. However, some

participants' accounts of experiential knowledge were far more fluid than this analysis suggests; for these participants, experience was conceptualised as unstable, continually evolving and changing, and subject to revision at any point. Moreover, experiential knowledge could also be rendered obsolete by the valuing of particular responsibilities or through a personalisation of this knowledge; by rendering experiential knowledge as entirely individual and context-dependent, the relevance of experiential knowledge to reproductive decisions is called into question. These considerations will now be addressed.

Experiential knowledge as Unstable/Obsolete

Experiencing SMA, as has been discussed in Chapter 5, is a complex, multi-faceted and ever-changing phenomenon. The different ways in which SMA is experienced may alter over time and context, and personal responses to these changes may additionally be dependent on social, psychological and emotional factors. Despite an emphasis thus far on the way in which these experiences become condensed, summarised and attributed meanings in the context of reproductive decision making in order to negotiate the complex tensions and issues surrounding genetic risk, not all participants felt satisfied with this approach, and the strategic processing of their experiential knowledge associated with it. Some highlighted the personal nature of their experiences and the impossibilities of generalisation, whilst others presented their experiential knowledge as obsolete in order to give weight to all-encompassing responsibilities, to preservation of life and to God.

The Instability of Experiential Knowledge

Chapter 5 has demonstrated the ‘slipperiness’ of the experiences of SMA described by participants in this study. Despite attempts by medical professionals to capture and define particular ‘types’ of SMA, according to physiological characteristics, the way in which lives with SMA are actually experienced defies such classification, and instead appears characterised by movement between different experiential categories. The range of experiences associated with SMA, and the emphasis placed by participants on the social context of these experiences as well as the personal strategies developed by individuals to counter-balance negative consequences of SMA render the use of experiential knowledge in reproductive decision making highly complex; it suggests that the unique nature of each experience with SMA negates the possibility of drawing generalisations.

Eileen is in her 40s, was diagnosed with SMA type III in childhood and has gone on to marry and has two (now adult) children. Eileen has a brother who is also diagnosed with SMA (type II) which was confirmed shortly after her own diagnosis, having initially been diagnosed as having DMD. Eileen’s brother has never been able to walk. However, she is able to walk with a walking aid, but describes herself as tiring quickly. Eileen’s adult children do not have symptoms of SMA, but are presumed to be carriers of the condition by the medical profession and the family, given that all children of people diagnosed with SMA inherit one copy of the damaged SMN1 gene, making them carriers of SMA. Eileen’s account of her life with SMA, the strategies she employed to make her reproductive decisions in her twenties,

and her current reflections on these decisions demonstrates the fluid nature of experiential knowledge, and its unreliability in the context of reproductive decision making:

When they were born, I mean my children are 21 and 23 now, there was no testing for it [SMA] at all so really it was pot luck, we didn't know what we were going to do if...but we were quite prepared to take that chance at that time because SMA didn't seem that scary to us. I was 20 and I was still quite active, much more so than now, my brother was much stronger then, although he was still in a chair, so you don't think about what happens 20 years down the line, as you age and you go downhill. You just see how you are at that particular time when you make your view of what SMA is like and even with that you can probably never know enough because with any condition, every person's different, the condition changes, so you can only get a brief idea of what's going to happen and I mean 20 years down the line it could go...[pause]You can have a brief idea about what's going to happen, but you don't know exactly and even the doctors can't tell you that, because with SMA particularly there's so much individual variation.

Whilst Eileen described her experiential knowledge of SMA accumulated in her twenties as reassuring, on the basis that she felt her symptoms to be mild- *'I guess I just thought that I wasn't that bothered by it, it could be lived with'-*

the context of growing older with SMA and experiencing deterioration altered Eileen's view of the stability and reliability of past experience as a resource with which to imagine the future. For Eileen, her changing perception of her SMA affected the value she attributed to experiential knowledge in the context of reproductive decision making. Whilst firstly describing it as an important factor in her decisions to have her children in her early 20s, now in her 40s, Eileen's perceptions of SMA have evolved. Having experienced a decline in her abilities over time, and witnessing the same with her brother, Eileen now questions the reliability of experiential knowledge in guiding reproductive decisions and shaping a 'view' of what SMA is like. In the same way that the medical profession cannot accurately predict an individual's future experiences with the condition, so one's own sense of what SMA is like may prove to be unstable; continually expanding to incorporate new experiences and circumstances, with certainty only ever being provisional.

This change in the meaning attributed to experiential knowledge of SMA, however, was not always based on embodied experiences, and could instead be based on empathetic knowledge or expansion of information, as Nathan, whose sister has been diagnosed with SMA type II commented:

I must admit, my perspective on SMA did start to change when I had grown up and started to learn more about it. I suppose I just thought of SMA meaning that you can't walk, you're weak, but when I found out that chest infections could like...kill my sister, they could be fatal, I think I started to see it differently. I hadn't realised when she'd been

ill when we were children that that could have been so serious. So yeah I was quite shocked by that part of it, it made me rethink things a bit.

As Eileen and Nathan's accounts highlight, the negotiation of parental and reproductive responsibility may be approached differently when experiential knowledge is discounted as a reliable indicator of future lives with SMA.

Eileen's reflections on the nature of her experiences with SMA, however, suggest an additional influence on the interpretation of experiential knowledge as a reliable or unreliable resource. Her reproductive decisions may well have been presented differently had her children been born with SMA. Reproductive status, therefore, as well as the perceived status of experiential knowledge, could alter the way in which knowledge of SMA and reproductive responsibilities were experienced and recounted. Perceived accountability to ideas about parental responsibility restrict the number of possible readings of experiential knowledge retrospectively, as either a stable or unstable resource, as a strategy to justify reproductive decisions already made as well as those not yet taken.

The Limits of Experiential Knowledge

In addition to the continual processes of re-negotiation and re-naming of experiential knowledge over time and context, Eileen's account additionally touches on another issue in the uses of experiential knowledge in reproductive decision making. By referring to the individual variation in experiences of SMA, Eileen highlighted one of the limits of experiential knowledge

acknowledged in the literature in relation to the rise of the so-called ‘expert patient’, that is, its particularity (Prior, 2003). Whilst SMA may be a label assigned to a large group of individuals, a disparate and broad range of experiences are nevertheless described by those living with SMA. Indeed, as well as physiological differences between individuals (which account for some of the variation in these experiences), peoples’ lives with SMA are also intersected by social, economic and political circumstances which affect the way in which they live with SMA. These factors as well as the strategies individuals use to manage SMA, as has been discussed in Chapter 5, may account for these differences in experience of SMA as much as the medicalised taxonomy system currently used to make sense of them.

Lily, who is in her late twenties, has been diagnosed with SMA type II and recently had her first child commented about her views of the classification of her own experience of SMA:

I always thought with type II I was safe...you know how nasty type I can be, so I always thought you know ‘I’m out of the water’ sort of thing, but it wasn’t until I went to the [JT SMA] conference for the first time and I saw the type IIs there and I started to see the amount of variation...a lot of them are very twisted and have respiratory problems, and when my mum saw them as well we could just not believe that they’ve got what I’ve got. So whilst I’m classed as type II, that’s only ever been just a label to me, I don’t really

think that it's quite the same as what the others have got. I'm
a very good type II.

Prior (2003), in his discussion around the status of experiential knowledge accumulated by patients vis-à-vis medical knowledge has suggested that experience, as a basis for knowledge is ‘...invariably limited and idiosyncratic. It generates knowledge about one instance, the one case, the single ‘candidate’” (Prior, 2003: 53). Indeed, Lily’s experiences with SMA were contained by the circumstances of her own life and her embodiment; she had intimate knowledge of what SMA meant for *her*, but these experiences did not necessarily translate to the lives of others. Her reflections on meeting others with SMA for the first time in her twenties reveal the distance she perceived between her own experiences of SMA and those who shared her diagnosis. For Fae, diagnosed with SMA type II in childhood, this lack of similarity and the individualised nature of SMA meant that experiential knowledge was of limited use in reproductive decision making. When considering the relevance of her experiences to the decision someone might make about having a child with type II SMA she commented:

...this is my thing with disability, is that just because I’m
disabled and just because the person round the corner is
disabled, doesn’t mean that we’ve got anything in common
...and I read the Jennifer Trust stuff about people who have
ventilation and things and I’ve been one of the lucky ones,
I’ve never needed that...so they could have a child, type II
SMA who could be like that. Equally, they could be ten

times stronger than I am. Nobody can say. You know SMA is like an umbrella and there is so much under that, but a lot of people aren't aware of it. I know that the last time that I went to the conference, there was a photograph taken and people looked at it afterwards and said 'why do you look different to the rest of them?' [other people diagnosed with SMA] and I said 'I don't know, do I?' and it is true, we don't look the same. You can have the same condition, but what they go through can be nothing like what you go through... And a person who was pregnant with an SMA baby could look at someone with type II and think 'ok, I could cope with that' but that doesn't necessarily mean it's going to go that way. You just don't know. Equally you could have someone who has just the same level of SMA me but who just can't handle it, you know, people are different.

Whilst Fae's knowledge of SMA had been expanded by her contact with other people with SMA, she nevertheless recognised the difficulties in assuming a universality of experience based on a genetic identity, which may emerge from 'biosociality' (Rapp et al., 2001). Indeed, participants cited a range of factors which they deemed to have been particularly central in their ability to manage their lives with SMA which minimised any negative impact of the condition including the positive attitude and upbringing of parents, the maintenance of networks of social support, the receipt of adequate health and social care as well as acceptance into the worlds of education, employment

and parenthood without discrimination. These findings are similar to the coping strategies of people with SMA outlined in Lamb and Peden's (2008) study. These relative factors are extremely important in that, like differences with muscle tone and strength, they have the capacity to substantially influence the way in which SMA is experienced in individuals' lives. They are also factors, as Lily points to, which are highly personal and variable, calling into question the relevance of the experiential knowledge accumulated by individuals with SMA themselves to reproductive decision making.

Conclusions

This chapter has presented the intersection of experiential knowledge with various forms of reproductive responsibility in the context of families affected by SMA. Through the presentation of the accounts of individuals living with, and alongside SMA in various capacities, it has been argued that experiential knowledge can be both a strategic tool and a complicating factor in the negotiation and presentation of reproductive responsibilities in the decisions faced by families affected by SMA. The experience of SMA was reflected on and attributed meaning in different ways depending on nature of the experience as well as the point in reproductive decision making at which participants were being interviewed. As Nelson and Nelson (1995) have argued, decision making is a factor in identity construction in that the decisions we make contribute to the way in which we are perceived and evaluated by others. Maintaining an identity as a responsible decision maker in the context of reproduction, where powerful discourses around parenthood

(and more specifically, motherhood), disability and inheritance both exist and conflict, therefore, is an arena in which experiential knowledge can be strategically mobilised to justify and support complex reproductive decisions, as well as to deflect the felt and articulated criticisms of others. This chapter has demonstrated that through defining experiential knowledge as ‘warning’ or ‘reassurance’ and also as a form of expert knowledge inaccessible to others, participants are able to negotiate the tensions and conflicts between these discourses whilst simultaneously maintaining their identities as responsible (prospective) parents, irrespective of what decision is taken. However, experiential knowledge is not a straightforwardly useful resource and for many participants considering reproduction, such detailed and intimate knowledge of the realities of life with SMA could both heighten and personalise the tensions in reproductive decisions, contributing to painful and impossible dilemmas, rather than offering a safeguard against future regret or accusations of irresponsibility which had informed its use in the reproductive decisions of others. For other participants, however, the *destabilisation* of experiential knowledge as a reliable source of information on which to ground such decisions was the most accessible means of negotiating these tensions and ensuring the maintenance of participants’ identities. These various ways of using and abandoning experiential knowledge in strategic ways suggest that even in instances where its applicability and relevance were questioned, participants’ lives and their entanglement with the experience of SMA were always factored in reproductive decision making; they could never simply be ignored.

Chapter 7

Discussion

In this discussion chapter I will draw together the analysis of the previous chapters and discuss the implications of the findings for wider debates and practices. The findings of this research have implications firstly for debates around the nature and status of ‘experiential knowledge’ in a variety of contexts, and secondly to debates within the disability rights literature around the role of experiential knowledge in relation to prenatal testing and screening decision making. In this chapter I will highlight the contribution of my research to these fields, as well as the points of intersection between them. Furthermore, I will consider the practice and policy implications of my findings, particularly around screening, testing and genetic counselling practices. In order to achieve these aims, firstly I will restate my research questions (as set out in Chapter 2), before moving on to present a summary of the key points of the research which address these questions. Finally, I will present an analysis of these findings within the context of the pre-existing literature in order to highlight and demonstrate the contribution of my research to it, as well as suggesting areas for further and future research.

The Research Questions

My central research question is:

In what way does experiential knowledge of SMA inform conceptualisations of genetic risk and reproductive decision making in families medically defined as ‘at risk’ of transmitting SMA to future generations?

I have also considered the following secondary research questions:

1. What are the main concerns and challenges faced by families and individuals affected by a diagnosis of SMA?
2. Is SMA perceived differently by those with a diagnosis of SMA, in contrast to the perceptions of their family members?
3. How do families and individuals affected by SMA relate to medical definitions of SMA?
4. To what extent are the concerns of disability rights activists about prenatal testing and selective termination reflected in the views and concerns of families affected by SMA?
5. How do families and individuals interpret the value of medical knowledge vis-à-vis their experiential knowledge in reproductive decision making?
6. How are notions of reproductive and relational responsibility negotiated in the context of experiential and medical knowledge of SMA in reproductive decision making?

Summary of the Key Findings

****Medical Knowledge of SMA is Uncertain and Ambiguous***

Despite recent developments in clinical research which have been posited as offering researchers new opportunities to understand the mechanisms by which SMA occurs (Yu et al., 2008), SMA is still a relatively poorly understood condition. Indeed, whilst research into the condition has gathered

pace in recent years, and has become systematically organised through the development of organisations such as Treat-NMD (a registry of all those affected by SMA to facilitate trials) and the establishment of universal outcome measures for SMA clinical trials, the outcomes of such developments are yet to offer any benefit to those living with SMA. Stem cell researchers have recently highlighted the possibility of developing treatments which will eventually replace the inadequately functioning motor neurone cells deemed to produce SMA (Kerr et al., 2000; DiDonato, 2003; Corti et al., 2008). However, currently, for families and individuals affected by SMA, no effective cure or treatments are available to alleviate the symptoms of SMA, and for many who took part in this study, the concept of cure was no more than an abstract hope for the future; one that may, or may not, materialise. As writers such as Fleising (2001) have argued, this mismatch between expectations and delivery of genetic interventions can be understood as part of the ‘clinical genohype’ surrounding such genetic technologies.

Through an analysis of the emergence of SMA as a disease category, and the analysis of participants’ accounts of obtaining and making sense of a diagnosis of SMA, I have highlighted the instability of medical knowledge of SMA. Despite the authority attributed to diagnostic categories and the legitimating effect of such labels, the medical typology offered to classify those diagnosed with SMA can be experienced as alienating, irrelevant and confusing. Being unable to tally one’s experience of SMA with a diagnostic bracket caused feelings of displacement for some, whilst for others being given a prognosis of premature death which later proved to be inappropriate

had devastating consequences for the individual concerned and those around them. Whilst for many people affected by SMA, and also within the medical profession, it is becoming increasingly accepted that the medical typology for SMA is to be treated as a loose guide rather than a definitive classificatory system (Dubowitz, 1991), these diagnostic brackets nevertheless held considerable influence in terms of conceptualisations of SMA and the identities of those living with it. Medical knowledge thus influenced the way in which SMA was experienced by those diagnosed with it by carving out different forms of experience associated with it. The JTSMA's annual conferences continue to be organised around SMA's diagnostic types and individuals readily identify themselves as belonging to a particular 'type' of SMA and measure their abilities alongside others attributed this same diagnostic category: *'I'm quite a good type II'* (Georgia), *'I've noticed I'm a lot stronger than all the other type IIs'* (Lily), *'...even though [daughter's name]'s survived infancy, I still consider myself to be a mother of a type I'* (Trisha). The medical typologies of SMA, were, furthermore, used by participants as a shorthand to express the quality of life associated with the condition; type I, generally considered to be most the severe type was associated with the poorest quality of life, leading up to type III, with those so-diagnosed deemed to enjoy the best quality of life, as the symptoms are the least severe. However, following interviews with people diagnosed with types I, II and III SMA as well as its variant conditions, SBMA, SMARD and ADSMA, it became clear that these assumptions, embedded within the

medical typology of SMA, were a gross over-simplification of the lived reality of SMA.

These uncertainties around the medical typology of SMA become particularly problematic in the context of reproduction. It was clear, through an analysis of the interviews, that the medical typology of SMA held considerable sway in the way in which many participants conceptualised SMA as a condition, and thus approached their reproductive decision making, in spite of the acknowledged difficulties with its coherence. Participants did not speak of wishing to avoid SMA per se, but, more accurately, certain *types* of SMA, as a proxy for severity. Particular concern was expressed around the desire to avoid the birth of a child who was unlikely to survive infancy. However, the prospect of having a child who would be disabled, but nevertheless survive into adulthood, elicited a far broader range of viewpoints. These preferences were largely expressed in terms of the undesirability of a pregnancy affected by type I SMA, with varying degrees of concern associated with pregnancies affected by types II and III.

The desire to avoid particular forms of SMA that would lead to particular (presumed) experiences of life, however, is not matched by the current capacities of the medical technologies used for prenatal testing of SMA. Prospective parents and their doctors are not able to decipher the ‘type’ of SMA a foetus is affected by from a prenatal test, but rather only whether the foetus will develop into an infant displaying symptoms of SMA. The only available guide by which prospective parents may index the sort of life the prospective child could have is by reference to the type of SMA previous or

existing family member(s) were diagnosed with, as it is assumed that the recurrence of SMA within families groups will be of similar severity (personal correspondence with Jane Fenton-May, Associate Specialist in Medical Genetics, 2008). Despite this assumption, however, differences in severity of SMA within sibling groups have continued to be observed by both the medical profession (e.g. Dubowitz, 1991) and families living with SMA. In terms of reproductive decision making, therefore, the uncertainties around medical knowledge heightened many of the dilemmas and confusions experienced by prospective parents. Despite the rhetoric of ‘informed decision making’ that is emphasised by those working in the field of clinical genetics, prospective parents with SMA in their families approach reproduction with imperfect medical knowledge of what SMA might mean for future generations.

**** SMA is Associated with a Broad Range of Experiences: ‘Experiences of Disability’, ‘Embodied Experiences of Impairment and Disability’ and ‘Experiences of Illness, Death and Bereavement’.***

Whilst the medical typology of SMA carries with it an assumption that it is possible to index the sort of life an individual will experience within a particular ‘type’ of SMA, the accounts of those individuals living intimately with SMA revealed a far more complex picture of how life with SMA was experienced. The nature of each person’s SMA often defied medical classification, extending beyond the boundaries of the diagnosed type, in a far more chaotic manner than the medical classificatory system suggests. Some individuals diagnosed with SMA type II died earlier than medically predicted,

whilst those diagnosed with type I who were expected to die in infancy survived into childhood or adulthood, blurring the boundaries of, and throwing into disarray, the medical classifications of the condition. When analysing the accounts of the families and individuals living with SMA, it emerged that whilst the medical typology of SMA serves a useful heuristic function, a sorting device that can be used as a shorthand to reference SMA in its various guises, it was *particular types of experience rather than particular types of SMA* that were being described when participants conceptualised SMA. The reality of SMA was talked about in terms of different forms of experience that could not readily be contained within medical classifications, even as these classifications shaped conceptualisations of SMA.

In light of these different ways of describing experiences with SMA, in Chapter 5 I developed an ‘experiential typology’ of SMA and marked out the (collapsible) boundaries between the different ways in which individuals live with SMA. These different forms of experience are ‘experiences of disability’, ‘embodied experiences of impairment and disability’ and ‘experiences of illness, death and bereavement’.

1) I used the term ‘experiences of disability’ to describe those experiences of SMA that are shaped primarily by social and environmental factors. Whilst these experiences were typically explained by participants in terms of restriction and limitation on account of the inadequacies of social and environmental support, crucially, these experiences were also viewed as being amenable to manipulation. Indeed, adopting a ‘positive outlook’ or attitude,

together with the ability to ‘fight’ institutions/individuals to ensure adequate support and access were viewed as important strategies to overcome the difficulties associated with experiences of disability. ‘Experiences of disability’, furthermore, in line with social model of disability theorising, were viewed as *non sequiturs*; having SMA did not automatically disable a person and many participants, (particularly younger adults with SMA), spoke about witnessing the expansion of opportunities for their participation in a wider range of activities that had previously been inaccessible (e.g. various forms of sport, driving, job opportunities). This is not to say that participants did not often regard their experiences of disability as frustrating, isolating and discriminatory, but rather that these experiences were conceptualised as being, at least partially, negotiable. Experiences of disability were not regarded as primarily a product of having SMA; instead, they were externalised and regarded as evidence of the failings of society to cater for disabled people.

2) Through an analysis of the accounts of those affected by SMA, a separate experiential category, ‘embodied experiences of impairment and disability’ emerged. Key writers discussing the social model of disability have long wrestled with the theoretical distinctions between ‘*disability*’ and ‘*impairment*’ and, indeed, whether social model of disability theorists have any political interest, or obligation, to account for disabled people’s embodied experiences. I have placed embodied experiences of impairment and disability within a separate category on the basis that they were conceptualised in a different way to experiences of disability, although the two are intimately

connected. 'Embodied experiences of impairment' refers to those descriptions offered by those diagnosed with SMA of what it feels like to live in a body with weakened muscles, spinal curvature and, sometimes, compounding respiratory difficulties. Also within this category, I have included the embodied experiences of the family members of those diagnosed with SMA, and specifically those undertaking care work for their relative. Care work, as Meyer et al. (2007) have observed, is a thoroughly embodied activity, a form of 'bodywork' (Twigg, 2000), and this came across strongly in the accounts of carers, who experienced through their bodies, the physical exhaustion, stress and strain of '*being the muscles and strength*' (Rachel, mother of a child with SMA) of their relative affected by SMA. By highlighting these experiences, I have drawn attention to the conceptual differences between 'embodied experiences of impairment' and 'embodied experiences of disability'. This distinction has been highlighted in order to distinguish between experiencing the impaired body and the more *reactive* embodied experiences of disability: for example, emotional responses to experiences of discrimination. As Reeve (2002), as well as Paterson and Hughes (1999) have argued, disability can produce embodied experiences as well as social ones, and experiences which may not be accommodated by the social model of disability; oppression is not just in the social fabric, but in the 'flesh and bones' of disabled people (Paterson and Hughes, 1999: 606). Unlike 'experiences of disability', the negative aspects of embodied experiences of impairment and disability were conceptualised as requiring a personal, rather than social, solution. Living in a body that deteriorates over time, or coping

with the demands of an intense care regime, required individuals to summon considerable personal resources in order to prevent them from becoming overwhelmed or distressed. Participants thus elicited metaphors of ‘fighting’ and ‘battling’ to communicate the idea that these embodied consequences, where experienced negatively, had the potential to be (at least partially) reconciled, through personal resilience and stamina. However, the possibility of these experiences being potentially too difficult or overwhelming for an individual to overcome was also emphasised. Embodied experiences of impairment and disability, therefore, carried with them the potential of suffering and distress less amenable to social intervention.

3) The final area that participants described when narrating their experiences with SMA were ‘experiences of illness, death and bereavement’. The overlaps and critical distinctions between conceptualisations of ‘illness’ and ‘impairment’ have been the subject of much debate within and between the disciplines of medical sociology and disability studies (Thomas, 2007). The presence of ‘suffering’ has been suggested as a potential differentiating characteristic between the two concepts by De Wolfe (2002), with ‘illness’ constituting a term reserved only for those experiences marked by wholly negative states of embodiment. Whilst ‘impairment’ may be viewed as a neutral or positive characteristic, and indeed *is* perceived so by many people living with impairment, illness conversely, does not allow such room for the negotiation of meaning (De Wolfe, 2002). De Wolfe (2002) has argued that illness is a state of being that everyone would seek to avoid. Similarly, when

describing experiences of chest infections, spinal fusion surgery, pneumonia and the deaths of relatives from SMA together with the consequent bereavement of the family, the notion that certain experiences *invariably* involve physical and psychological suffering was used as a means to distinguish them from experiences of impairment and disability, where personal and social interventions may alleviate negative consequences to varying degrees. Whilst the point at which an embodied experience of impairment became an experience of illness appeared to be subjective, and variable from individual to individual, there nevertheless appeared to be a unanimous opinion that *particular experiences of SMA caused people to suffer*, and, further, that these experiences traversed medically defined ‘types’ of SMA. All people with SMA live with the constant possibility of a serious chest infection and pneumonia and this dimension of the condition, together with the experiences of death and bereavement in the family described by many, were regarded as universally negative experiences and ones largely immune to social or medical remedy.

By distinguishing between different forms of experience associated with SMA, I have demonstrated the way in which the different ‘types’ of SMA cannot be aligned with particular levels of severity or ‘quality of life’ in an unproblematic way. Whilst the different types of experience I have presented were attributed hierarchical values in the stories of those living with SMA in that experiences associated with suffering were perceived as wholly negative and experiences to be avoided, whereas those deemed to be primarily social experiences were more ambiguous, it was simultaneously recognised

that people affected by SMA could move between experiential categories, or exist in multiple experiential categories at any given point in time. Thus, by highlighting the intersecting experiential dimensions of SMA, and their relative susceptibility to personal, physical, or social manipulation I have highlighted the complexity of the lived reality of SMA which is not represented in medical typologies of the condition.

This complexity has important implications for reproductive decision making. Through demonstrating the dispersion of experiential categories across the medical typology of SMA, the link between types of SMA and quality of life, to which reproductive decisions are often anchored, becomes harder to sustain. The ability to predict and control the degree of suffering and distress a future child would experience with SMA was extremely important for prospective parents to determine where the boundaries of their reproductive responsibilities lay. Prospective parents were keen to protect their potential offspring from avoidable or intolerable suffering. However, as has been discussed, reference to the medically defined type of SMA was not always the most reliable basis for such predictions, and many participants had experienced the unreliability of medical knowledge through the disjuncture between the prognosis they were given and their actual lived experiences of SMA. It was in this context, the realisation that medical science has limited abilities to predict and control the nature and course of SMA in future generations, that experiential knowledge of SMA became particularly important for prospective parents. Whilst experiential knowledge in itself was, at least in part, shaped by and formed through medical knowledge, these two

forms of knowledge were often presented as juxtaposed in participants' accounts in order to justify particular perspectives in reproductive decision making. Prior experience with SMA, and the challenges and transformations these experiences entailed were thus crucial in reproductive negotiations. It determined the contours of what participants defined as an acceptable life, even as this experiential knowledge was, in itself, both limited and idiosyncratic; bound to the specificities of individuals' lives with SMA and the instability of constant revision.

**** Experiential and Medical Knowledge of SMA Mediate the Negotiation of Reproductive Decision Making for Families Affected by SMA.***

Responsibility is a key theme that has been used to explore the experience of gender, reproduction and also the uses of genetic information (e.g. Reed, 2009; Hallowell, 1999). Through interviewing families and individuals affected by SMA, it was apparent that norms of parental responsibility, disability and genetic responsibility coalesced, contradicted and informed one another at different points of reproductive decision making. Experiential knowledge of SMA, as described above, not only constituted the context of reproductive decision making, providing a basis from which to predict the likely suffering of future generations with SMA, but was also mobilised strategically to negotiate the tensions inherent in discourses of (genetic) responsibility and disability, whilst simultaneously maintaining participants' identities as moral social actors.

As previous research has demonstrated, couples and individuals rarely approach reproductive decision making as autonomous individuals (Roberts and Franklin, 2006), but instead, as Cox (2003: 262) has argued, as the ‘mothers and daughters, fathers and sons, sisters, brothers, aunts, uncles, cousins, spouses, life partners and friends’ of others; as social beings we exist ‘in and through our social and familial ties with others’ (Cox, 2003: 262) and with an obligation to maintain our moral identities in relation to these connected others. Kenen (1994), through the notion of ‘genetic responsibility’ has demonstrated the way in which genetic medicine has augmented these responsibilities by suggesting new facets of obligation in intimate relationships, but also obligations to wider society, through the ‘quality control’ of future generations (Rapp, 2000). These notions of responsibility take on particular meanings in the reproductive decision making of families where the notion of having a child with a serious condition is not based in abstract wonderings, but instead is grounded in a lived reality. As Cox (2003) has highlighted, for families affected by inheritable conditions, the ‘unfavourable outcome’ in reproductive decision making, the ‘worst case scenario’, is not a depersonalised notion of disability or disease, it is a member of the family. For such families, then, responsibilities and reproductive risks are highly personalised. However, relatively few studies have explored how such families conceptualise and negotiate the personal, moral, familial and political questions that the possibilities of genetic medicine highlight (Kelly, 2009).

Through my interviews with families and individuals affected by SMA, experiential knowledge emerged as a key mediator of the responsibilities associated with reproduction. Participants in this study approached reproductive decision making in the context of having lived with someone with SMA or having SMA themselves, through which they assessed their genetic risk and formulated their reproductive decisions. For some participants, experiential knowledge was mobilised strategically to justify particular reproductive decisions. By drawing on the suffering of previous or existing family members with SMA and defining this experience as a warning as to the certain suffering of future generations, participants were able to justify their decisions (anticipated or taken) to prevent further lives with SMA and exonerate themselves from anti-abortionist discourse which they perceived as situating their decisions as irresponsible.

Similar strategies were adopted by those participants who opted to avoid all use of genetic technologies designed to prevent the lives of future generations with SMA. However, in these instances, by drawing on (embodied) experiences of disability and impairment, participants presented SMA not as a condition that involved suffering, but as a disability that could be 'overcome', and around which a happy and fulfilling life could be established. By recourse to their experiential knowledge of SMA, these participants were able to circumvent the notions of parental responsibility that would position them as irresponsible for risking the birth of another child with SMA in the family. The use of experiential knowledge in this way had novel benefits: it was presented as securing epistemic privilege for those who laid

claim to it. That is, by defining experiential knowledge as a resource only accessible to those living with SMA, participants were able to discredit the views of those who would criticise their decisions and present their experiences as irresponsible. The claim to ownership of experiential knowledge additionally had other protective qualities; for many individuals diagnosed with SMA, steps taken by other family members to prevent the birth of another child with SMA could be extremely painful. Thus, by claiming a privileged insight into life with SMA which was inaccessible to their family members, and possessed exclusively by those who had been diagnosed with the condition themselves, they could dismiss such reproductive decisions as uninformed or misguided and thus create an emotional buffer against the existential anxieties they experienced on account of their relative's actions.

Whilst the mediation of reproductive decision making by participants' experiential knowledge of SMA allowed for a less fraught negotiation of the tensions inherent in reproductive responsibility for some participants, for others, this mediation introduced new complexities and anxieties. Whilst for the former group of participants, intimate knowledge of life with SMA presented new possibilities for the justification of perceived problematic reproductive decisions, for the latter, this knowledge conversely sealed off such routes of justification and thus became experienced as burdensome. For such participants, feeling unable to cope with the support needs of another child with SMA but simultaneously witnessing their existing child thrive in spite of their SMA meant that it became harder to justify a decision to prevent

the recurrence of SMA in terms of protecting the best interests of the child, a justification which is frequently referenced by prospective parents in such circumstances (Kaplan, 1999). In such instances, prospective parents felt trapped by their knowledge of SMA and their belief that to prevent another child with SMA would be tantamount to the rejection of their existing child, a dilemma not expressed by those prospective parents who had more negative experiences with SMA such as acute illness and death. Indeed, the difficulties of prospective mothers, in particular, in articulating their own emotional and physical needs in relation to these decisions points to the uneven way in which the responsibilities of reproductive decisions were experienced. As Reed (2009) and Dragonas (2001) have highlighted, women are more heavily implicated in reproductive decision making than men, are held more accountable for pregnancy outcomes and bear the brunt of work for caring for disabled infants (Kelly, 2009). In this way, the experience of genetic responsibility is necessarily gendered (Hallowell, 1999; Reed, 2009), which constrained the way in which women made, and accounted for their reproductive decisions in relation to genetic risk.

Finally, for another group of participants, experiential knowledge introduced a further layer of complexity to reproductive decision making; whilst on the one hand it promised a window through which to glimpse future lives affected by SMA, the fluidity of its very nature simultaneously rendered it an unstable basis for knowledge claims and projections. The high degree of individual variation in experience of SMA and its constant evolution over time and context meant that some participants felt apprehensive about using

their experiences as a 'yardstick' by which to measure the anticipated quality of life of imagined others. This perspective was reinforced by the existence of different experiences with SMA between family members, within diagnostic categories, and indeed within the life of a person diagnosed with SMA. On the one hand experiential knowledge appeared to be useful resource, offering insight and familiarity; on the other however, it was also highly uncertain and unstable. The limited and idiosyncratic nature of the knowledge as well as its continual revision and reframing (Abel and Browner, 1998), meant that any decisions grounded in such knowledge were invariably contingent.

Experiential knowledge mediated reproductive decision making in more or less helpful ways for families and individuals affected by SMA. It was mobilised strategically to justify and support very different reproductive decisions, and was placed on an epistemic pedestal to discredit the views of those who would judge them as irresponsible. An examination of the uses of experiential knowledge sheds light on the intricate web of familial, interpersonal, social and moral responsibilities woven into the reproductive decisions of families in the context of a known inheritable condition.

Responsibilities were not only felt toward future generations of the family, but also past and existing relatives, the enactment of which, in recent years, have taken on new moral dimensions in the context of increased availability of genetic information and testing (since the 1990s for SMA). Experience of the condition within a family is a crucial mediator of these moral decisions.

Even for those participants who felt that their experiences of SMA were largely irrelevant to the lives of future generations affected by SMA, for all

groups of participants, experiential knowledge of SMA personalised and heightened the tensions and conflicts of responsibility. Experiential knowledge was used and abandoned strategically to navigate responsibility, and a family's entanglement in the experience of SMA was always factored into reproductive decisions; it could never simply be ignored.

Discussion

The findings of my research contribute to the literature that deals not only with the status, constitution and uses of experiential knowledge, but also that which surrounds the disability rights responses to prenatal testing, as set out below.

Abel and Browner (1998) in their discussion of experiential knowledge, have marked out a distinction between 'embodied' (i.e. that based on sensory experience) and 'empathetic' (i.e. that emerging from close relationships with others) experiential knowledge, which has been an important point of departure for much subsequent work on experiential knowledge, including my own. Whilst D'Agincourt-Canning (2003), developing this concept, recognised the possibilities of these two forms of knowledge becoming 'intertwined' as individuals experience caring for a relative with cancer whilst simultaneously having the condition themselves, this distinction is nevertheless used to mark out two different ways in which people come to know cancer.

This distinction between 'embodied' and 'empathetic' knowledge of SMA, however, proved difficult to sustain in relation to my own work. As

D'Agincourt-Canning (2003) has argued, people can occupy both positions, as embodied subject and empathiser simultaneously. Through the analysis of the accounts of people living with and around SMA, it became apparent that not only were the experiences of those caring for others with SMA thoroughly embodied, but that also people with SMA themselves had an acute empathetic understanding of the effects of SMA on those around them. Indeed, it is over-simplistic to assume that people living with relatives with SMA approached their understanding of SMA entirely through an emotionally based empathy, a 'commonality of feelings and experiences' (Keller, 1985: 117). Writers such as Morris have indeed been keen to disentangle such emotionally charged words from the arena of personal assistance for disabled people on the basis that it implies a form of emotional dependency of disabled people on those who care for them (Morris, 2002), and many participants were keen to disassociate the care work they received from either family members or privately employed personal assistants from any emotional relationship with that individual. Indeed Forbat (2002) has set out the range of emotions that 'carework' for a disabled person in the home may invoke within familial relationships including stress, sadness, bitterness, and jealousy as well as empathy. Participants thus constructed their understandings of SMA through this range of physical, emotional, and psychological experiences and responses, as well as by reference to broader professional, medical and social knowledges that construct SMA in particular ways (Good, 1994). This finding suggests that whilst a distinction between 'embodied' and 'empathetic' knowledge may be useful heuristically to

contrast the very different types and intensity of experience associated with having, or living alongside SMA, to suggest that these categories can adequately account for the range of conflicting, overlapping and contradictory experiences contained within them may be misleading and overlook the complexity of the experience of living with SMA. Moreover, as D'Agincourt-Canning (2003) suggests, the separation between embodied and empathetic forms of knowledge can be seen to imply that they are of contrasting status. Whilst it might be assumed that those diagnosed with a given condition themselves are 'closest' to the experience of cancer, D'Agincourt-Canning (2003) is keen to emphasise that empathetic knowledge can be just as 'poignant' or 'real' as embodied knowledge of cancer (p. 151).

Debates around the status of different forms of knowledge have formed the basis of standpoint feminist theorising. Smith (1987) and Hartstock (1983) have been among the chief proponents of feminist standpoint epistemology which posits women's experience as the basis for knowledge claims. Through women's subjugated position in society, they argue, unique insights into the subjective experience of oppression can be accessed, ones that offer 'truer' accounts of the internal workings of patriarchy than those obtained from 'above'. Within my own research, the hierarchical ordering of different ways of knowing SMA emerged as an important strategy by which individuals could position their accounts as 'authentic'.

The contrasting ways of accessing experiential knowledge have been addressed by previous analyses of experiential knowledge. Indeed, Etchegary

et al. (2008) have employed a quasi-visual metaphor to explore ‘vivid’ (personal), and ‘vague’ (more distant) forms of empathetic knowledge that pregnant women mobilise in their decisions about the uses (or otherwise) of prenatal screening and testing technologies. For D’Agincourt-Canning (2003), these different positions from which a person could ‘know’ cancer took the forms of ‘tangible knowing’ (or the knowledge derived from physically living with someone affected by cancer), ‘recent’ and ‘accidental’ knowing (which account for the more distant ways in which people come to know about cancer in their family, e.g. through the stories about unknown relatives). The concepts of distance and closeness as part of the nature of empathetic experiential knowledge, moreover, have been outlined in the work of Kay and Kingston (2002) in their exploration of the reproductive decision making of female carriers of X-linked conditions. Through conducting interviews with women who had ‘close’ relatives (e.g. first degree relatives) with an X linked condition and comparing the accounts of their reproductive decisions with women who had more distant relatives affected by an X linked condition (e.g. cousins, uncles), they suggest that proximity to the experience of disability may be associated with higher levels of guilt and anxiety around reproductive decision making.

My own research supported the findings of these studies in that distance from, and closeness to, the experience of SMA appeared to inform the way in which narratives were conceptualised and presented. Importantly, however, and a factor resolutely absent from previous accounts of experiential knowledge, is that this ordering of closeness and distance in

experiences of SMA went beyond mere descriptions of the type of experience, and instead was a means by which participants could check the *authenticity* of each other's accounts of SMA, and affirm the validity of their own. Whilst Etchegary et al. (2008) have asserted that the different types of experiential knowledge accumulated by women approaching reproductive decision making were not necessarily ordered according to which provided 'better' accounts or versions of reality, there was evidence within my own research that the hierarchical organisation of different ways of knowing SMA was an important strategy in the justification of certain standpoints and decisions about reproduction, as well as the discrediting of others. Being able to claim 'closeness' to SMA, and thus the ability to assign oneself the status of 'knower', was an important means of establishing authority on the condition and bolstered the legitimacy of reproductive decisions, whether anticipated or taken. Even as experiential knowledge was, by its very nature, partial, limited and uncertain, the positioning of this knowledge as 'authentic' was an important means by which participants could claim a right to define their own reality, as well as to discredit or deflect the (anticipated) judgements of others.

The ordering of different knowledge claims according to their proximity to so-called 'authentic' experience has been met with criticism from within and beyond feminism itself. Writers such as Haraway (1988) and Tuana (1993) have criticised feminist standpoint theorists' preference for the favouring of particular perspectives over others, and pointed to the dangers of defining certain perspectives as 'privileged' rather than simply different.

Critics have argued that it may be an impossible task to ever distinguish 'truth' claims from their discursive construction without falling into the trappings of relativism (Ramazanoglu and Holland, 1999), and indeed it may not be desirable to do so. Haraway (1988) has suggested a solution to these dilemmas in feminist epistemology by pointing to the possibility of documenting 'situated knowledges'; knowledges marked by the social locations of those who produce them, but not necessarily organised hierarchically. For Haraway (1988), the task of feminist research is not to decide which accounts of reality are 'better' than others, but to understand how complex subjectivities are expressed in the production of knowledge; knowledge which is both marked by, and born out of, our positioning in society.

Haraway's (1988) theorising on situated knowledges is particularly useful in making sense of the experiential knowledge of families affected by SMA. My own research has suggested that whilst experiential knowledge is 'real' in the sense that experience has a material reality, a being, beyond our descriptions of it; as researchers, all we have to understand this reality are the various accounts of these experiences which our research methods elicit, which, as Haraway (1988) has suggested, are thoroughly embedded in, or 'marked' by the social locations of those who produce them, both researchers and participants. Experience may be remembered or presented differently in various social contexts, it may be re-interpreted according to new information, and is always as fluid, partial and incomplete as other forms of knowledge. Haraway's (1988) work, therefore, is useful in highlighting the

need to treat experiential knowledge as situated knowledge and not a 'pure' account of reality.

Despite these various criticisms, feminist standpoint theorists have responded by arguing that, despite the difficulties associated with the ordering of accounts of reality, there may be ethical, social or political justifications for the privileging of particular vantage points in certain contexts (Hartstock, 1997). By this, Hartstock (1997) had in mind the possibility of setting aside such objections to standpoint epistemology if it is possible that more equal social relations could be envisaged by doing so. Indeed, in the context of my own research, this ordering of experiential knowledge could be interpreted as a political project. Privileging particular experiences over others was not simply about claiming a right to define their own experiences, or a means to visualise the operation of power and disablism as they interacted with their lives for families affected by SMA; rather many used their experiential knowledge as a way to circumvent and challenge these very values. Fisher (2007), through her study of parents of disabled children has argued that experiential knowledge of disability acquired in the private sphere is central to the creation of 'counter-hegemonic discourses' which challenge individualised notions of disability and ideas of 'normality'. The knowledge parents acquire of their children's abilities, through living intimately with them, is the foundation on which parents construct alternative conceptualisations of disability and independence as defined by dominant discourse. In a similar way, families affected by SMA used their experiential knowledge to co-create alternative narratives which redefined discursively

constructed ideas of disability, parental, and genetic responsibility in ways that incorporated their own lived realities. By reference to this experiential knowledge as a bounded and privileged form of knowledge, they were able to deflect the judgements imbued in dominant discourse and present their own versions of reality. The possibility of counter-hegemonic discourses to emerging through and out of experiential accounts of disability, impairment and illness reinforces calls by disabled feminists for the incorporation of these so-called private experiences into social model of disability theorising (Crow, 1996; Morris, 1991), and indeed, research has already pointed to the way in which these experiential aspects of disability and impairment may already run contrary to discursive constructions of them (Albrecht and Devlieger, 1999; Young and McNicoll, 1998). Thomas (2007) has highlighted that whilst medical sociology has been concerned with the experiential aspects of chronic illness, social model of disability theorists have tended to write out this crucial aspect of disablism. Further research is thus indicated that bridges this ideological gulf between experience and the social structures and mechanisms that disable people, between which experiential knowledge, and the counter-hegemonic discourses and constructions of disability that emerge from it, may provide a crucial link.

As well as having political consequences in terms of the discursive challenging of ideas of parenthood, disability and genetic responsibility, claims to ownership of a bounded body of experiential knowledge of SMA also had important emotionally protective qualities for families affected by SMA. Being able to substantiate particular reproductive decisions by recourse

to a body of experiential knowledge that they alone could claim and possess, individuals could exclude the interpretations and judgements of others in the midst of reproductive decisions fraught with difficult emotions and uncertainties. For individuals diagnosed with SMA, claims to a body of experiential knowledge of greater authenticity than their family members' was an important strategy by which to neutralise the emotional harm arising from family members' steps toward preventing SMA in future generations, which many interpreted as a negative evaluation of their own lives. Whilst the existence and theoretical basis of the expressivist objection to prenatal testing and selective termination by disabled people has been described in the literature and media (Asch, 2000; Atkinson, 2008; Kent, 2000), there is a lack of research detailing how this objection, and the emotional consequences associated with it, are managed in families affected by inheritable conditions, where, as I have argued, the stakes may be heightened by the experiential knowledge of the condition family members possess. In such situations, claims to lack of knowledge by prospective parents as to the reality of life with a given condition, may be an emotional safeguard unavailable to individuals with SMA whose relatives pursue methods of preventing its recurrence. The sense of responsibility that emerged in family members' accounts, moreover, towards the emotional well-being of their relative with SMA additionally suggests that the expressivist objection has consequences not only for those individuals diagnosed with SMA, but that it can also constrain the responses of families affected by inheritable conditions. Further research is indicated to explore the way in which the expressivist objection is

negotiated within families affected by other conditions to determine how experiences of disability, impairment and illness are conceptualised in such instances and presented in relation to prenatal testing and selective termination.

Whilst my attention has focused so far on the way in which participants esteem and privilege accounts of experiential knowledge, it is important to note that a similar strategy was, paradoxically, also used by those who adopted a more critical stance towards the relevance of their experience with SMA to future lives with the condition. Indeed, just as Etchegary et al. (2008) discovered that a perceived *lack* of experiential knowledge and gaps in knowledge could be referenced in reproductive decision making in a similar way to those who possessed experiential knowledge, so the uses of experiential knowledge by those who regarded it as an unstable or obsolete resource mirrored the strategies of those who privileged such knowledge claims. Whether experiential knowledge was considered a concrete resource on which to base reproductive decisions, or an unstable basis for knowledge claims, it was the privileging of *particular versions* of experiential knowledge (whatever these may be) to the exclusion of others as a means by which to negotiate tensions in reproductive decisions that characterised the use of experiential knowledge in these decisions. Being able to present experience in particular ways and at particular points was a means for participants to tell those stories which may be difficult to tell in a society which, through the use of medical technologies and discourse, more

or less explicitly supports some reproductive decisions over others. Cox (2003) in her analysis of the testing decisions within families affected by Huntington's disease (HD) has furthermore suggested that the point in the decision making process that participants are positioned at the time of interview also influences the story they are able to tell. As she interviewed people who had already decided to undergo testing for HD, and most were awaiting their results, the stories she heard were far less imbued with the 'weighing of options, the to-ing and fro-ing, and the wonderings of 'what if'' than she had initially anticipated to hear (Cox, 2003: 261). The interview, for these participants, was thus an 'occasion for justifying, rather than re-living' their decisions, as they were reflecting on actions already undertaken (Cox, 2003: 261). As the participants in my own research were at differing stages of reproductive decision making, and indeed, some considered themselves to have not yet made any such decisions, this context invariably affected the way in which their thoughts were presented. As Cox (2003) similarly discovered, it may be unsurprising to find more certainty in the accounts of those reflecting on reproductive decisions already taken, in comparison to the oscillation between different possible decision outcomes that were rife in the accounts of those participants who had not yet enacted reproductive decisions. All were acutely aware that once taken, the decisions (and the knowledge and experience arising from their enactment) could not be undone, only re-represented.

A further finding of my research relates to the relationship between experiential and medical knowledge. The literature around experiential

knowledge, as has been discussed, has tended to focus on its status vis-à-vis medical knowledge and dominant constructions of disability, to demonstrate the way in which it has become an alternative source of identity construction, as well as information in decision making and risk perception, particularly where medical knowledge is contradictory or incomplete (Abel and Browner, 1998; D'Agincourt-Canning, 2003; Etchegary et al., 2008; Lippman, 1999; Fisher, 2007). The research conducted by Abel and Browner (1998) in relation to pregnancy and the experiences of carers for relatives with dementia, as well as D'Agincourt-Canning (2003) in relation to breast and ovarian cancer, for example, have suggested that experiential knowledge of a given condition is a filter through which medical knowledge is passed, and either accepted and incorporated into pre-existing conceptions, or else rejected.

Markens et al., however, in an attempt to move beyond these dichotomous constructions of medical and experiential knowledge have suggested a 'dynamic and synergistic' (Markens et al., 2010: 52) relationship between the two, whereby both medical and experiential knowledge contribute to one another (p. 39). My own research confirms that experiential knowledge was a means by which families affected by SMA could interrogate, challenge and expand on, medical knowledge, but that further, this medical knowledge also informed and structured experiential accounts of SMA. For example, whilst many participants challenged the medical typology of SMA through their experiential accounts e.g. stating they were 'good' or 'poor' compared to others with the same diagnosis, their very understanding

of what constituted a person with any given type of SMA was nevertheless grounded in the medical definitions they sought to challenge. In a similar way, the experiential knowledge of participants also fed into, and contributed to, medical knowledge of SMA. As documented in Chapter 3, the emergence of SMA as a disease category was characterised by the challenging of, and expansion of, medical diagnostic categories on the basis of experiential accounts of SMA that surpassed clinical expectation (Dubowitz, 1995b). Whilst expert and experiential knowledge are frequently presented as oppositional within research on experiential knowledge, and indeed, there may be political motivations to maintain such a distinction (e.g. the juxtaposition of medical and experiential accounts of impairment by disability rights supporters), my findings have highlighted the way in which these knowledges were constructed and maintained in continual reference to one another, in a fluid, ever-evolving relationship. Experiential knowledge, whilst often used as a means by which to challenge, or even invalidate medical knowledge, was nevertheless conceptualised through dialogue and exchange with this very knowledge. This finding highlights the way in which experiential and medical knowledge, rather than rigid, diametrically opposed structures, may instead be more usefully conceptualised as mutually constituting knowledges, existing by and through their reference to one another.

Aside from the ontological and epistemological status of experiential knowledge, Abel and Browner (1998) have furthermore highlighted the tendency within research on experiential knowledge to romanticize the

accounts of individuals that are grounded in everyday experience and used to challenge medical authority (p. 322). As Markens et al. (2010) have suggested in relation to women's experiences of prenatal testing, this has frequently been interpreted by researchers as a cue to validate experiential knowledge and in particular, to assert its value as a means of reassurance. In contrast to medical knowledge from which has emerged uncertainties, insecurities and the 'tentative pregnancy' (Katz Rothman, 1986), experiential knowledge is deemed to be the site in which women come to trust their own bodies and subjective understandings. However, my research has pointed to the way in which experiential knowledge, akin to medical knowledge, can similarly be experienced as highly uncertain, imperfect, and even distressing. Indeed, whilst experiential knowledge was a resource sought out by many families and individuals affected by SMA, for some, the knowledge they acquired through the stories and lives of others with SMA was a deeply unsettling experience, and threw into doubt what they had previously accepted as certainty. Whilst the possibility of challenging medical knowledge was sometimes welcomed, experiential knowledge could also threaten the stability that families felt they needed following diagnosis. As Abel and Browner (1998) discovered, at points where people feel particularly vulnerable, being able to 'cling' to the perceived certainty, trustworthiness and reliability of medical knowledge was an important coping mechanism (p. 322). As D'Agincourt-Canning (2003) notes in relation to women at risk of hereditary breast/ovarian cancer, women both accepted and supported the 'privileged discourse of science' because it responded 'where they felt most

threatened' (p. 157). The importance attached to medical knowledge, therefore, should not be under-estimated despite the challenges of experiential knowledge. As Henwood et al. (2003) have argued, despite rhetoric of patient empowerment in the context of health, people do not always prefer to have the control (and consequently the responsibility) over what happens to them. Whilst genetic technologies have increased the number of reproductive decisions presented to would-be parents, this control is not always welcome, and sometimes self-determination includes the choice to relinquish that control (Fisher, 1986). Accepting the authority of medical knowledge and associated advice can be seen as one of the ways in which prospective parents relinquished control over their reproductive decisions in contexts that were perceived as highly uncertain and risky, as acts both of self-preservation, by avoiding the guilt and anxiety associated with making the decisions solely themselves, and of self-determination, by accepting advice from those deemed to be in positions of authority (Fisher, 1986).

As has been described in Chapter 3, the development of the JT SMA into a 'genetic advocacy group' (Novas, 2007), points to the centrality of the future promises of medicine and genetic research, not only at the point of reproductive decision making, but also in the daily reality of life with SMA. Being able to hope for a future in which the difficulties associated with living with SMA are resolved by medical science was an important facet of daily coping mechanisms of many families. Moreover, medical knowledge provided families with a definitive diagnosis, on the basis of which identities and collectivities could be forged. Whilst experiential knowledge of SMA

often called these diagnoses into question, medical classifications nevertheless provided families with socially sanctioned labels with which to understand their experiences; the importance of having a seemingly stable diagnostic badge with which to explain and understand one's experiences should not be underestimated, and could offer a sense of security to families living with SMA.

Families and individuals affected by SMA, therefore both challenged and endorsed medical knowledge of SMA even as that knowledge contributed to, and disrupted, their conceptualisations of the condition. The relationship between medical and experiential knowledge of SMA is complex and multi-faceted; experiential knowledge did not offer an alternative to medical knowledge in any straightforward way. Instead the two forms of knowledge existed as parallel sources, crossing, intersecting and weaving together at various points, but nevertheless representing two distinct bodies of knowledge that were strategically drawn upon in different contexts to construct and maintain ways of approaching, and conceptualising, SMA.

Practice and Policy Implications

My research has implications for practice and policy on a range of different issues. Firstly, my research has contributions to make to the call, made primarily by disability rights supporters, for more knowledge about conditions that are currently screened and tested for in the UK to be made available to would-be parents (Fletcher, 2002; Patterson and Satz, 2002). As the number of conditions which are screened and tested for increases, these

demands seem set to increase, and moves to make such information available have already appeared (AnSWeR, 2009). Whilst population screening for SMA is not yet offered in the UK, there have been moves more recently to make this screening available in the USA ('new born screening programme', FSMA, 2008; Prior, 2008), and it seems feasible that this type of intervention will at some point be available in the UK. Indeed, a similar programme to identify those babies affected by, but, critically, also those who are carriers for, CF has already been implemented in the UK through the newborn heel prick test (2007, although no carrier screening for adults is currently available). This screening will mean that an increasing number of individuals will be growing up with the knowledge that they are a carrier of a recessive condition, without necessarily having a known affected family member, which previously was the means by which a family came to know about CF (NHS Choices, 2008). These developments have implications for a discussion of experiential knowledge, and its usefulness as a resource for prospective parents identified as being genetically at risk by antenatal screening tests (Patterson and Satz, 2002).

In the first instance, as Kaplan (1999) has argued, one of the key motivators for people to use prenatal testing and selective termination is an erroneous belief that disability is a necessarily negative trait and one associated with a poorer quality of life than might otherwise have been expected for an able-bodied child (p. 132). Whilst this motivation may be set alongside others, Kaplan (1999), along with other disability rights supporters, has argued that this assumption is highly problematic. She observes that this

justification for using prenatal testing and selective termination is frequently deployed as it can easily be presented in terms of altruism; prospective parents can claim they are preventing harm to a future life, rather than acting out of self-interest, and many participants in my study mobilised this justification in accounting for their own reproductive decisions. Whilst some writers in the field of medical ethics have argued strongly that disability *does* inflict some degree of harm on future lives and consequently would-be parents are justified in enacting their responsibility to protect future lives from such an outcome (Purdy, 1996; Harris, 2000; Harris, 1998), there have been many dissenting voices in the field of disability studies who have questioned this assumption and presented alternative visions of life with disability (Edwards, 2001; Asch, 1999; Saxton, 1984; Hubbard, 2006). Whilst for some writers, the negative assumptions described above are seen as emerging from a medical model of disability (Asch, 2001) and the presentation of unbalanced information about conditions in medical encounters at the point of prenatal testing and selective termination decisions (with an emphasis on medical complications rather than opportunities for successful living (Williams, 2002)), it has been suggested that an appropriate way to challenge them is for more information about the experiences of those living with disability to be made available to would-be parents (Fletcher, 2002; Patterson and Satz, 2002). Etchegary et al. (2008) amongst others (Kay and Kingston, 2002; Chandler and Smith, 1998) have argued that women's experiential knowledge of, and associated attitudes towards, disability, illness and impairment may have a significant impact on their attitude towards

selective termination; and indeed there appears to be evidence to suggest that those individuals who have a genetic condition in their family, and thus experience of the condition, may approach the issue of selective termination in a different way to those for whom the condition is simply a medical label (Gow, 2000; Wertz et al., 2002; Green, 1993). A key concern of disability rights supporters, therefore, who draw on the positive experiences reported by many of the families of disabled children to support their argument (Asch, 2001), is how to make the sort of knowledge about life with disability acquired through close contact (such as within a family) available to those who are making such decisions without this insight. This knowledge is sometimes presented as a counter-balance to medical information about disability (Fletcher, 2002; Patterson and Satz, 2002), with the implicit suggestion that this experiential knowledge can challenge prevailing assumptions about disability.

The findings of my own research contribute to these suggestions by writers from within the disability rights community, as well as the movements to introduce novel ways of presenting information about different conditions to would-be parents (such as AnSWeR, 2003). My research supports the findings of other research such as that by Gow (2001) and Kelly (2009), which document the way in which attitudes towards disability can be transformed by the birth of an affected family member, and that this attitude can be a guide in future reproductive decisions. Indeed, it is noteworthy that nearly all of the able-bodied siblings of people diagnosed with SMA reported that their experiences with SMA made them feel more at ease with the

prospect of having a child with SMA themselves than may otherwise have been the case. Moreover, it was also reported that their experiential knowledge of SMA not only had an impact on attitudes towards future pregnancies diagnosed with SMA, but also towards other disabilities, impairments and illnesses. As was argued in Chapter 4, the experience of SMA was conceptualised in terms of the experiential categories of ‘experiences of disability’, ‘embodied experiences of impairment and disability’ and ‘illness death and bereavement’ rather than according to diagnostic label, which may account for the transferability of this knowledge to other disabilities, impairments and illnesses. This finding calls into question the way in which information is currently presented to prospective parents on websites such as AnSWeR (on the basis of medical diagnoses), and suggests that people draw on multiple sites of experience when evaluating a prenatal diagnosis including that of both the diagnosed condition as well as other conditions, particularly if there is concordance with the associated experiential categories between the two conditions. Future research is indicated that would explore the reproductive decisions of those with inheritable conditions in their family and the uses of this knowledge in prenatal screening and testing decisions relating to conditions other than the one affecting their family.

As well as highlighting the transferability of experiential knowledge in the context of prenatal screening and testing decisions and the insights this offers to an understanding of the innovative ways in which prospective parents approach and use their experiential knowledge in this context, the

research I have conducted also points to the constraining effects of experiential knowledge. Whilst disability rights supporters have been keen to emphasise the reassurance or change in attitude which can be derived from experience with disability, it appears that through the championing of experiential knowledge, such writers have the promotion of *particular* reproductive decisions in mind. For those participants who chose to opt out of prenatal testing or carrier testing to prevent the recurrence of SMA in their family, there was evidence that such participants found their experiential knowledge to be a valuable resource with which to challenge prevailing discourses of disability and responsibility. However, for other participants, this knowledge heightened existing dilemmas and even introduced new ones.

As has been highlighted in Chapter 6, experiential knowledge can constrain the responses of those making reproductive decisions in families affected by SMA by removing the possibilities of using the justification of altruism, suggested as a common motivation by Kaplan (1999) for undergoing prenatal or carrier testing. For individuals whose experiences with SMA were largely positive, the notion that prenatal testing and selective termination prevents future suffering becomes harder to sustain. Disability rights supporters have celebrated the removal of this assumption as progressive. However, my research suggests that for would-be parents, and particularly for would-be mothers facing reproductive decisions, this knowledge can paradoxically be experienced as oppressive, increasing uncertainties around reproduction and rendering them trapped between competing concerns and demands. Having insight into what life with SMA

could be like, therefore, was not always a helpful resource and had the potential to disrupt relationships within the family, particularly with members diagnosed with SMA themselves. My research points, therefore, to the difficulties as well as the benefits associated with the advancement of experiential knowledge as a means by which to 'give voice' (Bricher, 1999) to the perspectives of those with disabilities in the prenatal testing and screening debate, highlighting the agenda underpinning the advancement of such information in this context. It highlights the high degree of sensitivity to the needs and concerns of would-be parents that is required in the advancement of such information, as well as those of people diagnosed with inheritable conditions who may require support to manage the emotional consequences of the reproductive decisions of others.

Despite the disputes about the coherence of the expressivist objection to prenatal testing (Shakespeare, 2008; Sparrow, 2008; Edwards, 2004; Stainton, 2003), individuals diagnosed with SMA nevertheless reflected on the emotional consequences of the reproductive decisions made by their relatives in ways that highlighted the possibilities for, and actual emotional harm that resulted from the perceived suggestion that others would want to prevent the birth of another person with the same condition as themselves. Many experienced this as a personal rejection on the basis of their impairment. The possibility of emotional harm emerging from the expressivist objection (Asch, 2001) to prenatal testing in families affected by inheritable conditions has largely been ignored in the genetic counselling literature, and my research points to a need to acknowledge this experience,

not only in terms of offering support to those diagnosed with the condition, but also acknowledging the constraints the possibility of an expressivist objection may place on the reproductive decisions of others. Wishing to avoid the perception of rejection on the part of an affected relative was indeed described as a motivating factor for many participants who opted to decline carrier and prenatal testing for SMA. Despite the importance of the relationship to family members diagnosed with the condition, genetic counselling research has largely overlooked the attitudes and responses to genetic testing of those with disabilities and genetic conditions (Chen and Schiffman, 2000). Little is known about the way in which the expressivist objection to prenatal, and also carrier testing, is experienced and expressed within families affected by inheritable conditions and the consequences it has for perceived reproductive options and responsibilities. My research has highlighted the emotional consequences of the expressivist objection within families and the way in which it can both constrain and clarify reproductive decisions. However, further research is indicated to inform future genetic counselling practices and highlight the need for sensitivity in dealing with this issue.

Despite the emphasis on informed decision making, rational choice and patient autonomy, which currently preside over clinical practice in the fields of genetic medicine and beyond, research is increasingly calling into question individualised notions of autonomy, and underlining the importance of understanding the relational nature of responsibility and the way in which experiences shape medical decision making (Burgess and D'Agincourt, 2001;

Cox, 2003; Werner-Lin, 2007; Babb et al., 2002; Hallowell, 2006; Roberts and Franklin, 2004; Shiloh, 1996), an element Fischhoff et al. (1978) refer to as the 'human factor' in decision making. The consequences of the expressivist objection are but one example of the way in which 'relational responsibilities', or those grounded in experiences of everyday life (Burgess and D'Agincourt, 2001), unevenly shape and constrain reproductive autonomy within family groups, and point to an interdependent notion of self emerging from mundane everyday experience. The reproductive decisions made by those families affected by SMA cannot be usefully understood as rational, outcomes-based decisions made by autonomous and isolated individuals. The decisions made by the families affected by SMA who took part in this research were instead born out of, and marked by shared familial experience with SMA, by wider social discourse around disability, genetic disease and parental responsibility and the complex interplay between these factors. An emphasis on rational decision making overlooks the highly emotive relationship individuals have with their history with the condition, which can transform, or even render obsolete, statistical measures of genetic risk (Shiloh, 1996; Werner-Lin, 2007). These findings suggest that it may be useful for genetic counsellors to explore experiential knowledge when counselling families affected by inheritable conditions, including the experiences of those diagnosed with the condition, the experiences of caregivers and the resulting conceptualisations of the condition. Such an exploration, as highlighted by Etchegary et al. (2008) may help counsellors identify those individuals most likely to experience distress in the prenatal or

carrier testing counselling process and who might benefit from additional support (p. 123). Furthermore, such an exploration may verify such experiential knowledge as a valid form of knowledge about SMA which can be drawn upon to make sense of, and challenge medical knowledge.

Conclusions

My research has addressed the question of how experiential knowledge and responsibility are negotiated in the reproductive decisions of families affected by SMA. From an analysis of the accounts of the anticipated, and completed, reproductive decisions of 64 people from such families, it was apparent that the negotiation of such responsibility, and the accompanying moral, social and ethical dilemmas, could be found in the processing, and accounting for these decisions, rather than in the decision outcomes themselves. Whilst much research has focussed on the different decisions people make according to a variety of variables (Wertz et al., 1992; Sagi et al. 1992), those facing genetically 'risky' reproductive decisions may reach the same end decision through very different means, and it is these accounting processes which are the most useful for examining the negotiation of competing norms and values.

Experiential knowledge was of particular importance to families affected by SMA, a condition for which medical knowledge is both imperfect and uncertain, and the prospect of cure or treatment remain distant. Medical classifications of the condition remain unsatisfactory in their ability to account for all experiences of SMA, and thus medical knowledge of SMA, in its current form, has limited potential in predicting the sort of life a future

person with SMA may have. The families who took part in this study thus existed in a space where genetic risks and the possibility of disability became highly personalised; they were not considering a disability unknown to them, but instead the recurrence of a disability that affected their sibling, their parent, their child, themselves, and consequently many considered themselves to be the best experts on the impact of SMA for their family. Experiential knowledge of SMA, therefore, could not be ignored when approaching reproductive decision making; it was at the very core of how genetic risk was conceptualised. Medical statistics of risk were not objectively evaluated, but instead interpreted through this lens of experience, and through interpretations of the responsibilities and ‘moral risks’ that emerged from these experiences: to family members past, present and future, to partners, to other families affected by SMA, to wider society and to themselves. Experiential knowledge, moreover, was not static, but instead in a constant state of flux, meaning that genetic risk could be conceptualised differently at different times and contexts; it could be mobilised retrospectively to account for previous reproductive decisions, or projected into the future to justify decisions not yet made; presented as either a certainty or an unstable resource respectively to solidify particular versions of genetic risk at particular times, and furthermore to present them as conforming to overarching notions of relational, and reproductive responsibility. These strategies highlight the highly precarious, and often impossible, position of such families and individuals approaching reproduction. Positioned within the uncertainties of experiential and medical accounts of SMA and prevailing expectations of

reproductive responsibility, such individuals must weave a path through the various moral, social and ethical dilemmas surrounding reproduction and genetic risk, which, as this research has demonstrated, may ultimately never be resolved, only re-represented

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Appendices

Appendix I: Research Leaflet

INFORMATION SHEET FOR PARTICIPANTS

You are being invited to take part in a research study. The research is being conducted myself, Felicity Boardman, a PhD research student at the University of Warwick under the supervision of Dr. Hannah Bradby and Dr. Karen Throsby and with the co-operation and support of The Jennifer Trust.

I am a doctoral student and have an interest in the implications of disability and genetics for the individuals and families who directly experience them.

This information sheet will explain why the research is being done and what it will involve. Please read this information carefully to help you decide if you would like to take part. My contact details and those of my supervisors are provided at the end should you wish to ask any questions.

What are the aims of the study?

The research aims to explore the experiences of people with Spinal Muscular Atrophy and their families. In particular, the research will look at how families affected by these conditions think about having children and their experiences and ideas about genetics and genetic testing. The research will make a contribution to our understanding and knowledge about the impact

How can I get involved?

If you would like to take part in this study, you may express your wish by sending me an e-mail to [e mail address] or by telephoning [number]

If you decide not to participate yourself, you may wish to pass this information leaflet on to a relative or friend who could take part instead.

of disability and genetics. By taking part, you will have the opportunity to discuss your thoughts and experiences on these topics and for these to be included as a part of this broader contribution.

Why have I been asked to participate?

You are being asked to take part because I understand that you are over 18 years of age and either have Spinal Muscular Atrophy yourself, or one of your family members has. I am interested in speaking with the parents, sisters and brothers, step brothers and sisters, aunts and uncles, partners or spouses to people with one of these conditions.

What does the research involve?

The research involves an interview with me for a minimum of 20 minutes. This may be done over the telephone, on the internet through e-mail or I may be able to come and visit you at a suitable location.

After the first interview you may be asked to take part in a longer interview, which will last about an hour. You partner or spouse is invited to take part in this with you. This interview can be done in the same way as the first and organised at a time and place that is convenient for you. You will be asked questions about your experience of the condition that affects your family. You will also be asked about how you feel about having children and your experiences and thoughts about genetics and genetic testing.

Who can I contact for more information?

For more information regarding this research, or if you would like to take part, please contact:

[name and address]

Supervisors' contact details:

[names and addresses]

Once the interview has taken place, you will be given a transcript (write up) of your interview to keep and to add any further comments.

Ethical considerations

All information given during the research will be treated as strictly confidential. Your real name will not be included, nor will any personal details that could identify you. However, if another member of your family is interviewed as well as yourself, then they may be able to recognise your answers even if your name has been changed. The data will be written up as part of my PhD thesis. This thesis will be stored at the University of Warwick and will be available for the public to read. Sometimes the results of PhD theses are also published in journals or books. It is important to remember that if you agree to take part in the research, you are still free to drop out at any point, even if an interview has been arranged or already carried out. You do not have to give a reason for this and *no information will be included in the study without your permission.*

Who is funding the study?

The research is being funded by the Economic and Social Research Council. For more information about this organisation see www.esrc.ac.uk

DISABILITY AND GENETICS RESEARCH PROJECT

For individuals and families affected by Spinal Muscular Atrophy



This leaflet will explain why the research is being conducted, what it will involve and who is being asked to participate

Appendix II: Consent Form



CONSENT FORM

DISABILITY AND GENETICS RESEARCH PROJECT

Name of Researcher: Felicity Boardman

Name of Participant:.....

I confirm that I agree to participate in the above study. I understand that my participation is entirely voluntary and I am free to withdraw at any time without giving a reason.

Signature

Date

.....

Appendix III: Interview Guide

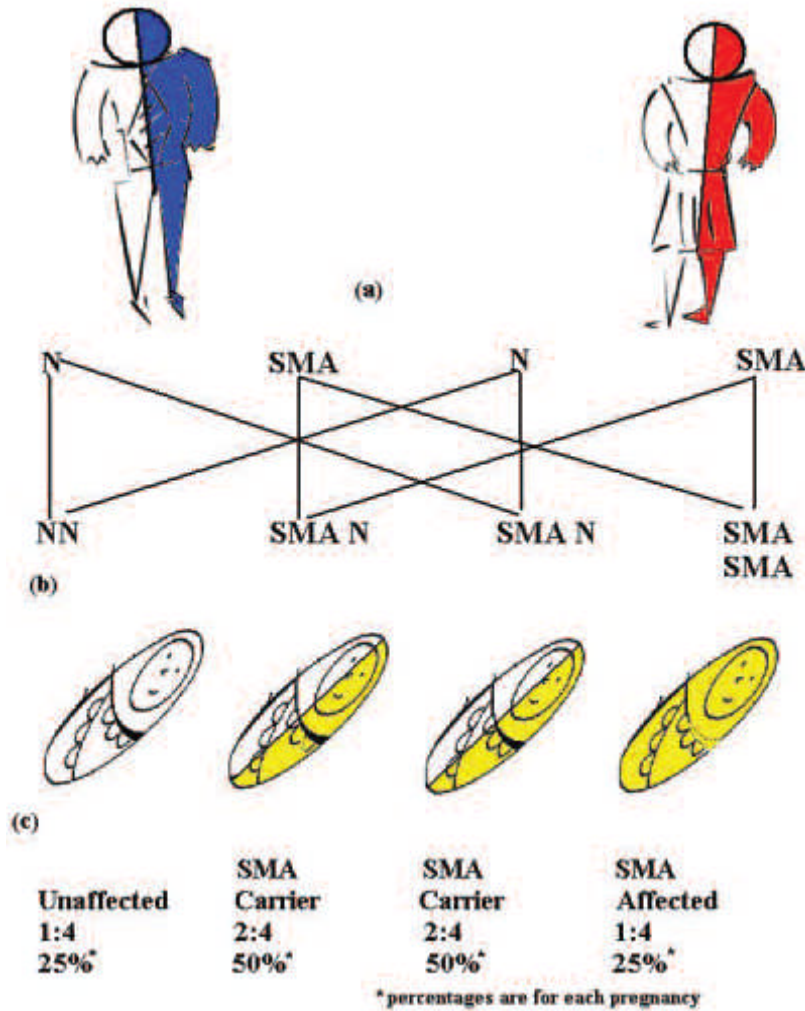
Interview Guide

N.B. These questions were used as a loose guide and were adapted as appropriate for each interview. Participants further had considerable freedom to direct the interviews.

- 1) Can you tell me a bit about yourself? (Age, occupation, living arrangements, family, hobbies/interests, diagnosed with SMA or relative of someone with SMA?)
- 2) How would your friends/family describe you?
- 3) If I was a biographer writing up the story of your life, what would I write as being the important events/experiences that have shaped it?
-Prompt for story of diagnosis and responses to genetic diagnosis including responses of family
- 4) How have these events/experiences affected who you are today and your life in the present?
- 5) How would you describe SMA to someone who has never heard of the condition before?
-prompt for descriptions of different experiences associated with SMA
- 6) What do you understand by the term 'quality of life'? What do you think you need to have to enjoy a good 'quality of life'? Can people living with SMA enjoy a good 'quality of life'? Why (not)?
- 7) Are you a member of the JTSMA? Why? What involvement, if any, have you had with the JTSMA? Do you find it helpful? Why?
- 8) Do you think genetic testing for families affected by SMA is useful/important? (Carrier testing, prenatal testing, PGD) Why? Do you see any draw backs or difficulties with testing? Why?
- 9) Do you think prenatal testing and screening for other conditions, e.g. Down's Syndrome is a useful option to have? Why?
- 10) How/when might you/have you used genetic testing yourself? Why (not)?

- 11) In what situations would you consider genetic testing to be particularly appropriate? When is it less useful?
- 12) How do you feel about the possibility of carrier screening of the whole population for SMA? Do you see it as important? Why?

Appendix IV

Diagram Showing the Inheritance of SMA for Two Carrier Parents

(Taken from 'The Genetics of SMA' information sheet and reproduced with the kind permission of 'Families of SMA' (FSMA), 2010)

*Appendix V***Forms of Genetic Testing Available to Families
Affected by SMA***Forms of Genetic Testing Available to Families Affected by SMA*

Since the linking, in the early 1990s of SMA to particular genetic mutations, there has been a rapid expansion in the number of options available for families affected by SMA to gather information about the genetic status of their members, present and future. In 1990, through the use of DNA markers, prenatal testing became available, to determine whether or not future generations of families affected by SMA would similarly display symptoms of SMA (Zerres et al., 1997). This testing could be done through either chorionic villus sampling (CVS) at 10-12 weeks of pregnancy or amniocentesis at the 15th week of pregnancy (Simard, 2007). CVS involves the extraction and genetic testing of samples of the chorionic villi that form the placenta, whereas amniocentesis involves the extraction and testing of amniotic fluid to determine whether or not the foetus has two copies of the deleted SMN1 gene. Following the results of prenatal testing, either by amniocentesis or CVS, prospective parents have the option of terminating or continuing with the pregnancy.

Diagnostic, Prenatal and Carrier Testing

In early 1995, the gene candidate SMN was identified, which further expanded the possibilities for genetic testing of families and individuals (Zerres et al., 1997). In the first instance, the sequencing of the gene led to the possibility of genetic testing for diagnostic purposes. As the vast majority of instances of SMA are deemed to be linked to mutations and changes in the SMN1 gene,

molecular analysis could supplant more invasive diagnostic tests that were being used prior to this date, such as muscle biopsy (Van Der Steege et al., 1995). Furthermore, the possibility for carrier testing became available. This test was of particular use to the siblings of individuals affected by SMA, as well as extended family members who wished to ascertain their likelihood of having a child expressing symptoms of SMA and the results could be obtained through a simple blood sample. This form of testing is designed to detect the number of copies of the SMN1 gene a person has on each chromosome, but due to the possibility of an individual having two copies of the gene on one chromosome and none on the other (making them a medically defined 'carrier'), carrier testing is unable to detect 2-3% of carriers with this variation (Simard, 2007). If prospective parents are both known to be carriers, they may opt to undergo prenatal testing of their pregnancy, however if only one prospective parent is a carrier, they are unlikely to have a child displaying symptoms of SMA, they may, however, have a child who is also a carrier of SMA. All children born to parents where one parent is diagnosed with SMA will be carriers of SMA, whilst children born to parents who are both diagnosed with SMA will similarly have SMA themselves (Ogino and Wilson, 2002).

Carrier screening of the general population is not currently available for SMA in the UK, however there have been recent moves to promote such screening in America (Prior, 2008), and patient organisations such as the JTSMA support the development of such programmes in the UK context. It has also been suggested that neonatal screening for SMA may become justifiable as therapies become available (Wirth et al., 2006).

Pre-implantation Genetic Diagnosis

In 1998, a further possibility became available for prospective parents known to be carriers of SMA. A Dutch team, led by Dreesen (1998), conducted the first pre-implantation genetic diagnosis (PGD) of SMA for a couple whose first child had died at 9 months of age from SMA type I. A pregnancy was established after the second cycle of PGD, and this development was heralded in the medical community as offering an alternative to prenatal testing and the possibility of selective termination for families affected by SMA (Dreesen et al., 1998). PGD works by undertaking genetic testing at the embryonic stage of foetal development, rather than 10-14 weeks into an established pregnancy, as is the case for CVS and amniocentesis. This testing is made possible by the creation of embryos through in vitro fertilisation (IVF), whereby gametes are taken from the couple and fertilisation occurs extra corporeally. The resulting embryos are subsequently tested for the specific genetic mutation which the prospective parents are known to be carriers of, and embryos found not to carry the genetic mutation are transferred into the woman's body for gestation. As Franklin and Robert's (2006) ethnography has shown, for some couples who are carriers of SMA, the possibility of undergoing PGD can be experienced as a welcome alternative to prenatal testing and selective termination (p. 118), in spite of its low success rate. Indeed, Guys and St. Thomas' clinic in London is one of the main sites for PGD for SMA, and between 1997 and 2000, 51% of referrals for single gene disorders to this PGD clinic were for couples who are carriers of SMA (Pickering et al., 2003), compared to 38% for Cystic Fibrosis (a more common condition), suggesting that PGD is an option that is being

taken up such families. Whilst PGD is not currently available for all of the variants of SMA, e.g. SMARD and ADSMA, in 2001, PGD also became available for SBMA (Georgiou et al., 2001).

Amniocentesis

Amniocentesis is a diagnostic prenatal test that can be carried out between 15 and 21 weeks of pregnancy. It is offered to women whose pregnancies are considered to be at 'high risk' of being affected by a particular condition. The procedure involves removing a sample of amniotic fluid (the fluid surrounding the foetus) via a needle. This fluid contains foetal cells which can then be tested for chromosomal disorders such as SMA. Amniocentesis is associated with a slightly increased risk of miscarriage (1-2%) (NHS Choices, 2009).

Chorionic Villus Sampling (CVS)

Chorionic villus sampling is a diagnostic prenatal test carried out between 10 and 12 weeks of pregnancy. It is offered to women whose pregnancies are considered to be at 'high risk' of being affected by a particular condition. The procedure involves the removal of a small sample of the developing placenta (the organ that connects the woman's blood supply to the foetus), which is then tested for chromosomal disorders. The procedure is associated with a slightly increased risk of miscarriage (1-2%) (NHS Choices, 2009)

*Appendix VI***Variant forms of SMA***Spinal Muscular Atrophy with Respiratory Distress (SMARD)*

SMARD is a condition affecting infants which has very similar features to severe SMA, however it is not linked with the SMN1 gene. Whilst having many overlapping features with severe SMA including generalised muscle weakness, SMARD is associated with diaphragm paralysis (not seen in children and infants with severe SMA), and the weakness begins with the distal muscles (those furthest away from the body), moving inwards. In severe SMA however, the muscle weakness presents in reverse order with the proximal muscles (those closest to the body) being affected first, and more severely. With SMARD, there may also be sensory disturbances which have been excluded from the diagnostic criteria for SMA. SMARD is inherited in the same way as SMA, through an autosomal recessive inheritance (see appendix IV).

Spinal Bulbar Muscular Atrophy (SBMA)

SBMA is an adult onset form of SMA (with onset generally between 20 and 40 years of age) which shares many features with 'classical' SMA, including generalised muscle weakness, however it is not linked to the SMN1 gene. Instead, SBMA is X linked, which means it affects males, but the gene is transmitted by females. Women who are carriers of SBMA may experience mild symptoms of the condition. SBMA may furthermore involve symptoms

which are not associated with SMA including hormonal changes (Fischbeck, 1997).

Autosomal Dominant Spinal Muscular Atrophy (ADSMA)

Autosomal Dominant Spinal Muscular Atrophy is an adult onset form of SMA with onset between 30 and 50 years of age. It is considered to be milder than the 'classical' forms of SMA, but involves what have been described as the key features of SMA including generalised muscle weakness. This adult onset form of SMA is not linked with the SMN1 gene and is inherited in an autosomal dominant manner, which means that only one copy of the gene is needed to pass on ADSMA, unlike recessive SMA which requires both parents to be 'carriers'.