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## **Experiential Knowledge of disability, impairment and illness: the reproductive decisions of families genetically at risk**

### Introduction

The number of conditions that may be detected by reproductive genetic technologies (RGTs) has risen steadily in recent years, paralleled by a corresponding increase in the number of testing and screening decisions to be made by would-be parents. A large body of research exists documenting the wide range of factors that inform the way decisions around the use of RGTs are perceived, arrived at and experienced by such parents; factors including (but not limited to) ethical beliefs (García et al., 2008), social norms (Lippman, 1991; Markens et al., 2010), familial relationships (Downing, 2005), perceptions of the technologies and pregnancy termination (Green et al., 1993; Markens et al., 1999), attitudes of medical professionals (Press and Browner, 1997) and the perceived impact of parenting a child with a disability (Lawson, 2001). 'Experiential knowledge', that is, knowledge derived from everyday stocks of knowledge, subjective interpretations and meanings accumulated through daily life (Abel and Browner, 1998) has also been acknowledged as an important factor in these decisions. Researchers exploring the influence of experiential knowledge have focused their attention on women's experiential knowledge of pregnancy, and the way in which this informs their usage of RGTs (Markens et al., 2010; Lippman, 1991; Etchegary et al., 2008). However, 'experiential knowledge of disability' (France et al., 2011) has also been noted as having a significant role to play in such decisions (France et al., 2011; Kay and Kingston, 2002; Wertz et al., 1992; Lafayette et al., 1999). Research on the influence of this type of experiential knowledge, however, has been extremely limited, and the findings often conflicting; some researchers found greater support for, and use of, RGTs by individuals with experiential knowledge of disability (Frets et al., 1990; Kay and Kingston, 2002; Evers-Kiebooms et al., 1990), whilst others found less support and usage (Wertz et al., 1992; Lafayette et al., 1999), still others reported ambivalence and mixed usage amongst those with this type of knowledge (France et al., 2011; Etchegary et al., 2008). In spite of these inconsistent findings, there nevertheless appears to be agreement in the literature that experiential knowledge with a given condition has a key role to play in reproductive decisions regarding the (non) use of RGTs.

A key omission of such studies, however, is a detailed exploration of the nature and content of 'experiential knowledge of disability', with researchers tending to treat 'disability' as a homogenous experience across

different conditions. Indeed, descriptions of the experiential knowledge of disability have typically been limited to a presentation of the diagnosis (e.g. 'experience with Down's Syndrome', 'experiences with Cancer'), or very broad experiential categories (e.g. 'experience with disabled children'). This lack of detailed exploration of experiential knowledge of disability is striking given that the range of experiences subsumed under categories such as 'Cancer' or 'disability' is vast. Research has demonstrated, for example, the significant differences that often exist between the experiences of families with children with learning disabilities and those living with primarily physical disabilities. **Von Gontard et al. (2002) for example reported much higher degrees of psychopathology and familial stress in families with children affected by Fragile X Syndrome (which is characterised by both mental and physical impairment) when compared with families living with a child with Spinal Muscular Atrophy (a primarily physical condition). The authors attribute the reported differences between the families to the cognitive and behavioural implications of Fragile X Syndrome for the children living with it which are absent in the example of Spinal Muscular Atrophy. Similarly, the language of 'disability' has limitations when applied to the experiences of those families in which the child's condition involves frequent illnesses or hospitalisations, or is terminal (Tong et al. 2010; Carnevale et al., 2006).** Yet, within the literature on experiential knowledge, these fundamentally different types of experience, subsumed under the title of 'disability', are not explored or accounted for.

Using the conceptual apparatus developed by social model of disability theorists and medical sociologists as a point of departure, this paper will contribute to debates on the role of experiential knowledge of disability in decisions around the uses of RGTs. Drawing on an interview study around reproductive decisions and uses of RGTs with 64 individuals living with an inheritable condition, Spinal Muscular Atrophy (SMA) in their family, an experiential typology: 'experiences with disability', 'embodied experiences of impairment' and 'embodied experiences of illness, death and bereavement' will be developed to highlight the various ways in which experiential knowledge came to be defined by participants in the context of reproduction. By highlighting the way in which participants presented SMA, not in terms of its sub-diagnoses (types I-III), but in terms of the *types of experience* associated with them, this paper will contribute to debates addressing the status and value of this type of experiential knowledge, by pointing to the collapsible and porous nature of diagnostic categories in this context.

Disability, Impairment, Illness and Experiential Knowledge

The fluid nature of 'disability', 'impairment' and 'illness' means that they may be difficult to entirely differentiate, and indeed, life with a particular condition may involve a continual oscillation between all three states. However, for social model of disability theorists, the complete separation of disability from impairment has political significance. As Hughes and Paterson (1997), Morris (1991) and Crow (1996) note, the distinction between 'impairment' (the corporeal aspects of disablement) and 'disability' (its social product) dominated early conceptualisations of the social model of disability and made a neat (albeit reductionist) separation between those aspects of disablement which were of socio-political origin, and those which fell beyond the remit of the social model. For a political project which sought to locate the 'problem' of disability as being exclusively within the social and environmental set-up of society, to draw attention to the experiential aspects of disablement and impairment was to risk confirming that impairment or 'impairment effects' (Thomas, 2004 p.42) were really the problem all along (Shakespeare, 1992: 40), and so give ground to medical models of disability.

Feminist disability scholars, such as Morris (1991), French (1993) and Crow (1996) were amongst the first to make calls to 'bring the body back into' the social model of disability, and to acknowledge the significance of experiential aspects of impairment in shaping disabled people's lives. Further, as Shakespeare (2006) highlights, some impairments effects-such as intractable pain or discomfort- cannot be accounted for, nor indeed removed by, social arrangements, suggesting their incompatibility with social model thinking (Shakespeare, 2006). **Moreover, a burgeoning qualitative literature developed by clinical researchers, has documented the lived reality of family life with children with complex disabilities and conditions which highlights the centrality of bodily experiences ( e.g. Woodgate et al., 2012; Carnevale et al., 2006).** By demonstrating the centrality of impairment to many disabled people's lives, these writers called into question the utility of the heuristic distinction between 'impairment' and 'disability' around which the social model of disability was constructed. Oliver (1996), in response to these debates, tentatively suggested the development of a 'sociology of impairment' as an arena in which to address this omission and interrogate the experiential aspects of impairment whilst simultaneously retaining a critical distance from the medical model of disability (Oliver, 1996: 49). However, for many disability theorists, the separation of impairment and disability is no longer considered as politically progressive (Hughes and Paterson, 1997: 334-5) and as such, social model of disability

theorising has taken new directions in recent years, with increasing acknowledgement of the intersection of impairment and disablement (Shakespeare, 2006).

Whilst the theoretical distinctions between disability and impairment are thus widely contested, the conceptual differences between impairment and 'illness' have been relatively under-theorised by both medical sociology and disability studies. Indeed, theoretically robust distinctions which account for the differing contours of both impairment and illness are still lacking (Oliver, 1996; 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 and strategies of adaptation to it (Barnes and Mercer, 1996). Social model of disability theorists, however, as discussed, largely dealt with the experiential aspects of impairment by bracketing them off in favour of an analysis of the structural origin of disability. As some, albeit limited, dialogue between these two fields has developed however, the similarities, particularly between what medical sociologists term 'chronic illness' and disability theorists term 'impairment' have been emphasised (Mulvany, 2000: 592), and sometimes the terms have been used interchangeably (Oliver, 1996: 40). However, tensions remain, as evidenced in the difficulties reported by those with chronic illnesses in identifying their experiences as an 'impairment' or 'disability' (De Wolfe, 2002: 257). For writers such as De Wolfe (2002), any attempts to subsume the experience of chronic illness into social model thinking will necessarily be inadequate as 'illness' is different to 'impairment' in fundamental ways (De Wolfe, 2002: 262). For De Wolfe (2002), the key difference between 'impairment' and 'illness' rests in the absolute presence of suffering. Unlike impairments, which may be experienced as static and unobtrusive in particular contexts (Shakespeare, 2006), illness, De Wolfe argues, is always experienced negatively and, as such, is a state of embodiment most people would seek to avoid. Whilst impairments may, through the adaptation of society, be transformed into neutral characteristics through their dislocation from disability (Oliver, 1990), the experience of illness may not so easily be transformed or remedied through social change.

Within the literature on the experiential knowledge of disability, and its use in reproductive decisions, however, the experiential constituents of 'disability', 'impairment' and 'illness' are not referenced or considered. This oversight may in part be explained by the general lack of inter-disciplinary dialogue between medical sociology (from where many studies of experiential knowledge of disability emanate) and disability studies, where a differentiation between the concepts has been critical to the advancement of a political

agenda. Indeed, France et al. (2011), in their study of the influence of experiential knowledge of disability in prenatal testing decisions amalgamate a broad range of conditions under the category 'experiences with disability' for their analysis (including Sickle Cell Disease, congenital heart conditions and neural tube defects). Similarly, Etchegary et al. (2008) simply refer to 'experiences of disability' in their exploration of the role of 'vivid' and 'vague' forms of experiential knowledge. Yet, the experiences reported by their participants varied hugely from accounts of life with a child with Down's Syndrome (p. 18) to descriptions of conditions where the child did not survive for more than a few hours (e.g. Trisomy 18) (p. 19). Although within Etchegary et al.'s (2008) study, both of these experiences were analysed as 'experiences with disability', the content and meaning attributed to the experiences by the participants were ultimately very different, and as such and were used to inform contrasting reproductive decisions (Etchegary et al., 2008: 118-9). A lack of exploration of these nuances within participants' accounts of their experiential knowledge overlooks the complexities that they bring to reproductive decisions, and its absence in previous analyses may also (at least in part) account for the conflicts within the body of evidence around the uses of experiential knowledge of disability in RGT decisions (e.g. Wertz et al., 1992; Evers-Kiebooms et al., 1990).

This paper will address this oversight in the literature by presenting an analysis of the ways in which 'experiences with disability', 'embodied experiences of impairment' and 'embodied experiences of illness, death and bereavement' emerged in families' accounts of living with, and making reproductive decisions around, Spinal Muscular Atrophy (SMA). By drawing on in-depth qualitative interviews with 64 individuals with SMA in their family, it will be argued that distinctions between experiences with disability, impairment and illness were key to the way that experiential knowledge of SMA was presented, and consequently the ways in which reproductive decisions were justified.

### Spinal Muscular Atrophy and Reproductive Genetics

After Cystic Fibrosis, SMA is the most common (potentially fatal) autosomal recessively inherited condition in the UK (Dreesen et al., 1998). It is a neuromuscular condition characterised by degeneration of the anterior horn cells of the spinal cord leading to generalised, and often severe, muscle weakness. SMA has been sub-categorised into three distinct clinical 'types' (I-III) with different presentations, ages of onset, severity of muscle weakness and prognosis (ranging from early infantile death in the case of type I to late onset muscle

weakness in adulthood in type III). In spite of these categorisations, however, the boundaries of the types of SMA are widely contested within the medical community and there is a vast overlap in symptoms between them (Dubowitz, 1991, 2008).

It is estimated that one in forty people in the general population are carriers of the deleted SMN1 gene thought to cause SMA (Wirth, 2000). For each pregnancy, two carrier parents have a 25% chance of having a child with SMA and a 50% chance of having a child who will be an asymptomatic carrier. Both prenatal testing and Pre-Implantation Genetic Diagnosis (PGD) are licensed for use in the UK for SMA, and relatives of a person diagnosed with SMA (and their partners) can undergo carrier testing on the NHS to assess their risks of transmitting it. Whilst prenatal testing is also available for SMA, the test cannot accurately predict the type and severity of SMA to be expected. The type of SMA in existing relatives is sometimes cited by genetic counsellors as a rough guide as to the likely prognosis, although there are many cases of different 'types' of SMA being diagnosed within one family (Dubowitz, 1991).

### Methods

Interviews were conducted between 2007 and 2009 with 64 participants who all had at least one person (living or deceased) diagnosed with SMA in their family. Participants were recruited to explore the role that experiential knowledge of SMA played in reproductive decisions and attitudes towards having children with SMA (**Boardman, 2010**). Firstly, participants' stories of life with SMA were elicited before the interviews moved on to a discussion of views around, and uses of, RGTs. Stories of life with SMA, however, were often interwoven with accounts of reproductive decisions. Family members with differing levels of proximity to the diagnosed person (e.g. parent, sibling, grandparent) were included to allow an analysis of different types of experience with SMA and their impact on reproductive decisions (Table 1).

Participants were recruited into the study through the main advocacy group for SMA in the UK, the Jennifer Trust for Spinal Muscular Atrophy (JTSMA, 2011). Recruitment occurred through a variety of channels: through the JTSMA annual conference (n=16), through advertisements placed in their publications (n=16), personal contacts (n=3) and snowball sampling (n=22). Recruitment was also attempted outside the JTSMA, to allow for the possibility that people with SMA might not necessarily identify primarily with their diagnosis. Attempts were made to recruit through disability organisations (Motability and DaDa) as well as personal websites/blogs set up and run by families and individuals, however, these methods only led to the successful recruitment of

four participants. Participants were provided with an information leaflet outlining the aims of the research and the nature of the interview before being asked to participate and were asked to sign a consent form prior to the interview. The research was conducted in line with guidance from the **University of Warwick's** ethical guidelines for research with human participants.

As the participants were geographically dispersed within the UK, interviewing took place through a variety of channels: over the telephone, via email and face-to-face (Table 2). Telephone and face-to-face interviews lasted on average for one hour and ten minutes, whereas email interviews took place over periods lasting from three weeks to eight months. The method of interviewing employed was determined primarily by participant preference, but also took account of their geographical location and the constraints of the research budget. The ethical considerations associated with using these different interview techniques (particularly in the context of being a researcher with a visible disability) are discussed elsewhere (**Brown and Boardman, 2011**). All of the data presented in this paper are derived from telephone interviews. **Whilst data from only five participants are presented in this paper, the experiential categories within which their accounts are located: 'experiences of disability' 'embodied experiences of impairment' and 'experiences of illness, death and bereavement' were developed through an analysis of the whole dataset (64 participants). The participants whose interview excerpts are included within this paper were selected on the basis of their markedly lucid and eloquent portrayals of the overarching themes and experiential categories that cut across the whole dataset.**

Interviews were transcribed verbatim (with names and identifiers removed or changed), and the text responses from the email interviews were compiled into single documents for analysis. A constructivist approach to grounded theory data analysis was used. Initially, 'open coding' (Gibbs, 2007) of the data was carried out which was largely descriptive, before hierarchical coding was undertaken through the use of qualitative data analysis software, Nvivo 7. A process of coding, refinement of concepts (through data interpretation), followed by re-coding and further sampling were carried out over a period of eight months until 'theoretical saturation' had occurred (Glaser and Strauss, 1967).

## Results

### *Disability and the 'Dys-appearance' of Impairment in Reproductive Decisions*



Participants' stories of life with SMA were interwoven with accounts of their reproductive decisions; indeed, the way in which participants conceptualised SMA was critical to shaping how they portrayed their reproductive decisions. Current life with SMA was the 'yard stick' by which to measure, and imagine, the likely impact of SMA. Whilst prenatal genetic testing for SMA is currently unable to predict its severity, participants nevertheless relied heavily on their experiential knowledge of SMA when considering reproduction. For many participants, these accounts were primarily demarcated by experiences with disability. Despite Lamb and Pedon's (2008) focus on SMA as an 'illness' and consequently participants' strategies of 'symptom management' in their exploratory study of SMA, individuals diagnosed with SMA who took part in this study, rarely spoke of their experiences in these terms. Instead, the emphasis was on the way in which their social and physical environment shaped their lives. Rhona, a woman in her twenties diagnosed with SMA type II, described her experiences of disablement 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 mean when I think back to times when [...]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 [something]... 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, disability emerged at specific 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, SMA, was not experienced as disruptive, and only came into her awareness through interactions with a disablist society. 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 as a way of being in the world. For Rhona, the presentation of her experiences with SMA in these terms was key to views around reproduction. She and her husband were hoping to have a child in the near future and decided to forgo carrier testing when it was offered to them by a geneticist:

We decided not to go for it [carrier testing] in the end because we both feel very strongly that someone with SMA can have a good quality of life, and so we were going to go ahead whatever. We've made the same decision about screening for Down's Syndrome, as well, in that we're not going to have it...[...]... They're just disabilities at the end of the day, they don't end your life, they can be overcome if you've got the right sources of support around you and you can access everything you need to. The only time it ever gets hard is when those things aren't in place....but they should be, and so we don't really think that's a good enough....um, that it's *necessary*... to go down the testing route. I've coped with everything, and I'm fine, that's how I look at it anyway.

In spite of the discourses of maternal responsibility which urge compliance with medically prescribed testing of pregnancies (Lippman, 1989; Press and Browner, 1997), as well as the powerful discourses which position disabled women as inadequate mothers (Boardman, 2011; Kallienes and Rubenfeld, 1997), by presenting her experiential knowledge primarily in terms of socially-created disability and minimising both her impairment and the psycho-emotional consequences of disability (Reeve, 2002), Rhona was able to define disability as a situation which could be 'overcome' with appropriate resources, and around which a successful and fulfilling life could be built. **Through the application of this definition of disability, the diagnostic boundaries between conditions became less relevant for Rhona and her husband's decision-making: the testing and screening decisions were not about a diagnosis of SMA or Down's Syndrome per se- but were instead transformed into decisions about life with *disability*. Indeed by framing her reproductive decision in these terms, Rhona was able to present her (and her husband's) refusal of genetic testing for SMA (and her avoidance of screening for Down's Syndrome) as both responsible and positive.**

However, for some participants, their embodied sense of living with SMA was less dormant than that presented within Rhona's account. Whilst the literature on experiential knowledge has made a key distinction between 'embodied' forms of experiential knowledge (a term commonly used to describe the experiential knowledge of those people diagnosed with a particular condition) and 'empathetic' experiential knowledge (referring to the experiences of those close to that individual- typically those doing care work for them) (Abel and Browner, 1998), for families living with SMA, the boundaries between embodied and empathetic experiential knowledge were porous. Indeed, 'impairment effects' (Thomas, 2004) associated with SMA, including fatigue and pain, were felt not only by those individuals diagnosed with the condition themselves, but also by other family members. Family members came to know and understand the effects of SMA, not only through empathic understanding, but through their own bodies, as Sarah, the mother (and primary carer) of a 12 year old girl, Ava (diagnosed with **type II SMA**) commented:

**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...Ava'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....she can't support her weight, so I lift her. You become their muscles and their strength, in effect, for them....you do what they can't...so it is very tiring and I end up thoroughly exhausted at the end of the day if I'm honest.**

For Sarah, caring for daughter meant the blurring of bodily boundaries. Through not just observing, but *becoming* the 'muscles and strength' for another body, Sarah became two bodies in one in a quasi 'Möbius strip' (Grosz, 1994), highlighting the inseparability of embodied and empathetic experiential knowledge and the 'inflection of mind into body and body into mind' (Grosz, 1994: xii). Stress, exhaustion and pain featured heavily in many family members' accounts of living alongside SMA, highlighting the way in which 'impairment effects' (Thomas, 2004) may not be experienced solely by those diagnosed with SMA, but also by those providing care. For Sarah, unlike Rhona, the bodily effects of Ava's SMA were critical in shaping her experiential knowledge of SMA and consequently her reproductive decisions, as Sarah commented:

I think I'm just getting tired now, and I see Ava getting more tired with it too as she gets older, I definitely think it's affecting us more.... Mark [husband] and I don't really know where to go

from here. At first we thought we didn't want any more [children] but I now feel it's something I'd like to look at again...but it's just....I guess we're torn, because we have to think about what we can cope with. It's not just about 'having a child in a wheelchair', you know, I think I could have coped with that... it's the...tiredness, the back pain you get with it, that makes it hard for all of us. So yeah, no idea what we do with that one! [laughs]

Whilst participants' accounts of life with SMA were often framed by an oscillation between descriptions of disability and embodied experiences of impairment, when accounting for reproductive decisions, descriptions of SMA came to be more clearly defined in terms of experiences of disability or those of impairment. For those participants who described SMA primarily in terms of disability (and consequently in terms of social and environmental factors amenable to manipulation) reproductive decisions were clarified and bolstered through experiential knowledge. Experiences of disablement were linked with SMA but were presented as a non sequitur- when physical and social barriers were removed, or at least minimised, SMA could be successfully lived with. However, for those participants whose experiences with SMA were presented in terms of embodied experiences of impairment, reproductive decisions were described as much more uncertain. Whilst Sarah acknowledged that Ava's SMA could be experienced as unobtrusive in particular contexts, *'you do get those days when everything's in place and you're able to forget about the SMA'*, she also acknowledged that SMA goes beyond an experience of disability: *'It's not just about having a child in a wheelchair'*, and is instead felt in the bodies of all the family members.

#### *The Presence of Suffering: Illness, Death and Bereavement in Reproductive Decisions*

For some participants living with SMA, their experiences were described as invariably, and inevitably, involving suffering. The 'suffering' associated with SMA, furthermore, transcended medical classifications. Rather than being confined to the severe types of SMA, suffering was more closely correlated to specific embodied experiences. Some of the physical complications and medical interventions associated with SMA, including chest infections, difficulty breathing, going through, and recovering from, spinal surgeries as well as the deterioration of abilities over time were all experiences that participants aligned with suffering. However, these experiences were experienced across the spectrum of different types of SMA. Indeed, seven participants

with type II in their families had experienced the death of a relative from SMA due to respiratory difficulties, in spite of premature fatality usually only being associated with type I SMA, according to medical classifications.

For families who experienced the death of a relative with SMA, the suffering was both innate and intractable. Unlike those experiencing embodied impairment or disability, this suffering could not be displaced by social or environmental changes, and was not context-dependent. Patricia and Fraser are in their 50s and experienced the deaths of two of their children, Max and Annabelle, from SMA type I at 8 and 10 months of age respectively. For Patricia, the deaths of their children in early infancy marked the end of a relentless and futile battle with the condition which she 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? ...[...]...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...[pause]...because I thought they were suffering.

(Patricia)

For Patricia and Fraser, the experience of watching Max and Annabelle suffer and eventually die from SMA type I had a profound impact on their reproductive decisions:

The ability to undergo prenatal testing [for subsequent pregnancies] was a godsend to 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 ...[...]...The test is there and I think every parent should take advantage of it....I mean they were such beautiful wee children but they lived their lives in such pain, there was no quality there...no, it was an easy decision for us.

(Fraser)

For Patricia and Fraser, as for all of the parents in the study who had witnessed their child die of SMA, the suffering associated with SMA was immense, undeniable and experienced by the whole family. For them, as for Rhona who presented her experiences of SMA in terms of disability, their experiential knowledge was key to

confirming where their reproductive responsibilities lay, even though it led them to an entirely different reproductive decision.

Acute illness and subsequent death, however, were not the only embodied experiences associated with SMA that came to be presented as aligned with suffering in participants' accounts of their reproductive decisions. Acute periods of degeneration in symptoms of SMA over time could also prompt an individual to re-classify their previously static impairment as an 'illness'. Emily is in her late 40s and was diagnosed with **type II** SMA in infancy. Whilst Emily's SMA remained relatively static for the majority of her life, a gradual decline in her abilities caused her to re-conceptualise SMA:

I'm much more aware of my condition now than I was when I was growing up...like my skin's breaking down now... [...]...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...[...]...before it was just how I was, you know, it was in the background....It used to just be a disability, but now I'm increasingly aware that I'm living with a death sentence.

Whilst SMA is not defined as a degenerative condition within the medical literature, there is acknowledgement that people living with SMA may nevertheless experience decline in their abilities over time as a consequence of the endurance of their SMA symptoms combined with natural ageing processes (De Groot and De Witte, 2005). For Emily, this deterioration brought SMA into the forefront of her awareness- 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'. Unlike Rhona, whose sense of impairment and physical difference arose from her interactions with a disablist society, for Emily, it was her own sense of 'dys-embodiment' that kept SMA continually in her awareness. This marked decline caused her to re-evaluate her views of SMA, from 'just a disability' to 'a death sentence' within which suffering was inevitable. This change in conceptualisation of the condition was inextricably linked to her decision to forgo childbearing:

I used to get upset sometimes that I'd never had a child myself, you know, when I had the chance, but the way I see it now, knowing what I know now about it [SMA], it was the best thing....I couldn't risk that they'd get it [SMA]. If I had a child with SMA, I'd just....crucify myself forever... because I'd have to watch them suffer and I'd know that I'd done it to them, and I don't think I could live with that.

For Emily, her re-conceptualisation of SMA as a condition which causes people to suffer made her clear that she had a responsibility to prevent its recurrence in future generations. The process of deterioration (and the resulting oscillation between dys-embodiment and attempts at re-embodiment) caused Emily to describe SMA in terms of an illness, rather than the resulting level of impairment per se. Indeed, participants diagnosed with the (medically defined) most severe form of SMA (**type I**) and yet had experienced their SMA as a static impairment since birth or early infancy, did not invoke references to suffering in their accounts of reproductive decisions in the same way as those who had experienced deterioration. The degree of suffering experienced by an individual with SMA could thus not be 'read off' from their level or severity of impairment in an unproblematic way, but rather, particular types of experience (including deterioration in health and abilities, death and bereavement) came to be more closely aligned with suffering than others and were presented as such in decisions to prevent SMA in future generations.

### Discussion

Within this paper, the key experiential categories within which participants posited their lives, 'experiences of disability', 'embodied experiences of impairment' and 'experiences of illness, death and bereavement' have been presented in order to demonstrate their strategic uses within accounts of reproductive decisions for families living with SMA. For many participants, and particularly those who were diagnosed with SMA, 'experiences with disability' were presented as predominantly shaping their lives. Disability in this context was defined as experiences of restriction and oppression that arose from the social and spatial environment, and, specifically, the points at which this environment failed to cater for their needs. By defining the 'problem' of SMA not as physical impairment, but instead as the inadequacies of a disablist society, participants who described their experiences in such terms were able to present their decisions to refuse the use of RGTs as responsible, irrespective of discourses of maternal responsibility which demand compliance with the medical

management of pregnancies (Press & Browner, 1997). Societal failings were described as the 'real' issue to be addressed, not the presence, or absence, of SMA in the foetus. By presenting their reproductive decisions and experiential knowledge in these terms, participants such as Rhona diminished the significance impairment in shaping their lives.

For other participants, however, experiences with SMA were less easily attributable to the inadequate adaptation of society. For these participants, embodied experiences of SMA were described as (at least potentially) problematic in themselves and highly context-dependent. For Sarah, for example, life with SMA was marked by a frantic oscillation between her daughter, Ava's, impairment being in the background and foreground of their lives, depending on both the physical and social environment as well as the changeable nature of impairment effects, which were felt both in Sarah's, and Ava's body. For these participants, reproductive decisions were marked by higher degrees of uncertainty than those of participants who presented their experiences with SMA primarily in terms of (socially and environmentally pliable) disability. The inability of genetic tests to diagnose the severity of SMA, together with a lack of guarantee of a suitable environment for a child with SMA meant that Sarah felt uncertain where her reproductive responsibilities lay, particularly in a context where maternal responsibility in pregnancy is culturally aligned with compliance with medical testing (Press & Browner, 1997), leaving her trapped between competing concerns and demands.

For the participants whose experiences with SMA were characterised primarily by illness, pain, deterioration in abilities, death or bereavement, however, reproductive decisions were presented as a more clear-cut process, with responsibility lying with prospective parents to prevent the suffering associated with SMA in future generations. Unlike embodied experiences of impairment, and experiences with disability, the participants who presented their lives with SMA in these terms suggested that the suffering associated with SMA was both intractable and inevitable. Unlike impairment effects, such as the fatigue described by Sarah, this suffering could not be manipulated or alleviated, whether by medical, social or environmental intervention, and consequently the certainty of the suffering was presented as a key justification to prevent the recurrence of the condition of future generations, and all participants who described SMA in this way reported their reproductive decisions in this way.



Whilst in terms of the political advancement of disability politics, neat divisions between ‘disability’, ‘impairment’ and ‘illness’ may not be desirable (Shakespeare, 2006), by highlighting the way in which participants come to *privilege* particular experiential categories, or versions of their experiential knowledge when accounting for their reproductive decisions, draws attention to the powerful discourses of parental responsibility and disability that govern such decisions (Kallienes and Rubenfeld, 1997) as well as the need to consider the vastly different types of experiential knowledge that can exist around one particular condition. Indeed, previous research on the role of experiential knowledge of disability in reproductive decisions has produced conflicting results which can be in part explained by a focus on medical diagnoses or overly broad categories such as ‘experiences with disability’ (France et al., 2011) to characterise this type of knowledge, rather than a detailed analysis of the nature of the experiences subsumed within these groupings. This type of analysis is crucial, particularly as many tested-for conditions such as SMA present in highly variable ways and are often associated with an uncertain prognosis, which invariably shapes experiential knowledge of the condition. Moreover, as France et al. (2011) highlight, individuals sometimes also draw on their experiential knowledge of conditions other than the one being tested for when considering their ‘experiences with disability’ in reproductive decisions. Whilst France et al. (2011) do not explore the possible reasons for this, this paper highlights the way in which experiential categories can transcend diagnostic boundaries (in this instance between the vastly different types of SMA). For would-be parents, the key to being able to imagine (and consequently appraise) a child’s life in the context of a congenital condition may not necessarily be contained within a specific diagnosis, but instead within the type of experience associated with it, e.g. whether it is a condition with which the child will be disabled, experience pain or die prematurely. Diagnostic boundaries may thus become collapsible in the assessment of experiential knowledge of disability.

This finding has implications for the way in which would-be parents are counselled in the context of reproductive decisions and RGTs. Indeed, such parents may benefit from an exploration of their experiential knowledge, not only of the condition being tested for, but also of other conditions they have experiential knowledge of, with similar experiential properties to the one tested for.

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### Declaration of Competing Interests

None

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