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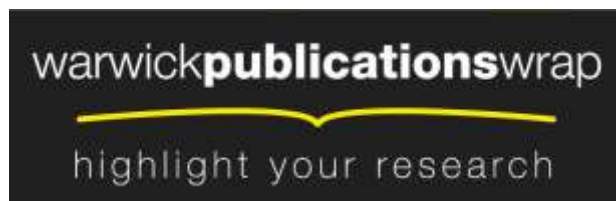
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# The expressivist objection to prenatal testing: The experiences of families living with genetic disease

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## ABSTRACT

The expressivist objection to prenatal testing is acknowledged as a significant critique of prenatal testing practices most commonly advanced by disability rights supporters. Such writers argue that prenatal testing and selective termination practices are objectionable as they express disvalue not only of the foetus being tested, but also of disabled people as a whole, by focusing exclusively on the disabling trait. While the objection has been widely critiqued on the basis of its theoretical incoherence, this paper highlights the way in which it, nevertheless, is a significant mediator in decisions around the use of reproductive genetic technologies. By drawing on 41 in-depth qualitative interviews (drawn from a sample of 61) conducted in the UK between 2007 and 2009 with families and individuals living with a genetic disease, Spinal Muscular Atrophy (SMA), this paper highlights the ways in which expressivist objections feature prominently in the reproductive decisions of families living with SMA and the significant emotional burden they represent. While the literature on the expressivist objection has focused on the reproductive decisions of those undergoing prenatal testing for a condition of which they have little (or no) prior knowledge, the context of intimate familial relationships and extensive experience with the tested-for condition fundamentally alters the nature and impact of expressivist objections within families living with an inheritable condition. By focussing on the reproductive decisions of families living with SMA and their strategic management of the expressivist objection, this paper will address the call, made primarily by disability rights supporters, for 'experientially based' (as opposed to medical) information about the tested-for disability to be made available to would-be parents considering selective termination. It will be argued that parents' experiential knowledge of the tested-for disability can, in fact, amplify expressivist objections to prenatal testing, and thus paradoxically constrain, rather than facilitate, reproductive decisions.

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## 1. Introduction

As the capacities of reproductive genetic technologies have expanded in recent years, so too have the number and nature of reproductive decisions facing would-be parents. While such technological advancements have been heralded as increasing the reproductive autonomy of such parents, the ways in which these decisions are actually made and experienced have come under scrutiny by researchers, who have problematized the notion of 'choice' in which the technologies are couched, instead pointing to the potentially constraining effects of the technologies (Lippman, 1991). The so-called 'expressivist objection' has been amongst such critiques of prenatal testing. The term 'expressivist objection'

(hereon referred to as the 'EO') was coined by Buchanan (1996) and refers to:

The claim...that...the commitment to developing modes of intervention to correct, ameliorate, or prevent genetic defects expresses (and presupposes) negative, extremely damaging judgements about the value of disabled persons.

(Buchanan, 1996: 28)

As such, the EO, as a critique of genetic testing practices, has been most often advanced by disability rights supporters (e.g. Parens and Asch, 2000). Such disability rights supporters have questioned prenatal testing and screening practices (and the subsequent offer of selective termination of pregnancies where genetic differences are detected) on the basis that they not only express a negative valuation of the foetus being tested, but also of the lives of

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people living with disabilities more generally (Parens and Asch, 2000; Saxton, 2000). Following this argument, the selective termination of a pregnancy is considered objectionable on the grounds that the particular trait (the disability) comes to represent the entire foetus, rendering immaterial the complexity of all its other traits (Asch, 2000; Saxton, 1997).

While the theoretical coherence of the EO, and in particular its claim that prenatal testing practices are capable of 'expressing' any message, has been called into question by various scholars, both within and without the disability rights community (Shakespeare, 2006), there remains a lack of empirical evidence exploring its impact, particularly amongst people with disabilities (Madeo et al., 2011: 1779). A select number of studies have explored the attitudes of people with the inheritable disabilities to genetic testing (e.g. Middleton et al., 1998; Gollust et al., 2003), and more recently, researchers have considered how the EO may be shaped by, and managed within, the broader context of the disabled person's family, and familial experiences with the condition (Barlevy et al., 2012; Raspberry et al., 2011; Helbig et al., 2010; Kelly, 2009). While critics have argued that prenatal testing and termination decisions are usually made in the private sphere and, as such, are incapable of communicating negative messages to disabled people (Murphy, 2011), families can be described as arenas of 'reproductive accountability' and 'public' reproductive decision-making (Burgess and D'Agincourt-Canning, 2001). They are sites in which reproductive decisions may be subject to family scrutiny, and where there can be a (felt) need to justify decisions taken (Downing, 2005; Cox, 2003). Moreover, disabled people and their families usually approach genetic testing decisions equipped with extensive 'experiential knowledge' (Abel and Browner, 1998) about the condition being tested for (Kelly, 2009; Raspberry et al., 2011). This experiential knowledge, as well as the familial context of reproductive decision-making fundamentally alters the nature and power of the perceived messages expressed by prenatal testing technologies. Yet, a clear distinction between *this* context and that of testing decisions made by the general population with no prior knowledge of the tested-for condition has not been widely acknowledged within the EO literature, in spite of the different implications these scenarios have for experiences of the EO. Indeed, the experiences and views of disabled people and their families living with an inheritable condition have a lot to offer debates around the EO. Where this issue has been explored in the literature, the focus of the studies has either exclusively been on those individuals diagnosed with the condition themselves (e.g. Helbig et al., 2010), or their family members (e.g. Raspberry et al., 2011; Kelly, 2009) with very few studies addressing both (e.g. Barlevy et al., 2012). Consequently, the question of how differing ways of knowing genetic disease inform reproductive decision-making and perceptions of the EO within families has been hitherto neglected. Such an analysis can address the question of how differing levels of experiential knowledge of the condition being tested for can impact on perceptions of the expressive potential of testing decisions, and add to policy debates regarding the value of such knowledge in prenatal testing decisions.

This paper will address the above outlined gap in the literature by drawing on an in-depth interview study (61 interviews with 59 participants) with people living with an inheritable condition, Spinal Muscular Atrophy (SMA) in their family (Boardman, 2010). Through the accounts of a sub-sample ( $n = 41$ ) of these participants, I will argue that while the theoretical basis of the EO may be disputed, its existence and 'felt presence' amongst families living with SMA significantly influences the way they approach- and manage- prenatal testing decisions. For such families, the EO not only has a significant- and sometimes burdensome-emotional impact, it was also described as having a constraining effect on

reproductive decision-making, which had to be carefully negotiated.

## 2. The expressivist objection

Disability rights supporters have been amongst those who have most passionately advanced the EO as a way to critique the discriminatory attitudes and beliefs they deem to underpin prenatal testing practices as well as the messages they perceive to be sent by them to disabled people (Parens and Asch, 2000; Saxton, 2000; Wendell, 1996). In particular, disability rights supporters have critiqued the incompatibility of prenatal testing technologies and the practices surrounding them with the unconditional acceptance of children, irrespective of their genetic traits. To test for disabling traits in the foetus, and then to base selective termination decisions upon this information, is, according to Asch (2000), to allow the disabling trait to 'trump' all other (as yet unknown) characteristics of the foetus. For many disabled people, this prioritising of disability over and above all other traits is echoed in their daily experiences in a profoundly disablist society:

As with discrimination more generally, with prenatal diagnosis a single trait stands in for the whole, the trait obliterates the whole. With both discrimination and prenatal diagnosis, nobody finds out about the rest. The tests send the message that there's no need to find out about the rest.

(Asch, 2000:13)

For disability rights supporters, the medical profession (inadvertently or otherwise) reinforces and recycles these negative messages through the processes of prenatal diagnosis and selective termination by counselling prospective parents (following a positive prenatal diagnosis) only on the medical complications associated with that disability, while usually ill-equipped to offer insight into the daily realities of life with that particular disability, as Williams et al. (2002) demonstrated in relation to Down's Syndrome counselling. For feminist writers as well, the very existence and consequent routinisation of prenatal testing technologies suggests an implicit responsibility to use them (and thereby to test and terminate affected fetuses) (Lippman, 1991; Markens et al., 2010; Clarke, 1991), potentially making it harder for those parents who wish to continue with an affected pregnancy to justify their decision as 'responsible' (Barlevy et al., 2012: 36).

These critiques of prenatal testing and screening practices, however, have not been universally accepted. Within the field of disability studies itself, the EO has come under harsh scrutiny and there is an acknowledgement that the EO is not supported by the whole of the disability community, many of whom see the EO as an over-simplification of their diverse views on this topic. However, writers such as Shakespeare (2006) have argued that to assume that prospective parents wish to terminate pregnancies affected by disability primarily on the basis of ignorant, prejudiced or otherwise negative attitudes towards disability is to simplify what are often highly complex and emotionally charged decisions. Indeed it is not, Shakespeare (2006) points out, a contradiction to both terminate a pregnancy affected by disability (for example, if a person feels that they could not provide the additional resources required to raise a disabled child), and to simultaneously uphold respectful and supportive views of disabled people within society. Indeed, for many disability rights supporters, the concerns of EO proponents with the decision-making processes of prospective parents and the 'messages' these decisions are deemed to send not only vilifies would-be parents but also diverts attention away from the social and political context in which reproductive genetic

**Table 1**  
The relationship of participants to diagnosed member, gender and number spontaneously mentioning the expressivist objection during interview out of whole sample  $n = 61$ .

Participants	Numbers	Gender		Mentioned EO
		Female	Male	
Diagnosed with SMA	25	21	4	19 (76%)
Sibling of person with SMA, without SMA themselves	7	4	3	5 (71%)
Parent of person diagnosed with SMA	24	21	3	16 (67%)
Grandparent of person diagnosed with SMA	1	1	0	0 (0%)
Son or daughter of person diagnosed with SMA	2	1	1	0 (0%)
Partner or spouse of person diagnosed with SMA	2	1	1	1 (50%)
Totals	61	49	12	41 (67%)

technologies are developed, advanced and supported (Shakespeare, 2006).

In spite of these debates surrounding the EO, there remains a notable lack of empirical evidence exploring its conceptualisation and impact amongst those who use prenatal testing technologies, as well as amongst disabled people themselves. While other studies have demonstrated the existence of a philosophical concept through empirical data e.g. Katz Rothman's exploration of the 'tentative pregnancy' (1986), relatively few exist around the EO. A recent attempt to ground the EO in empirical data has been made by Klein (2011) who used data on the medical profession's handling of prenatal testing and termination practices to conceptually delineate the EO. However, the views of would-be parents and/or disabled people were not included in this empirical exploration. Indeed, many writers on the EO appear to have in mind testing decisions made by people with no prior experience of the condition being tested for, with little attention paid to the families living closely with that condition (who are also the intended users of prenatal testing technologies) and in spite of the acknowledged significance of 'experiential knowledge' to prenatal testing decisions more broadly (Etchegary et al., 2008; Cox, 2003; Abel and Browner, 1998; Markens, 2010; Katz Rothman, 1986; Rapp et al., 2001; Lippman, 1999). Moreover, disabled people often appear within the literature on the EO primarily as the recipients of the negative messages sent by prenatal testing as opposed to the prospective parents making active decisions about technology usage in their own reproductive decisions. This is in spite of media stories and reflective accounts suggesting that the EO is indeed a significant consideration for many disabled people (e.g. Barford, 2011; Atkinson, 2008; Boardman, 2011).

This paper will address some of these shortcomings of the EO literature by presenting in-depth interview data from a sample of 41 participants either diagnosed with, or with a family member diagnosed with, the genetic neuromuscular condition, SMA. The study explored participants' conceptualisations of their genetic risk and attitudes to reproductive decision-making. While the EO was not specifically asked about within the interview schedule, data pertaining to the objection emerged spontaneously within participants' accounts and it is these data that will be presented.

### 3. 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). All infants with Type I SMA, and some people with Types II and III SMA,

require respiratory support (either through a ventilator or BiPaP machine) due to weakness of the muscles used to support breathing. All people diagnosed with SMA are at risk of complications from chest infections due to this compromised respiratory function. Most people with Type II SMA are full time wheelchair users from early childhood and commonly experience spinal curvature and joint contractures (usually requiring surgical correction and intensive physiotherapy). People diagnosed with Type III SMA may remain ambulant up until the third decade of life when a wheelchair usually becomes necessary. In spite of these categorisations into types, however, the boundaries of the diagnoses 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 to assess their risks of transmitting it.

While prenatal testing is available for SMA, the test cannot accurately predict the type (and consequently the severity) of SMA to be expected in the child. The severity of SMA already diagnosed within the family is sometimes used by genetic counsellors as a 'rough guide' as to the likely severity of SMA likely in future generations. However, there are many instances of different 'types' of SMA being diagnosed within one family that have been reported in the clinical literature, e.g. Dubowitz (1991), as well as within the sample of participants in this study. As Raspberry et al. (2011) acknowledge, prenatal testing decisions are highly contingent on the specifics of the condition being tested for, with conditions involving early infantile death (e.g. Tay Sachs) resulting in very different attitudes and decisions than prenatal testing for relatively milder impairments e.g. hereditary deafness (p. 997). However, SMA has been described as a neurological condition with a vast variability in symptom expression (Dubowitz, 1991) and can present anywhere in the spectrum from a terminal condition in infancy to a relatively mild adult-onset impairment. This variability, together with the inability of prenatal testing technologies to accurately predict where on this spectrum a future child's SMA will lie, makes decisions around prenatal testing particularly fraught and heightens the associated social and ethical dilemmas, of which the EO is one.

### 4. Methods

Interviews were conducted between 2007 and 2009 with sixty-one 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 decision-making 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, reproductive technologies. 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 decision making (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, through disability organisations (Motability and DaDa) as well as personal websites set up and run by families, 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. Ethical approval for the research was granted by The University of Warwick's Research Ethics Committee.

As the participants were geographically dispersed within the UK, interviewing took place through a variety of channels: over the telephone (75%), via email (17%) and face-to-face (8%). 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 into account 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).

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). While the EO to prenatal testing and selective termination was not specifically asked about within the interview schedule, it emerged spontaneously in 41 (67%) of the participants' accounts (see Table 2). All data pertaining to the EO were coded together during analysis, and it is the analysis of these data (41 transcripts) that is presented in this paper. The participants whose voices are quoted within this paper

all opted to participate in a telephone interview (although the full range of interview techniques – email, telephone and face-to-face – were used within the sample of 41 participants included in this analysis). Their accounts were selected for presentation as they articulately encapsulate the key themes that emerged from the analysis of the EO data (41 transcripts).

## 5. Results

### 5.1. The expressivist objection and reproductive decision-making

Within participants' accounts of their views around reproductive decision-making, the EO was frequently mentioned in various forms. For some participants, prenatal testing was described as a form of 'genocide' (Hannah, diagnosed with Type I SMA) which communicated very clear disparaging messages about the value of people with SMA, while for others, expressivist concerns – though considered valid and relevant – were conceptualised as an unfortunate 'by-product' of processes that were 'necessary' in facilitating the reproductive decision-making of families living with SMA (Thomas, able-bodied sibling of Demi, diagnosed with Type II SMA). Moreover, the EO was most frequently referenced by those individuals who were either diagnosed with Type II SMA themselves, or who had Type II SMA in their family, with 56% of the EO sample belonging to this group (see Table 2 for information regarding the Type of SMA affecting the EO sample). What was clear from all of the interviews however, was that expressivist concerns around prenatal testing took on a different character and role within families living with an inheritable condition to those described in the literature on the EO. Indeed, the accumulated wealth of experiential knowledge about the condition within the family appeared to alter the nature and content of the message sent by prenatal testing in a fundamental way. It was common, for example, for participants in this study to entirely align the identity of a (hypothetical) foetus diagnosed with SMA with their existing family member with SMA, so much so that they were often conceptualised as the same person. Beth is the able-bodied sibling of Melissa, who has SMA Type III. During her interview, Beth discussed her feelings about undergoing prenatal testing for SMA during her pregnancy with her daughter (now 18 months):

I thought long and hard about whether or not I wanted to have the testing, and in the end I just thought, why would you want to stop yourself from having another Melissa? You know, she's absolutely fantastic, she's got such mental strength, you know she's intelligent, she's bright, she's got a fantastic personality...she does have physical problems but there isn't anything else wrong with her and so in the end I decided it [prenatal testing] wasn't something I was really bothered about.... I'd be perfectly happy having another Melissa!

While critics of the EO have been at pains to demonstrate the way in which disability cannot be considered an 'identity constituting' feature – an observation that undermines the very logic of the EO (Edwards, 2005; Baily, 2000) – within the accounts of families living with SMA, the possibility of having another affected family member could not be disentangled from their experiences and knowledge of their current family member's SMA. Even though it was evident in Beth's comments that she did not view her sister's identity entirely in terms of SMA and was keen to highlight all of Melissa's positive personality traits, the hypothetical foetus diagnosed with SMA that Beth referenced was nevertheless so closely associated with Melissa that it became 'another Melissa', even being assumed to possess all of her personality traits. In spite of the medical profession being unable to accurately predict the severity

**Table 2**  
Type of SMA in family and gender of those mentioning the EO  $n = 41$ .

Participants	Type I	Type II	Type III	Female	Male
Diagnosed with SMA	1	12	6	17	2
Sibling of person with SMA, without SMA themselves	1	3	1	3	2
Parent of person diagnosed with SMA	7	7	2	14	2
Partner or spouse of person diagnosed with SMA	0	1	0	0	1
Totals	9	23	9	34	7

of SMA through prenatal testing, the severity of an existing family member's SMA was nevertheless conceptualised as the yardstick by which future lives with SMA could be assessed. This was so much so that the (hypothetical) foetus diagnosed with SMA came to be conceptualised as an exact replica of that family member (Boardman, *in press*); the foetus became 'another Melissa' or 'another David' etc. As Cox (2003) has highlighted, for families living with genetic conditions (unlike women undergoing antenatal screening tests) 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. This 'personalisation' of genetic risk perceptions in relation to the family member with SMA has implications for the way in which the expressive potential of prenatal testing was understood and responded to; when a hypothetical foetus with SMA is conceptualised as an existing family member, then decisions surrounding the selective termination of that foetus take on new meanings, both for the person with SMA, and the prospective parents.

Throughout the interviews, it was apparent that participants were acutely aware of the possible ways their reproductive decisions could be interpreted by others, and many described it as impacting on the reproductive choices they felt able to justify. When prospective parents from the general population face selective termination decisions following a prenatal diagnosis of a screened-for condition, they may manage this highly emotional decision by reference to the intolerable suffering associated with the condition being tested for, and thus invoke altruistic justifications for termination of affected pregnancies (Kaplan, 1999). This argument, however, proved more difficult for some families living with SMA to sustain, particularly if they had witnessed a family member (or themselves) live a fulfilling and successful life in spite of the difficulties associated with their SMA. Natalie is in her early thirties and has Type II SMA. She described her competing sense of responsibility to both adequately parent a child with SMA, but also uphold the value of life with SMA, in the context of a society in which it is devalued:

It'd be so hard if I had a child with SMA, because I genuinely believe that an SMA child needs two able-bodied parents to do all the care that they need.... and yet if I, of *all* people, had a termination for SMA, I'd be sending completely the wrong message about life with SMA, wouldn't I? I mean I live with it day in day out, so if I rejected a child with it, it'd be me saying.... (sigh) I just feel like I spend a lot of my life trying to say to people that SMA's not that bad you know, you *can* live with it, you *can* have a normal life with it... so if I did that[termination], I would feel like I was, I guess, de-valuing my life, in a way.... And not only *my* life, but everyone's life who's got SMA. It'd be saying 'yes this is the right thing to do for babies with SMA', and people might take that seriously, you know, because I should know! [Original emphasis]

Natalie appeared to struggle to reconcile her concerns about raising a child with SMA (while having SMA herself) with her feelings about the need to affirm the value of life with SMA, not only for herself, but for all people living with the condition. For Natalie, the responsibility to consider the expressivist potential of prenatal testing and selective termination is heightened for people who have SMA: her decision about whether to use the technologies could be interpreted as an authoritative statement on what life is like with SMA, and as a template of action for others to follow. This sense of heightened responsibility for reproductive decisions in the context of living with the condition being tested for is reflected in the accounts of other disabled people considering reproduction

(Barford, 2011; Atkinson, 2008), and points to the way in which expressivist concerns can add a further layer of complexity to such decisions, both for disabled people, and their family members, leaving them trapped between competing concerns and responsibilities (Boardman, 2011, 2014). Some participants, however, used strategies to actively negotiate these competing responsibilities within their reproductive decisions, and it is to the accounts of these decisions that I shall now turn.

## 5.2. Negotiating the expressivist objection in reproductive decision-making

While some participants described their felt sense of the EO within their reproductive decision-making (particularly those who described hypothetical or anticipated reproductive decisions), other participants used strategies to actively manage the implications of the EO, for themselves and for their family members, when accounting for their decisions. These strategies were most often employed by participants describing reproductive decisions already taken. Kate is in her late 30s and has a seven year old son, Daniel, diagnosed with SMA Type II. Kate described her desire to have another child, but also her unwillingness to 'test and terminate', a process which she differentiated from Pre-Implantation Genetic Diagnosis (PGD), of which she was undergoing her third cycle at the time of the interview. PGD involves the creation of embryos through IVF technologies, with genetic testing undertaken at the embryonic stage. Embryos affected by SMA are discarded, with only unaffected embryos being transferred into the woman's uterus for gestation.

We always knew we wanted a second child... and we also knew we couldn't handle having another Daniel on our hands because we just pour all our lives into looking after him and we couldn't do that with another one, with Daniel as well. But the only way we could know that it wouldn't happen again would be to um test and terminate, and... I just didn't think I could do it....it'd be like getting rid of Daniel, that's how I'd look at it. And how would I explain that to him, you know, when he's older? How could I say that we got rid of your brother or sister because they were like you? It would just be like saying.... well wiping him away really. Saying he wasn't really what we had wanted. So when we couldn't conceive naturally and we had to go down the route of IVF and the PGD anyway, well that was great because it took away that issue for us. It wasn't like we were terminating for SMA anymore because it was out of our hands...the fertility problems meant that we didn't have to go down that route of having to end a pregnancy affected like Daniel.

Kate's account appears to mark a conceptual distinction between disposing of an affected embryo (in the case of PGD) and an affected foetus 'like Daniel' in the case of prenatal testing (which usually takes place between 10 and 15 weeks of pregnancy for SMA, depending on the technique used). Such prenatal testing and selective termination, Kate believed, would have communicated a negative message to Daniel about the value of his life. While PGD is not positioned within the medical and scientific literature as an infertility treatment (Franklin and Roberts, 2006), by presenting her transition to using PGD not as a decision, but as something Kate *had* to do in order to conceive, she emphasises her lack of real choice in the process, and in doing so, eliminates the possibility of Daniel later interpreting her actions as a rejection. By doing this, Kate could navigate the complex terrain of the EO and maintain her 'relational responsibilities' to her son (Burgess and D'Agincourt-Canning, 2001). While the existence of prenatal testing and PGD, as well as critiques of the EO, are often justified by reference to the

need to prioritise reproductive autonomy for prospective parents over and above the need to protect against the emotional harm done by them (Edwards, 2005; Murphy, 2011), as Kate's account reveals, expressivist concerns, can, paradoxically, constrain the reproductive agency of would-be parents by closing down particular reproductive options, or ways of accounting for them. Indeed, for Kate, when taking into consideration the EO and the significant emotional impact her decision to test and terminate could have, both for herself and her son, the ways she could present her reproductive decision as responsible and also as upholding the value of her son's life, became severely limited. The only way Kate could manage this complexity was to conceptually remove herself as the decision-maker, and consequently absolve herself of the responsibility for a decision associated with great emotional, ethical and practical implications, whichever way it was taken.

While thus far, the data presented in this paper have been derived from interviews with people living with Types II and III SMA, for some families living with Type I SMA, and particularly those who had experienced the premature death of a family member, the expressive capacity of genetic technologies to prevent the recurrence of SMA, while acknowledged, was presented as less significant in the context of reproduction. Fraser is in his fifties and experienced the deaths of his first two children, Ciaran and Eve, to SMA Type I before their first birthdays. He described his feelings about the use of prenatal testing and selective termination to ensure that his third child did not have SMA by contrasting it with the decisions facing families affected by Type II SMA:

I think that if you've got Type II then there's a big dilemma in the family. If you've got a Type II that's ten years old and you're expecting again, you know I've heard Type IIs say 'would you have gotten rid of me then if you'd had the chance?', you know? It's different when it's Type II in the family because...they live. They can lead perfectly full, happy lives...which was just not going to be the case for my children. Ciaran and Eve both suffered and died....I don't think anyone could see a dilemma for wanting to stop that. No one could say to me that if I did [prenatal testing and selective termination] that I didn't love and want my children to live because they had SMA. No, of course I did....but the fact is that they weren't going to live, so yes, although I wanted Ciaran and Eve, I also wanted rid of the...horror of what happened to them...and not go through that again. I don't see that anyone could take issue with that...not even a Type II.

Fraser's comments strike at the heart of one of the major criticisms that has been levelled at the EO: namely, its inconsistent applicability. As Shakespeare (2006) has argued, there are often certain circumstances in which disability rights supporters 'waive' their objection to prenatal testing, with the severity of the condition affecting the foetus being one such circumstance (p. 93). Fraser's conceptual distinction between Type II SMA, which, as he points out, can be lived with successfully, and Type I SMA which, for his children, involved grievous suffering and premature death, highlights the point to which Shakespeare (2006) refers. Indeed the lack of evidence for the EO in Barlevy et al. (2012)'s study of families living with QT Syndrome (a potentially fatal condition involving cardiac arrhythmia) suggests that EO concerns may have less resonance within families where the condition to be inherited is- or is at least potentially- fatal. Prenatal testing technologies are currently unable to accurately distinguish between different types of SMA in the foetus, making reproductive decisions particularly fraught, given the vast spectrum of severities associated with the condition. However, Fraser's presentation of the *certainty* of his future children having Type I SMA (and the undisputed suffering

that Type I entails) enabled him to circumvent the reproductive 'dilemma' that faced him- the management of the EO- and consequently justify his decision to use prenatal testing and selective termination.

## 6. Discussion

This paper has presented some of the key ways in which the EO was experienced and negotiated within families living with SMA. While bioethicists have been amongst those to highlight some of the theoretical flaws within such an objection (Shakespeare, 2006; Edwards, 2004; Malek, 2010), by drawing on the experiences of those with SMA in their family, this paper has highlighted that the emotional consequences of the EO are, nevertheless, a significant mediator in the reproductive decision-making of such families, and that they may also constrain the ways that prospective parents are able to present and justify their reproductive decisions.

As Cox (2003) has shown in relation to Huntington Disease, families are arenas in which reproductive decisions come to be scrutinised, and within which there may exist multiple, and potentially competing, sites of accountability for decisions taken. We make reproductive decisions not as autonomous individuals Cox argues, but as the 'mothers and daughters, fathers and sons, sisters, brothers, aunts, uncles, cousins, spouses, life partners and friends' of others; as social being we exist 'in and through our social and familial ties with others' (Cox, 2003: 262). It is in this familial context that prenatal testing and selective termination practices take on particular meanings; meanings that are augmented by the family's history with the condition, where the notion of having a child with SMA is not based in abstract imaginings or medical information, but instead in the lived reality of family life.

Unlike women approaching antenatal screening decisions, for families living alongside an inheritable condition like SMA, the experiential and intimate nature of their knowledge about the condition being tested for fundamentally alters the message conveyed by testing and selection procedures- by aligning future lives with existing family members with SMA. Within families, wherein people with SMA are linked to other (potential) people with SMA, not solely through their shared SMN1 deletion, but also through blood ties, the way in which biological kinship relations are conceptualised may become re-configured (Featherstone et al., 2006). As has been demonstrated, SMA became conceptualised as being synonymous with the family member diagnosed with it, meaning that reproductive decisions become transformed from decisions about life with SMA into highly personal appraisals of the quality and nature of *that* individual's life and experiences. For those diagnosed with SMA, the emotional consequences of this alignment were significant: the decision of a family member to terminate a pregnancy affected by SMA was often interpreted as a devaluation of their own life, with this judgement being more acutely felt if that family member had had had extensive involvement and was considered to have 'authentic' insight into what life is *really* like with SMA (Boardman, 2011). Indeed, participants frequently spoke in terms of their 'relational responsibilities' (Cox, 2003); to prevent emotional harm to their family member with SMA, as well as to other people living with it. For some, the emotional risks and potential harms to relationships were so great, that they described forgoing particular reproductive options (usually termination of pregnancy affected by SMA), highlighting the potential of expressivist concerns to constrain reproductive decisions for such families.

While some participants were able to navigate the implications of the EO and still present their decisions as responsible, for example by conceptually removing themselves as the decision-maker when recounting decisions (Kate), or by using experiential knowledge of



SMA to highlight the certain intolerability of suffering in future lives (Fraser), that these participants nevertheless presented their decisions in terms of the EO highlights its significance as a mediator of their reproductive decision-making. Indeed, 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, this research, in line with other studies, calls into question individualised notions of agency in the context of reproductive decision-making and underlines the importance of understanding the way that relational responsibilities shape reproductive decisions (Burgess and D'Agincourt, 2001; Hallowell et al., 2003). An emphasis on rational, calculated reproductive decision-making, both in genetic counselling, as well as within debates around the EO (e.g. Malek, 2010) overlooks what Fischhoff et al. (1978) refer to as the 'human factor' in decision-making: namely, the highly emotive relationship individuals have with their history with a condition, their relationships to family members and significant others, as well as wider social discourses around disability, genetic disease and parental responsibility. Thus, while the theoretical coherence of the EO may indeed be imperfect, its emotional reverberations throughout families living with genetic disease were nevertheless profound. Even for those participants who rejected the notion that EO concerns could justifiably be made in relation to their own reproductive decisions (e.g. Fraser), these participants nevertheless framed and validated their reproductive decision by reference to the EO ('No one could say to me that if I did [prenatal testing and selective termination] that I didn't love and want my children to live because they had SMA' - Fraser), highlighting the perceived need to be accountable to the EO, even by those who rejected it.

The findings of this research have a contribution to make to the call, made primarily by disability rights supporters, for more knowledge about the conditions that are currently screened and tested for to be made available to would-be parents approaching testing decisions (Fletcher, 2002; Patterson and Satz, 2002). Such disability rights supporters are keen to highlight that experiences of disability within families are often more positive than might be supposed by the medical professionals who counsel couples (Asch, 2000), as well as the general public. The implicit assumption of this argument is that access to experiential knowledge might challenge prevailing attitudes to disability (and consequently their anticipated role in selective termination decisions). This research, however, has highlighted the tangible effects such direct experiential knowledge can have on the way in which the EO is formulated, felt and experienced, within the context of families living with genetic disease. For the participants in this study, having knowledge of what life with SMA was 'really' like heightened the emotional distress associated with the EO, and invalidated the emotional 'buffer' that may be used by people living with conditions that are routinely screened for in the general population (e.g. Spina Bifida, Down's Syndrome): the idea that such screening and selective termination decisions are made in the context of ignorance, or even prejudiced ideas, about the reality of that particular condition. Indeed, for families living with inheritable diseases such as SMA, these decisions around selective termination were not being made by unknown others, but by members of their own family.

This finding thus highlights the need to carefully consider the support needs of disabled people living with tested-for conditions in the context of increasing genetic testing, and echoes Peterson's (2012) call for an engagement of the genetic counselling community with the EO. It may be useful for genetic counsellors to explore conceptualisations and experiences of the EO when counselling families affected by genetic disease where appropriate, and importantly, the experiences of those diagnosed with the condition themselves. Such an exploration 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 (Etchegary et al., 2008: 123).

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