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Enacting genetic responsibility: experiences of mothers who carry the fragile X gene

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Abstract

A woman who carries the gene for fragile X syndrome (FXS) has a 50 per cent chance per pregnancy of passing the gene to her sons and daughters. In this paper we analyse interview data from mothers who are carriers of the FX gene, and who have at least one child with FXS, to examine how their understandings and enactments of reproductive options, obligations, and responsibilities support an expanded notion of genetic responsibility. Accounts of 108 women from across the United States show that the majority of mothers chose not to have another biological child once they learned their carrier status. They discussed genetic responsibility and reproductive agency in terms of an obligation not to risk having another child who carried the gene, although their accounts reflected the tensions that arose from managing oneself as a genetically at-risk actor. Another 22 mothers either purposefully became pregnant or continued an unplanned pregnancy after finding out their carrier status. These mothers' accounts reflect an expanded version of genetic responsibility that incorporates ideas and values beyond managing risk in what it means to act responsibly in light of genetic knowledge.

Keywords

responsibility; fragile X syndrome; reproductive decisions; genetic citizenship

Introduction

> We're sitting on the edge of the genetic revolution and we're gonna basically choose not to have these children or we're going to manipulate our genes so that we all become intellectual people. Diane (all person names are pseudonyms)—mother of three boys with fragile X syndrome.

In the current `age of genomics', individuals increasingly encounter and use genetic tests and explanations of genetic risk to assess and manage their own and their family's health. Following detection of a genetic disorder, especially one caused by an alteration in a single gene where inheritance patterns and penetrance are relatively well understood (*e.g.* fragile X syndrome, Huntington's, Tay-Sachs), an individual faces an array of difficult decisions, such as to determine one's responsibility to inform relatives, make reproductive decisions, and monitor individual and family health, using genetic information. The purpose of this paper is to examine how women who carry the gene for fragile X syndrome (FXS), the most

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common inherited cause of intellectual disability, construct and enact such notions of genetic responsibility on personal, familial, and social levels.

Genetic responsibility and citizenship are concepts discussed in a growing body of social science literature on how individuals are impacted by genetic risk, technologies, and surveillance. In much of this literature, the genetically responsible citizen is presented as one who makes use of genetic information to manage her own and family's health. This exemplary genetic subject informs relatives who may also be at risk, chooses not to take a chance of passing on a genetic disorder, and incorporates personal risk information into a sense of being genetically at risk (Fitzgerald 2008, Kerr and Cunningham-Burley 2000, Lemke 2004, Nelkin 1996, Robertson 2000). In this framework, genetically at-risk individuals have an obligation to actively manage the self in light of knowledge revealed by genetic tests and explained by medical/clinical genetics (Novas and Rose 2000). An apparent `choice' to use genetic information and diagnostic testing is thus experienced as an obligation, as the responsible citizen feels compelled to use this information to engage in self-surveillance (Etorre 2002, Kerr 2003) and to manage her extended family's present and future health (Polzer 2005, Polzer *et al.* 2002, Raspberry and Skinner 2007).

This sense of genetic responsibility becomes most salient at times of making reproductive decisions, when the genetically at-risk individual is faced with the possibility of passing on the defective gene (Parsons and Atkinson 1992). Several studies suggest that for most people, genetic responsibility is enacted by not passing the gene to their children—either by using genetic testing to select for an unaffected embryo or foetus, or by choosing not to have children (Arribas-Ayllon *et al.* 2008a, Downing 2005, Hallowell 1999, Kelly 2009). Carriers who unintentionally pass on a genetic condition often hold themselves responsible for causing the problems of those affected by it. While this responsibility may translate into feelings of guilt or blame (Hallowell *et al.* 2006, Kay and Kingston 2002), at least initially, it may also translate into a form of genetic citizenship as individuals become lay experts in their particular genetic condition and join with others to advocate for targeted research and services (Heath *et al.* 2004, Rose and Novas 2005).

Other studies on gendered dimensions of reproductive decision making show how women are held primarily accountable for the health of the foetus, and for making definitive decisions about prenatal testing and childbearing (Ivry 2007, Locock and Alexander 2006, Markens *et al.* 2003, Rapp 1999, Reed 2009, Saukko 2004). This research coupled with discussions of genetic responsibility suggest that the genetically responsible woman uses diagnostic information in a preventative fashion both for her own and her extended family's health and reproduction.

While these studies portray the genetically responsible person as one who singularly prioritises genetic risk and identity in everyday health and life decisions, others present more complexity and a broader notion of biocitizenship. For instance, Plows and Boddington (2006) critique an exclusively biomedical version of biocitizenship and argue in favour of a more varied conceptualisation that locates genetic citizenship within an interaction of diverse and multiple identity claims and responsibilities. Also, Novas and Rose (2000: 507), though writing on the significance of genetic information for individuals, note that people formulate decisions and strategies within a complex ethical field, informed by multiple perspectives and practices that go beyond biological and medical ones.

A number of empirical studies demonstrate this complexity. For example, Arribas-Ayllon *et al.* (2008b) examine how nuanced family bonds affect the disclosure of genetic information. In their research on late onset Alzheimer's disease, Lock and colleagues (Lock *et al.* 2006, Lock *et al.* 2007) show how genetic risk information does not override, but rather

supplements already existing causal explanations for disease and inheritance. Kelly (2009), who notes the paucity of studies on the reproductive choices of parents who already have a child with an inherited condition, found that prenatal decisions made by these parents offers a version of `responsible parenting' in which they refused to structure their reproductive choices solely on risk estimates and diagnostic tests. Rapp (2000) also compellingly portrays how genetic diagnosis is only one element in the ways that parents make sense of, contest, and participate in biomedical discourse on Down Syndrome. She writes that their understandings reveal how `[r]eligious orientations and practices, informal folk beliefs, class-based and ethnic traditions as well as scientifically inflected counter discourses also lay claim to the interpretation of extra chromosomes' (Rapp 2000: 206).

Taken together, this literature indicates the complex and situationally-dependent aspects of genetic responsibility. We add to this literature an empirical investigation of the ways in which 108 mothers of children with FXS enact genetic responsibility. These women constitute a group for whom reproductive decisions are highly salient and an object of their own and others' surveillance. FXS is not life threatening but it poses some challenges to those who have it and to their families. FXS is a single gene disorder caused by mutation of the FMR1 gene on the X chromosome. A female carrier has a 50 per cent chance per pregnancy of her children inheriting the gene, either in its premutation or full mutation state. Males with FXS have moderate to severe intellectual disabilities, a range of language disorders, and social and behavioral difficulties, including problems with attention, impulsivity, anxiety, and arousal. About one-third also meet the diagnostic criteria for autism (see Bailey *et al.* 1998, Rogers *et al.* 2001). Females with FXS are overall less severely affected, but may also exhibit intellectual impairments and other characteristics of FXS.

In the following analysis, we explore through women's accounts how they talked about responsibility in relation to their decisions to oppose, consider, or actively pursue having another biological child and the ambivalences they felt in making such choices We conclude with a discussion of how women's varied and sometimes contradictory understandings of what it means to be a responsible reproductive actor, mother, and citizen bring to light the complex and heterogeneous enactments of genetic responsibility in everyday life.

The study

The data analysed here are drawn from a series of semi-structured interviews conducted with 108 families from across the United States who participated in a mixed-methods study designed to assess family adaptations to FXS (see Bailey *et al.* 2008, Roberts *et al.* 2009, Wheeler *et al.* 2008). Selection criteria were that the mother had to have at least one biological child under the age of 15 diagnosed with full mutation FXS (children do not inherit the full mutation from the father though all daughters of a male carrier are also carriers). Fifty-six per cent of the families were recruited through existing studies at the University of North Carolina-Chapel Hill and the university's online FXS participant research registry; 16 per cent were recruited from a FXS parent listserv and family support groups; and 28 per cent were recruited through other investigators in the field of FXS who gave our recruitment materials to participants in their studies. Recruitment material invited participation in a study that examines how families' lives are affected by having a child with FXS to learn more about sources of support, perceptions, resources, and family adaptations. Interested families contacted the study co-ordinator via a toll-free telephone call, e-mail, or return of a pre-paid response card. Mothers provided written informed consent for their own and their child's participation. This recruitment strategy resulted in families from 29 states in the final sample. All regions of the US were represented.

Mothers were the primary respondents. All but four women learned of their carrier status when one of their children was diagnosed with FXS. Most women were still of reproductive age at the time of finding out their carrier status, which made this genetic information particularly relevant for continued family planning. Of the 108 women, only 21 had made an explicit choice not to have another child before finding out their carrier diagnosis. Two-thirds (66%) of the women had only one child with the full mutation FXS, 29 per cent had two children with the full mutation, and six per cent had three children with the full mutation. Of the 108 children who were the focus of the assessments, 91 (84%) were male. Approximately one-third (32%) of the families were low-income (defined as below 200 per cent of poverty guidelines). Eighty-four (77%) mothers were white, 21 (19%) African American, two (2%) Hispanic, and one (1%) was of Middle Eastern descent.

Methods and analysis

Research assistants conducted three semi-structured interviews with the 108 mothers from 2003 to 2007. Mothers were interviewed initially as they joined the study, and again after 18 and 36 months about topics of interest to the larger project, including mothers' understandings of FXS, their perceptions of their own and their children's quality of life, child-rearing strategies, and sources of support. These interviews, each of which lasted approximately 90 minutes, focused on how FXS affected decisions and family life and included explicit questions about reproductive choices and plans. Interviews were conducted in the family's home and were digitally recorded and transcribed verbatim.

To analyse how mothers talked about reproductive decisions and responsibility, both authors thoroughly read and reread the first semi-structured interview to chart mothers' accounts of the ramifications of learning about FXS, especially as related to reproductive options. For each mother, the first author then systematically recorded and summarised in data display matrices (Miles and Huberman 1994) the different ways in which their reproductive decisions were influenced (or not) when they found out their carrier status, how they talked about personal responsibility, and other experiences and meanings that came into play in their discussions and decisions about reproduction and parenthood. The two authors then reviewed all subsequent interviews for information pertaining to reproductive decisions and women's reflections on and explanations of those decisions. We charted the reproductive trajectory for each woman and used this information to classify women into the groups described below. We compiled interview data on how women with different reproductive trajectories negotiated and talked about reproductive responsibility and familial obligations: how they reflected anew on their reproductive decisions, and how they justified their choices with and against imagined audiences and perceptions of societal expectations. We extracted themes within each mother's account and compared these to other women who shared the same reproductive trajectory and to those who did not. We chronicled our evolving interpretations of how women were constructing themselves as responsible actors using the constant comparative method, modifying our interpretations in light of each account (Strauss and Corbin 1990). In our analysis below, we report themes that were predominant in women's accounts and select excerpts representative of these themes.

Enacting responsibility

In the following sections we examine women's accounts for the complexities, contradictions, and ambivalences embedded in their experiences of being a responsible citizen, mother, and member of an extended family. A related paper (Raspberry and Skinner 2010) focuses specifically on the different reproductive paths that these women forged, which included non-mutually-exclusive decisions to not have more biological children (83), adopt (6),

undergo assisted reproductive and genetic technologies (6), use prenatal testing (17), and have children irrespective of FX status (22).

Our aim in this study is to analyse women's understandings of responsibility as formulated in their discussions with us about their reproductive choices and how they enacted being a responsible reproductive actor. We first present how mothers who did *not* have more children after carrier diagnosis constructed genetic responsibility. These women discussed responsibility in terms of managing genetic risk by not taking the chance of passing on the FX gene, though this decision was not without the complicated tensions that arise from managing oneself as a genetically at-risk actor. We then turn to the accounts of 22 women who, conscious of their 50 per cent chance of passing on the FX gene, either purposefully initiated an unmediated pregnancy (10), or continued an `at-risk' unplanned pregnancy (12). By `unmediated pregnancies' we refer to pregnancies in which prenatal testing and other reproductive and genetic technologies were either not used, or were not used to avoid passing on the FX gene. We find that in contrast to their counterparts, this latter group of women did not frame responsibility in terms of monitoring genetic risk, but evoked other forms of responsibility, such as the responsibility to love, care for, and for some, procreate all children, regardless of their genetic status.

Not passing the gene

After finding out their carrier status, 77 per cent (83) of the mothers in this study chose not to have a biological child who might carry the FX gene.[1] They constructed genetic responsibility at least in part as a commitment not to pass on the FX gene, either by not reproducing, or by using reproductive and genetic technologies (preimplantation or prenatal) to have a child who would not carry the gene.[2] For example, Pamela, mother of two young daughters who both have FXS, decided not to have more children when she found out she was a carrier. She consciously talked about this decision as a `responsible' one, albeit one that limited her envisioned family: `I love my children and I probably would love to have a whole houseful of them but I said `You know what? I'm not going to do that. I'm going to be responsible. Take care of the two I have'. Similarly, Katie, who adopted a daughter after her son was diagnosed, characterised as reckless the act of having a biological child after knowing one's carrier status. She explained, `I would have felt – I think it's – what's the word? I think it's irresponsible. I would have felt irresponsible maybe if I had known this was a possibility and maybe not considered it seriously and just plunged ahead'. By increasing her family size through adoption rather than unmediated pregnancy, Katie viewed herself as acting responsibly and thoughtfully.

Other mothers wanted more biological children, but discussed responsibility in terms of using prenatal testing to prevent passing on the gene. Mary, who has two sons with FXS, expressed a strong sense of individual responsibility to stop transmission of FXS:

> My first husband was like, `Get sterilized. Get your tubes tied. Just forget it. The way to deal with this is to not have any more kids. People should get diagnosed, and the ones who have it shouldn't have children. That will get rid of it. That will exterminate it from the population'. I always thought of that as being a little bit Naziistic, but it is true. If you diagnose people, and if they are aware they have this problem, and they don't have children, then, you will stop it.

[1]The 83 mothers includes 21 who said they were already done having children at the time of diagnosis, three who adopted after diagnosis, and two who successfully used assisted reproductive technology (ART) techniques to have a biological child without passing on the gene (IVF with PGD and IVF with donor eggs).

[2]In discussing the possibility of passing on the FX gene, mothers did not distinguish between full mutation FXS and carrier status. In other words, the risk of passing on the gene was implicitly referred to as a risk of having a child with full mutation FXS.

Although Mary recognised the eugenic potential of knowing one's carrier status—a prospect particularly cautioned against by disability rights advocates and others—she also characterised herself as having acted responsibly when she terminated an unplanned pregnancy after testing indicated the foetus had the full mutation.[3] For her as for the majority of these mothers, being `responsible' included a commitment to prevent passing on the FX gene.

Familial responsibility

In a similar vein, many of these women's understandings of genetic and reproductive responsibility extended to a sense of obligation to immediate and distant relatives as well as to potential future generations. Most participants identified the issue of whether and when to inform relatives about the presence of the FX gene in the family as being a key facet of a familial sense of genetic and reproductive responsibility. For many like Valerie, acting responsibly entailed telling everyone in the family that two of her three sons had inherited the FX gene that she carried, and letting them decide for themselves how to use the information. She explained, `I just wanted everybody to be informed so I wouldn't feel guilty later. They had a right to know'. While some mothers felt a moral obligation to inform relatives that they may also be carriers, others believed it was their responsibility to protect certain family members from that information. For example, Dawn did not want her mother to know she was a carrier because she `would start blaming herself and would never get over it'. Anne, who has one young son with FXS, told her mother but felt that she could not tell her grandmother: `You have these grandchildren that are mentally retarded and you're the reason why'. At the same time, Anne acknowledged that not disclosing this information meant that her grandmother's siblings, their children, and grandchildren might remain uninformed that they too could carry the FX gene and pass it on.

This sense of obligation to living relatives at times also extended to those as yet unborn. Vicky, who was pregnant when her two sons were diagnosed, expressed this sense of awareness and responsibility:

> And that even if they are just a carrier and don't show any signs, that I think it's a parent's responsibility to keep the wellbeing of your family for even generations to come. So even if you're going to have a child that's just a carrier, are you going to be able to deal with the fact that your grandkids are going to possibly have it?

Like the other mothers interviewed, Vicky articulated a parent's role as including an obligation to ensure and protect `the wellbeing' not only of current family members, but also of future relatives. In enacting familial responsibility, these mothers had a role in shaping their extended families as well as their immediate one, through their management of genetic information and personal reproductive decisions.

Our discussion to this point has centred on monitoring aspects of genetic risk and reproductive responsibility for oneself and one's family. However, these mothers' experiences of genetic and reproductive responsibility also included a complex negotiation of doubts, tensions and allegiances.

Conflicted reflections

As the above examples illustrate, the majority of women made personal decisions not to have more biological children after finding out that they carried the FX gene. However, in

[3]Although we do not have conclusive data for the entire sample, pregnancy termination does not appear to have been a commonly used method by these women to avoid passing on the FX gene. Out of the 17 women who talked about their use of prenatal testing, five terminated following a positive result, while six continued a positive result pregnancy.

discussing their decisions in retrospect, some also voiced concerns and ambivalence about the implications of their choices. In arriving at this decision mothers had to adjust deep-seated desires and values (see also Raspberry and Skinner 2010). They found themselves revising their reproductive imaginaries to fit their new reproductive circumstances, but often not without some distress and conflict. Carrie underscored these tensions when she reflected on how having two sons with FXS had impacted on her:

> I think the largest thing that has really affected me, just me, is the fact that my husband has always wanted a lot of children. And you know we have the two. […] So that's the biggest thing because you know you're carrying it and you think, `Well if he had married someone else, he could have had a houseful of kids'.

Carrie's account of her experience, and the divergence in the reproductive path taken and that imagined, convey the contradiction that many women grappled with after finding out their carrier status. On the one hand, Carrie articulated the difficulty of letting go of the vision of having many children, but on the other, she was equally clear that she would not risk passing on the FX gene to make a bigger family.

Some women, while resolute about their own choice not to have more children, indicated they would not impose this on others, recognising different notions of what is responsible behaviour and `doing what's right'. Rachel, whose only child had FXS, refused to judge women who consciously take the risk of passing on the gene, though she personally would not do so. She explained, `And there are people out there that wouldn't mind, that I know have just gone ahead and had kids, and if they have fragile X, that's great. If they don't, great. But I'm not at that point, and I don't know if I ever will be'. In her reluctance to criticise women who chose to follow reproductive paths that she could not imagine for herself, Rachel acknowledged that there were diverse ways of enacting responsibility, perhaps different from her own, but equally valid.

Another tension in some women's accounts was their recognition that by not reproducing or in some cases using tests and technology to select against children like the ones they already had, they might be viewed as devaluing lives of individuals with disabilities. One mother's account particularly encapsulates this conflict. While Diane resolved not to have more children after her three sons were diagnosed with FXS, at the same time she expressed unease about the possible societal consequences of such genetic testing-based choices over time. Her concerns led her to envision a future in which individuals increasingly base reproductive decisions on their potential genetic predispositions for disorders, and in so doing create a society where `special children' are not wanted. She explained:

> And the idea being: `Why should I have a disabled child when I can make a princess', and this whole sort of genetic realisation and manipulation of our culture. Like I have a couple friends who – I think they have like a 20 per cent chance of passing on a gene that has something to do with the nervous system or muscular system, and they decided not to have kids. They're going to adopt, and I'm like `Hey, adoption's great and everything'. But it's going to get to a point where if people are predisposed to a certain gene and they know about it, they're not going to have those kids, or they're going to go in vitro, and so in a way a lot of these special kids like our kids I think will be sort of taken out of Western culture countries.

Diane speculated that with increased availability of genetic testing, more people will choose not to risk passing on a genetic disorder, or even a genetic predisposition. She worried that a predominance of such genetically based reproductive choices could ultimately result in less tolerance and acceptance for difference, a possibility that seemingly follows from a risk-management version of being a responsible genetic citizen. The tensions evident in her

account and those of other women who decided not to have biological children indicate that a risk-management-based notion of genetic responsibility does not encapsulate the complexities that arise in practice. To further illustrate the varied dimensions and interpretations of what it means to be genetically responsible, we now turn to the experiences of mothers who knowingly chose to risk passing on the gene in order to have biological children.

Choosing reproduction

Twenty-two women consciously decided to have a baby after finding out that they had a 50 per cent chance of passing on the FX gene with each pregnancy. These mothers refused to choose *not* to have the families they had imagined raising with their partners before they knew they were carriers. Their accounts of why they made this decision reveal how they incorporated different senses of what was responsible and moral behaviour in the light of genetic information.

This group of 22 women does not include the few families who initiated or continued a pregnancy with the intent of using prenatal testing and terminating if results came back positive for FXS, nor does it include the two families who used assisted reproductive and genetic technology techniques to conceive and continue a non-affected pregnancy.[4] Ten of the 22 women planned a non-mediated pregnancy, willing to risk having an affected child. The other 12 decided to continue an unplanned non-mediated pregnancy. While differing in this respect, the two groups spoke similarly of why they went ahead with their `risky' pregnancies. Both groups knew and accepted that they had a 50 per cent chance of having a child who would either be a carrier of or have fragile X syndrome.[5]

Overall, the mothers who chose to continue planned and unplanned `risk' pregnancies talked about their reproductive decisions primarily in relation to a moral framework that values parenthood and honours difference. In defending or situating the choice they made to risk having a child with FXS, these 22 mothers explicitly constructed responsibility within this context of valuing a child's `specialness' and the importance of family. For example, Lynn, who decided to have a second child after her first son's diagnosis of FXS, and subsequently gave birth to another son with FXS, expressed a sentiment commonly shared among these mothers: `And as far as I'm concerned, a child with a disability is no less of a worthy person to live'. Similarly, Beverly, a mother of five children, also reflected that she deliberately chose to have more children in part to emphasise that she did not negatively value her first two children who had FXS. Beverly saw her reproductive choices as testament to the value and beauty of all her children, despite sensing public disapproval of her actions. She related: `He's a beautiful kid. They're beautiful and I mean that's why I went on to have more kids. We get a lot of criticism for that'. Like Beverly, other women sensed or were confronted with societal censure for acting irresponsibly by knowingly taking the risk of having a child with intellectual disability, and they responded with their own accounts of what constituted responsible and moral acts (see also Landsman 2003, 1998 for discussion of reproductive choices and valuing the personhood of `less-than-perfect' children).

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[4]This group of 22 women does not include three mothers who explicitly stated that they used prenatal testing with a planned or unplanned pregnancy in order to avoid passing on the FX gene, nor does it include the two mothers who successfully used ART to have an unaffected child. Three of the four women who knew their carrier status before having children are counted in this group of 22 mothers. Two chose to have two children, one woman adopted, then continued an unplanned pregnancy. The fourth woman who knew she was a carrier is not in this group because she is one of the two women who used ART to avoid passing on the gene (she used IVF with PGD to have three children, due to a lab error her first child has full mutation FXS).

[5]The 22 women who continued planned and unplanned unmediated pregnancies gave birth to a total of 31 children, 20 were born with the FX gene. Of the 10 mothers who planned a `risk' pregnancy, three of them did so twice, and two of these three subsequently continued unplanned pregnancies. Of the 12 women who continued unplanned pregnancies (this does not include the two who first planned a pregnancy), two women did so twice.

These women did not experience knowledge of carrier status as an imperative not to reproduce, nor as a basis on which to calculate genetic risk. Instead they enacted forms of responsibility predicated on valuing difference and investing in parenthood. For example, Catherine explained that she and her husband's commitment to expand their family outweighed the risk of having another child with FXS:

> We decided to have another child. Went through the whole PGD process. That didn't work. So we just decided that it was important for [son] to have a sibling regardless, and if we had two kids with fragile X, then at least they would have each other. That's kind of how we looked at it. So we decided to do that'.

Catherine, whose second son did not inherit the FX gene, framed her and her husband's decision to have another child as a conscious evaluation of what was best for their family: giving their son a brother or a sister.

In talking about their roles as mothers, these women also emphasised the importance they attached to being their children's best advocates as well as being the best parent possible in an array of situations. For some, an enhanced sense of life purpose or fulfilment came out of these parenting and advocacy efforts. Karen, who chose to have two more children after the first was diagnosed, conceptualised her role as mother to three children with FXS as a positive one: `There was a purpose that I was supposed to fulfil that I wasn't fulfilling and I was given my children to help me become the person that I'm supposed to be'. Like other mothers, Karen saw herself as an advocate for her children, and further reflected that the presence of FXS in her family made her feel `a greater responsibility to be a good parent'. This construction of being a `good parent' often also included knowing one's parental limits, and being aware of emotional and financial restraints on having more children. Lynn also defined her reproductive choices as based on her abilities to be a good parent and the value she placed on all children: `I mean it didn't keep us from having another child. We did know. It's not that we felt that a child with special needs is not a worthy human being. It's just you do as a parent needs to be able to provide what they need'. Lynn decided to have a second child only after making a personal calculation that she could responsibly parent two children even if they both had FXS. The decision to risk having another child with FXS was made by balancing a calculation of available financial and emotional resources with a privileging of motherhood and wanting more children.

These accounts indicate that women who continued to have children did not talk about obligations to manage risk, but rather explicitly situated and described their decision to have children within a framework of choice, affirmation, and love. Like their counterparts who stopped reproducing, these mothers did not make their decision lightly. It is important to note also that these women's knowledge of the risk of transmitting FXS was not different from the women who did not have another biological child (see Raspberry and Skinner 2010), nor did they express different feelings of self-blame or guilt associated with being a carrier (see Skinner and Raspberry 2010). Nor did we find any significant differences in age, education, income, ethnicity, religious affiliation, or religiosity between the women who stopped reproducing and those who continued.[6]

On the other hand, there are identifiable differences in how these women talked about their choices to have another biological child. While all the mothers in this study emphasised the

[6]A statistician ran t-tests comparing the two groups. All differences were nonsignificant. In a paper based on interview data from women in the same study, Michie and Skinner (2010) concluded that the majority of mothers used a religious framework in talking about their children having FXS, with some explicitly saying it was a part of God's plan and purpose for their lives. While religious faith and practice clearly played a role in how mothers in this study talked about their children, there were no significant differences in religious affiliations nor in the degree of religiosity between women who chose not to have more children and those who had more biological children with unmediated pregnancies.

unconditional love they felt for their children with FXS, women who continued to reproduce talked about this choice by referencing a framework of values of motherhood and family as integral to their understandings of being responsible. For these 22 mothers, particular dimensions of responsibility were more salient than others, and genetic responsibility did not primarily revolve around whether to risk passing on a gene or not. Through their choices, actions, and discourse these women enacted and emphasised different facets of being responsible which included affirmation of the worthiness of all children, valuation of `difference', investment in parenthood and family, a sense of increased empowerment and life purpose, and an awareness of emotional and financial limits to parenting. It is notable that many of these themes were also raised by the women in this study who decided not to have more biological children. However, those mothers were more likely to discuss the value and important of their children and family while describing their child's characteristics and development, or in relation to everyday circumstances. In contrast, the women who continued to reproduce brought up these themes in the context of talking about responsibility and why they made the reproductive decisions they did.

In summary, for the women who continued to reproduce, `doing the right thing' was defined within a framework that extended beyond risk calculations of transmitting a genetic disorder. In rejecting a prescriptive construction of themselves as genetically at-risk individuals, these women instead emphasised their social roles as mothers and advocates for their families and communities. They framed themselves as responsible actors within a discourse that encompassed love and appreciation for their children, a rejection of value distinctions between those with disabilities and those without, feelings of increased empowerment and life purpose, and a heightened awareness of their own parenting strengths and limits.

Conclusion

Diagnosis of an inheritable condition reveals health information of consequence not just for the individual but for kin through the generations. While other studies have documented the complex nature of genetic responsibility, constructions of genetic responsibility based primarily on risk management suggest that the responsible individual is one who would choose *not* to `roll the dice' and chance passing on a condition such as FXS (Ekberg 2007, Hallowell 1999, Novas and Rose 2000). The majority of women in this study indicated through their accounts of their reproductive trajectories that they were constructing themselves in line with this notion of the `good genetic citizen' as one who does not engage in `risky reproduction', and who through disciplining the self and family contributes to the wellbeing and health of society. They `did the right thing' by choosing not to have another biological child who could carry the gene. They also performed what they saw as their moral obligation to disclose genetic information to relatives, thereby possibly shaping the kinds of families they would reproduce. At the same time, these women's accounts also reveal the conflicts and ambivalences that arise in enacting this circumscribed form of responsibility as they reconciled desires to have more children with a sense of oneself as genetically at-risk. Some of them voiced tensions engendered by the intersection of contrary societal stances: a sense of being censured and criticized if they passed on a gene known to cause intellectual disability, as well as concerns that the nature of their choice, if extended and endorsed more broadly by others, could have societal consequences of reducing the number of children who have genetic problems or predispositions for undesired characteristics, and thus lead to a devaluing of individuals with disabilities or imperfections.

Paediatric genetic testing was used to determine the children's and mothers' FX status (*i.e.* full mutation for the former and either full mutation or carrier status for the latter), and the genetic information resulting from these diagnostic tests was powerful for shaping most

families' subsequent reproductive decisions. However, very few families used prenatal testing for selective purposes. In this regard they were similar to families of affected children in Kelly's (2009) study who chose not to put themselves in the position of having to decide whether to terminate a pregnancy based on prenatal testing. By not getting pregnant, they circumvented the biomedical interventions and moral dilemmas involved in making that choice. Those families who had more children through unmediated pregnancies present an interesting counterpart to the argument that the availability of genetic testing leads to a liberal eugenics (Duster 2003). In respecting and investing in difference within their own families, these mothers add their unique voices to debates over `appropriate' uses of genetic testing and risk information. These mothers' multifaceted accounts, like those reported for example in Downing (2005), Kelly (2009), and Rapp (1999), underscore the point that whether or not to have children is not a straightforward decision based on circumscribed notions of genetic risk and responsibility, but rather involves a complex negotiation of personal desires, family values and diversity, religious faith, and financial constraints. The stories told by women who chose not to have more children as well as those who did, evocatively attest to the complexity of reproductive decision making in the context of inheritable conditions.

One of the strengths of this study is the relatively large sample homogeneous by type of genetic disorder but diverse by race, education, income and region. That we found no differences in these factors between those who did not risk having another child with FXS and those who had more biological children without the use of reproductive and genetic technologies, suggests that the difference between these two groups is how responsibility is reckoned—with risk management as priority for the non-reproducers and other notions of responsibility for those who had more children.

Our focus on how women who know they carry the gene for FXS make reproductive choices and construct themselves as responsible social actors provides evidence for the nuanced ways in which genetic responsibility is enacted in everyday situations by those who have experience with a specific genetic condition. It is likely that these enactments would be somewhat different in families who have other types of inherited conditions. Variations in inheritance patterns, uncertainties of probabilities and prognosis, and level of severity of associated characteristics make a difference in how carriers think about and act upon reproductive decisions. While raising a child with FXS may present challenges and stressors for a family (Bailey *et al.* 2008, Wheeler *et al.* 2008) and for some include the stigma associated with intellectual disability, these challenges are probably experienced as less disruptive than those that accompany other inherited diseases, such as Tay-Sachs, which brings profound suffering and early death. While most families would not purposely choose that their child has FXS, they speak about loving all their children and the joy and sense of purpose that having a child with FXS brings to their lives (see Michie and Skinner 2010), and so for some families, the possibility of having another child with FXS is not a reason not to have more children. In addition, the `manageability' of living with FXS makes it more probable that some families will risk passing on this gene, while they might be less likely to do so in cases of a fatal genetic disorder, an issue for further research.

To conclude, we suggest that the ways that these women construct themselves as responsible mothers provide evidence of the complex and dynamic ways that genetic responsibility is enacted and requires a refashioning of this notion to be inclusive of a variety of values and the inherent ambivalences of being a genetic citizen.

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