

# “It’s Something I Need to Consider”: Decisions About Carrier Screening for Fragile X Syndrome in a Population of Non-Pregnant Women

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Population carrier screening for fragile X syndrome can provide women with information about their risk of having a child with fragile X syndrome and their risk of fragile X-associated primary ovarian insufficiency and fragile X-associated tremor ataxia syndrome. Few studies have explored women’s decisions when offered carrier screening for fragile X syndrome. Interviews were conducted with 31 women who participated in a pilot study offering carrier screening to non-pregnant women. A qualitative approach was used to gain an in-depth understanding of women’s experiences and examine their decision-making processes, including women who were tested and those who decided not to be tested. The decision-making process occurred in two phases. In the first phase, the participant’s reproductive stage of life and experience with illness and disability were major factors influencing whether she would consider screening. In the second phase of decision-making, participants’ perceptions of the value of knowing their carrier status was the most notable factor for influencing whether a woman actually had the carrier test. Some women appreciated having time for deliberation and those who were tested did not express regret about their decision. Our findings support offering carrier screening for fragile X syndrome to non-pregnant women and suggest that women from the general population will have specific informational and counseling needs when offered carrier testing. This study highlights the unique challenges encountered by women from the general population when making a decision about testing for fragile X syndrome carrier status and illustrates the importance of understanding how women make decisions.

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**Key words:** carrier screening; fragile X syndrome; health belief model; health behavior; decision-making; qualitative research

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## INTRODUCTION

Current guidelines on population carrier screening for fragile X syndrome (FXS) recommend offering screening only as part of a clinical research protocol [Sherman et al., 2005]. This is due to concern about the broad phenotype of *FMRI*-related conditions and the need for adequate education, counseling, and resources for individuals identified as carriers [McConkie-Rosell et al., 2005; Sherman et al., 2005].

FXS is the most common known cause of inherited intellectual disability. An expansion of a CGG trinucleotide repeat in the 5’ region of the *FMRI* gene is the mutational basis for the condition

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[Verkerk et al., 1991] in more than 99% of cases [Brown, 2002]. The normal range of CGG repeats is between 6 and 44 while those affected by the condition have over 200 repeats (full mutation). Individuals with a pre-mutation (carriers of FXS) have between 55 and 200 repeats. In addition to their risk of having children with FXS, carriers are at risk of fragile X-associated primary ovarian insufficiency (FXPOI) [Allingham-Hawkins et al., 1999] and fragile X-associated tremor ataxia syndrome (FXTAS) [Hagerman et al., 2001]. Research studies are currently investigating the possible significance of the gray zone (45–54 repeats). It is estimated that 1 in 157 females [Berkenstadt et al., 2007] and 1 in 755 males [Rousseau et al., 1996] are carriers of the pre-mutation.

The current approach to identifying carriers of FXS is to offer genetic testing to individuals with a family history of FXS or undiagnosed intellectual disability [McConkie-Rosell et al., 2005; Sherman et al., 2005]. However, this approach is limited by issues around dissemination of genetic risk information in families [van Rijn et al., 1997] and its reliance on the diagnosis of an affected individual to make relatives aware of their risk. An alternative approach of population carrier screening has the potential to identify more people who are carriers.

Studies have shown that there is support for making carrier screening for FXS available to women in the general population [Spence et al., 1996; Wildhagen et al., 1998; Ryyanen et al., 1999; Pessa et al., 2000; Toledano-Alhadeef et al., 2001; Cronister et al., 2005; Musci and Caughey, 2005; Fanos et al., 2006]. However, most research has focused on offering carrier screening in the prenatal setting rather than prior to pregnancy when women have a wider range of reproductive options.

Uptake of a carrier test has often been used as a measure of acceptability of FXS screening and has ranged from 7% to 85% in pregnant and non-pregnant women [Spence et al., 1996; Ryyanen et al., 1999; Cronister et al., 2005]. It is also important to understand what factors influence uptake. In a study investigating the benefits and risks of carrier screening for FXS in women of reproductive age, Anido et al. [2005, 2007] found that carrier testing should be offered prior to pregnancy if it is to be viewed as beneficial and that women from the general population may be unprepared for a carrier result. Their study highlights the importance of exploring experiences of women from the general population who participate in carrier screening for FXS. However, very little is known about women's decision-making in this context and to the best of our knowledge, no studies have included women who choose not to have carrier testing.

The data presented here are derived from a larger pilot study of non-pregnant women from the general population who were offered carrier screening for FXS [Metcalf et al., 2008]. The study was conducted at Family Planning Victoria (FPV), a reproductive and sexual health service in the Melbourne metropolitan region. The pilot study involved three phases: (I) a needs assessment exploring views of clients and staff about the possibility of offering carrier screening for FXS at FPV; (II) non-pregnant women from the general population were offered carrier testing for FXS through FPV. They were given an information brochure about the clinical features, genetics of FXS, reproductive implications (including termination of pregnancy and pre-implantation genetic diagnosis) of a carrier result, and the process of having a carrier test. Women were then required to return to the clinic on another day to have the

carrier test. They also completed a pre-test and post-test questionnaire measuring anxiety, knowledge, reasons for and against testing, and views regarding population carrier screening. Participants had access to genetic counseling at any stage. Phase III consisted of follow-up interviews with a sample of women who had participated in phase II, the findings of which are reported here. We describe in-depth women's experiences of being offered carrier testing for FXS and their decision-making processes.

## MATERIALS AND METHODS

### Ethics

This study was approved by Family Planning Victoria Human Research Ethics Committee.

### Recruitment of Participants

Participants were 18 years of age or older, not pregnant, and able to speak, read, and write English. All had participated in phase II and indicated that they would be willing to be interviewed. Purposive sampling, which involved selecting participants to represent the range of test results and decisions about testing, was used. To include a variety of experiences we recruited: (a) women who were tested (including those with gray zone or carrier results) and (b) women who were not tested (including some who had initially indicated that they wanted carrier testing at the time of recruitment but did not return to have the test).

Recruitment continued until analysis revealed that no new themes were emerging from the data indicating that the data had reached saturation. Therefore, not all participants who were willing to be interviewed were contacted.

### Interviews

Participants were offered a phone or face-to-face interview. Interviews were conducted by AA and were between 8 and 50 min in duration. Interviews were digitally recorded, transcribed verbatim, de-identified, and participants were assigned a pseudonym.

### Methods

A phenomenological approach was chosen to gain an in-depth understanding of women's experiences of deciding whether to have a carrier test for FXS. This is an inductive approach and involves exploring individuals' lived experiences of the phenomenon in question [Robinson, 2006]. Semi-structured interviews were used to allow women to "tell their stories." The interview guide was developed and informed by previous research in this area [Anido et al., 2005, 2007] and the outcomes from phase I [Metcalf et al., 2008]. Participants were asked questions relating to their decisions about carrier testing for FXS, including: what decision they made; factors influencing their decision; and how they felt about the decision.

Due to the limited amount of research regarding women's experiences of carrier screening for FXS, transcripts were analyzed using thematic analysis in order to allow a range of themes to emerge from the data. Analysis began with a process of initial coding

where transcripts were read through and themes were identified. This was followed by a grouping together of similar or related themes after which themes were compared to look for patterns, relationships or tensions [Glasser and Strauss, 1967]. Transcripts were coded independently by AA, SW, and SM, and themes were compared to reach consensus. NVivo 7 software (QSR International Pty Ltd, Melbourne, VIC, Australia) was used to manage the data and facilitate coding.

## RESULTS

Two hundred thirty-three of the 318 women who participated in phase II consented to be contacted for interview. The first 53 women who completed phase II were invited to participate in phase III and 31 consented. Of those interviewed, 13 women had been tested: 10 with a normal result and 3 with a gray zone or carrier result; 18 were not tested: 10 who initially wanted to have the test but did not return to have it and 8 who had indicated upon recruitment in phase II they did not want to have the test. Socio-demographic characteristics presented in Table I reflect those of the larger phase II group.

During data analysis four main categories of themes emerged from the interviews: (1) wanting to know more about carrier testing for FXS; (2) considering having carrier testing—*influencing factors*; (3) making a decision—*influencing factors*; and (4) reflecting on the decision.

### Wanting to Know More About Carrier Testing

Participants indicated that the invitation to take part in the pilot screening study had sparked their interest and they had found the

concept intriguing. Their initial reaction was to seek more information about carrier testing for FXS:

I thought it was really interesting . . . [*it's*] really peculiar that you can have someone take some of your blood and tell you what you do and don't have and what you could pass on . . . [Ellen, 20, not tested]

. . . I hadn't heard of [*FXS*] before . . . so I was interested to find out more . . . [Sophie, 35, tested]

Many participants reported that at this stage they had not determined whether they wanted to have the test. They discussed approaching the concept with an open mind:

. . . initially I was really interested . . . I didn't initially think, 'no I won't have the test' . . . I thought 'oh, now that's one more thing that I know about' . . . [Ella, 20, not tested]

### Considering Having Carrier Testing—Influencing Factors

While many participants showed an initial interest in carrier testing, the woman's life experience related to health and her reproductive stage of life influenced whether she went on to consider having the test.

**Experience with health problems, genetic conditions, or disability.** Participants discussed a range of health-related experiences including their own health-related problems, those of family members, or friends, or experience gained through their occupation. The following examples demonstrate how prior health experiences can motivate consideration of having the carrier test:

I have thalassaemia, just a like a silent thing, carrier. I was kind of interested, just in the whole genetics thing.

[Gina, 21, tested—normal result]

One of my friend's children has autism . . . so that would make me concerned, because I know a child with autism . . .

[Michelle, 20, not tested]

Though this participant, and others, did not go on to have carrier testing for other reasons, she was motivated to consider it because of her prior health-related experiences. While familiarity with health issues often influenced women to subsequently be tested, one participant commented that lack of awareness of health issues might be a barrier for some to even consider carrier testing:

. . . I think it all comes back to life experience, what you've been through or what you've seen people go through . . . I think when people don't live with it they don't really have the same level of insight. [Natasha, 28, not tested]

**Reproductive life stage.** Participants associated the carrier test for FXS with reproduction and many who were at the stage of planning to have children considered having the carrier test:

. . . you know [I] hope to fall pregnant next year . . . so I think that [the test] was really appropriate.

[Amanda, 28, tested—normal result]

TABLE I. Socio-Demographic Characteristics of Participants

Characteristic	Participant, total N = 31 (%)
Age range (years)	
18–25	12 [38.7]
26–30	6 [19.4]
31–35	4 [12.9]
36–40	4 [12.9]
41–45	4 [12.9]
>45	1 [3.2]
Highest level of education	
Year 11 or less	1 [3.2]
Secondary school	9 [29.0]
Trade/apprenticeship	0
Tertiary: college certificate/diploma/university	21 [67.8]
Other	0
Relationship status	
Living with partner/married/de facto	13 [41.9]
Divorced/separated	3 [9.7]
Partner, not living together	6 [19.4]
Single	9 [29.0]
Widowed	0
Other	0
One or more children	8 [25.8]

In contrast, participants who chose not to have the carrier test often perceived the test as irrelevant because they were not planning to have a family in the near future:

I think because I am quite young, and at this stage in my life I'm not thinking about having children, so it's [the carrier test] not really something at this stage that's really important to me. [Abby, 22, not tested]

The two factors mentioned above, experience with health-related problems and reproductive stage of life, appeared to influence participants' initial perception about whether carrier testing for FXS was relevant to them. The quote below illustrates how this participant has chosen to defer her consideration of carrier testing because it is not relevant to her current stage of life:

[*carrier testing for FXS is*] something I need to consider, I'm not ready to have a child at the moment, it's not something I've thought much further than, 'oh at some point I need to think about these issues'. [Natasha, 28, not tested]

However, while reproductive stage of life and experience of health-related matters provided the initial motivation to consider testing, not all women chose to be tested because other factors influenced their decision.

## Making a Decision—Influencing Factors

For those women who did consider screening the process of deliberation involved a weighing up of factors that were grouped into four categories: perceptions of the value of knowing their carrier status; perceptions of risk; uncertainty about the features of FXS; and practical issues.

**Perceptions of the value of knowing carrier status.** There were three major themes relating to participants' perceptions of how valuable they felt it would be to know their carrier status. The first theme related to curiosity about one's genes and a commonly held perception that carrier testing would provide more information about themselves:

...yeah, curiosity, to know more about myself. [Lucy, 29, tested—normal result]

A second theme related to information for making future decisions:

... the more information I have, it would better help me and benefit me in the future, in relation to my decision-making and my partner's decision-making... [Amanda, 28, tested—normal result]

Participants talked generally about using the information for future decisions but they did not specify what reproductive choices they would make in the future if they received a carrier result. As this participant explains, people may not know what decisions they would make:

... it's hard. You can say 'okay I'll make these decisions' or 'I might make these decisions' but until you're in that position who knows what you would do.

[Louise, 34, tested]

FXPOI was rarely discussed, however, those who did mention it reflected that knowing their risk of FXPOI would be useful as it could inform future decisions. Less commonly women discussed that knowing their carrier status would help them to prepare for having a child with FXS. No participants referred to FXTAS in their discussions about wanting to know their carrier status.

The third theme related to concerns that carrier testing could create anxiety during pregnancy and impact on future plans. For this reason some participants did not believe it would be worthwhile knowing their carrier status:

... being a worrier and I thought about it, you know, would I go through nine months of pregnancy worrying, thinking, 'I've got the gene, I've got the gene, I wonder how it's going?' [Ella, 20, not tested]

... I've always wanted to have kids. What if I did it [*had the carrier test*] and it said yes? I mean that would really sway the wanting to have children. [Ellen, 20, not tested]

Only one person specifically stated that they did not have carrier testing because they would not terminate an affected pregnancy.

**Perceptions of risk.** For some participants there was a perception, and misconception, that the presence of family history should be a major factor influencing their decision to have carrier testing:

... if I had a history of it in my family I would definitely want the test... [Miranda, 39, not tested]

Additionally, some participants' decision to have carrier testing was guided by a higher perception of risk due to their life experiences of other health-related issues:

... my parents had me who was quote unquote [sic] totally normal and then the next two children were deaf through recessive genes, so that's a real motivating factor. I've got this perfectly normal baby, who knows what could happen with the next two or three... [Laura, 25, tested—normal result]

**Uncertainty about the features of FXS.** Participants appeared to struggle to understand the features of FXS, and uncertainty in this area may have impacted on their decision-making. The genetics and inheritance of FXS seemed to be understood, and while the variability of FXS was mentioned in the brochure some participants clearly wanted more detailed information about the condition:

... you know, whether there's a more severe form of fragile X or whether there's a minor form... you can get some people that have disabilities yet they can communicate, yet some can't... [Caroline, 25, tested—normal result]

A lack of prior experience with FXS may have made it difficult for participants to understand the clinical features of the condition:

... I suppose it would be good to actually meet some children that actually have it. So I think the fact that I don't

know any children that are affected makes it something I can't perhaps relate to. [Sarah, 42, tested—gray zone result]

**Practical issues regarding carrier testing.** There was one main practical issue that influenced women's decision-making arising from the requirement (by the Human Research Ethics Committee) that participants had to return to the clinic on another day to provide a blood sample for testing. For some participants this was a barrier:

... actually to be honest I wouldn't mind having [*the test*] done but I really couldn't be bothered organizing the appointment [Michelle, 20, not tested]

On the other hand, for others, having to return to have the test was not perceived as a barrier as it provided time for deliberation:

... rather than just being put on the spot when I got to the appointment... it was good that I had the information to think about it for a little bit before I went in. [Christina, 20, tested—normal result]

... I was thinking, oh wow, I can know this... then I was hesitant and I was, I don't really know if I would really want that. And I don't know if I really want to know [Ella, 20, not tested]

Having the time to consider the carrier test gave participants an opportunity to discuss the test with others and to further weigh up the pros and cons:

I guess my immediate response was yes... but then after that initial response my second response was, well actually I need to discuss this with my husband... if it had been a case of well you get the testing done now or never at all I probably would have got the test done but I would have had a bit of a bad feeling about it... [Charlotte, 31, not tested]

There were no other practical issues related to carrier screening mentioned by participants. Having to provide a blood sample was not considered a barrier and, because the test was provided free of charge, cost was not an issue.

## Reflecting on the Decision

Participants who were tested did not express any regrets about having the test and were content with their decisions:

I mean I'm happy that I did it and I'm happy that I made the decision that I did [Amanda, 28, tested—normal result]

I think it was a good idea... I feel like it's in control now, it's gonna reduce the risk of being passed down to the generations and it's gonna save a lot of problems for later in life as well so I think it is a good idea that I did do it.

[Jenny, 19, tested—carrier result]

I'm feeling quite pleased that I had the testing done... I'm happy with the decision that I made

[Bec, 40, tested—gray zone result]

When asked by the interviewer about how she felt about her decision not to have the carrier test, one participant responded:

... although I was very close to [*being tested*]... [*my decision*] was well thought out but it was always, since it was first offered, it was 'nah'. [Ellen, 20, not tested]

One participant with a gray zone result had entered the study to help research. At the time of being tested she may not have fully understood the implications:

I think research is good and so for the purpose of research yeah, I'm happy to perhaps to do it but no, I don't really think that it benefited me. No. I'd say not.

[Sarah, 42, tested—gray zone result]

## DISCUSSION

The most interesting finding of this study is that participants utilized a two-step process of decision-making, which required time and deliberation. In the first stage, when participants were offered carrier testing, their stage of life and their experience with health-related matters influenced whether they would consider having the test. These two factors could occur together or separately. In the second stage, participants who felt the test was relevant to them went through a process of deliberation. This deliberation was influenced by: perceptions of the value of knowing carrier status; perceptions of the risk of being a carrier; understanding the features of FXS; and practical issues relating to carrier screening, with the value of knowing their carrier status being given the most consideration. The results support offering carrier testing for FXS to women who are not pregnant and suggest that women from the general population may face unique challenges when offered carrier testing for a condition with which they are not familiar. This is the first study to explore in-depth the process of decision-making in women who declined carrier screening for FXS as well as those who accepted. It provides valuable insight into how and why individuals from the general population make decisions regarding FXS carrier testing.

One factor influencing the first stage of making a decision about carrier testing was participants' reproductive stage of life. The increased likelihood that those planning families will take up the offer of carrier testing has also been observed in studies exploring population carrier screening for cystic fibrosis [Tambor et al., 1994; Honnor et al., 2000]. Additionally, the time in a woman's life when she is offered carrier testing for FXS may influence how beneficial she perceives the information to be [Anido et al., 2005, 2007]. In agreement with Anido et al., women may prefer to be offered screening when they are planning to have children rather than during the prenatal period [Metcalf et al., 2008].

Experience with health issues, genetic disorders, and disability also played a role in the initial stage of decision-making. It has been suggested that prior experience may be an important factor in an individual's decision-making about screening [McClaren et al., 2008], and these experiences may shape the decision-making process [Brown et al., 2002; Etchegary et al., 2008; Ziebland and

Herxheimer, 2008]. In the absence of any experiences with FXS itself, participants may incorporate other health-related experiences into their decision-making. Clearly, this process would be different for women with a known family history of FXS.

In a review of studies investigating influencing factors regarding population carrier screening for cystic fibrosis, perceived benefits of screening were related to an increased likelihood of having carrier testing [Chen and Goodson, 2007]. Our data showed that participants saw value in knowing their carrier status because they could use this information to inform family planning, whereas few discussed a motivation to learn their risk of FXPOI, and none mentioned FXTAS. This interesting finding suggests that even though FXPOI and FXTAS were in the information brochure women viewed carrier testing as primarily providing them with information related to their reproductive rather than personal health. While the information brochure stated the reproductive options available to carriers, aside from one person, references to termination of pregnancy were not clearly articulated. Although some women had carrier testing to aid family planning, they did not specify which reproductive options they would choose should they receive a carrier result. There are a number of possible explanations for this finding. For some participants termination of pregnancy did not appear to be a factor in their decision-making. For others, it appeared that they preferred to consider carrier testing in a step-wise fashion: first have the test and learn their result; then, if they received a carrier result, consider reproductive options. These women viewed carrier testing primarily as a way to provide information that may be useful for future decision-making, perhaps reflecting an inherent difference between screening in this setting and prenatal screening. Finally, it is possible that participants may have felt uncomfortable discussing termination of pregnancy. Given the participant-guided interview approach, they were not specifically asked about their views on termination of pregnancy and how that affected their decision-making.

Some women who chose not to be tested mistakenly felt they were not at risk of having a child with FXS because they had no family history of this condition. In the general population, a lack of family history may be perceived as minimizing a person's risk of being a carrier of a genetic condition [McClaren et al., 2008], yet people are often not aware, or may not have, a prior family history of FXS due to its complex mode of inheritance. Furthermore, it has been suggested that health beliefs are associated with family narratives and that absence of disease in a family can lead to individuals and families believing that they are not at risk [Gustafson et al., 2007].

Women in this study did not have personal experience of FXS and therefore they lacked specific knowledge about the condition. They relied on the study brochure to provide information about FXS but some found it difficult to relate to the list of clinical features of FXS. Similarly, Ryyananen et al. [1999] found that participants would have liked to have received more information about FXS and the meaning of a carrier result. In contrast, when exploring views of obligate carriers of FXS, McConkie-Rosell et al. [1997] reported that most carriers viewed FXS as a serious problem suggesting that those with a family history may be better able to formulate a perception of the severity of the condition.

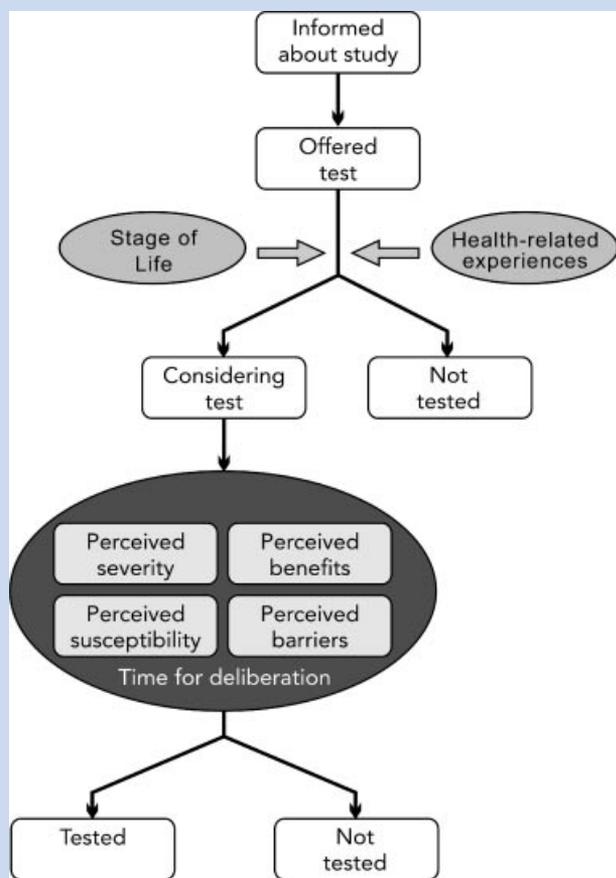
The requirement to return on another day for testing was reported to be a barrier by some, while most women reported that

it was important to have had more time to deliberate the pros and cons of testing. The time needed to consider the carrier test varied considerably and while some said they would be able to make a decision on the day they were offered carrier testing, others valued having more time. In contrast, Anido et al. [2005] described that women from the general population displayed a "why not" attitude when offered carrier testing for FXS. Providing time to consider the test may counterbalance this "why not" response. Brown et al. [2002] reported women's preference for an active involvement in decision-making about their health. Providing time for deliberation may allow women to actively seek information, talk to others, examine the test with respect to one's own values and beliefs, and consider the implications of a carrier result thus minimizing the likelihood that a woman will regret her decision. Our results suggest that it is important to recognize that each individual will require different timeframes when considering carrier testing.

One woman, who received a gray zone result, acknowledged that she had chosen to have the test to support research and reflected that knowing she had a gray zone result had not benefited her. It is important to ensure participants have fully explored the implications of undergoing genetic testing rather than basing their decision to participate on altruistic motivations.

The categories of factors influencing deliberation about carrier testing (perceptions of the value of knowing carrier status; perceptions of risk; uncertainty about the features of FXS; and practical issues) can be reframed using the Health Belief Model (HBM). The HBM has been applied widely to explain and interpret health behavior; it involves four components: perceived benefits; perceived susceptibility, perceived severity; and perceived barriers [Janz and Becker, 1984]. Each component influences the likelihood that an individual will adopt a health behavior. Of all models explaining health behavior the HBM appeared to be the best fit for our data because it focuses on the determinants of the health behavior [Bartholomew et al., 2006] and does not necessarily rely on a prior awareness of the illness and intervention under consideration. However, the HBM does not incorporate stage of life or prior health-related experiences. We are not aware of a single model that encompasses all the factors we identified as influencing the decision-making process. We suggest an alternative framework (Fig. 1) that integrates the various elements in decision-making that we have identified.

The findings in this study have implications for the practicalities of implementing a population carrier-screening program for FXS. Given that women from the general population may have a lack of awareness of FXS and will not have the lived experience of having a family member with FXS, there is a need to develop genetic counseling guidelines specific to this group. The guidelines should include: (1) discussion of prior health-related life experiences and how these experiences might influence decision-making; (2) exploration of perceived benefits of knowing carrier status to ensure a thorough understanding of the implications of a possible carrier result; (3) recognition that a flexible approach is needed regarding the amount of time required for decision-making; and (4) evaluation of informational needs to ensure an accurate understanding of the clinical features and inheritance of FXS, emphasizing that most carriers of FXS do not have a family history. Furthermore, women from the general population will need in-depth information about



**FIG. 1. Participants' decision-making process.** This figure illustrates the process participants went through when making a decision about carrier testing for FXS. The process appeared to occur in two stages. In the first stage participants determined whether the test was relevant to them and, if so, in the second stage participants considered the test in more detail.

FXS. The inclusion of vignettes of individuals with FXS might be useful in printed educational materials. Providing other sources of information [McConkie-Rosell et al., 2007] such as websites with videos of individuals with the condition and their families may also help to make the information more relevant.

Our study findings suggest that FXS carrier testing needs to be offered at a time in a woman's life considered by her to be "relevant" to actively engage women in a consideration of the pros and cons of testing. Screening may need to be offered to a woman at multiple stages of life in conjunction with raising awareness about genetic conditions in the general population. There is an inherent tension between efficient and effective service delivery and meeting the needs of the individual that will need to be balanced in a population carrier-screening program for FXS.

## LIMITATIONS OF THE STUDY

We progressively interviewed women until no new themes were observed in the interview transcripts. However, due to the

qualitative nature of this study our results should not be generalized to all genetic screening programs. Nevertheless, our study does identify a range of factors that influenced decision-making and could be used to inform the development and evaluation of other programs.

In this study we only spoke to women and did not interview their partners, and therefore we can only report from the women's perspective. A number of participants indicated that they discussed the carrier test with their partners, and it would be worthwhile including partners in future research.

## CONCLUSIONS

This study highlights the complexities involved in decision-making regarding carrier testing for FXS in women from the general population. The importance of the woman's reproductive stage of life, the association of carrier testing with relevance to family planning, and the importance of providing time for decision-making are all factors which support offering screening to women prior to pregnancy. Additionally, the findings that participants required more information about FXS, misconstrued that a family history was necessary to be a carrier, and reflected on health-related life experiences suggest that making a decision about carrier screening for FXS may be different for women from the general population than for those with a family history of FXS. More research into the process of how individuals from the general population make decisions about their health and family planning will inform the design and implementation of population carrier-screening programs for FXS.

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