Disclosure of a Fragile X Syndrome Diagnosis to Symptomatic Females: A Qualitative Study of the Parental Approach

Master’s Thesis

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Gretchen Schneider, MS, CGC, Advisor

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By
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ABSTRACT

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A thesis presented to the Graduate Program in Genetic Counseling

Graduate School of Arts and Sciences
Brandeis University
Waltham, Massachusetts

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Discussing the implications of a genetic condition with an affected child is challenging; it may be even more so in conditions like fragile X syndrome (FXS) where the child is cognitively impaired and at risk of passing the same condition to their offspring. Previous research has not examined the communication of a FXS diagnosis with symptomatic girls with *FMR1* full mutations. This study explored the experiences of parents discussing the diagnosis of FXS with their symptomatic daughters. We conducted semi-structured interviews with six mothers we recruited through the National Fragile X Foundation, FRAXA Research Foundation, and Facebook. We analyzed and coded interview transcripts using grounded theory and Atlas.ti software. The mothers’ ages ranged from 43 to 54 years. The daughters ranged in age, from 3 to 14 years at the time of diagnosis, and 14 to 26 years at the time of this study and the average age of disclosure of the daughter’s diagnosis was 13 years. Mothers in this study stated they used open communication and tried to give their daughters a positive outlook when discussing FXS with them. We found that most mothers used their daughters’ learning difficulties to initially explain their FXS diagnosis and the majority did not focus their conversations on the reproductive risks associated with FXS. Some mothers explained to their daughter that she has
the same condition as a male relative but not as severe. The parental challenges that emerged from this study were guilt felt by mothers for being carriers, trouble determining if daughters understood the information, and needing more guidance from health care professionals on how to have these discussions. The most common piece of advice for other parents was to find more resources, including help from health care professionals and support groups, when preparing to have these discussions.

Keywords: Family communication; fragile X syndrome; genetic counseling; genetic risk; adolescents; qualitative research
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INTRODUCTION

Communication about inherited genetic conditions among family members can be highly complex. Factors that contribute to the complexity include the need for parent to child communication about risks of inheritance and implications the disorder has for the individual’s children, their future health, and reproductive decisions. Discussing genetic risk information with children can be particularly challenging and parents face the dilemma of when, how, and what to tell them about the risks associated with their specific condition (Metcalfe et al., 2008). A number of research studies have investigated how parents discuss genetic risk information with their children. Gallo et al. (2005) examined how parents shared information with children who had one of the following conditions: phenylketonuria, sickle cell disease, cystic fibrosis, neurofibromatosis, hemophilia, thalassemia, Marfan syndrome, or Von Willebrand disease. They found that parents shared information based on their assessment of the child’s developmental readiness and interest, and parents stated they viewed information sharing as a continuum. Metcalfe et al. (2011) explored communication of genetic risk in families with the following conditions: cystic fibrosis, neurofibromatosis, Duchenne muscular dystrophy, hemoglobinopathies, familial adenomatous polyposis, and Huntington’s disease. Their study found that, in families where children had a better understanding of and more insight into their disorders, it was usually because parents had discussed the condition throughout childhood. Another study by Gallo et al. (2010) explored the opinions of health care providers regarding parental communication with their affected children. Gallo and his colleagues found that health
care professionals believed that starting the learning process early and being open and matter of fact helped children understand their disease and cope with symptoms. The findings from these studies support additional research on family stress and coping which have found that positive communication patterns can help develop resilience and minimize stress. This can lead to improved family functioning (Patterson, 2002). Research has also found that how individuals communicate genetic information to children may have a direct effect on future communication and adjustment to the diagnosis (Sullivan & McConkie-Rosell, 2010).

Discussing the risk of passing on a genetic condition may be even more challenging with children who are cognitively impaired, yet the challenges faced by families in this situation have not been well explored. A recent study by Faux et al. (2012) of eight parents of children with chromosome 22q11.2 deletion syndrome, who can have developmental delays and cognitive deficits, investigated the factors that influenced how parents informed their children of the diagnosis. Their study found that six out of the eight parents had disclosed the diagnosis to their child. Half the families who disclosed the diagnosis reported that the child brought up the topic by asking questions while with the other half, the parent initiated the conversation. The parents in this study reported the motivational factors for sharing this information with their child were to work towards the goals of coping, responsibility, independency, and self-sufficiency. These caregivers also reported that factors influencing them to share the diagnosis were being able to provide their child with an explanation for their differences and not hiding the diagnosis from them. Although this study examined the factors that influenced parents to share the diagnosis, it did not examine what specifically parents shared with their child.

Another model for understanding parental communication about genetic conditions associated with cognitive impairment is fragile X syndrome (FXS), which is the most common
single gene cause of inherited autism and intellectual disability. FXS is an X-linked condition with reduced penetrance (Abrams et al., 2012) and a prevalence that is estimated to be 1 in 4,000 (Turner et al., 1996). In the vast majority of cases, FXS is caused by a CGG trinucleotide repeat expansion in the 5’ untranslated region of the Fragile X Mental Retardation 1 (FMR1) gene. Repeat sizes fall into one of four major categories, with the normal range most commonly seen in the general population varying between 6 to 44 CGG repeats. Most males and females with FXS have a FMR1 allele with more than 200 CGG repeats, known as a full mutation. The large repeat size causes hypermethylation of the FMR1 gene leading to transcriptional silencing and subsequent absence or reduction of the fragile X mental retardation protein (FMRP). The premutation allele contains between 55 and 199 repeats and is usually unmethylated. Premutation carriers are at increased risk for fragile X-associated tremor ataxia syndrome (FXTAS), and approximately 20% of female premutation carriers have fragile X-associated primary ovarian insufficiency (FXPOI). Intermediate alleles contain between 45 and 54 CGG repeats, and at this time, are not thought to have clinical implications except the potential to expand to a premutation in future generations (Abrams et al., 2012).

The first clinical indication of FXS in children with the full mutation is usually delayed developmental milestones, including motor and/or language delays. Autistic-like behaviors, such as hand flapping, avoiding eye contact, and tactile defensiveness, may also be seen (Maes et al., 2000). Characteristic physical features, including tall stature, macro-orchidism, relative macrocephaly, prominent forehead, long narrow face, mid-face hypoplasia, highly arched palate, prominent mandible and large ears, are apparent but tend to become more pronounced with age. Personality characteristics include anxiety, hyperactivity, and attention deficit. The IQ values in males with full mutations range from 20 to 70, and females with full mutations exhibit a
spectrum of cognitive deficits that range from no obvious impairment to as cognitively impaired as males with FXS. About one third of females with an \textit{FMRI} full mutation have a behavioral phenotype that includes shyness, poor eye contact, anxiety, and social avoidance (Terracciano et al., 2005; Powell, 2003). Females without intellectual disability can experience relative deficits in math achievement and problems with attention and executive functioning (Keysor & Mazzocco, 2002). Males with FXS do not typically reproduce, presumably due to severe intellectual disability. Females with full mutations however, are more likely to reproduce, leaving these women at risk of transmitting the full mutation to their offspring (Crawford et al., 2001).

Research on FXS genetic testing and disclosure has focused on attitudes regarding carrier testing in children at risk for FXS, communicating genetic risk to asymptomatic daughters in families with FXS, and self-concept of asymptomatic females in families with FXS (McConkie-Rosell et al., 2011, 2009, 2002, 2000, 1997). McConkie-Rosell et al. (2011) explored parental perspectives on discussing genetic risk with daughters at risk of being a FXS carrier. At the time of the study, the adolescent daughters of the study participants were either confirmed carriers or non-carriers or had not been tested but were at risk for being carriers. Carrier females included premutation carriers and full mutation carriers with no obvious symptoms of FXS. Obvious symptoms of FXS were defined as the presence of IQ below 80, a diagnosis of autism or Asperger syndrome, or inpatient treatment for mental health issues. All parents whose daughters were carriers reported discussing FXS in frequent and multiple conversations using a resilient communication style which was defined as “any aspect of the conversation that emphasized the importance of honest, truthful, open communication between parent and daughter, an effort at reassurance, optimism, or as attempt to normalize the situation”. This was different from parents
who only knew their daughter was at risk to be a carrier. These conversations about the possibility of being a carrier included fewer resilient characteristics, suggesting that parents may struggle with how to communicate uncertainty such as carrier risk. Parents of the daughters who were carriers reported remembering an actual conversation with their daughters and these parents frequently planned the timing of this conversation. These parents reported that the content of the conversation focused on the daughter and what being a carrier meant for the future and family planning. The majority of parents reported that they believed their daughters had understood the message they were trying to disclose.

The purpose of this research study was to explore the experiences of parents with discussing the diagnosis of FXS with their symptomatic daughters. We examined how parents disclosed information about FXS, at what age the conversations started, what specifically parents shared with their daughter about the diagnosis and associated genetic risks, what guidance parents received, and advice for other families. Understanding these experiences can provide insight to genetic counselors and other health care professionals on how to better support and inform families that are going through these processes.
METHODS

Brandeis University’s Institutional Review Board approved this study.

Recruitment and Selection of Participants

We recruited participants through the National Fragile X Foundation (NFXF), the FRAXA Research Foundation, and the “I Love Someone with Fragile X Syndrome” Facebook page. We provided the NFXF with a recruitment notice (Appendix A) to post on their website under the Research Opportunities section, and sent the recruitment notice to all members of the FRAXA Research Foundation’s Fragile X listserv. Additionally, we posted the recruitment notice on the “I Love Someone with Fragile X Syndrome” Facebook page. We encouraged interested individuals to contact us directly by email. We then completed a brief questionnaire by phone (Appendix B) to assess eligibility. Subject inclusion criteria were that individuals must be 18 years or older; must speak fluent English; must have a symptomatic daughter, age 13 years or older, with an FMR1 full mutation; and must self-identify as the parent who had informed their daughter of the diagnosis and had major conversations with her about FXS. Symptomatic was defined as having a diagnosis of a learning disability or autism spectrum disorder.

Twelve individuals responded to the recruitment notice and six respondents were eligible for study participation, and we scheduled phone interviews at their convenience. The other six that were not enrolled in the study were found to be ineligible mainly due to the age of their daughter. We emailed a copy of the consent form (Appendix C) to each of the participants prior to the interviews and obtained verbal consent from each parent before the interviews began. We
offered the participants a $25 gift card to Amazon.com as a gesture of our appreciation for their time.

**Interviews**

We designed a semi-structured interview guide with open-ended interview questions (Appendix D). The questions in the interview guide focused primarily on how parents disclosed information about FXS to their symptomatic daughter, what age the conversations started, what specifically parents shared with their daughter about the diagnosis and associated genetic risks, what guidance parents received, and advice for other families. Additionally, we gathered information about each participant’s family and their symptomatic daughter. We used previous literature regarding communication of genetic risk information to minors for question design. Experienced genetic counselors and a parent of a daughter with FXS reviewed the interview guide. We used the same interview guide for each participant to ensure consistency; however the number and type of questions asked of each participant were dependent upon the issues explored during the interview. Interview duration ranged from 40 to 65 minutes.

**Data Management and Analysis**

We digitally recorded the interviews, and a professional transcriptionist transcribed them verbatim. We stored documents containing identifying information on a password-protected computer. The study’s author analyzed the data using ATLAS.ti (version 7.0), a software package for qualitative analysis of textual data, and a methodology based in grounded theory (Glaser & Strauss, 2009). The study’s author coded the interviews to condense the data into analyzable units, and assigned codes to sections of text, phrases, or paragraphs. As the themes emerged, the study’s author combined or discarded codes, which enabled her to reduce the number of codes. The study’s author consulted with a statistician who specializes in qualitative
research about the coding and themes. The thesis committee reviewed themes and subthemes, and one of the committee members with qualitative research experience looked over the transcripts and codes. The committee members considered the conclusions of the themes but did not analyze for content.
RESULTS

Characteristics of Study Participants

All six participants recruited for this study were mothers who ranged in age from 43 to 54 years and were from five different states. The participants’ symptomatic daughters ranged in age, from 3 to 14 years at the time of diagnosis, and were 14 to 26 years at the time of this study. Participants reported that all of their symptomatic daughters with FXS received services in school and had learning disabilities as well as behavioral or mental health issues. All of the daughters had anxiety, three had depression, and one had attention deficit hyperactivity disorder (ADHD). None of the daughters had a diagnosis of an autism spectrum disorder. Two of the six participants had more than one daughter with FXS and, for consistency, interviews were based only on discussions with the oldest daughter with FXS. Table 1 shows further demographic information about the participants, Table 2 provides information about their children and other family members with FXS, and Table 3 provides additional information about the symptomatic daughters with FXS.

Table 1: Summary of participant demographics

<table>
<thead>
<tr>
<th>Mother</th>
<th>Age (years)</th>
<th>Current Age of Daughter (years)</th>
<th>Age of Daughter at Diagnosis (years)</th>
<th>Age of Daughter When Diagnosis was Discussed (years)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>49</td>
<td>22</td>
<td>7</td>
<td>16</td>
</tr>
<tr>
<td>2</td>
<td>54</td>
<td>24</td>
<td>9</td>
<td>13 or 14</td>
</tr>
<tr>
<td>3</td>
<td>52</td>
<td>19</td>
<td>14</td>
<td>15</td>
</tr>
<tr>
<td>4</td>
<td>45</td>
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<td>10 or 11</td>
</tr>
<tr>
<td>5</td>
<td>51</td>
<td>26</td>
<td>2 or 3</td>
<td>10</td>
</tr>
<tr>
<td>6</td>
<td>43</td>
<td>16</td>
<td>14</td>
<td>15</td>
</tr>
</tbody>
</table>
Table 2: Summary of participant’s families

<table>
<thead>
<tr>
<th>Mother</th>
<th>Total Number of Children</th>
<th>Son(s) with FXS</th>
<th>Son(s) without FXS</th>
<th>Daughter(s) with FXS</th>
<th>Daughter(s) without FXS</th>
<th>Other Family Members with FXS</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>2</td>
<td>1</td>
<td>0</td>
<td>1</td>
<td>0</td>
<td>Nephew</td>
</tr>
<tr>
<td>2</td>
<td>2</td>
<td>0</td>
<td>1</td>
<td>1</td>
<td>0</td>
<td>Three nieces</td>
</tr>
<tr>
<td>3</td>
<td>2</td>
<td>0</td>
<td>0</td>
<td>2</td>
<td>0</td>
<td>Brother</td>
</tr>
<tr>
<td>4</td>
<td>2</td>
<td>0</td>
<td>1</td>
<td>1</td>
<td>0</td>
<td>Maternal aunt</td>
</tr>
<tr>
<td>5</td>
<td>2</td>
<td>1</td>
<td>0</td>
<td>1</td>
<td>0</td>
<td>Maternal aunt</td>
</tr>
<tr>
<td>6</td>
<td>4</td>
<td>1</td>
<td>0</td>
<td>2</td>
<td>1</td>
<td>-</td>
</tr>
</tbody>
</table>

Table 3: Summary of daughters

<table>
<thead>
<tr>
<th>Daughter</th>
<th>Current Age (years)</th>
<th>Age at Diagnosis (years)</th>
<th>Received Services in School</th>
<th>Learning Disability</th>
<th>Type of Classroom Attended</th>
<th>Diagnosis of Autism</th>
<th>Type of Behavioral or Mental Health Issues</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>22</td>
<td>7</td>
<td>Yes</td>
<td>Yes</td>
<td>Typical</td>
<td>No</td>
<td>Anxiety, depression</td>
</tr>
<tr>
<td>2</td>
<td>24</td>
<td>9</td>
<td>Yes</td>
<td>Yes</td>
<td>School for children with learning disabilities/inclusion/typical</td>
<td>No</td>
<td>Anxiety</td>
</tr>
<tr>
<td>3</td>
<td>19</td>
<td>14</td>
<td>Yes</td>
<td>Yes</td>
<td>Typical</td>
<td>No</td>
<td>ADHD, anxiety</td>
</tr>
<tr>
<td>4</td>
<td>14</td>
<td>4</td>
<td>Yes</td>
<td>Yes</td>
<td>Resource classes/typical</td>
<td>No</td>
<td>Anxiety, depression</td>
</tr>
<tr>
<td>5</td>
<td>26</td>
<td>3</td>
<td>Yes</td>
<td>Yes</td>
<td>Resource classes/typical</td>
<td>No</td>
<td>Anxiety, depression</td>
</tr>
<tr>
<td>6</td>
<td>16</td>
<td>14</td>
<td>Yes</td>
<td>Yes</td>
<td>School for children with learning disabilities/typical</td>
<td>No</td>
<td>Anxiety</td>
</tr>
</tbody>
</table>

Themes

Three main themes that emerged from the six interviews were logistics of disclosing FXS diagnosis to daughter, challenges parents face when disclosing the diagnosis to their daughter, and advice for other parents. Using codes to categorize the participant’s remarks under those areas, we identified recurring subthemes.
1. Logistics of disclosing FXS diagnosis to daughter

*Linking daughters’ learning difficulties to their diagnosis of FXS*

We asked parents to describe what they told their daughters about their FXS diagnosis, and found that the majority of the participants focused their initial conversation on their daughter’s learning disability or clinical features associated with FXS to explain what FXS is, thus making the connection more concrete for their daughters. All of the daughters in this study reportedly had learning disabilities and behavioral or mental health issues such as anxiety, depression, and ADHD. One mother disclosed her daughter’s diagnosis by saying,

“‘You have this Fragile X. This is part of why you have this-- some of these learning challenges. It’s going to be harder for you to do X, Y or Z thing sometimes. And that’s understandable.’ It sort of is a way to sort of say it’s okay. ‘It’s not like you’re making some mistake, this is genuinely hard for you.’ Not as a way to sort of pound it in and sort of we’re shaming you for this. It’s sort of as a way to sort of explain why things have sometimes been hard.”

Mother age 52; daughter age 19

Another mother said,

“I kept it very simple. Like, I said, you know, you have learning disabilities. But she knows about-- she accepts the fact she has learning disabilities because she went to a school for kids with learning disabilities. And so she’s comfortable with that. And so I said this explains why you have-- where the learning disability comes from and why you have such a hard time with certain things in school.”

Mother age 43; daughter age 16

A third mother added,

“And so we decided to share her diagnosis with her. But, just to give it a name so that it wasn’t her fault that it was hard for her to learn. She wasn’t doing anything wrong, and we told her that she had Fragile X. Tried to explain that it’s something that wasn’t her fault, that it’s something that it’s just part of her like she has brown eyes and she has Fragile X.”

Mother age 54; daughter age 24
**Little focus on reproductive risks**

We asked participants what they explained to their daughters about the genetic risks associated with FXS and whether their discussions included the risk of them having children with FXS. We found the majority of the mothers did not center their discussions with their daughters on reproductive risks. In fact, two mothers had not yet told their daughters, ages 14 and 16 at the time of the interviews, about the genetic risks associated with FXS. One mother reported she did not talk about how FXS would affect her daughter’s life in the future until her daughter began to ask questions. This mother stated,

> “We did not tell her until later how it would impact her life going forward. But when she started to ask more questions on her own, such as what if I ever want to have a child, what would happen? And those discussions happened in high school.”

Mother age 54; daughter age 24

Another mother waited until her daughter was in health class at school to explain reproductive risks. This mother said,

> “We explained to her when she was learning-- going through school and going through health ed. and learning about, obviously, sexual activity and all that, we explained to her that what she has, this Fragile X gene that she has, can cause her to have children as well with this disability.”

Mother age 51; daughter age 26

However, one mother did initiate discussions about the genetic risks associated with FXS by talking to her daughter about birth control when disclosing her daughter diagnosis to her. This mother stated,

> “And the reason why I did was because I wanted to find out whether or not she felt that she needed to be on birth control... And then that’s when I told her that whenever she does have children, she will have children with Fragile X. And she didn't seem like she understood. And so the way I explained to her, I said, ‘You’re going to have children like [her brother],’”

Mother age 49; daughter age 22
Use of own family to illustrate how FXS is inherited

We asked participants to talk us through how they explained the genetics or inheritance of FXS with their daughter. The majority of the mothers discussed how FXS was inherited by giving their daughter examples from their family. Many of the mother’s in this study had other family members with FXS and many of them had been tested themselves so they knew which side of the family FXS came from. One mother said,

“I tried to explain to her about how my father passed it to me and my youngest sister, but my two older sisters, they are not carriers. And certainly my brother is not, and I explained how he could not be. But that I gave, but through me, she has a full mutation and she, too, will give it to her children.”

Mother age 49; daughter age 22

Another mother added,

“And I explained to her that as simply as I could that I passed on these symptoms to her because she knew what the word symptom meant. And that [your grandmother], who is my mother, has a cousin who has the same disability. I did call it a disability then. I didn’t want to say a problem, I didn’t want to say a disorder. But she had heard the term in school, she had heard me use the term disability on the phone.”

Mother age 45; daughter age 14

In addition to using their own families to illustrate FXS inheritance, some mothers used other family members to normalize their daughter’s condition. These mothers wanted their daughter to know that she was not the only one in their family to have this. One mother said,

“We did talk a little bit about the genetics of it, like it came from mommy’s side of the family type of thing. And that’s why she has cousins who have it also and we talked about that, that she wasn’t the only one in the family who had it.”

Mother age 54; daughter age 24

Open communication

A common sub-theme that emerged when exploring how mothers disclosed their daughter’s diagnosis to her was open communication between them. Several mothers described
their conversations about FXS with their daughters to be open and straight-forward. One mother said,

“I like to play it very straight and I don’t tiptoe. That's just the way I am. I didn't sugar coat anything with her.”

Mother age 45; daughter age 14

Another mother added,

“Well, because the door was open from that point on, we’d have periodic conversations about it, generally brought up by her. And by the time she was in high school, we were talking about what would happen when she got married and had kids.”

Mother age 54; daughter age 24

The two mothers who each have two symptomatic daughters with FXS told them about their diagnosis together. One mother explained her reason for taking this approach was that their younger daughter would find out due to the open discussions about FXS within their family. She stated,

“Well, it’s just that we knew-- communication is pretty open in our house and I think as soon as one child had this information, the question would have-- it would have been obvious.”

Mother age 52; daughter age 19

One mother explained that her daughter also expresses her feelings about FXS openly with her. This mother said,

“I was sitting by her and I said to her that, I said, '[Daughter’s name], we don’t have to think about Fragile X every day.' And she said to me, ‘But I do.’ And I never heard her say anything like that before. And it was a very defining moment when she let me know that she’s the one that lives with this disorder and I can try to talk to her all I want about how it doesn't have to be about her disability. But she said to me, ‘But Mommy, I'm the one that lives with it. I have to live with it.’”

Mother age 45; daughter age 14
Mothers made discussions positive

Positive outlook was another sub-theme that emerged when mothers were disclosing their daughter’s FXS diagnosis. The majority of the mothers wanted their message to their daughter to be positive and offered their daughter reassurance she will have a full life. One mother stated,

“...Everyone has something. There is not a person that doesn’t have something. Your something is Fragile X and we all live with whatever our something is, for better or for worse. And that’s who you are. It doesn’t mean that it fully defines you, but it’s what makes you up. You have brown eyes and brown hair and you have Fragile X. So just make the best of it, move on. If you don’t like how things are to an extent you can change them, change them. Work harder, we can try some of these new medications if you want and see if they help, whatever you want we can try. But, you have to live within what you have.”

Mother age 54; daughter age 24

Another mother added,

“Is that it doesn't dictate her life. She will have a complete, full, wonderful life. That I don’t want her to think about Fragile X first when she gets up in the morning but that she is a nice person, a generous person, and someone that loves life regardless of roadblocks that will come our way.”

Mother age 45; daughter age 14

A third mother said,

“I just wanted her to know that it doesn’t mean that she can't do anything like anybody else. She can do whatever-- it’s not going to stop her from doing anything she wants to do. If she wants to go to college, if she wants to have a job, if she wants to have a family, she can do all that.”

Mother age 43; daughter age 16

Some mothers made their message about reproductive risks positive by telling their daughter about technology to help them have children without FXS in the future. One mother stated,

“We told them that it was going to play a role in their decisions about having children down the road, and that was going to be a big important thing for them to pay attention to. That there were going to be technologies developed over time that we’re going to be able to help them have children if they decided they want to have children without having to worry about passing on the Fragile X to their sons or daughters.”
Helping daughters understand that she has FXS but not like her male relative

Many of the mothers in this study had a male relative with FXS, including a son or brother. We found that three of the six mothers stated they wanted their daughters to understand that she has FXS but it is different than their male relative with FXS. One mother said,

“[My daughter] was immediately upset by the idea because I think she equated Fragile X, as much as we wanted to calm them or to reassure them, she associated Fragile X with [her uncle]. And I think the implication in her mind was that we were telling-- and she was very aware of her own ADHD and the struggle she was having at school. And I was always afraid that she was-- she thought the implication was that she was disabled, too. And we tried to discourage that thought and to encourage her to realize all of the great--her great strengths and the things she was doing well and I reminded her that I had it and very likely my mom had it to some extent.”

Another mother stated,

“She knew that she was a part of that and we had told her that she had it, but it was different. So when he would act up, she's like, ‘Am I going to be like that? Is that going to happen to me?’ And we would always say to her, ‘No, no, no, that’s not going to happen to you, it’s different.’”

One mother explained the genetics to help her daughter understand the difference between how FXS affects males and females differently. The mother said,

“I drew it out for her the x, two xx, and showed her how the repeats repeat itself so much and then it turns off this protein in your brain that is responsible for certain things. And I showed her how she still has another x chromosome that could compensate and that’s why she doesn't have it so severe. It’s not like her brother, who has more symptoms. She only has, you know, learning disabilities and shyness.”
Beginning discussion of FXS around adolescence

We asked study participants how old their daughter was when conversations about her diagnosis started. All mothers disclosed the diagnosis to their daughters during adolescence. The age ranged from 10 to 16 years. The average age was 13.3 years. The amount of time between their daughter’s initial diagnosis and the time of disclosure ranged from one to nine years. Some mothers reported events that prompted the disclosure, such as their daughter was changing schools or going onto a research study, while other mothers did not report a certain event leading to the disclosure. Two mothers reported they just decided it was the right time to tell their daughters. One mother who did report an event leading to the disclosure said,

_The very first conversation I had with her was not until she was ten and we were getting to go see [a doctor who specializes in FXS] to be part of a study. And so I had to explain to her why we were going and what we were going to be doing there. I explained to her that she was different from other kids, different from there brother. And I explained why we were going and why she was different than other kids. And I explained to her that as simply as I could that I passed on these symptoms to her because she knew what the word symptom meant._

Mother age 45; daughter age 14

2. Challenges

_Mothers feel guilty_

We asked study participants what was the most challenging aspect of discussing their daughter’s diagnosis with her. We found that the majority of the mothers expressed guilt about being a carrier and passing FXS on to their children. One mother stated,

_“And so, I talk about those kind of things with [her daughter], that that's some of my guilt even though it shouldn't be, my head tells me that I shouldn’t feel guilty because I had no- - how do you know that you're going to pass that to your child if you don’t even know you’re a carrier of it? We’ve had conversations where I’ve told her how I sometimes have a little pity party that I couldn’t give them the typical family life. But she doesn't know anything different, really. So this is typical to her. But that makes me sad, that that’s typical to her.”_
Mother age 49; daughter age 22

Another mother added,

“Honestly, I felt guilty. I mean, I know it’s not rational, but I really feel-- it’s hard to get past feeling bad that I passed that on to them.”

Mother age 52; daughter age 19

A third mother expressed that she feels like she let her partner down. She said,

“I remember when the kids were diagnosed, me as a woman, all of a sudden you feel like you let your partner down. And I know this sounds probably silly because God knows my husband would tell me all the time, ‘Oh, stop it. I love them just as much.’ But you say because this is my disability, you know what I’m saying? I’m the one with the gene, I’m the one with this gene inside me that gave it to them. So I don’t ever want her to have to feel what I felt like that, because I did. I was like, it came from me. He was fine. I brought this into the family.”

Mother age 51; daughter age 26

**Difficulty determining if daughter understands**

Another sub-theme that emerged was parents having trouble determining if their daughter understood the information they had given them. This sub-theme also came up when we asked parents how they determined if their daughter understood what they explained. Several mothers expressed uncertainty about whether their daughter understood how FXS would affect them in the future, in some instances because of their daughter’s reaction to this information. One mother said,

“It’s hard to assess whether she understands what you’re telling her, because she will say she does... She’s always afraid to show that she doesn’t understand something. I think that’s all part of-- she’s just been sort of struggling with these learning disabilities and I think she’s very chained by it. So I think the disability itself interferes with understanding it, you know?”

Mother age 52; daughter age 19

Another mother stated,
“But even though I talked about it, and told her that her children were going to be like [her brother], that she wouldn’t really understand that. You know, maybe I’m completely wrong. Maybe she does understand it and is just not bothered by it, you know? I don’t know.”

Mother age 49; daughter age 22

*Parents need more help*

We asked study participants if they received any guidance about how to talk with their daughter about FXS during or before they started these conversations with her. We found that all mothers expressed a need for more help, and many disclosed that they did not receive any guidance on how to have these conversations with their daughters. Many felt that some physicians did not know about or understand FXS. One mother stated,

“But when you’re learning about that as sort of a struggling pre-teen, you know, that’s a tough time. You’re so self absorbed and so awkward and so unsure of yourself, to learn information like that about yourself is hard. It was frustrating that a pediatrician couldn’t understand that. So I didn’t feel like I had a lot of people to go talk to about this.”

Mother age 52; daughter age 19

Another mother added,

“And so I just feel like this is so important to help people, more genetic counselors and more families, to help people understand it and talk about it because-- I mean, when we tell-- we rarely tell people, but when we do tell people, like professionals like doctors or-- nobody knows much. It’s crazy.”

Mother age 43; daughter age 16

One mother, who also has a son with FXS, said the schools had less information about girls’ learning and behavioral problems than for boys’. This mother stated,

“The school systems did not support it. No one understands this Fragile X. That was always my biggest issue with my son, especially for a girl, it's worse. Oh my God, it's worse.”

Mother age 51; daughter age 26
Several mothers suggested having a TV program or a special segment on a TV show about FXS, like they have previously done for other conditions. One mother said:

“You know, right now I think I wish that the Rugrats would have had one of their specials on it, you know? Because she would have understood it better if Barney sang a song about it or Rugrats did a, “My brother has Fragile X syndrome” cartoon at that time because she related to those things so much better. So, something like that would have helped.”

Mother age 54; daughter age 24

3. Advice for other parents

Find more resources

We asked study participants if they had any advice for other parents for discussing the diagnosis of FXS with their symptomatic daughter. The majority of the mothers would advise future parents to utilize as many resources as possible to help prepare themselves for discussions with their daughter about FXS. One mother said,

“Keep knocking on every door until you find people that can help you. Don’t give up like I did... I would say that would be my advice, is just keep getting answers and work with them and make them feel that it isn’t a death sentence -- I don’t want to say death sentence, but that’s not -- you know, that they can be this beautiful being and that they can prove everybody wrong.”

Mother age 51; daughter age 26

Another mother said,

“I would say they should take better advantage -- or to do their best to find resources to help them before they have the conversation and to plan to let their kid lead them as far as what they feel like they want to know at the time and to plan on allotting -- to plan on it being a process rather than an event.”

Mother age 52; daughter age 19

Some mothers reported that the information they did receive from genetic counselors or geneticists was helpful to them. One mother said the guidance she received from a genetic counselor about what initially to tell her daughter about FXS was very useful. She stated,
“Oh, it was very, very helpful. Like, it was so-- I wouldn't even have known where to-- how to begin or what to tell them or anything. So it was so, so helpful to have her tell me what to tell them.”

Mother age 43; daughter age 16

Tell daughters about their diagnosis when they are young

In addition to asking study participants if they had any advice for other parents for discussing the diagnosis of FXS with their symptomatic daughter, we also asked if they would have changed anything about how they approached these conversations with their own daughters. Three of the six mothers either advised to tell symptomatic daughters shortly after diagnosis or when they are younger that they have FXS, or questioned whether they should have told their own daughter at a younger age. One mother stated,

“You know, there's no good time. Sometimes I think would it have been-- and it all depends on the circumstances. Where in the family the child is, what birth number. I think anyway. But, sometimes I think that maybe we should have let it be known right from the beginning.”

Mother age 54; daughter age 24

Another mother gave advice to tell daughters as young as possible. She said,

“To explain things to them as young as possible and that their child is special and they're only really different on the inside, not on the outside. And that it doesn't make them different.”

Mother age 45; daughter age 14

However, one mother said the opposite. She said not to tell children too early. This mother stated,

“I would definitely not do it too early. Like, I don't think young girls, before they're, for sure, 12, need to know. And then like I had-- and just tell them the minimum they need to know and then let them ask questions when they're ready to ask questions.”

Mother age 43; daughter age 16
Two mothers did not comment on what age they thought was best to disclose a FXS diagnosis to a symptomatic daughter. However, participants were not directly asked what they believed would be the best age to disclose a diagnosis to symptomatic daughters.
DISCUSSION

The six interviews provided an in-depth view of parents’ experiences and issues surrounding disclosing a FXS diagnosis to a symptomatic daughter. All participants self-identified as the person who had major conversations about FXS with their daughter.

Logistics of disclosing FXS diagnosis to daughter

One of the main purposes of this study was to investigate how parents approached informing their symptomatic daughters of their FXS diagnosis. The theme that mothers used their daughter’s own struggles to initially explain their FXS diagnosis came up often in the interviews. Many mothers used their daughter’s learning challenges as a way to make the FXS diagnosis more concrete. Mothers wanted to give their daughters a name or reason for their difficulties. This was similar to a finding reported by some parents in the Faux et al. (2012) study of children who have chromosome 22q11.2 deletion syndrome. Those parents indicated that some of the factors that influenced how they shared the diagnosis with their child included being able to provide their child with an explanation for their differences and not hiding the diagnosis from their child. Most mothers in the current study focused their conversations on their daughters and her behavioral or learning difficulties. The mothers were trying to normalize the diagnosis by relating it to something the daughter was already aware of having. It is possible parents use this approach to first discuss their daughters’ FXS diagnosis, rather than focusing on future genetic risk since they might find this topic more challenging to discuss.
This study found that the majority of mothers did not focus on reproductive risks in their conversations about FXS. In fact, two mothers had not yet shared any information about reproductive risk with their daughters, ages 14 and 16 years old, at the time of their interviews. It is possible that mothers in the current study felt their daughters were too young or immature for this type of information. Additionally, the daughter’s learning disabilities may have played a role in the parent’s decision to not focus on this aspect of their diagnosis. This is similar to the approach reported by many parents of daughter who are asymptomatic carriers of FXS in the McConkie-Rosell et al. (2011) study. Those parents typically informed their daughters, at some point in their conversations about FXS, that being a carrier meant they could have an affected child, but it was not the focus of their initial conversation. In contrast to those who did not focus on the reproductive risks with their daughters, one mother in the current study led her initial discussion about FXS with her daughter by discussing birth control and the risk of having an affected child. Her daughter was 16 when this initial discussion took place, and the mother reported initiating this conversation to find out whether her daughter thought she should be on birth control.

Another theme that came up often was participants using their own families to illustrate the inheritance of FXS. The mothers gave examples of either carriers or affected family members to show how FXS was passed down from generation to generation and how their daughters can pass FXS onto their children. This was also observed in parents of girls who were carriers or non-carriers in the McConkie-Rosell et al. (2011) study. Similar to what has been previously reported, mothers in this current study stated that they used the family to illustrate and normalize the diagnosis by pointing out that other individuals are affected and that their daughters are not the only ones in their family to have FXS. It has been suggested that having this condition
portrayed as something that runs in the family as part of a “family story” can help to define the family identity making it easier for the daughter to accept her diagnosis (Sullivan & McConkie-Rosell, 2010).

 Mothers in this study frequently reported using open communication when talking about FXS, and several mothers described their conversations with their daughters to be honest and straightforward. In addition, all mothers reported having multiple conversations about FXS with their daughters. This is similar to the conclusion that open communication was a common finding reported among both adolescent girls, and parents of daughters, who were carriers, non-carriers and at-risk to be carriers of FXS (McConkie-Rosell et al., 2011, 2009). However, this is also different from another study by Gallo et al. (2005) who found that only approximately half the parents of children with various single gene disorders reported openly sharing information about their child’s condition and its inheritance with them. This difference may be due to the severity of several of the conditions including some that are life-limiting. Additionally, most of the children in that study who were not informed were under age 8. Parents in the current study may have used open communication because they wanted to be honest and did not want to keep information from their daughters. This would encourage their daughters to feel free to ask questions and have open discussions about FXS with them.

 Interestingly, both mothers in this study who have two symptomatic daughters with FXS informed their daughters of their diagnoses together. One mother chose to tell her daughters at the same time due to open communication in their family and the concern that if one daughter had known this information, the other daughter would have found out soon after. This may have been the rationale of the other mother with two daughters as well. Both of these mothers contemplated whether it was the right choice to tell her daughters together, and one mother
wondered if her daughters would have asked more questions if told separately. The other mother expressed her reason for telling her daughters together was because she treated them as a unit. However, she was unsure if this was a good reason to wait as long as she did to tell her oldest daughter. In the end, both mothers did not think they would have changed how they disclosed their daughters’ diagnoses. In families with more than one daughter with FXS, genetic counselors or other health care professionals may want to discuss with parents ways they can disclose the diagnosis with their daughters whether it be together or separately. Families may want to consider telling the daughters together but then having separate conversations with each of their daughters afterwards. These separate conversations can be age appropriate and tailored to what information the daughter is capable of understanding. This will also provide each daughter with individual time to ask questions she may have.

Many mothers offered optimism for the future and a positive outlook on FXS when disclosing their daughters’ diagnosis. One of the main messages mothers in this study wanted to get across to their daughters was that FXS is a part of them but does not define them. These mothers want their daughters to have happy and fulfilling-lives that are not limited by having a diagnosis of FXS. This was similar to a finding in the McConkie-Rosell et al. (2011) study that reported parents of daughters who were carriers of FXS offered reassurance and provided a positive outlook to their daughters. Many adolescent girls and young adult women in the McConkie-Rosell et al. (2009) study felt very strongly that information should be positive and the majority endorsed an open communication approach. This advice was consistent with a resilient family communication pattern which Sullivan and McConkie-Rosell (2010) suggests lead to improved family functioning. Additionally, some mothers in the current study provided positive information about technologies available to help their daughters have children without
FXS in the future. This is also similar to the McConkie-Rosell et al. (2011) study which reported that parents of daughters who are carriers of FXS provided positive information about reproductive technology.

A novel finding in this study was the subtheme ‘helped daughters to understand she has FXS but not like her male relative’. Some of the mothers in this study explained to their daughter that she has a condition that is the same as one of her male relatives but it is not as severe. It seems that some daughters had a difficult time understanding this concept because they would either get upset or still express fear that they would have similar behaviors as their male relatives. This unique perspective, not previously described in past studies, may help other parents and genetic counselors best approach conversations of this nature if the family also has a male relative with FXS, such as a son, especially if daughters ask why there is a difference when they both have the same condition. Parents may want to prepare how to answer questions like “Am I going to be like my brother?” a head of time. Genetic counselors and other health care professionals can help parents prepare for these conversations by working with them to develop a plan and discussing how to answer questions that may arise during these discussions. More data is needed to determine if daughters comparing themselves to male family members is truly a common issue.

Mothers in this study typically started their conversations about FXS with the daughter’s learning disability to give their daughter a name or reason for their difficulties. In later conversations, some mothers talked about the reproductive risks and inheritance of FXS. The ages when mothers disclosed their daughter’s diagnosis of FXS ranged from age 10 to 16 years, with an average age of 13.3 years. The Faux et al. (2012) study found a younger age of diagnosis disclosure of 9.6 years for children with chromosome 22q11.2 deletion syndrome. However, the
average age from that study excluded two of the eight parents who had not yet informed their child of their diagnosis, ages 13 and 15 years. A factor that may have influenced their earlier age of diagnosis could have been children asking questions sooner because of the medical issues seen in chromosome 22q11.2 deletion syndrome. In the McConkie-Rosell et al. (2011) study, the majority of the girls at risk to be a carrier of FXS reported learning this possibility by age 13 years. This slightly older average age of disclosure in this study may be due to the daughters’ learning difficulties and immaturity as well as later ages at initial diagnosis, which ranged from 3 to 14 years.

Challenges

A recurrent sub-theme in our study participants was that mothers feel guilty about being a carrier of FXS and passing this condition onto their children. Several mothers in this study reported that the feelings of guilt made it challenging to disclose their daughter’s diagnosis to her. In a previous study of mothers who were carriers of FXS, most women reported that learning they were carriers changed the way they viewed themselves. These women did not specifically report feeling guilty, but they did report feelings of anger, depression, or lowered self-esteem related to having an affected child (McConkie-Rosell et al., 1997). This finding of their negative emotions impacting mothers was slightly different from the current study in which mothers specifically stated feeling guilty for being a carrier. Another study with findings more similar to ours found that guilt and grief inhibited mothers’ communication with their child if they had a serious maternal X-linked recessive condition (Metcalfe et al., 2008). Feelings of guilt could prevent parents from having open conversations or could hold them back from sharing some information about FXS with their daughter. Health care professionals should be aware of the feelings that parents experience, such as guilt for passing this condition to their child, and
provide advice and support so parental emotions do not hold them back from sharing information about their child’s diagnosis with them.

Several mothers reported difficulty in determining if their daughter understood how FXS would affect them in the future. Some of the barriers mothers faced in assessing their daughters’ understanding were their daughter not reacting, not asking questions, or their perception that the daughter would always say she understood information as a defense mechanism. This differed from the findings that in a previous study that the majority of parents of daughters who were carriers, non-carriers, and at-risk to be carriers of FXS reported that they believed their daughter had understood the message they were trying to disclose (McConkie-Rosell et al., 2011). This difference in the current study may be due to the daughters’ learning disabilities and behavioral issues, which could have impeded their understanding of what their parents were discussing with them. In addition, symptomatic daughters may be more likely to refrain from asking questions due to their anxiety.

All mothers in this study expressed a need for more help. Many disclosed that they did not receive enough guidance on how to discuss their daughter’s diagnosis with her. Gallo et al. (2005) also found that parents felt they did not receive enough direction about how to share genetic information with their children. Metcalfe et al. (2008) also reported the finding that parents felt a lack of support or advice from health care professionals about discussing genetic conditions with their children. It is important to note that the majority of parents in the current study disclosed their daughters’ diagnosis over four years ago including one mother who disclosed her daughter’s diagnosis 16 years ago. For the participants in this study, one issue may have been the availability of fewer resources at the time of their daughters’ diagnosis. However, the more current studies illustrate that lack of guidance for families continues to be a challenge.
even in an age when information about genetic conditions and better-educated health care professionals are much more readily available.

Advice for other parents

The majority of the mothers would advise other parents to access more resources to better prepare themselves for discussions with their daughter about their FXS diagnosis. The mothers that did receive guidance from genetics professionals said the information they received on how to disclose their daughter’s diagnosis was helpful. Future parents should seek genetic professionals, like genetic counselors, as well as other parents for guidance on conducting these discussions with their symptomatic daughter. Additionally, as our study demonstrated, there may be variation in the length of time between a daughter’s initial diagnosis and when parents begin discussions with her about her condition, making it important to re-contact genetics professionals in cases where a significant amount of time has passed.

Half of the study participants either suggested other parents should tell their daughters at the time of initial diagnosis or beginning at a younger age, or considered whether they should have told their own daughter at a younger age. Studies of health care professionals have shown that they believe that starting the learning process early and being open and matter of fact will help children to understand and cope with their symptoms (Gallo et al., 2010). However, one mother in this study advised the opposite. She suggested parents do not tell their daughters too early. Her reasoning behind this may have been that girls with FXS may not understand the complexity of FXS and how it may impact them in the future at a younger age. Every family will have to determine when it is the right time to start these discussions with their daughter, and there are many factors that come into play, including how many children are in the family, birth order, and the number of other relatives with this condition.
Limitations and future research

This was an exploratory qualitative study designed to gain insight into parents’ experiences with sharing the FXS diagnosis with their symptomatic daughters. The study was limited by small sample size, consisting of only six participants. However, the participants were not localized to one geographic area of the United States and we enrolled parents from multiple sources to help minimize the sampling bias and provide a broader perspective. In addition, while there was a range in the ages of daughters who have FXS, many of the daughters had high cognitive functioning. All the participants were mothers; no fathers chose to participate. The parents also self-identified as having an interest in participating in this study and therefore may not represent the views and experiences of all families who have disclosed a FXS diagnosis to symptomatic daughters.

Another limitation was that all six interviews were via telephone. While this maintained consistency, the anonymity may have affected responses. Additionally, the interviewer could not observe the participant’s nonverbal cues, and this may have had an effect on rapport building. Some of the initial discussions about FXS that participants had with their daughters occurred over ten years ago, which may have affected the participants ability to recall their experiences. In addition, as with all self-reports, there is a potential for social desirability bias. Finally, since the study used qualitative methodology, the study’s author subjectively created all coding and emergent themes. However, this is the first examination of the diagnosis disclosure to symptomatic daughters with FXS and so provided valuable insight into this parental experience.

Further studies should include a larger sample size, incorporating fathers, and including families who have daughters with lower cognitive ability. Additional studies should also delve into the daughters’ perspective of this process. This may help parents and genetic counselors
better understand the feelings, expectations, and understanding of adolescent girls and young adult women with FXS.
CONCLUSION

This study explored mothers’ experiences with disclosing a FXS diagnosis to their symptomatic daughter. Overall, the mothers in this study used open communication and tried to give their daughter a positive outlook when discussing FXS. Most mothers used their own daughters’ awareness of her clinical features of FXS to initially explain their diagnosis. As previously reported and demonstrated by this study, a diagnosis disclosure is a process that will typically happen over many conversations. During discussions, participants often used their own families to illustrate the inheritance of FXS and normalize the diagnosis in their daughter. However, some mothers reported they needed to explain to their daughter that she has the same condition as a male relative but not as severe. The majority of mothers did not focus their conversation about the reproductive risks associated with FXS. The biggest challenges reported by mothers were feeling guilty about being a carrier, trouble determining if their daughters understood the information, and needing more help from professionals. The most common piece of advice for other parents was to find more resources when preparing to have this discussion with their daughter.

Parents of children with a genetic condition such as FXS are faced with questions such as when, how, and what to tell their children. Families frequently turn to genetic counselors, medical geneticists, primary care providers and other health care professionals for help with these questions (Metcalf et al., 2008; Sullivan & McConkie-Rosell, 2010). The results of this study provide insight for health care professionals, such as genetic counselors, who are meeting
with families that have symptomatic girls with FXS. Genetic counselors who are more aware of the approaches parents take when talking to their daughters about FXS and the challenges they face can better help families come up with a plan on how to disclose their daughter’s diagnosis.

As seen in this study, most discussions between health care professionals and parents occur at the time of diagnosis and information sharing by parents with their children occurs many years later. Providers therefore should remind families to re-connect with them in the future for guidance and resource information as they prepare to talk to their child. Another approach to helping families with disclosing a FXS diagnosis would be staged genetic counseling sessions. Multiple sessions planned over several years may be helpful for parents to form parent-practitioner partnerships that can help them to build the steps to positive communication about genetic information with their family. Families should be encouraged to seek additional resources, develop a plan for their family, and to take the opportunity to explore and practice what they will share with their children (Sullivan & McConkie-Rosell, 2010; O’Daniel & McConkie-Rosell, 2010). Other parents may also learn from the experiences shared by the mothers in this study and be better prepared to speak with their daughters about FXS as they get older.
REFERENCES


Do you have a daughter with fragile X syndrome?

I am a graduate student in the Genetic Counseling Program at Brandeis University. I am seeking volunteers to participate in a qualitative research project. The goal of this study is to explore the experiences of parents of symptomatic girls with \textit{FMR1} full mutations who have discussed the diagnosis with their daughter. Only parents who have gone through this process, like you, have this knowledge. It is our hope that the knowledge we gain from this study will inform future parents, genetic counselors and other health care professionals.

Participation in this study is voluntary and open to parents who:

- Are 18 years of age or older
- Parent who self-identifies as the parent who had major conversations with their daughter about FXS
- Daughter must be a symptomatic \textit{FMR1} full mutation carrier and 13 years or older
- Must have informed their daughter of the FXS diagnosis
- Speak fluent English

Following a brief phone questionnaire to assess eligibility, participants will be asked to take part in one audiotaped telephone interview that will last approximately 30 to 45 minutes. All participants will be given a $25 gift card to Amazon.com as a token of appreciation for donating their time. I hope to have all interviews completed by February 2014.

All identifying information of participants will be kept confidential and will be destroyed after completion of the study. Identifying details will be changed to protect the privacy of the participants.

If you are interested in participating in this study, please contact me by email at lstobie@brandeis.edu by January 15, 2014.

I appreciate your willingness to participate in this study and look forward to hearing from you.

Sincerely,

Lindsey Stobie
Genetic Counseling Graduate Student
Brandeis University
Waltham, MA
APPENDIX B
ELIGIBILITY QUESTIONNAIRE

Demographics and Eligibility Screening:

Name: _________________________
Phone Number: (____)______-_________
Age: ____
City and State you live in: _____________________________
Number of children:____ How many have FMRI full mutations:__ Males:__ Females:__
Age of symptomatic daughter with FMRI full mutation:_____ 
Age of diagnosis:_____
Other family members with FXS: ______________
Pedigree:

Do you self-identify as the parent who has had major conversations with your daughter about FXS?  Yes   No
Has your daughter been informed of her FXS diagnosis?  Yes   No

Participant’s symptomatic daughter with FMRI full mutations: (Could you tell me about your daughter that has the FMRI full mutation?)
Receive services at school:   Yes    No
Has learning disability:   Yes    No
Has IEP:   Yes    No
Type of classroom she attends:___________________________
Diagnosis of autism or Asperger syndrome:   Yes    No
   Please specify:____________________________________________________________
Behavioral or mental health issues:    Yes    No   (Does she have anxiety?)
   Please specify:________________________________________________________________

Informed Consent:

I.  Conduct informed consent process and discuss confidentiality
   a.  The aim of the study is to learn more about the experiences of parents sharing the
diagnosis of fragile X syndrome with their symptomatic daughters who have
FMRI full mutations. I will be asking you questions about when and how your
daughter was told about fragile X syndrome and her diagnosis, how you assessed
their daughter’s understanding of the information, advice you may have for future
parents, and if you received any guidance. Any experiences you can share will be valuable.

II. Explain the presence and purpose of recording equipment
   a. With your consent, I will be audiotaping our interview so that I don’t miss anything. Afterward, the interview will be transcribed by a confidential transcription service and your name will not be associated with it. Any potential identifying information that might come up as we talk will be deleted from the transcript before data entry.
   b. Explain that the informed consent will be emailed to them
      I will be emailing you a copy of the informed consent after this conversation. We asked that you read the consent form before the interview. You will be asked if you understood all the information in the informed consent form, whether you have any questions, and if you voluntarily agree to participate in this study, before we begin the interview.
   c. Any questions so far?

When would be a convenient time to schedule the interview?

Date and time for interview: ________________________________
Appendix C

Informed Consent Form

Brandeis University
Department of Biology
Genetic Counseling Graduate Program

Informed Consent to Participate in Research

Communication of Fragile X Syndrome Diagnosis with Symptomatic FMR1 Full Mutation Females: The Parental Perspective

Principal Investigator: Gretchen Schneider
Student Researcher: Lindsey Stobie

Introduction

Gretchen Schneider is the Co-Director of the Clinical Training for the Brandeis University Master’s Program in Genetic Counseling Program. Lindsey Stobie is a Masters candidate in the Genetic Counseling Program at Brandeis University. This is a research study exploring the experiences of parents of symptomatic girls with FMR1 full mutations who have discussed the diagnosis with their daughter.

You are being invited to participate in this study because you are a parent of a symptomatic daughter with Fragile X syndrome.

Taking part in this research study is completely your choice. You should not feel any pressure to participate. You can decide to stop taking part in this research study at any time for any reason.

Please read all of the following information carefully. Ask any questions that you have about this research study. Do not consent to this study unless you understand the information in it and have had your questions answered to your satisfaction.

If you decide to take part in this research study, you will be asked to give verbal consent at the time of your interview. The verbal consent will be audiotaped and transcribed for documentation. You should keep this consent form for your records. It has information, including important names and telephone numbers, to which you may wish to refer in the future.

Purpose of Study

The purpose of this study is to explore the experiences of parents of symptomatic girls with FMR1 full mutations who have discussed the diagnosis with their daughter. Specifically we plan to investigate how parents approach disclosing information about FXS to their symptomatic daughter, what age the conversations started, what specifically do parents share with their daughter about the diagnosis of FXS and associated genetic risks, and what guidance during or before this process did parents receive. We hope that the experiences shared by the parents...
participating in this study will provide valuable insight for future parents, genetic counselors and other health care professionals.

**PROCEDURES TO BE FOLLOWED**
You will be asked to participate in a 30 to 45 minute audiotaped phone interview. During this interview, you will be asked questions regarding your experience with sharing the diagnosis of fragile X syndrome with your symptomatic daughter. The interview will be audiotaped, transcribed, and studied.

**RISKS**
Participation in this study presents no more than minimal risk. However, it is possible that taking part in the interview could cause distressing thoughts and feelings. Should that occur, Janet Rosenfield, a certified genetic counselor, is available to provide additional support. You may contact Janet Rosenfield at (617)413-2704.

**BENEFITS**
There will be no direct benefit to you for your participation in the study. We hope that in the future, information obtained from this study will help us gain a better understanding of the experiences parents have with sharing the diagnosis of fragile X syndrome with their symptomatic daughter.

**ALTERNATIVES**
An alternative is to not participate in this research study.

**PRIVACY AND CONFIDENTIALITY**
All records containing identifying information, such as names, email addresses, and telephone numbers, will be kept strictly confidential during the study. All study related documents and materials (including eligibility questionnaires, interview transcripts and audiotapes) will be kept in a secure location accessible only to the Principal Investigator and student researcher, and any databases containing identifiers will be password protected using a password known only to the Principal Investigator and student researcher. Transcripts, interview notes, and audiotapes will be labeled with a coded ID number, which will be assigned to you upon enrollment into the study. If you are quoted or referred to in any written or oral reports of the study, you will be given an alternate name. You will never be referred to by your real name or any other identifying information in any written or oral reports based on the interview.

**PAYMENT**
You will receive a $25 gift certificate to Amazon.com for participation in the research study as a gesture of appreciation for your time.

**COST**
There will be no cost to you to participate in the study, other than the time it takes to conduct this interview.

**WHOM TO CONTACT**
If you encounter any problems related to study participation or have questions about the study, you may contact the student researcher, Lindsey Stobie, at lstobie@brandeis.edu or (978)430-6175.

You may also contact the Principal Investigator for this project, Gretchen Schneider, at gretchen@brandeis.edu.

If you have questions about your rights as a research study subject, contact the Brandeis Committee for Protection of Human Subjects by email at irb@brandeis.edu, or by phone at 781-736-8133.
APPENDIX D
INTERVIEW GUIDE

Below is a general guide that we will use to lead our individual interviews. We may modify this guide as needed.

Participant Name:  
Date:  
Time:  

Background Information from Eligibility Screening:  
Daughter’s age of diagnosis:_____  
Other Children: Yes  No  Number:___ How many have FMR1 full mutations:___ Males:___  
Females:___  
Pedigree:  

III. Introduction  
a. Thank you for agreeing to talk with me about your daughter and experiences with talking to her about fragile X syndrome (FXS). We appreciate you sharing your insights. Only parents who have had the experience of sharing the diagnosis of FXS with their daughters, like you, have this knowledge. It is our hope that the information we gain from these interviews will provide valuable insight for future parents, genetic counselors and other health care professionals.  
b. I wanted to remind you that all identifying information will be kept confidential and will be destroyed after completion of the study. I will be audiotaping our interview so that I don’t miss anything. Afterward, the interview will be transcribed by a confidential transcription service and your name will not be associated with it. Any potential identifying information that might come up as we talk will be deleted from the transcript before data entry. Also, you can take a break or terminate the interview at any time for any reason.  
c. Did you receive and have a chance to look over the informed consent I sent you? If so, I will be turning on the audiotaping now. Do you understand everything discussed in the informed consent? Do you have any questions? Do you voluntarily consent to being a participant in this study?  
d. Any other questions before we begin?  

IV. Interview Questions  
1) I would like to start off by asking you if you could tell me a little more about your daughter. What are her strengths?  
   1a. What are her weakness?
Prompt: School, anxiety, social

1b. How old was she when you or her doctor first had concerns about her?

2) Could you describe how your daughter was diagnosed with FXS?
   2a. Who told you?
       Prompt: Pediatrician, geneticist, genetic counselor
   2b. How long ago was this?
   2c. Were the genetics of FXS explained to you?

3) Can you talk me through the initial conversation you had with your daughter about FXS in general, not necessarily her diagnosis?
   3a. What age was she when you first told her about FXS?
   3b. Can you discuss how the conversation was initiated?
       Prompt: Daughter asked, doctor recommended, parents thought it was time, family member with FXS

4) Who was the first person to tell your daughter about her diagnosis of FXS?
   Prompt: Parents, doctor, genetic counselor
   4a. If you were the person who told your daughter about her diagnosis, did you plan what you wanted to say before the initial conversation? How did you prepare?
   4b. Did you plan a certain age to tell your daughter? What age?
   4c. Did a certain event lead to the initial discussion about FXS? Can you describe that?

5) Could you describe what your daughter was told about her FXS diagnosis?
   5a. Can you discuss how the conversation was initiated?
   5b. What age was she when this conversation started?
   5c. What do you think having FXS means to your daughter?
   5d. How did she react to the initial diagnosis?
       Prompt: upset, angry, confused, indifference

6) Describe the message you wanted your daughter to take home from your discussion(s) about FXS?

7) What was explained to your daughter about the genetic risks associated with FXS?
   7a. Was the risk to have children with FXS explained to her?
   7b. If you were the person to explain the inheritance of FXS with your daughter, could you talk me through how you explained the genetics or inheritance of FXS to your daughter?
   7c. How would you describe her reaction to this information?
       Prompt: Confusion, anger, indifference

8) How did you determine if she understood what you explained?

9) Could you estimate how often you talk about FXS with your daughter?

10) If you have had more than one conversation with your daughter about FXS, have you found that your conversation has changed over the years?
    10a. In what way?
10b. Did you find the conversation changed after puberty?
11) Did you receive any guidance about how to talk with your daughter about FXS during or before you started these conversations with her?
   11a. From who? What kind of medical professional?
   11b. How did you feel about any guidance you received from medical professionals?
12) What was the most challenging aspect of discussing your daughter’s diagnosis with her?
   12a. Were there any factors that held you back from having these conversations with your daughter?
13) Do you have any advice for future parents that are in a similar situation as you for discussing the diagnosis of FXS with their symptomatic daughters?
14) Is there anything you would have changed about how you approached these conversations with your daughter?
15) Is there anything about this process that you think is important that I did not ask about?

V. Closing Comments
   a. Thank you for participating in this interview.
   b. How was the interview experience for you?
   c. I wanted to remind you that Gretchen Schneider and Sharyn Lincoln are available as a resource if you experience any emotional distress following the interview.
   d. I again wanted to reassure you of the confidentiality of responses.