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ORIGINAL RESEARCH

# Genetic Risk Communication: Experiences of Adolescent Girls and Young Women from Families with Fragile X Syndrome

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**Abstract** Little is known about how and what genetic risk information parents communicate to their children and even less is known about what children hear and remember. To address this void, we explored how genetic risk information was learned, what information was given and who primarily provided information to adolescent girls and young adult women in families with fragile X syndrome. We explored three levels of risk knowledge: learning that fragile X syndrome was an inherited disorder, that they could be a carrier, and for those who had been tested, actual carrier status. These data were collected as part of a study that also explored adolescent self concept and age preferences about when to inform about genetic risk. Those findings have been presented separately. The purpose of this paper is to present the communication data. Using a multi-group cross-sectional design this study focused on girls ages 14–25 years from families previously diagnosed with fragile X syndrome, 1) who knew they were carriers ($n$=20), 2) noncarriers ($n$=18), or 3) at-risk to be carriers ($n$=15). For all three stages of information the majority of the study participants were informed by a family member. We identified three different communication styles: open, sought information, and indirect. The content of the remembered conversations varied based on the stage of genetic risk information being disclosed as well as the girls' knowledge of her own carrier status. Girls who had been tested and knew their actual carrier status were more likely to report an open communication pattern than girls who knew only that they were at-risk.

**Keywords** Fragile X syndrome · Risk communication · Genetic testing in children · Genetic counseling · Carrier testing · Adolescents · Risk and resiliency

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## Introduction

Families may encounter multiple barriers to communication when they are informing relatives about genetic risk. Some of these barriers include not being believed, encountering negative emotions such as anger, difficulty in determining who is at-risk, uncertainty about how to explain the diagnosis and its inheritance, worry over providing the wrong information as well as concerns about when and how to inform (Forrest et al. 2003; Holt 2006; McConkie-Rosell et al. 1995; Tercyak et al. 2002; Tercyak et al. 2007). When the at-risk relative is a child, additional concerns may arise regarding the possible effect genetic information may have on the other's perceptions of the child and the child's own ability to understand and positively utilize the information.

Little is known about how and what genetic risk information parents communicate to their children and even less is known about what children hear and remember. The majority of prior research on how genetic information has been communicated has focused on adults who learned about family genetic information in childhood. Holt (2006) reported two distinct patterns of family communication about genetic risk for Huntington disease (HD), disclosing and nondisclosing. Regardless whether the adult children in their study were members of disclosing or nondisclosing families, all expressed a preference to learn about the

inheritance of HD and their own genetic risk early in life, preferably by a parent. More recently, (Bradbury et al. 2008) interviewed adult children who learned a parent's BRCA test result in late adolescence. The majority of the adult children in this study did not report being emotionally distressed by the disclosure and felt, in general, there were benefits to early communication of hereditary risk. In one study that did enroll adolescents, Sparbel et al. (2008) interviewed teens (ages 14–18 years) growing up in families where there had been a diagnosis of HD. The teens in this study also emphasized the importance of open honest communication about the diagnosis. In order to develop strategies to appropriately communicate genetic information to children, it is critical to explore with young people who are members of families with genetic disorders to explore their experiences as well as their preferences for how genetic risk information is communicated in their families.

Family communication is a major determinant in how families manage tension, stress, and strain and develop or maintain family functioning, adjustment and adaptation (McCubbin, Thompson, and McCubbin 1996b). Positive open communication leads to improved family functioning and resilience (Walsh 2003). Communication within a family may be directed to the individual for whom it is intended or communicated indirectly, though overheard or unintended exposure (Miller et al. 2000). In some families, information is not freely discussed as there are family rules about what can and cannot be openly discussed (Boss 1988). Families have different styles of communication, which may be influenced by both the family customs and responses to crisis as well as influenced by the meaning the family makes of the diagnosis and the importance and implications of the information.

The major purpose of this study was to describe the relationship among adolescent girls' self-concept, coping behaviors, and adjustment associated with knowledge of genetic risk for fragile X syndrome. We also explored the remembrances of the adolescents and young adults about how, when, and by whom genetic information was communicated to them. We previously reported our findings regarding self concept (McConkie-Rosell et al. 2008) and age preferences about when to tell and offer testing (Wehbe et al. in press). We found that, for many of the participants in this study, the emotional response previously thought to be associated with carrier testing may be related to disclosure of risk of the possibility of being a carrier, not to outcome of the carrier test, and for some girls, may be as emotionally difficult as learning one is a carrier. We also found that the study participants endorsed ages less than 18 for offering carrier testing and they felt that decisions regarding the timing to inform about genetic risk and offer testing should be tailored to the individual needs of the child and his/her family.

Fragile X syndrome is a common X-linked disorder with an estimated frequency of the full mutation of 1/4000–1/6000 (Crawford et al. 2002; Morton et al. 1997; Turner et al. 1996) and a carrier frequency of the premutation in North America of approximately 1 in 250 females and 1 in 800 males (Dombrowski et al. 2002; Rousseau et al. 1995). The full mutation causes a range of cognitive disabilities and autistic-like behaviors (Hagerman and Hagerman 2002). Individuals who carry the premutation are at increased risk for fragile X associated tremor and ataxia (FXTAS) (Hagerman and Hagerman 2004) and fragile X associated primary ovarian insufficiency (Sherman 2000).

We report here our findings regarding the experiences of the adolescent girls and young women about how they were informed about 1) inheritance of fragile X, 2) communication style/approach utilized and 3) what they remember being told.

## Methods

Using a multi-group cross-sectional design this study focused on adolescent and young adult women from families previously diagnosed with fragile X syndrome who knew they were 1) carriers, 2) noncarriers, or 3) at-risk to be carriers.

### Sample Recruitment and Data Collection

After review and approval of this study by the Duke University Health System (DUHS) Institutional Review Board, we recruited females aged 14–25 years. To be included in the study those adolescents ≤18 years must have had knowledge of her particular genetic risk status for at least six months prior to study participation. Young adults between the ages of 19 to 25 years must have learned their status prior to the age of 19 years. We expected variability in the length of time each had knowledge of her status and in the manner in which she learned this information.

We excluded girls with obvious symptoms of fragile X syndrome, defined as the presence of any of the following: IQ below 80, a diagnosis of autism or Asperger syndrome, or inpatient treatment for mental health issues. Genetic risk status was confirmed through review of medical records for those participants who had been tested using DNA analysis. At-risk status was confirmed through standard pedigree analysis and each at-risk participant had a 50% chance of being a carrier. Carrier testing was not offered as part of this study.

The study sample was recruited through the Fragile X Clinic at DUHS, the family support groups from the National Fragile X Foundation, and postings on the

FRAXA listserv. After completing the prescreening to ensure subjects met enrollment criteria, researchers traveled to the participants. All interviews were conducted by the study's principal investigator, AMR.

### Structured Interview

The structured interview was adapted for adolescents from one used previously in a longitudinal study of adults going through the carrier testing process (McConkie-Rosell et al. 2002; McConkie-Rosell et al. 2000, 2001; McConkie-Rosell et al. 1997). The structured interview was composed of both open and closed ended questions. The interview was piloted with 10 adolescents who knew they were carriers of fragile X syndrome and revised to help ensure that participants could understand all questions. We were interested in exploring how genetic information is communicated in a family, and for this reason, we also explored three specific stages of knowledge about fragile X syndrome and the associated genetic implications with the study participants. The girls and young adults were asked to describe when and how: 1) they first (*initially)* found out that fragile X runs in the family (i.e., that fragile X was inherited); 2) they learned that they could be a carrier (could have a child with fragile X syndrome); and for those girls who had carrier testing, 3) they learned their test result. Each question was followed with probes, asking them to describe their memories of what they were told, how old were they, and who told them or how they learned the information. We also asked participants' for advice about ways families and health care professionals can discuss genetic information with children. The interviews were audiotaped and transcribed verbatim.

### Qualitative Method

The transcribed interviews were uploaded into ATLAS Ti 5.0. We used directed content analysis, a qualitative method that is guided by theory or prior research (Potter & Levine-Donnerstein 1999) to analyze the interview data. We utilized family communication theories (Walsh 2006) as well as prior research on genetic risk communication (Holt 2006; Sparbel et al. 2008; Tercyak et al. 2002) to guide the development of our initial coding categories. The interview transcripts were repeatedly read and then coded considering the communication style, what information was shared, and who provided it or how it was learned. The emerging themes were identified and categorized and new codes developed as needed. The responses were then tabulated for each specific question and sorted based on genetic risk status. The interviews were first independently coded by AMR and EM or GAS then jointly reviewed until consensus was reached.

## Results

### Study Sample

Fifty-three adolescents and young adults, 20 who were carriers (mean age 18.35 years s.d. 2.5), 18 who were noncarriers (mean age 17.78 years s.d. 2.69), and 15 who knew they could be a carrier (mean age 17.87 s.d. 3.18) from 13 different states in the US were recruited into the study between 2003 to 2006 (Table 1). The majority of the participants were in high school at the time of their participation, had at least one sibling affected by fragile X syndrome and some had multiple affected relatives. Many of the participants reported learning that fragile X syndrome was an inherited disorder and that they could be a carrier before they were 14 years old (Table 2). Twenty-three percent of the participants could not remember a specific age that they learned about the inheritance of fragile X syndrome. Forty-two percent of the participants who had been tested knew their actual carrier status by age 13 years. There were no identified demographic differences among the three groups.

### How Participants Learned Genetic Information

Regardless of which stage of information was being provided, the majority of the participants in this study

**Table 1** Demographics of Study Participants

| | | |
|---|---|---|
| Ethnicity | | |
| | Caucasian | 50 (94.4%) |
| | African American | 2 (4%) |
| | Hispanic | 1 (2%) |
| Religion | | |
| | Baptist | 8 (15%) |
| | Protestant/Christian non-denominational | 27 (52%) |
| | Catholic | 9 (17%) |
| | Jewish | 2 (4%) |
| | No formal affiliation | 6 (11.3) |
| | No religious beliefs | 1 (2%) |
| Year completed in school at the time of the interview | | |
| | 7–8th grade (Middle School) | 6 (11.4%) |
| | High School (9–12th) | 31 (58%) |
| | Some College | 16 (30%) |
| Closest relative affected by fragile X syndrome | | |
| | Sibling/parent* | 36 (68%) |
| | 1st Cousin | 9 (17%) |
| | niece/nephew | 5 (9.%) |
| | Uncle/aunt | 1 (2%) |
| | Greater than 3rd degree | 2 (4%) |
| Multiple relatives affected | | 29 (55%) |

There are no significant differences among the three groups ($p>.1$ Pearson Chi-Square)
*One girl had an affected mother

**Table 2** Participants Remember Age of Learning Each of the 3 Stages of Genetic Risk

| | | Carrier | Noncarrier | At-risk |
|---|---|---|---|---|
| Learned fragile X was an inherited disorder | | | | |
| | 0–10 years | 3/ 15% | 7/ 39% | 7/ 47% |
| | 11–13 years | 6/30% | 2/ 11% | 5/ 33% |
| | 14– <18 years | 6/ 30% | 3/ 17% | 0 |
| | ≥18 years | 2/ 10% | 0 | 1/ 7% |
| | Don't know/can't remember | 3/10% | 6/ 33% | 2/ 13% |
| Learned could be a carrier (could have an affected child) | 0–10 years | 1/ 5% | 6/ 33% | 3 / 20% |
| | 11–13 years | 7 /35% | 6 / 33% | 7/ 47% |
| | 14–<18 years | 8/ 40% | 3/ 17% | 2/13% |
| | ≥18 years | 3 / 15% | 0 | 1/ 7% |
| | Don't know/can't remember | 1/ 5% | 3/ 17% | 2/ 13% |
| Learned actual carrier status | 0–10 years | 2/ 10% | 3/ 17% | na |
| | 11–13 years | 5 /25% | 7/ 39% | na |
| | 14– <18 years | 7 / 35% | 4 /22% | na |
| | ≥ 18 years | 4 /20% | 1 / 6% | na |
| | Don't know/can't remember | 2/ 20% | 3/ 17% | na |

*Ages are the remembrances of the participants. There is no significant difference in mean ages of the girls ($p$=.72, Kruskal-Wallis Test) or remembered stages of knowledge ($p$>.1 Pearson Chi-Square) among the three groups

remembered being informed by a relative, usually their mother.

*My mom, I know my mom was the one who told me. She sat me down. Um, I'm guessing I was about 6, maybe 5. I don't really remember the conversation we had, but I do know my mom was the one who told me.*

*19 years (noncarrier)*

Others reported they learned the information simultaneously as their family learned it.

*...it was two years, two or three years ago. Yeah, and um, I learned about it first because (my nephew) had it, we learned his diagnosis. And my sister explained it to me, so and it was really easy for me to understand And the more we thought we talked about it, she said, well, you could be a carrier, too.*

*16 years (carrier)*

Some of the participants who are carriers and one girl who is at-risk remember learning that fragile X was an inherited disorder either as part of a clinic appointment or at a fragile X related family conference. They were not, however, the focus of the visit, and reported conversations that were not directed to them.

*Well, I think because my brother was 10, so I would have been 11 or 12 and my mom would have told me and I remember when we were at the, we were at like some doctor, I don't remember what doctor it was. I don't think I was at the original meeting when they found out or something, but my mom was really having a rough time with it and like I was in the office with them and she started to cry because she didn't know anything about it. Well, so at that point, she didn't think the diagnosis was necessarily a good thing, so, but I was too young to really be just like, so I was like, okay, they have Fragile X. Now we know what's wrong with them. So what? But I was, like I felt bad for my mom because she has never been like, been like one to like break down or anything and that really took a lot out of her. So it's what I remember the most.*

*16 years (at-risk)*

A few of the participants who are carriers and noncarrier remembered going to a clinic to discuss the possibility of "being a carrier" and 25% of the participants who are carriers and one noncarrier were seen by a health professional to talk about their test result. If the participant remembered talking with a health professional about the possibility of being a carrier or her test result, the focus of the counseling session was the participant herself, not an affected relative. All but one of these participants reported they were initially told their test result by a parent.

*I was 17. I mean, my mom just said that we could be carriers, like we already knew whatever we knew about Fragile X and then she said we could be carriers of it and then when we went to speak to the specialist, um, she gave a full overview of it like charts and lots of stuff. It was great. It was helpful, I mean, it was a lot of stuff I didn't really understand, because science is not my thing and it was like DNA and like cells and stuff. Not things I like.*

*20 years (carrier)*

Communication Styles

Three different communication styles were identified: open, sought information, and indirect.

*Open*

*Always Openly Discussed.* The diagnosis of fragile X syndrome, clinical features, as well as inheritance was reported to have always been openly discussed within the family by the majority of participants. Some also reported that they had difficulty remembering exactly how and when they learned that fragile X was an inherited disorder.

> *I've always known because it's always been in my family line, so thing is I think I've always realized that I could be a carrier. Now it could miss me, and I could be a carrier and not miss me, but my brother could have it.*
>
> *17 years (at-risk)*

Approximately one third reported that knowledge about the possibility of being a carrier had always been openly discussed and a few felt they had "always known".

> *I can't remember an exact date because it's…we didn't have like a big sit down conversation. It's always talked about. It's always been out in the open. They've never tried to hide anything from us. If we have any questions, they're always willing to answer it, so as far back as I can remember since my cousin was diagnosed, I don't remember a time that I didn't know about it, so…*
>
> *20 years (carrier)*

Only three (two carriers and one noncarrier) reported that their actual carrier status had always been openly discussed.

> *No, I don't remember….well I kind of do. I've heard my mom saying like a million times, but I don't remember them saying "You're not a carrier". I don't remember that at all. I just remember knowing.*
>
> *18 years (noncarrier)*

*Actual Conversation.* About 60% of the participants remember having an actual conversation about fragile X syndrome being an inherited disorder. However, a few of those who remember that there was a conversation cannot remember what was said.

> *…my mom just…I knew that something was different about my cousin, and it was just sort of told to me just like someone would have said, "Your cousin's name is…. his eyes are blue." It was told to me in that way. Like, this is who he is, and this is how it is. Not…it wasn't broached to me like, "(sigh) listen, we got to…" You know…so, I just know about him having fragile X…..I was really, really young, like 10 or 11 or something, and my mom just told me that it was inherited and that…like how people get blue eyes and some people get brown eyes, but that this was from on my mom's side of the family, and there's something called a carrier, and the carrier maybe doesn't express the gene like…like, a mom may have brown eyes, but she carries a gene for blue eyes, and so she gives her child blue eyes, even though that's not expressed in her. So that's how it was told to me, and that's how I understood.*
>
> *21 years (noncarrier)*

Fewer than half of the girls remember an actual conversation about the possibility of being a carrier. However, of those who remember a conversation they almost all remember the content.

> *Yeah, they told me. Well, you know what, I don't know if my mom told me everything. She might have told me about my cousin and then my aunt, my aunt explained the whole carrier thing to me later, so maybe that did happen in two parts. Yeah, I knew about it from the beginning, early. She said that, she explained the differences between being a carrier and having it and how it's less severe in boys and girls because of the XX and XY and, yeah, she just talked about how she was a carrier, but she didn't have it, yeah, she just explained that to me I might be a carrier.*
>
> *24 years (at-risk)*

Of the girls who had been tested and knew their actual carrier status, almost all of the girls who were carriers and noncarriers remember having an actual conversation about their test result and most are able to remember the content of the conversation.

> *They came in the mail or my mom, I don't know how she got them, and it came back negative and she was all excited and then my sister had gotten tested and she just said our kids won't have Fragile X and that we're not carriers.*
>
> *16 years (noncarrier)*

*Told in stages.* Some of the girls remembered being told in stages.

> *It's just something that I just pretty much always remember knowing. Like I don't remember a certain day when they sat me down and said like this is what it is technically and explained it to me… I just remember*

*her telling me when I was really little, just explaining that my brother was different. Just like some things would be like oh this bothers my brother. She said that he had Fragile X and I heard Fragile X growing up and she just pretty much explained it to me all my life, just what he had, but as I got older she said this is what it is and genetically and explained it to me like scientifically, but didn't like, there wasn't a set date. It was just like involved in my daily life and I just learned that he was different, but not in a bad way, which was good because it made it easier. You're just accepting when you're little like that...*

*16 years (noncarrier)*

*Told multiple stages at once.* A few of the participants who had been tested learned their actual carrier status simultaneously with learning about the inheritance of fragile X or were told about the possibility of being a carrier at the same time they remember being told their actual carrier status. For these girls there was never a time that they remembered wondering about the possibility of "being a carrier".

*Well, I guess when I was tested for it is when I found out, you know, I guess I just found out that what a carrier was and . . .Um, I just remember being told that it's just, not that you have it, but that you can give it to, you can pass it down in your family, so that's all I remember about that.*

*14 years (carrier)*

*I probably just said like, can my kids get this?*

*18 years (noncarrier)*

And a few of the girls asked their mother for their test result.

*Yeah, my mom just…I asked her one day because I was like I've not heard, and she said, "Oh yeah, you and your sister both got an 86." She was like…and I was like, "Is that bad?" She was like, "Well, you know, it's not, you know, as bad as it could be, but it's not where you're not affected or your kids could not be affected," so I was like ok. Yeah, she…I mean, she had forgot I think that somebody called her and told her.*

*18 years (carrier)*

*Figured it out on their own.* A few of the girls who are at-risk and one noncarrier felt that they had figured out for themselves that they could be a carrier through a combination of exposure to information in the family, researching information for a school project, attending fragile X related conferences, or reading resources on the internet.

*I just kind of figured it out, after I had done the research and figured out it was inherited, I just basically thought about it and especially after my aunt had um a child with Fragile X, that I realized, you know, my mom's family, a lot of them were carriers.*

*16 years (at-risk)*

*Sought Information for Themselves*

*Asked questions.* A few of the girls reported they remembered asking for information about how fragile X syndrome was inherited and asked about clinical features apparent in a relative.

*I think my mom has just always told me about, because you know when you're a little kid and you get curious and you're going to ask, "well why can't he talk?" and "why does he make those noises and he can't talk?" and "why does she learn slower than me?" and stuff like that. And I think she just told me that a thing called fragile X and it affects some people in different ways and it affects boys more than it does girls.*

*15 years (carrier)*

A few of the girls who are carriers or noncarriers and none of the girls who are in the at-risk group initiated the discussion themselves by asking questions regarding the possibility of being a carrier.

*Indirect*

Approximately one fourth of the girls who are at-risk, reported learning fragile X was an inherited disorder and a similar number learned about the possibility of being a carrier, indirectly, through overhearing conversations of adults around them, either at home or in a medical clinic. None of the participants who knew they are carriers or noncarriers reported learning any of the stages of risk information in this manner.

*I think it was my aunt again, because she saw all those doctors and stuff and she would come by and tell momma about it (whisper). I was listening. She said that the girls are the carriers and the boys really don't have no concern about, well they should be concerned but not like the girls should. And some people can be carriers and then some people just are not carriers at all. If you have a boy is a baby or whatever, he has a more risk than having a girl.*

*18 years (at risk)*

Information they Remember Being Told

*Learning Fragile X is Inherited*

Early remembrances about learning fragile X syndrome is inherited, for the majority of the participants, concentrated on fragile X syndrome itself, clinical features and how fragile X syndrome affected their siblings or other relatives' behavior or development.

*Um, my mom had told me, but that's it. She just told me a little bit about it. I was maybe 11 because I was actually old enough to kind of understand. She just told me like what my brother had and everything, and he would probably…what he might do or what he couldn't do and um that I would have to help her out with him and be here and stuff…um, things like that.*

*16 years (carrier)*

Over half of all the girls gave details of their family's diagnostic saga.

*I think it was in middle school and I think, wait, wait, wait, it was when, (my nephew), was born, it was when he was a toddler and I knew something was wrong with him and I remember (my sister) going to these doctors and stuff and trying to figure out what was wrong with him and then they found out that it was Fragile X and I remember her explaining it to the whole family and the reason why it was Fragile X and is because it was passed down by family members, not everybody was the carrier though, but it was just like you say, maybe that switch came on and all.*

*18 years (at-risk)*

Over one third included statements about the genetic status of relatives.

*I'd say right when my brother was born, probably when he was turning three we found out. I mean we diagnosed him with like ADD and all these things. I mean, I was five I knew that he had fragile X, but I didn't really know what it was so I guess until about, I don't know, like up until about a couple of years ago. We (my mom and I) were just, I mean, just like talking about my brother and everything, we just kind of got into it and we were writing on a piece of paper that thing with my grandpa. We started at the top and made like a family tree. And then in biology, I just did a huge report on it.*

*16 years (carrier)*

A few reported not understanding, being confused, or scared about what they were being told.

*Yeah. I guess I was confused then because I was only going into sixth grade I think and um, I was just, I didn't really know what it was or I didn't at all, but I was just told that it was um, a genetic disorder, and I didn't know what that meant either, but I just, they just told me that he just had like a learning disability kind of and just that um, he needed help with more things like he does, but um, I guess that's pretty much, I mean . . . I think I was probably 11.*

*14 years (noncarrier)*

*I think at first I thought it was some kind of fatal disease, but I've learned that it's not (laugh). I think at first I thought I was going to die, and then I got scared.*

*18 years (carrier)*

*Learning About the Possibility of Being a Carrier*

For those participants who remember being told that they "could be a carrier", the discussions frequently included statements about others in the family who were also carriers or could be a carrier.

*…after like we found out like that my brother did have Fragile X, because I know they (my parents) didn't tell me like all at once, …they just like explained that like my grandma was a carrier and then like my mom was and then so I have like the chance of being one.*

*14 years (at risk)*

The content of the discussions often included supportive and reassuring statements.

*We never made it a huge deal. Like, you know, we always say it could be worse. I could have cancer or something, so we've just put it in perspective where a lot of people are a lot more unfortunate than we are. I was probably 9 or 10.*

*18 years (carrier)*

Less than one quarter of the participants remember being told that "being a carrier" meant she could have an affected child.

*Um, I'd say I was around 12 or 13 again and, I mean, mom just said that we could all be carriers for it and that my kids, she just told me like my kids could have it, but I've never been tested for it, so I don't know if I'm a carrier or not yet.*

*14 years (at-risk)*

*Learning Actual Carrier Status*

The focus of the discussion about actual carrier status was on the girl herself and implications of the genetic information

for the future. Some of the girls simply reported the basic facts in response to the question about what they remember being told about their carrier test result.

*Yeah. She (my mom), because she had told me about it and I asked, did I have it? and she told me about it. That the results came back that I had it. I think that I was 12.*

*15 years (carrier)*

*My mom just was like "I have good news, you guys aren't carriers." I guess she got told and she told us.*

*17 years (noncarrier)*

Others, especially the girls who were not carriers, expressed relief and gratitude when learning their test result.

*I remember my mom just said, "You aren't a carrier for fragile X," and I was like, "Oh that's really great" and I understood that it was really great.*

*21 years (noncarrier)*

And those that were carriers expressed concern and acceptance.

*I remember mom said, "We got your test results back." And I had been dreading it. For some reason, I had just always had a feeling, like I just knew in my heart that I was. I just need somebody to tell me, and she told me then when I got home that day. ...I was kind of upset about it, but like I said, I always felt like I'd known in my heart that I was. I can't really explain that. I've just always felt like I was, but I was a little upset, but it wasn't like a dramatic "I've got to tell you something. You're life's going to be changed." You know. It wasn't anything like that. Yeah, that's how mom's always…she's not, you know, been like "it's going to change your life." She's always been like, "We'll be willing to help in any way. It's not going to ruin your future. You're still going to be able to have kids, so that's…that always made it easier.*

*20 years (carrier)*

Many of the participants who are carriers included statements relating to their shared family genetic identity. Only two of the girls who are noncarriers included these types of statements.

*Well, when I first like found out about Fragile X, it was when, I think about 2 years ago because my brother was like probably 2, and um, we kind of like, I think my mom discovered this Fragile X thing, and she said like that you know that she thinks that…that my cousin has Fragile X, and it may explain a lot. So, we all got tested for it, and you know, it all showed that we were all carriers, we all had it.*

*20 years (carrier)*

Approximately, one fourth of the girls who are carriers remembered being told that their positive test meant that they could have an affected child.

*Yeah, I remember because I had to have blood drawn. And I remember it was on this arm, so yeah, I remember…. I don't remember much other than I was a carrier and that whenever I had kids, it would affect them, and that was, you know, something that would come up at a later time when I was, you know, ready for that.*

*18 years (carrier)*

And a couple of the participants who carried the full mutation included statements about having a "touch" of fragile X.

*The doctor, the doctor's office called my mom at work and when she got home, she told me. She told me that I have a little bit of it. Like a small portion. Like I have Fragile X, but it's not bad like my brother has and that I am a carrier.*

*14 years (carrier)*

Those few who were seen in a clinic by a genetic counselor or a doctor (geneticist or pediatrician) remember being given more detailed information about the inheritance of fragile X syndrome.

*I think it was, I had went in there and I talked to the doctor and she told me that I didn't have to worry about it cause I didn't have it and if I was ever to get married or have children, that I wasn't a carrier or nothing like that. I was 10, or around 12.*

*14 years (carrier)*

## Advice

Participants were asked to give advice on how parents, genetic counselors, or other health professionals should approach informing young people (like themselves) about fragile X syndrome, how it is inherited, and the genetic risk. The majority of the participants endorsed an open communication approach and they stressed the importance of having an actual conversation, allowing for questions, and ensuring the discussion is open and honest.

*Well, I know that they should do it when they think that the kid is ready to know. It is different on your understanding level when you start talking to them about fragile X. You need to sit down and have an actual conversation rather than, oh, by the way. Don't leave anything out, be straightforward and honest, especially if they are a carrier and there is something that could happen to them in the future.*

*19 years (carrier)*

Over half felt strongly the information should be given in a positive light:

*Keep it positive. Let them know that they have options Stress the importance of just letting them know that it's not the end of anything, and I think early awareness is important also.*

*20 years (carrier)*

provided with reassurance,

*I think they should just treat it like it's normal. Don't treat it like it's like this huge, life-shattering thing. I mean, just broach the subject like it's…just like any other trait maybe, even though it's obviously not. Yeah, it's not, but just know that we all have things that are different, and they should definitely want to know as much as possible because it is their life that they're affecting, you know.*

*21 years (noncarrier)*

helping them to accept what couldn't be changed.

*I think that they (my parents) just, I think what really helped me was just accepting, you know, accepting either way what has been given to you. You cannot change your genetic make-up, you can't take the genes out. That is how you are made and you are made like that for a reason. Just accept it.*

*19 years (noncarrier)*

Some of the girls who are carriers and noncarriers felt that it is helpful to normalize the genetic information, by emphasizing there are other families facing the same situation.

*Just remind them that there's other people out there like that…you're not the only person that's a carrier and that there's things that can be done, and that, you know, there's worse things that could happen to a person, and just I guess that's about it…just, you know, let them know that they're not the only one with it..*

*18 years (carrier)*

The girls who are carriers, more so than the girls who are not carriers or who know they are only at-risk, emphasized the importance of the person informing to be knowledgeable and to know the facts before talking to a young person about fragile X.

*I think that sometimes people talk about it like it is taboo to people that deal with it. Like they don't want to ask questions but they really need to instead of just wondering about it and thinking incorrect things, so I think that it is important for family members to be really educated so they can answer those questions when they come up.*

*16 years (at-risk)*

Many of the girls who were carrier and noncarriers and over half of the girls who are at-risk felt it was important to be informed and knowledgeable about fragile X for themselves.

*Just, I think the most important thing would be just the actual information and facts without any sort of…I mean depending on the person you would have to deal with the more emotional side of it, I guess that affects different people, I think. But the most important thing would just be, that you know everything that you can know about it and want to know.*

*21 years (at-risk)*

Some stressed the importance of making the information developmentally appropriate

*I mean, probably not tell them anymore than they are ready to be told. I mean, and that's hard to determine, but I mean, everybody's…not everybody's as mature as everybody else, and it just depends on the person you are having to tell I mean, it just depends on that. And you know, they should take that…that should be one of the biggest things they take into consideration. You know, you're trying to figure out when you're going to tell somebody is…are they ready for it?*

*18 years (carrier)*

and to provide it in stages.

*I wouldn't keep it as a big surprise. Just kind of make them always aware that they've got a genetic thing. Just kind of phase it in, give them more information when they're old enough to handle more information. Because why just have a big stressful mountain of information.*

*18 years (carrier)*

## Discussion

The adolescent and young adult women in this study were informed about their genetic risk by a relative, usually their mother. Only a few remember a clinic visit and, for those that do, the focus of the clinic appointment was either on the affected sibling or was to clarify information already provided by their family. Some sought information for themselves by doing school projects on fragile X syndrome or found information on the internet. However, for the majority the source of their information was their family. Three different communication styles were identified and the content of the conversations remembered by the adolescent girls and young adults varied based on the stage of genetic risk information being disclosed as well as the girls' knowledge of her own carrier status.

An interesting pattern emerged regarding not only the type of communication remembered but also the content of

those discussions. The majority of all the girls, regardless of genetic risk status, were young when they learned about fragile X syndrome being an inherited disorder and some have no memory of not knowing. If they do remember an actual conversation they may not remember the content of family conversations. They described a family communication environment that was open and information was easily discussed when the topic was their affected sibling or other relatives resulting in the girl's perception of "always knowing". However, as the information became more personal, transitioning from the affected individual(s) to the girl herself, the family communication pattern changed. The girls were less likely to report that learning they could be a carrier or learning their actual carrier status had "always" been openly discussed, but were more likely to report that they remembered an actual conversation with one or both of their parents or another family member. Almost all of the girls who had been tested remembered a specific time when they were told their carrier test result. They were also generally able to remember the content and context of these latter conversations. These conversations were usually initiated by the relative who was informing.

There are several possible explanations for this difference. Although some of the girls reported learning the stages simultaneously, they were, on average, older when they learned their at-risk or actual carrier status. Therefore, the conversations may have been more recent, resulting in clearer memories. Also, information about the potential of being a carrier and actual carrier status may be more personally relevant as it is directly related to implications for self, rather than a concern for their sibling or other relatives. More personally relevant information may be more memorable to a child (Rubin 2000). It is also possible that talking generally about fragile X syndrome is an easier task for families than talking about specific genetic risk for the girl herself and her own future family.

Parents may also be waiting on cues from their daughters as an indication that they are ready to learn more. It is striking that most of the girls in this study did not ask for information. Only a few remembered asking about why their sibling was behaving differently or asking about the possibility of being a carrier. This finding suggests that, if parents are waiting for their daughter to ask questions as an indication that she is old enough to understand or ready to hear more information, she may not ask. Some of the girls who had been tested remembered having to ask for their carrier test result, sometimes years later, suggesting that parents were waiting for an appropriate time to inform her or were having difficulty in determining what and how to tell her.

In contrast to the open discussions and direct conversations reported by the girls who knew their actual carrier status, one fourth of the girls who knew only that they were at-risk to be a carrier reported learning about the inheritance of fragile X syndrome as well as their own "at-risk" status indirectly. These girls reported overhearing "adult" conversations in medical clinics, at family gatherings, and at home. In families identified with BRCA mutations Tercyak et al. (2001) found that the greatest exposure of children to genetic information was indirect through contact with family members who were affected or who were undergoing testing themselves. This finding was also true in this study, but only for the girls who were at-risk, not for those who had been tested. Additionally, the girls who reported these overheard conversations did not ask questions to clarify what they had heard. Children may remember and react to what they observe and the emotions with which information is said, as much as what is said directly to them (Koopman et al. 2004). Thus, it is important for families to consider not only directed conversation but also the family environment, the family response to the diagnosis, how information is being discussed, and their children's indirect learning.

It is possible that the girls whose parents initiated carrier testing are utilizing fundamentally different family communication or coping strategies that lead to facing concerns in a direct manner from those whose daughters have not had carrier testing. However, the girls almost uniformly reported that the clinical features of fragile X syndrome and how it affected relatives was always openly discussed. It is only when the outcome is uncertain (i.e., am I a carrier or not?) that information was not directly discussed. Parental uncertainty about what to say and how to respond to possible questions about actual carrier status may lead to a hesitancy to discuss genetic risk information.

Parents and other family members may also be trying to protect the child from painful or threatening information. A few of the girls in this study reported that they did not understand the information their family was telling them and were scared or confused by it. The combination of uncertainty, indirect conversations, and complex genetic information may be confusing to a child. Direct communication with trusted adults that provides the available information, acknowledges the uncertainties, and creates an open environment that encourages questions may be reassuring to children (Walsh 2006). It is also important to make sure that the information being provided is developmentally appropriate and that it is provided with concrete examples from the child's own life experiences.

The content of the remembered discussions was also interesting. Initially, the girls reported the focus of learning about the inheritance was on their affected sibling or other affected relatives. Many of the girls spontaneously gave very detailed stories of their family's own diagnostic saga. Some of the girls in this study were very young children themselves at the time of the diagnosis and were re-telling the family story as it had been told to them. Family stories play an important role in defining the family identity as

well as in making meaning of an event (Langellier and Peterson 2006). Family stories are a means to develop a shared understanding among family members and play a critical role in the process of making meaning of as well as helping with adaptation to a stressful life event (McCubbin et al. 1996a). For the children in the family, a genetic diagnosis may become part of the shared family story, a part of the family culture.

When the discussion was about the possibility of being a carrier as well as actual carrier status the focus shifted from the affected relatives to the implications of the diagnosis of fragile X syndrome for the girl, herself. As the information became more personally relevant, the participants remember being told in a reassuring manner. The participants also included statements of hope, optimism, and acceptance that genetic status could not be changed. Many of the girls included statements about "others" in the family who were also carriers. The frequency of statements that included who else in the family was a carrier suggests that their families presented the information to them in a manner that helped to normalize it by highlighting that "being a carrier" was part of a shared family genetic identity. We have previously described the importance of the family genetic identity in helping to incorporate genetic information for the individual (McConkie-Rosell et al. 2008).

It is interesting that only a minority of the girls remember that the discussion about the possibility of being a carrier as well as actual carrier status meant that they could have an affected child. This finding does not mean that they were not told or that they were uninformed. We have previously reported (Wehbe et al. in press) that the girls in this study were knowledgeable and knew that being a carrier meant that they could have an affected child. It is possible that the take away message being given by their parents was one of reassurance and identification with like "others" allows for social comparison and normalization of the information.

The advice offered by the girls is this study is consistent with a resilient family communication pattern (Walsh 2006). Participants felt that when disclosing risk related information to a minor child it was important to be open and honest, making sure to provide factual knowledge, as well as positive reassurance. Resiliency is defined by Froma Walsh as "the capacity to rebound from adversity, strengthened and more resourceful" (Walsh 2006) p. 5). Resiliency is more than surviving a stressful life event or a crisis. Resiliency is an active process, allowing for both positive and negative emotions, leading to growth, endurance, problem solving, and the overcoming of adversity (Walsh 2006). Resilient families are characterized by an optimistic bias that whatever happens can be overcome, acceptance of what cannot be changed, positive outlook, hope, and a sense of self efficacy (Walsh 2003). Resilient communication provides clear consistent messages in both words and actions, clarification of ambiguous information, with both truth seeking and truth speaking (Walsh 2006).

## Study Limitations

There are several limitations to this study that need to be considered. Fragile X syndrome is an X-linked disorder and the findings may not be generalizable to other disorders. Additionally, in this study, we did not include questions regarding the phenotype of the premutation. We plan to include exploration of specific concerns regarding possibly reduced reproductive lifetime and FXTAS in a future study. Study participants also self selected for the study and may either over or under represent different types of families or communication patterns. We attempted to obtain a representative sample through recruitment from multiple sources. This study is also limited by cross-sectional design and the remembrances are those of the participants. Memories of childhood events may or may not be accurately remembered and/or retold (Jack and Hayne 2007). However, given these limitations, the adolescent and young women in this study provide a unique perspective about how they learned, what they remember about how they learned the stages of genetic risk information, as well as the advice they gave regarding how to best approach talking with children about fragile X syndrome.

## Future Directions

Study findings highlight the importance of how genetic information is communicated, both directly and indirectly, to children in families with fragile X syndrome. It will also be important to explore communication patterns related to other disorders as inheritance, level of risk (for self or offspring), and the potential to alter risk (i.e. screening, prenatal testing, etc) may affect how families discuss a genetic diagnosis and ultimately how children manage the information. In her meta-analysis Peterson (2005) found that the family response to a genetic diagnosis is influenced by a number of variables, including knowledge of mutational status, inheritance, health beliefs, who in the family is affected, or a carrier or noncarrier, and family emotional support. Similarly, these different family variables would also be predicted to affect the communication of genetic risk information to the children in the family. Resilient communication patterns can be developed (Walsh 2006). Intervention based research, characterized by knowledge development, utilization, and design and development (Thomas and Rothman 1994), is needed to develop and evaluate the effectiveness of genetic counseling strategies designed to foster positive communication patterns.

## Conclusion

One of the major objectives of genetic counseling is to facilitate adaptive coping through interventions designed to provide families with the knowledge, skills, and resilient self-beliefs required to cope, adjust, and affect control over their lives (McConkie-Rosell and Sullivan 1999). How to accomplish these objectives when the focus is on children in the family is not clear. The meaning and utilization of the genetic information for both the child and the family will change as the child's ability to comprehend increases with age as well as the child's emotional maturation (McConkie-Rosell and O'Daniel 2007).

Genetic counselors can not only explore with the parents the family communication pattern but also help the family to consider both what is being said directly to the child as well as what the child might have overheard. Families will vary in their abilities to talk with their children about genetic risk. Genetic counselors can partner with the family to help facilitate resilient communication. Families need support and education about the genetic disorder as parents and other relatives are the ones primarily informing their children. An important component of the genetic counseling is exploring how parents plan to present information to their children. There is a need for ongoing family discussion and genetic counseling targeted to both the age of the child(ren) in the family as well as which stage (learning inheritance, possibility of "being a carrier", and actual carrier test result) of genetic risk information is being provided. Discussions also need to be sensitive to the emotional developmental age of the child as well as the type of information that is being provided.

Genetic counselors can provide an environment in which parents can practice what they might say to their children now and in the future. Just as children grow and change so must the genetic counseling be flexible and tailored to the current and future needs of the family (McConkie-Rosell and O'Daniel 2007). Genetic counselors can work with the family to develop a plan, considering the different genetic information stages, to provide ongoing support at critical developmental ages to facilitate good communication between the child/young person and the rest of the family. Staged follow up genetic counseling allows for the opportunity to address misunderstandings, respond to the children's own questions, and consider the family's current educational and psychosocial needs as well as to provide anticipatory guidance. The findings from this study suggest that it is how risk information is communicated not just when to offer carrier testing that is important to facilitation of a child's positive adjustment and understanding.

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