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Communication of Genetic Risk Information to Daughters in Families with Fragile X Syndrome: The Parent's Perspective

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Abstract

Parental approaches to communicating information about genetic disorders to their children may be an important determinant in how the children manage stress as well as their adjustment and adaptation to that information. We explored communication patterns through structured interviews with 46 parents of daughters who learned about their genetic risk status as minors. Three different levels of knowledge about fragile X syndrome were explored: 1) informing that it has been diagnosed in the family and is an inherited disorder, 2) informing about the possibility of a daughter being a carrier, and 3) if testing had been done, informing the daughter of her actual carrier status. Additionally, parental perceptions of their daughter's understanding of the information were explored along with frequency of discussions. We found that communication about genetic risk was initiated by the parents. Five disclosure patterns were identified with variations in style, content, and frequency of communication related to the information that was being disclosed. Aspects of resilient communication were present for all levels of disclosure; however, as the information became more personally relevant for the daughter such as disclosure about the possibility of "being a carrier" for fragile X syndrome and there was uncertainty regarding potential outcomes, the conversations included fewer resilient characteristics. Uncertainty about what and how to present information may negatively affect a parent's ability to include elements of resilient communication when disclosing genetic risk information.

Keywords

Communication; Genetic risk; Minor children; Fragile X syndrome; Resiliency; X-linked; Genetic counseling; Adolescents

Introduction

Little information is known about how parents discuss genetic risk information with their minor children. How information is communicated within a family is important. Family stress and coping theory suggests coping and adjustment to a stressful life event may begin with communication (Boss 1988). The parental approach to informing children about genetic risk may be influenced by the disorder itself; whether there is risk of a direct health concern to the child or the risk is of being a carrier of a genetic disorder; and the family's response to the diagnosis, which can include the personalized meaning and the importance and implications of the information (Sullivan and McConkie-Rosell 2010). Positive

communication patterns can help minimize stress and can help develop resilience while negative communication patterns increase stress (Patterson 2002).

A critical characteristic of positive adaptive coping is family communication that is sensitive, open, and direct (Rolland 2006). Thus, the way in which parents communicate information about genetic disorders to their children may be an important determinant regarding how the child manages tension, stress, strain, adjustment, and adaptation to learning about an inherited genetic disorder in their family. Additionally, communication within the family plays an important role in the development of shared family expectations and goals, and helps to facilitate core family functions (Patterson 2002).

Possible barriers to open communication about genetic risk may include uncertainty about when to inform, what to say regarding the genetics of the disorder, and current and future implications for the child, and the child's ability to understand, cope, and manage the information (Sullivan and McConkie-Rosell 2010). Parents tend to perceive that it is their responsibility to inform their children about their genetic risk and at the same time, find it challenging due to concern about facilitating a positive adaptation of the children to the information (McConkie-Rosell et al. 1999).

Prior research has focused mainly on whether genetic risk information is disclosed to minor children. Tercyak et al. (2001) explored whether mothers undergoing testing for BRCA1/2 informed their children about their test result and found that approximately 47% informed their children; however, this study did not explore what information children were told or how parents approached informing their children. In a separate study which focused on characteristics of families that disclose versus those that do not, investigators found that mothers were more likely to talk about their BRCA1/2 test result if: they had a prior history of discussing their own health concerns, were interested in exploring testing of their minor children, reported a more open communication pattern with their children and/or perceived that they had a strong parent/child relationship (Tercyak et al. 2002). The reasons given for disclosure included: feeling the child had a right to know, a sense of responsibility to inform about a potential future health concern that might directly affect the mother and child, to prevent the child from worrying, to establish an environment of open communication, or to provide reassuring news if the test was negative. Reasons given for nondisclosure were related to the developmental age of the child (i.e., too young to understand), or the mother felt the information would worry or make their child anxious. Findings from these studies may not be generalizable to other disorders in which the risks are for possible future affected offspring.

We have previously published findings regarding communication patterns with respect to genetic risk reported by adolescent girls and young women from families with fragile X syndrome (McConkie-Rosell et al. 2009). Fragile X syndrome is an X-linked triplet repeat disorder. The expanded triplet repeat can be inherited as either a premutation (typically between 55 and 200 unmethylated CGG repeats) or as a full mutation (greater than 200 hypermethylated CGG repeats) (Nolin et al. 2003). It is the hypermethylated full mutation, which leads to loss of production of the FMR1 protein and causes a range of cognitive disabilities and autistic-like behaviors (Hagerman and Hagerman 2004). We found 3 different communication styles: open communication, information provided if the girl asked for it, and indirect communication. The girls and young women in this study provided insight into how they would like to be informed about their own genetic risk. Consistent with a resilient family communication pattern (Walsh, 2006), they reported preferences for having an "actual conversation," the information to be staged, given with reassurance, normalized, and that parents be truthful, honest and knowledgeable about the genetic information. However, many of the girls and young women also reported they were very

young when they learned about fragile X syndrome and frequently did not have any memory of having been informed, stating only that they “always knew.” Thus, findings from the interviews of the adolescent girls and young women were limited by their own perceptions and memories. What remains unexplored is how parents approach communicating information about fragile X syndrome, the possibility of being a carrier, and actual carrier status to their daughters.

Because little is known about what parents tell their children about genetic disorders that have been diagnosed in a family, as part of our fragile X adolescent study we also enrolled the parent who self-identified as the person who informed their daughter about the genetic risk related to fragile X syndrome. The purpose of this paper is to present parents’ perspectives on communicating genetic information about fragile X syndrome to their daughters.

Methods

Design and Recruitment

Using a multi-group cross-sectional design, this study focused on adolescent and young adult women and their parents from families where a family member had been previously diagnosed with fragile X syndrome. After review and approval of this study by the Duke University Health System (DUHS) institutional review board, we enrolled parents and their daughters who were members of families in which fragile X syndrome had been diagnosed. A purposive recruitment strategy was used. We were interested in enrolling adolescent girls and young women who had been informed about fragile X syndrome as minor children in order to explore their experiences of growing up with genetic risk knowledge and to assess the effect this information had on their developing self-concept. To be enrolled, the daughters had to have been informed that they were either: a) carriers, b) non-carriers, or c) at-risk to be carriers before the age of 18 years. We also enrolled the parent who self-identified as the family member who primarily discussed fragile X syndrome with their daughter. The daughter and her parent were interviewed separately. The findings from the daughters have been previously reported (McConkie-Rosell et al. 2009; McConkie-Rosell et al. 2008; Wehbe et al. 2009).

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. Investigators traveled to families to allow enrollment of a diverse range of families. All interviews were conducted by the study’s principal investigator, AMR. Interviews occurred in a variety of locations such as local medical clinics or the family’s home.

Interviews

The parental interview was developed from that used by Miller and colleagues (Miller et al. 1994), which explored parental communication with children about marital divorce. We adapted this interview to incorporate questions about fragile X syndrome. Using this structured interview we explored parents’ communication patterns regarding the discussion of the three different levels of knowledge about fragile X syndrome: 1) that fragile X has been diagnosed in the family and that it is an inherited disorder; 2) informing about the possibility of the daughter being a carrier; and 3) if testing had been done, discussion about the daughter’s actual carrier status. Follow up probes included asking what was said, describing the message they wanted their daughter to take away from their discussions, and how the timing of communicating the information was chosen. Additionally, each parent was asked whether they felt their daughter had understood the information and how they assessed understanding. Parents were also asked to estimate how often they had discussions

with their daughter about fragile X syndrome, her genetic risk status, and how often she initiated the conversations. The parental interview was piloted with 6 parents of children with fragile X syndrome and revised based on their comments. Pilot data are not included in the analysis.

Data Analysis

The transcribed interviews were uploaded into ATLAS Ti 5.0 and analyzed using directed content analysis. This is a qualitative method of analysis in which the development of the initial themes are guided by theory or prior research (Potter and Levine-Donnerstein 1999). In the development of our initial codes we utilized family communication theories (Walsh, 2006) and prior research on genetic risk communication (Holt 2006; McConkie-Rosell et al. 2009; Sparbel et al. 2008; Tercyak et al. 2002). The interview transcripts were then repeatedly read and coded to consider: a) the communication style b) what information was shared, c) perceptions of daughters understanding, and d) frequency of communication of the information. The initial codes were developed by AMR, an experienced genetic counselor and researcher. The data were then independently coded by JDG and AMR, jointly reviewed, and discrepancies were discussed until consensus was reached. The emerging themes that were not able to be coded in the initial framework were categorized and new codes were developed as they were needed.

Each of the three levels of disclosure about genetic risk information were analyzed and coded for: how parents communicated, the focus or message of the conversation, and common aspects of resilient communication. We specifically coded for aspects of resilient communication as our previous findings from the adolescent girls and young women found a strong preference for genetic information to be communicated in this manner (McConkie-Rosell et al. 2009). Resilient communication 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 an attempt to normalize the situation. If any of these characteristics were identified parents were categorized as having used a resilient communication style. Responses were then tabulated for each specific code and sorted based on genetic risk status. Each interview was grouped according to how the parents presented the three levels of genetic risk information and each level was evaluated for common trends. The data were grouped based on carrier status of the daughter (carrier, non-carrier, and at-risk to be a carrier) and analyzed for group similarities and differences.

Results

Study Sample

Forty-six parents and 53 daughters from 13 different states were enrolled in this study from 2003–2006. Descriptions of the demographics of the daughters who took part can be found in previous publications (McConkie-Rosell et al. 2009; McConkie-Rosell et al. 2008; Wehbe et al. 2009). Briefly, the majority of the daughters were in high school at the time they were in the study and had at least one sibling affected by fragile X syndrome, and approximately half had multiple affected relatives. There were no significant differences in age or sample demographics among the daughters in the three groups. The majority reported learning that fragile X was an inherited disorder and that there was a possibility they were a carrier by 13 years of age. Forty-two percent of the daughters who had been tested reported they were informed of their test result by age 13 years and some reported having no memory of not knowing their genetic risk status.

The number of parent interviews was limited by the number of daughters in each risk genetic category who were recruited for the primary study. When reviewing the parental

interview data we observed that if there were two daughters in the family, especially if one had been tested and another had not, the parent's recollections included segments describing experiences with both daughters and it was difficult to differentiate which information was communicated to each daughter. Consequently, 7 interviews yielded only partial descriptions of the process for a specific daughter. Where possible we included these data.

There were 46 complete interviews that included: 18 parents of daughters who are carriers, 16 non-carriers, and 12 at-risk (knew that they could be a carrier). The sample included: 2 fathers (both carrier males) and 44 mothers (43 carriers of the premutation and one non-carrier). Two of the mothers were African-American and the remainder of parents were Caucasian. Parents' ages ranged from 36 to 70 years with a mean of 47.4 years ($SD=7.7$), and all were the biological parent of the daughter. Sixty-five percent of parents learned that fragile X syndrome was in the family because of the diagnosis of another of their children, and 80% learned that their daughters could be carriers at the time of diagnosis of their other child or at the time they learned about fragile X syndrome in the family. The reason provided for having carrier testing done and the manner in which parents learned their daughter's carrier status are summarized in Table 1.

Patterns of Disclosure

The type of information being discussed with the daughters varied based on the level of disclosure, and distinctive patterns were identified (Table 2). When talking about genetic risk for fragile X syndrome, some parents reported separate conversations when communicating each level of knowledge of fragile X-related information to their daughters, others reported providing multiple levels within the same conversation, and other parents omitted particular levels.

Communication Style: Level of Disclosure and Content of Information

Communication styles and content varied based on the type of information that was being discussed, and results are presented with respect to each level of information disclosure: 1) explaining that fragile X syndrome is in the family; 2) explaining to their daughter that she could be a carrier; 3) discussing the daughter's actual carrier status, if testing had been done.

Explaining Fragile X Syndrome is in the Family—During the first level of information disclosure, parents discussed the basic information about fragile X syndrome with their daughters. All the parents reported their daughters were told about fragile X syndrome in some manner, and the majority of parents recalled communicating this information in a specific conversation:

"I remember sitting down and trying to explain to [my daughters] that their brother had something called Fragile X and I tried to give them the best description of what it meant so that they would understand it."

Mom age 43; daughters, non-carriers ages 14 and 17

Some parents reported having multiple conversations, and these were more commonly reported by parents of daughters who were carriers. Other parents reported fragile X syndrome always being discussed and while they did not recall a specific conversation, they were confident that the information had been conveyed:

"I think it's just always been very open, but not in a big heavy kind of a way."

Mom age 47; daughter carrier age 18

Open communication about fragile X syndrome was frequently described and was reported to have occurred in a relaxed manner:

“It just happened. We were talking and it came up. I mean, we have never been where it has been a big deal.”

Mom age 38; daughter carrier age 16

For the majority of parents, conversations with their daughter about this first level of disclosure focused on an affected relative, usually a sibling, and the clinical features as they manifested in the affected individual, including behavioral and developmental differences; as well as how fragile X syndrome can be inherited. Some parents described focusing their conversation entirely around the affected family member.

“It was more about her sister. The term fragile X was more about ‘your sister needs this’, or that, ‘your sister has therapies’...”

Mom age 47; daughter carrier age 18

Other parents described including the affected member as an example of how fragile X syndrome presents:

“We just sat them down and told them that we need to talk to them about their brother and I think she said ‘I knew y’all were keeping something from us’.” And we told them that we weren’t keeping anything. That we had to wait until we found out what it was, but basically we told them that he had something called fragile X and it was a genetic disorder that affects, can affect the family”

Mom age 43; daughter non-carrier age 17

Still other parents recalled attempting to explain the genetics of how fragile X syndrome is passed on in families:

“‘Well, you know, it’s just genes.’ I said, ‘You know, you pass on eye color and hair color and all those things,’ and I said, ‘that has been passed to your brother, and so we have to make the best of what we’ve got and figure out how to help him’.”

Mom age 45; daughter at-risk age 17

Parents of the girls who were carriers and those who were non-carriers also used illustrations from the family to explain inheritance of fragile X syndrome.

“Well I went back to my grandmother. I went from there to, from my dad to me, being that how the men give it to or carry it to their daughters. Males can’t carry to male....I went through all of that with her and she just totally understood.”

Mom age 4; daughter non-carrier age 15

This strategy was infrequently used by the parents of the girls who knew only their daughter’s at-risk status.

The majority of parents included components of resilient communication when disclosing basic information about fragile X syndrome like having a specific conversation and allowing for open communication. In addition, we found that many parents of daughters in all three risk groups highlighted the reason they were informing their daughters was that they felt it was important to tell them the truth:

“I sat down and I told them the truth, I didn’t try to beat around the bush, I just told it like it was”

Mom age 58; daughter at risk age 18

Explaining That Their Daughter Could Be a Carrier—The second level of disclosure involved communicating that one could be a carrier of fragile X syndrome. The majority of parents of daughters who were carriers and non-carriers skipped this level. For the girls who were at-risk to be a carrier it was the final level of information disclosure at the time of study enrollment. When talking about the daughter's possibility of being a carrier for fragile X syndrome there was a general shift among the parents to begin to focus the conversation on the daughter and away from the affected relatives. Those parents who discussed the possibility of being a carrier with their daughters were able to recall having a specific conversation with her about her genetic risk.

"[I said] 'this is a possibility that you could someday have a child like your brother, but that it's not, we don't know that right now, that's not a definite.'"

Mom age 38, daughter at-risk age 14

However, few parents reported having multiple conversations to reinforce the information.

The parents of the girls who are carriers and non-carriers who recalled informing their daughters about the possibility of being a carrier focused on explaining the genetics by referring to inheriting and passing genes through the family:

"I told her that I was at first a potential carrier and then a carrier of something that could be passed on to her and that she had the risk of passing it on to a child of her own because of that, because we inherit things, you know genes, from our parents. And that you have a cousin who has this, you see what he is like, we need to find out now whether you run this risk also, so that you can make plans for your future."

Mom age 55; daughter non-carrier age 19

The parents of the at-risk girls typically informed their daughters that being a carrier meant she could have an affected child:

"I let her know right from the beginning, if you have children, you have the possibility of having a child like your brother."

Mom aged 58; daughter at-risk age 18

About a third of the parents of the at-risk girls reported not having had a conversation with their daughter about the possibility of being a carrier. In this case, some parents described utilizing other resources to communicate this information.

"I gave her the literature so she could read about it herself, and I figured that was the third party talking to her. And, she did read it and seemed to absorb it pretty well. And so that's how we presented the genetic information and possibilities to her."

Mom age 51; daughter at-risk age 23

One parent used enrollment in this study as an opportunity to talk with her daughter about the possibility of being a carrier while other parents of girls who are at-risk reported that they did not remember specifically discussing the information with their daughter:

"I don't think we did. I mean, it was more like, it's not like we ever sat down and said, 'now,' you know, 'this means that when you have', she just knew that."

Mom age 58; daughter at-risk age 24

Resilient communication patterns were less evident at this stage of disclosure. Some of the parents of the daughters who were carriers or non-carriers referred to themselves or other family members as also being carriers as a means to normalize the information.

“I’m sure it probably came about in the context of that I have been tested and that I was a carrier and that ‘now that I know that I’m a carrier, there’s a possibility that you all may be also’.”

Mom age 40; daughter carrier age 18

This strategy was rarely used by parents of the girls who were at-risk. Additionally, only three of the 12 parents of the daughters who were at-risk recalled offering a positive message to their daughters.

“[we said] whatever happens, you are equipped to handle it. It’s not going to be anything too difficult to cope with. We’re not in control of certain things, we have no control of certain, like our genetic makeup. We love each other the way we are, the way we are created’.”

Mom age 46; daughter at-risk age 19

Discussing the Daughter’s Actual Carrier Status—The third level of disclosure was discussion with the daughter about her actual carrier status, either carrier or non-carrier, for the 34 daughters who had been tested. While most reported having a specific conversation with their daughters about their carrier status, the discussions differed based on the whether the daughter was a carrier or a non-carrier.

Parents of daughters who were carriers reported remembering an actual conversation with their daughters and for some parents, this experience was remembered in vivid detail, including time, place, and intention:

“I sat her down at McDonalds and I told her, ‘I need to talk to you one-on-on.’ I got a little serious, I got her attention.”

Mom 42; daughter carrier age 16

These parents frequently discussed having planned the timing of that conversation for their daughter, choosing a time, or delaying informing until the girl was older.

“I never actually talked with her about it at the time because she was so small and I knew she wouldn’t understand. But when she got up in like 10–11 from then on, then I’d talk with her about it. I told her that it came from fragile X from [her grandfather] being a carrier and that comes down from him to her.”

Mom age 41; daughter carrier age 17

Multiple conversations were common, and parents reported that the content of the conversation focused on the daughter and what being a carrier meant for the future and family planning, including that being a carrier meant she could have an affected child. Almost all of the parents of the girls who were carriers included this piece of information in their discussion:

“Uh, just more that it was a positive. Knowledge is power and understanding, you know, this is a possibility that someday she could have a child, you know, that also is a carrier or has a full blown, you know.”

Mom age 51; daughter carrier age 17

Some parents referred to the affected relative as an example of what their children could be like.

“You carry it like I carry it. That means you could have a child like your uncle.”

Mom age 47; daughter carrier age 18

When talking with their daughters about their positive carrier status, the majority of the parents included statements of reassurance that frequently offered optimism for the future and a positive outlook on fragile X syndrome. Some simply reassured their daughters that no matter what they would be okay. Mothers tended to use the fact that they were carriers to relate to their daughters to let them know they were not alone and that fragile X syndrome was something they all shared:

“That is why I said ‘auntie and grandma and me,’ it is not a big deal, that it is okay. You are not alone because we all have it too.”

Mom age 39; daughter carrier age 16

Or mothers attempted to normalize and/or provide a positive perspective on fragile X syndrome being in the family.

“This is something our family has, but not something that we bear. You know, not...it’s not our cross to bear. It’s kind of what the role of the genes. It was the role of the gene pool, and that’s, you know, some people have a hearing problem, or some people, you know, have a vision problem or whatever. We have fragile X.”

Mom age 51; daughter carrier age 17

In some instances parents attempted to reassure their daughters that they would help to ensure she could have children who did not have fragile X syndrome.

“I was very clear I thought in telling her that there’s technology now and that we would make sure that she had normal children. We’d do anything we could.”

Mom age 42; daughter carrier age 16

Parents tried to frame the message as positively as they could:

“I just told her that we got the results back and that she, again, I kind of tried to keep it light, that you had some of those repeats that makes it possible that you could pass it to your children. I didn’t, I never said, ‘You’re a carrier.’ I was like, ‘Your labs look like that you have some repeats,’ and I even told them, and this may not have been the right way to do it, that their repeats were low. That on a scale of, because I had been on the internet, and up to 200, I said, ‘Yours is only 85.’ You know, so I tried to make it sound like, you know, your number could be a lot higher. So, I mean, just, I was trying to think of things that would immediately make them feel better knowing that more information could be forthcoming later, but the initial finding out, I wanted it to sound as good as I could make it sound.”

Mom age 40; daughter carrier age 18

“That we are a carrier of this and it’s nice for us to be aware of this, and we need to learn more about it and she needs to learn more about it and for when she has her own family, but it’s not a bad thing. It’s really not a bad thing and it can be good. And who knows, by the time she goes [on] to have a child, things might be out there to help her a little bit more, but it’s really, it’s not that bad, is what I’m trying to tell her, you know.”

Mom age 44; daughter carrier age 14

Others chose to reassure their daughters that being a carrier doesn’t make them different.

“I wanted to be as optimistic as I could be that you are who you are. To have this label on you doesn’t change who you are today.”

Mom age 51; daughter carrier age 20

Most of the parents of the non-carriers remember having an actual conversation; however, instead of being planned, the conversation almost always occurred at the time the test results came back:

“The genetic counselor called me and I was so happy I hollered up the stairs. I said, “Guess what? You’re ok!” and that’s how I told her.”

Mom age 44; daughter non-carrier age 18

As with carriers, multiple conversations were common among the parents of non-carriers to reemphasize their daughter’s knowledge of her status:

[I said] ‘I hope you understand that this is not something that you’re going to have to worry about with any child that you would have.’ And this is something that I’ve said more than once.”

Mom age 48; daughter non-carrier age 18

Two parents reported disclosing the information after the daughters had asked/requested it:

“We told her at the time, I believe that she asked me ‘Do I have what my sister has?’ and I said ‘No.’ I don’t think that I explained to her what carrier was, she wouldn’t have understood what that was at 9, but um. Since then we haven’t really, I mean I don’t ever remember having a discussion about [it] she knows that she doesn’t have it and I believe that I have told her that she cannot pass it on, but we have never really had the long discussion about that.”

Mom age 44; daughter non-carrier age 18

Many of the parents reported reassuring their daughters they were not a carrier and not at risk for having an affected child.

“I was trying to reinforce that she’s, that she doesn’t have to worry about this. That this is, she’ll have, you know, other things to worry about, like how to get her hair cut, but she’s not going to have to worry about having a child with Fragile X.”

Mom age 48; daughter non-carrier age 18

It was common toward the end of that conversation for the focus to switch to the affected family member.

“Alright you are not a carrier, you are not affected by it, you don’t have to deal with it in regards to your children. But you do, will also still have to deal with it in regards to your brother and your sister. And that will always be a part of your life, and that carries over to just the world in general.”

Mom age 47; daughter non-carrier age 15

Perceptions of the Daughter’s Understanding of the Information

Overall, the majority of parents, regardless of their daughter’s carrier status, reported that they believed their daughter had understood the message they were trying to disclose. Some of the parents of daughters who are carriers and non-carriers, reported they felt their daughters had only a basic understanding of fragile X syndrome, and for the parents of the daughters who are carriers, some felt that their daughter could not fully appreciate what being a carrier meant for future family planning or parenting a child with fragile X syndrome:

“I don’t think that she fully understands the magnitude of it, no. But yes, she has that awareness that it is in her family, but I don’t think she fully knows the magnitude”

Mom age 39; daughter carrier age 16

Similarly, parents of non-carriers felt their daughter's understanding had developed over time.

"At that time, she didn't understand, but now I think that she pretty much understands it. Whether she knows X-linked and all of that stuff, I don't know, but she understands it."

Mom age 44; daughter non-carrier age 18

Other parents reported that they knew their child understood because she studied it in school:

"She has taken some genetics in high school and they did do fragile X, so she does understand somewhat more of what the carrier means and stuff like that."

Mom age 44; daughter non-carrier age 18

It was common for parents of the girls who were carriers and some of the parents of the non-carriers to have checked back or to have had repeated conversations to make sure their daughters understood the information about their genetic risk:

"And I do remember thinking that, that she can't comprehend that, but I've said it again. I said it one more time and I'll say it again."

Mom age 47; daughter carrier age 18

For the parents of the daughters who were at-risk, only a few reported checking back with their daughters to make sure she understood that she could be a carrier of fragile X syndrome. The majority of parents of the girls who were at-risk and some parents of carriers and non-carriers either guessed at what their daughters understood, reported not knowing what they understood, or relied on indirect methods for assessing their daughter's understanding.

[Do you think she understood?] "I think, I hope so, I think so, yeah."

Mom age 40, daughter at-risk age 16

Frequency of Discussion

Parents were asked to estimate how often they initiated conversations about fragile X in general and how often they thought that their daughters initiated discussions. The majority of parents reported frequent conversations with their daughter about fragile X in general, regardless of risk status (Table 3). Similarly, we asked parents to estimate the frequency with which they initiated conversations with their daughters about their genetic risk status and how often she initiated a conversation (Table 4). Less frequent discussions (both parent- and daughter-initiated) were reported related to a daughter's genetic risk status.

Discussion

The predominant focus of research in this area has been to explore whether parents have disclosed specific genetic risk information to their children. This study investigated what information is disclosed and how parents approach informing their minor children about the genetic risk associated with fragile X syndrome. How parents approach disclosing this information to their daughters is a critical step in understanding genetic risk communication and the role of genetic counseling in facilitating positive resilient communication within the family. The results of this study provide valuable insight into how parents communicate

genetic risk information to their daughters. Patterns of disclosure and communication styles were influenced by which of the three levels of information parents were communicating.

All parents, regardless of their daughters' genetic risk status, informed their daughters about the diagnosis of fragile X syndrome using a resilient communication style and reported frequent and multiple conversations. The relative frequency of communication of this initial level of genetic information to children may occur because parents have a need to explain developmental or behavioral differences in affected relatives. It may also happen in this way because the focus of these conversations was on the affected persons and not on personal discussions of future genetic risk for the daughter, information which parents may find more difficult to communicate.

Communicating to the daughter that she could be a carrier and disclosing her actual carrier status began the process of personalization of the information as the focus of the conversations transitioned from the relative(s) with fragile X syndrome to the daughter, herself. Parents of daughters who were carriers, non-carriers, and at-risk who informed their daughters about the possibility of being a carrier, were similar in their approach. These parents tended to disclose this possibility in one conversation, had limited use of a resilient communication style (including expressions of optimism and/or reassurance), and they typically did not check back for understanding. Highlighting the difficulty in disclosing this level of genetic risk, several of the parents of the daughters who were at-risk reported not having had any or only very limited conversations with their daughters about their genetic status. Furthermore, given the important role that communication plays in coping and adjustment to a stressful event, parental difficulty with communicating about the possibility of being a carrier is especially problematic for these daughters who are at-risk, because this is their final stage of communication until they choose to be tested.

This communication style contrasts sharply to the parents' initial approach to introducing fragile X syndrome to their daughters. Additionally, parents of daughters who had been tested often choose to avoid informing about the "possibility of being a carrier" and instead informed their daughters about the genetic risk only when their carrier test results were available. Many of the same parents who experienced difficulty in informing their daughters about the possibility of being a carrier and those who avoided this stage completely, were able to incorporate aspects of resilient communication when providing carrier test results. These findings suggest parents may struggle with how to communicate the uncertainty inherent in informing their daughters of the "possibility of being a carrier." Uncertainty related to this level of genetic risk communication may have multiple sources. The uncertainty may be related to how to describe the possibility of being a carrier as well as concerns about how to inform about what being a carrier for fragile X syndrome means for future family planning. Parents may be uncertain about how to respond to questions from the daughter, such as, "Am I a carrier or not?". Uncertainty may also occur because actual carrier status is unknown. These possible barriers to open communication may also explain why parents of girls who are at-risk typically did not have follow up conversations with them about genetic risk. It is important to consider each of these possibilities and the role each may play individually and in combination, on the communication of genetic risk information to children. Parents may need support in developing ways to approach talking about being at-risk to be a carrier.

For the parents of the daughters who were carriers and non-carriers, a difference also emerged related to informing the daughter about her actual carrier status. Parents of the daughters who were carriers reported generally planning and taking their time to reflect on how they were going to tell their daughter about her positive carrier status. While talking with their daughter, they emphasized truth telling and offered reassurance through

normalizing (i.e., that everything is going to be okay and they are not unlike any other member of their family), or that the family will be there to offer support for the future. The focus of the content of the conversation remained on the daughter, and the conversation style included elements of resilient communication.

The parents of daughters who were non-carriers, on the other hand, recalled delivering the information immediately after receiving the test results and shifted the focus of the conversation from the daughter back to concerns for the affected relatives. There may be several explanations for the shift from the daughter back to the affected individuals in the family. One possibility is that once the personal component of the genetic risk is resolved, though learning that the daughter is not a carrier and has no future risk, the conversations shift back to the pattern that existed previously when informing about fragile X syndrome in general. However, it is important to consider that a possible unintentional outcome of this shift in conversation is that daughters who are non-carriers may not have adequate information to feel confident in their own carrier status. This shift in focus back to the affected relatives, with limited explanation about the carrier test result, may explain why some of the daughters who are non-carriers did not believe their carrier test result and wanted to be retested (Wehbe et al. 2009).

It appeared that parents of daughters who are carriers and non-carriers attempted to establish a sense of genetic identity in the family about fragile X syndrome through emphasizing that other relatives were also carriers. We have previously reported developing a “family genetic identity” as a means to normalize genetic risk information (McConkie-Rosell et al. 2009, 2000). While this strategy was commonly used by parents whose daughters had been tested; it was very infrequently used by parents of daughters who were at-risk. It may be that this coping strategy is not incorporated unless the daughter’s actual status is known and the family genetic identity is being used to help illustrate the inheritance. It may also be that parents of daughters who were at-risk were concerned that highlighting the inheritance using relatives known to the daughter may lead to increased worry about her own carrier status.

Implications for Genetic Counseling

An important component of the genetic counseling process is communication of genetic risk information. Communication of genetic risk occurs in many different ways as part of a genetic counseling session. Often, genetic risk is presented by the genetic counselor and discussed with the parents so the inheritance and its implications are understood and the risk is personalized (Shiloh and Saxe 1989; Veach et al. 2007). The findings of this study suggest it is also important to explore with parents not only their own understanding of the inheritance of a genetic disorder, but also how they are communicating genetic risk to their families, especially the children. Thus, a critical aspect of working with families is the inclusion in the genetic counseling process communication of strategies for parents to utilize with their children.

Families can be strengthened and develop resilience based on how they face problems and adversity (Walsh, 2006). This process begins with communication. Genetic counselors can facilitate resilient communication through partnering with parents to: explore the current family communication patterns and provide an environment to practice what might be said, pose possible questions that might be asked by a child, explore the child’s ability to understand the information, and identify ways to incorporate components of resilient communication that are tailored to the family. According to Walsh (2006, p. 131), communication components that are important to develop are: clarity (clear consistent messages that are honest and truthful), open emotional expression (sharing of feelings, empathy, tolerance, taking responsibility for ones’ own feelings, and finding the humor and pleasure in interactions), and collaborative problem solving (shared decisions, conflict

resolution, resourcefulness, developing and focusing on goals, building on success and learning from failures, taking a proactive stance to prevent problems).

Genetic counselors can help families to consider each level of genetic risk information and develop the messages related to each. Communication about genetic risk is a multi-staged process that includes learning about the diagnosis and clinical implications, that the disorder is inherited, about the possibility of being a carrier, and actual genetic status (Sullivan and McConkie-Rosell 2010). Each level represents specific tasks that have unique implications for the child and requires the communication of specific messages related to genetic risk. These levels of information may occur all at the same time or may be staged and occur at different times. The daughters commonly recommended staging the information (McConkie-Rosell et al. 2009); however, as this was a descriptive study it is not possible to draw conclusions regarding outcome of one approach versus the other.

Findings from this study also suggest that being uncertain about what to say and how to present information may negatively affect a parent's ability to include characteristics of resilient communication when disclosing genetic risk information. Genetic counselors can work with families to facilitate communication of each level of information. Each step builds a foundation for positive adaptation and coping for the child. As the following quote highlights, children may understand more than parents expect and providing information in an open, truthful manner may help to provide an environment which fosters communication.

“Don't try to hide anything from them (children) because they're too smart for that. They're old enough for an answer, even if it's a simple answer first and then try to work your way into more complicated issues as they grow.”

Mom age 40; daughter at-risk age 16

Study Limitations

This study focused on the communication process of genetic risk information in families affected by fragile X syndrome. Fragile X syndrome is an X-linked disorder and findings may not be generalizable to other genetic disorders with different clinical features and inheritance. We analyzed the communication patterns of the parents after we had completed analysis of the daughter interviews and utilized the findings from the daughters' interviews to help inform development of initial codes of the parents' interviews. In order to minimize the potential of bias, the initial coding was completed by a research assistant, JDG, who was not associated with the prior analysis and had not been part of the data collection. The interviews were retrospective and represent the parents' memories of conversations. These recollections may have been influenced by new knowledge about their daughters' response to the information overtime or by their daughters' carrier result. The parents enrolled in this study self-identified as having an interest in participating and may represent a unique group of families interested in communicating genetic risk information. Parents and their daughters were enrolled from multiple sources to help minimize this effect and to provide a broader perspective.

Conclusions

We identified that even a highly motivated sample of parents experienced difficulty in the communication of genetic risk information, especially as it related to the uncertain outcome and personal meaning of “being a carrier.” While aspects of resilient communication were present for all the parents in this study, as the information became more personally relevant to the daughter and uncertain regarding outcome (as found in the disclosure about the possibility of being a carrier), the conversations included minimal resilient characteristics. When the threat was confirmed, as it was for the parents of the daughters who are carriers,

they were able to disclosure with optimism, normalization, and reassurance. Compounding the difficulty parents encountered when discussing genetic risk information, the majority of the daughters, regardless of their personal risk status or parental communication styles, were reported by their parents to seldom or never initiate discussions about their genetic status. Thus, an important finding in this study is that communication about genetic risk was generally initiated by the parents, and if an open resilient communication style existed, it occurred because the parents established the pattern. Genetic counselors can help families develop a plan about what information is to be shared and when and to develop an environment that is open and honest and conducive to communication. It will be important to utilize outcome based research to assess the effect different types of communication approaches have on long-term outcomes for families facing genetic disorders so that effective genetic counseling strategies can be developed to help parents managing genetic risk disclosure to their children.

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Table 1

Manner in which parents learned their daughter's carrier status or reason given for testing

Manner/Reason	Carrier (n=18)	Non-carrier (n=16)
Father carrier male	2	
Parent wanted to know	7	2
Everyone tested at the same time	2	4
Doctor recommended	–	1
Tested as part of research study	2	2
Concern related to school (possible learning disability or ADHD/ADD)	3	3
Concern about planning for future	–	3
Daughter requested	1	–
Tested prenatally	1	1

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Frequencies of disclosure patterns used by parents communicating information about fragile X syndrome and carrier status to their daughters

Table 2

Result	Patterns of disclosure				
	1. Full staging	2. Partial staging A	3. Partial staging B	4. At the same time	5. No conversation about genetic risk
Carrier (n=18)	1	2	5	10	–
Non-carrier (n=16)	4	4	2	6	–
At risk (n=12)	3	Na	Na	6	3
Total	8	6	7	22	3

Full staging: Parents using this approach reported three separate instances in which they first informed their daughter about the basics of fragile X, then about her risk for being a carrier, and finally informed her of her actual carrier status

Partial staging A: In this approach daughters were informed about fragile X syndrome and about their own possibility of being a carrier in the same conversation. Girls who had been tested were told their actual carrier status at a different time.

Partial staging B: This approach involved daughters learning about the inheritance of fragile X syndrome and later being informed of their actual carrier status. There was not an interval step of being informed about the possibility of “being a carrier”.

At the same time: These parents told their daughters about fragile X syndrome and her carrier status (or at-risk status) within the same conversation or report that both pieces of information were always discussed.

No Conversation: These parents reported their daughters were informed about fragile X syndrome; however, parents do not remember having any conversation with her about the possibility that she “could be a carrier” or could have an affected child.

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Table 3

Frequencies with which parents reported either parent initiated or daughter initiated conversations about fragile X syndrome in general

Frequency	^a Parent initiated: How often do you talk with your daughter about fragile X in general?		Daughter initiated: How often does your daughter come to you with questions about fragile X?		
	Carrier (n=18)	Non-carrier (n=16)	At-risk (n=14)	Carrier (n=18)	Non-carrier (n=15)
More than several times per year	16	14	8	6	8
Yearly	1	–	–	1	–
Less than once a year/very infrequent	1	2	6	1	3
Never	–	–	–	10	4
Once/the time she was told	–	–	–	–	–

^aNumber of responses for each question varies as for some interviews it was not possible to distinguish a direct response related to a specific daughter

Table 4

Frequencies with which parents reported either parent initiated or daughter initiated conversations about genetic risk status for fragile X syndrome

Frequency	Parent initiated: How often do you talk with your daughter about her genetic status?			Daughter initiated: How often does your daughter come to you with questions about her genetic risk status?		
	Carrier (n=17)	Non-carrier (n=16)	At-risk (n=13)	Carrier (n=18)	Non-carrier (n=15)	At-risk (n=1514)
More than several times a year	7	2	2	6	–	1
Yearly	1	5	5	–	2	1
Less than once a year/very infrequent	6	4	3	3	7	4
Never	–	–	1	9	6	8
Once/the time she was told	3	5	2			

^aNumber of responses for each question varies as for some interviews it was not possible to distinguish a direct response related to a specific daughter