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ORIGINAL RESEARCH

Influence of Genetic Risk Information on Parental Role Identity in Adolescent Girls and Young Women from Families with Fragile X Syndrome

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Abstract Using a multi-group cross-sectional design, we explored self-concept related to parental role salience and enactment in 53 young women (14 to 24 years) with knowledge they were either carriers, non-carriers, or could be a carrier of fragile X syndrome (FXS). Parental role salience included the participants' desire "to be a mother" and the importance they placed on this role. Enactment focused on the participants' views regarding ways to become a mother (reproductive options), parenting a child affected by FXS, and the development of partner relationships (marriage). Participants completed the FXS Adolescent Interview and the FX-Visual Analog Scale. Participants' knowledge of their genetic risk status appears to have influenced both salience and enactment of the parental role, and the effect varied based on carrier status. For many, knowledge of genetic risk appears to have led to reappraisal, redefinition, and re-engagement with the goal of becoming a parent. This process was prominent in those who were carriers and less so in those who were at-risk, and it did not typically occur in those who were non-carriers. Findings offer valuable insight into the impact of genetic risk information on developing perceptions of the parental role and offer new directions for genetic counseling with adolescents and young women with a family history of FXS.

Keywords Fragile X syndrome · Self-concept · Parental role · Adolescent · Carrier testing · Genetic counseling

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Introduction

Self-concept is important to consider as a possible protective factor in response to potentially threatening information, such as learning carrier status for a genetic disorder, as well as an area of concern if information about self were to result in harm to self-concept. Importantly, how an individual defines themselves in the present as well as expectations for who they will become in the future are thought to influence behavior and responses to stressful events (Markus and Nurius 1987; Stein 1995).

Self-concept can be defined in many different ways, and outcomes of research on self-concept related to genetic testing have varied based on how it was defined (McConkie-Rosell and DeVellis 2000). Self-concept can be defined using Cognitive Behavioral Theory as a multi-dimensional, hierarchical sense of self and self perception related to identity, feelings, thoughts, behavior, appearance, and personal characteristics (Hattie 1992). Using this definitional approach, we have previously reported findings regarding self-concept in adolescent girls and young women who were members of families in which fragile X

syndrome (FXS) had been diagnosed (McConkie-Rosell et al. 2008). FXS is an X-linked triplet repeat disorder, which can occur due to inheritance of a full mutation (greater than 200 hypermethylated CGG repeats) (Nolin et al. 2003). The hypermethylated full mutation results in loss of the FMR1 protein leading to a range of cognitive and behavioral abnormalities that characterize FXS (Hagerman and Hagerman 2004). Females with the full mutation may have clinical features of FXS or can be unaffected. Females with the premutation (less than 200 repeats is not hypermethylated) are at increased risk for the FMR1 disorders of Fragile X-Associated Primary Ovarian Insufficiency (FXPOI) and Fragile X-Associated Tremor and Ataxia (FXTAS) (McConkie-Rosell et al. 2005). Carrier females may have either the full mutation or a premutation. We reported findings related to self-concept as it was measured by the Tennessee Self-Concept Scale (Fitts and Warren 1996), a self-rated visual analog scale, and descriptions from interviews with adolescent girls and young women in three groups with knowledge of their genetic risk status (carriers, non-carriers, and individuals who knew only that they could be a carrier for FXS, referred to hereafter as "at risk") (McConkie-Rosell et al. 2008). We found that although the mean scores on the Tennessee Self-Concept Scale were within the normal range for all three groups, differences in feelings about self were observed on the visual analog scale and in the interviews. On the visual analog scale we found that adolescent girls and young women who knew only that they "could be" a carrier for FXS reported diminished feelings about self related to their genetic risk status. These adolescent girls and young women also reported a significant negative effect on their relationships with friends compared to those who knew they were either carriers or non-carriers.

Using Identity Theory, self-concept can also be defined through the different roles an individual adopts. There are three assumptions of Identity Theory: 1) self-concept is developed from multiple internalized roles that have a shared personal and societal meaning, 2) there are specific behaviors or activities that are required to enact the role, and 3) the roles have a hierarchical structure based on the salience of a particular role to the individual and to society (Howard 1991; Stryker 1991). Salience includes the value, commitment to, and level of importance of a role to the individual, and the more salient a role the greater the effect of that role on the development of the individual's self-concept (Thoits 1991). Some roles are chosen by the individual (e.g., athlete) while others (e.g., sibling) are not. Some roles are also considered by the society in which the individual lives as a normal and attainable role (e.g., health professional) while others are not as commonly enacted (e.g., president). Identity Theory then predicts that a barrier to enacting a valued normative role may have an effect on a developing self-concept (Thoits 1991).

Based on these concepts, we have proposed that a possible mechanism by which self-concept might be altered based on genetic information is through a perceived barrier to the enactment of the parental role (McConkie-Rosell and DeVellis 2000). Genetic knowledge may challenge a "wished for" parental role several ways. First, learning carrier status may alter how a person defines him/herself in relation to reproductive expectations. For adolescents and young adults who know they are carriers of a genetic disorder, this changed perspective may lead to feelings of reduced desirability for marriage. The definition of the parental role and how the role is enacted may also be altered based on the possibility of parenting an affected child.

In a study of adult women who each had a 50% chance of being a carrier for FXS, we found some evidence that change in perception of the parental role occurs as part of learning about genetic risk for this disorder. These women expressed concern about what "being a carrier" meant for future reproduction for their children or grandchildren, or, if their families were not complete, for themselves (McConkie-Rosell et al. 2000).

Findings from the adult research may have limited applicability to adolescents because of differences in the stability of self-concept in these age groups. Unlike a mature adult whose self-concept is generally considered stable, an adolescent's self-concept is developing and is affected by tasks and challenges (Baumeister 1998). Adolescents are also trying on new roles and identities, developing their sexuality, and thinking about the future, while simultaneously seeking approval from peers and family and asserting their independence (Balk 1994; Erikson 1963). There is growing evidence that personal identity is a developmental continuum which includes exploration, choices, and commitment to those choices (Meeus 2011).

Because conceptualization of future roles may begin prior to their enactment (Markus and Nurius 1987), an adolescent who knows either she is a carrier or is at-risk to be a carrier may not explore a future parental role identity because of a perceived barrier. For those adolescents for whom this identity is already developed, a perceived barrier to the goal of becoming a parent may result in distress. Additionally, a barrier that is perceived to be insurmountable may result in identity foreclosure. Identity foreclosure occurs when the individual no longer imagines enacting a future role (Balk 1994; Markus and Nurius 1987) or abandons efforts to achieve that specific goal (Brandtstadter and Rothermund 2002; Rothermund 2011). Because of these concerns we specifically explored concepts related to the importance of becoming a mother and plans for the enactment of the

parental role as part of the adolescent fragile X study. We report herein our findings regarding parental role.

Methods

Sample and Procedures

Using a multi-group cross-sectional design, this study focused on adolescent and young adult women and their parents from families in which an individual had been diagnosed with FXS, herein referred to as the Fragile X Adolescent Study. FXS was chosen because it is an X-linked disorder; therefore females who are carriers face a risk of having affected children. We recognize that some individuals with expanded CGG repeats are not asymptomatic carriers as this term has traditionally been defined in medical genetics. However, for the purposes of this study we refer to the participants with expanded CGG repeats as carriers.

After review and approval of this study by the Duke University Health System (DUHS) institutional review board, we enrolled adolescent girls and young women (ages 14–24) and their parents who were members of families in which FXS had been diagnosed. We were interested in enrolling adolescent girls and young women who had been informed about FXS in childhood in order to explore their experiences with growing up with genetic risk knowledge. In order to be enrolled in the study, the adolescent girls and young women knew they were either: a) carriers, b) non-carriers, or c) at-risk to be carriers. The adolescent girls (ages 14–18) had to have this knowledge for a minimum of 6 months prior to being enrolled in the study. Six months was chosen in order to allow the study participants some time to have considered their genetic risk status before taking part in the research. The young women in the study (ages 19–24) had to have learned their genetic risk status prior to the age of 18 in order to be enrolled in the study. Girls with clinical characteristics suggestive of the fragile X full mutation were excluded. The study sample was recruited through the Fragile X Clinic at DUHS, family support groups from the National Fragile X Foundation, and postings on the FRAXA listserv.

Investigators traveled to families to allow enrollment of families who had a variety of experiences with FXS. All interviews were conducted by the study's principal investigator, AMR, using a structured interview method. Interviews were audio-recorded and transcribed verbatim. The interviews were conducted over a 2 year interval and typically lasted 60 to 90 min. All participants were interviewed independently. Time was allowed after completion of the study measures to discuss with the study participant and her parent(s) questions or concerns that were raised as part of being in the study or general questions about fragile X syndrome and associated disorders.

Measures

The Fragile X Adolescent Interview

The Fragile X Adolescent Interview was composed of 70 open and closed ended questions. It was adapted to include critical components of the parental role for use in this study from one previously used with adults (McConkie-Rosell et al. 2002; McConkie-Rosell et al. 2000; McConkie-Rosell et al. 2001). For the purposes of this study, we explored parental role related to the importance placed on "being a mother," enactment of the parental role (including biological children and reproductive options including adoption, prenatal testing, and pregnancy termination), parenting children affected by fragile X syndrome, and the development of serious partner relationships (marriage).

The Fragile X-Visual Analog Scale

The Fragile X-Visual Analog Scale (FX-VAS) was designed to work in tandem with the interview. A visual analog scale was used because of its sensitivity in measuring feelings (Areskog et al. 1984; Axworthy et al. 1996; Bond et al. 1995; Crawford-Little and McPhail 1973) and the ability to detect small differences that allows for increased statistical power with small sample sizes (DeVellis 1991). Related to parental role, we asked participants to rate: 1) the importance placed on being a mother, and 2) level of upset they thought they might feel if they did not have a biological child. The measure ranges from 0–10 and is scored to 1/10 cm. Each response is indicated on a 10 cm line with positive and negative anchors and a midpoint of 5 indicted. Responses are scored by measuring the distance of each response along the line between 1 and 10 cm.

Data Analysis

Statistical Methods

Power calculations for the main study were based on the FX-VAS pilot data on adolescents and from findings of our previous study of adult women (McConkie-Rosell et al. 2000). We had 80% power to detect mean differences among the three groups if the two most extreme means differed by at least 1.2 standard deviations with a minimum sample size of 15 per group. For pairwise group comparisons, we had greater than 95% power to detect differences in the two most extreme means (two-tailed test), but less power to detect an

intermediate difference if one mean was between the other two. Descriptive statistics were obtained for the three groups defined by carrier status. Statistical analyses were performed only on data obtained from the FX-VAS. Assessments of group differences associated with carrier status were made using nonparametric procedures and a 0.05 level of significance.

Qualitative Analysis

The transcribed interviews were uploaded into ATLAS Ti 5.0 and analyzed using a directed content analysis (Potter and Levine-Donnerstein 1999). In the development of our initial codes we utilized Identity Theory and our model regarding parental role (McConkie-Rosell and DeVellis 2000). The transcripts were then repeatedly reviewed and codes developed as needed. The data were first independently coded by EMH and AMR, both genetic counselors with experience in qualitative analysis and FXS, and then jointly reviewed. Discrepancies were discussed until agreement was reached. The structured format of the interview allowed for tabulation of responses into three major categories related to the reported effect genetic risk knowledge had on each component of the parental role. These categories were positive/yes, negative/no, and no perceived effect or uncertain, depending on the question. Responses were then tabulated for each specific code and sorted based on genetic risk status. The data were grouped based on carrier status (carrier, non-carrier, and at-risk to be a carrier) and analyzed for group similarities and differences.

Results

Socio-Demographics

Fifty-three adolescent girls and young women were enrolled in the study from eleven different states in the US. There were no significant demographic differences among the three groups (carriers, non-carriers, and those who knew only that they could be a carrier (at-risk). Study sample demographics are summarized in Table 1.

Table 1 Sample demographics (N=53)

	Carrier (n=20, 16-premutation 4- full mutation)	Non-carrier (n=18)	At-risk (n=15)
Age at the time of the interview			
	Mean=18.35 years	Mean=17.78 years	Mean=17.87 years
	Median=18 years	Median=18.5 years	Median=17 years
	(s.d.=2.52)	(s.d.=2.69)	(s.d.=3.18)
Ethnicity			
	Caucasian		50 (94%)
	African American/Hispanic		3 (6%)
Religion			
	Protestant/Christian non-denominational		27 (52%)
	Catholic		9 (17%)
	Baptist		8 (15%)
	No formal affiliation		6 (11%)
	Jewish		2 (4%)
	No religious beliefs		1 (2%)
Highest year completed in school at the time of the interview			
	Middle School (7th-8th grade)		6 (11.4%)
	High School (9–12th)		31 (58%)
	College		16 (30%)
Closest relative affected by FXS[a]			
	Sibling		35 (66%)
	1st Cousin		9 (17%)
	Niece/Nephew		5 (9%)
	Parent (mother)		1 (2%)
	Uncle/Aunt		1 (2%)
	Greater than 3rd degree		2 (4%)
	Multiple relatives affected		29 (55%)

There are no significant differences between socio-demographic characteristics among the three groups (p>0.1, Pearson Chi-Square)

Importance of "Being a Mother" (Salience of Role)

On the FX-VAS, participants who were carriers and non-carriers rated "being a mother" as significantly more important than those participants who knew they were at-risk to be a carrier (Table 2). There were no significant differences in level of upset reported in how they thought they might feel if they did not have a biological child.

To further explore parental role salience, we asked study participants how they felt knowledge of their genetic risk status may have or had not influenced their desire to have children. Findings are summarized in Table 3. Many of the study participants reported that knowing their genetic risk status had no effect on their desire for children, and typically these individuals did not provide a reason. A few of the adolescent girls and young women who were carriers and at-risk reported that they felt this information had a positive effect. These participants felt that knowing they either were carriers or faced the possibility of being a carrier made them think more about having children, which resulted in wanting them more:

> "Actually, I think it might have made it stronger. I mean… I've always been very strong in that I want to have children, but it made me think about it in a different way, you know, which brought up a lot deeper issues than, you know, just the fact of having kids. It's made me think about it, you know, a lot deeper, and it's made me actually want it more."
>
> Carrier, 22 years

Some of the participants who were non-carriers felt knowing they were not carriers resulted in relief:

> "Yeah I mean it makes you kind of less like that's like one less thing that could go wrong. So you can kind of rest a little."
>
> Non-carrier, 17 years

Some adolescent girls and young women who were carriers and those at-risk reported their knowledge of genetic risk had a negative effect on their desire to have children, but typically they did not give a reason for this impact. A few were uncertain whether they wanted children at all, regardless of genetic status, and some were simply uncertain about how they felt:

> "You would definitely be more cautious, but it may lessen (desire to have children). Or like, or be like the same, be like 'Okay, I can handle it'."
>
> Carrier, 14 years

Enactment of Parental Role

We explored with the adolescent girls and young women their perceptions of how knowledge about genetic risk for fragile X syndrome may have affected their plans about developing partner relationships and becoming a mother. We also explored their views about reproductive options, including reproductive technologies and adoption, and the possibility of parenting a child with FXS. Findings are summarized in Table 4.

Development of a Partner Relationship (Marriage)

The majority of adolescents and young women in all three groups did not feel their genetic risk status would affect their plans for getting married/having a long term partner. For those who felt it would affect their plans, these participants felt that although their carrier status has not altered their desire to be married, it has influenced the type of person they think they would marry. These participants commented that whoever they do marry would need to be someone special who would be able to manage issues related to FXS in their own children:

> "Yeah, I think it has because whoever I marry is going to have to be someone who I think, you know, could go through this with me and could handle it, you know, and has the right, you know, personality to handle this. You know, because I want to have kids, and I'm going to have kids (laugh). So it's, you know, the person I marry is going to have to be willing to accept that."
>
> Carrier, 22 years

Table 2 Fragile X Visual Analog Scale (FX-VAS) ("being a mother" and biological children)

FX-VAS item	Carrier (n=20)	Non-carrier (n=18)	At-risk (n=15)
In your overall life plan, how important is it to you to be a mother?	Mean=2.4	Mean=1.9	Mean=4.6*
(0 = very important 10 = not important)	(s.d.=3.1)	(s.d.=3.1)	(s.d.=3.2)
If you did not have your own biological children how would you feel?	Mean=5.0	Mean=5.1	Mean=5.8
(0 = very upset; 10 = not upset)	(s.d.=2.6)	(s.d.=2.9)	(s.d=2.7)

*p=0.037 (Nonparmetric Kruskal-Wallis)

Table 3 Fragile X interview items regarding maternal role salience (desire to be a Mother)

Desire to be a mother	Carrier (n=20)	Non-carrier (n=18)	At-risk (n=15)
How do you think knowledge of your carrier status *(at-risk: knowing you could be a carrier)* may influence (or may have influenced) your desire to have children?			
No effect	60% (12/20)	72% (13/18)	33% (5/15)
Positive effect (made me want them more)	10% (2/20)	27% (5/18)	13% (2/15)
Negative effect (made me want them less)	25% (5/20)	0	20% (3/15)
Uncertain	5% (1/20)	0	26% (4/15)

Some of the non-carriers felt a partner would also have to be someone special who could accept responsibility of their affected relative(s).

> "No, not at all. I mean, I know that I just have always told my mom. I'm like, 'I get the boys when I'm older'. That's just, I want to have them in my house with me, and whoever I get married to, it's just like they're mine. You get me, you get my brothers, and that's just the way it is, and I love them and they're my best friends, and that's just how it's going to be."
>
> Non-carrier, 16 years

Some also highlighted challenges in considering the timing during a relationship to inform their partner of their carrier status and the possibility of having a child with fragile X syndrome, and that it should be disclosed to someone who was close to them:

> "I guess yeah, it has changed [plans for marriage] because when I do, you know, meet the person, who I'm going to spend the rest of my life with I have to you know tell them that, you know, I have this disease and that, you know, there's a chance that when we have kids that our kids might have it. So it yeah, it does change that."
>
> Carrier, 20 years

Participants also commented that disclosing fragile X carrier status may be a test of a relationship:

> "I'd be kind of scared about how they would react but then I'd be like, you know what, if he doesn't accept it, then I don't need him in my life anyway."
>
> Carrier, 18 years

A few of the participants who are at-risk worried about finding this special person:

> "I am wondering if they will want to be married to me if I have, if they know that I might have that gene or carry that."
>
> At-risk, 14 years

Plans for Having Children

Carrier

For the participants who were carriers and who felt knowledge of their carrier status had not affected their plans for having children, the majority did not offer a qualifier. Although some had initially considered not having children, they later concluded that being a carrier should not prevent them from having biological children:

> "I mean at first I probably…I remember thinking, I'm never going to have kids and I'll just adopt, but I mean now, there should be nothing…I mean even this thing. There should be nothing holding me back from wanting to have a kid."
>
> Carrier, 18 years

For the participants who were carriers and reported knowledge of their fragile X carrier status had affected their plans for having children, some expressed more caution or hesitation about having biological children:

> "Um, maybe in the fact that I'll be a lot more cautious now. I'll definitely do genetic counseling. If there's that great of a factor or if it's that great of a risk, maybe I'll adopt children."
>
> Carrier, 16 years

Others would consider reproductive options:

> "I'm still going to have all of my children. The WAY of having my children is the only thing that's changed, you know, naturally, or in vitro, or anything like that. That's the only thing that's really changed. I'm still having my 5 kids."
>
> Carrier, 18 years

Non-carrier

Many of the non-carrier participants who indicated this knowledge had not changed their reproductive plans included a qualifier stating they might have felt differently if they had been a carrier. Those who included these statements often said they had

Table 4 Fragile X adolescent interview items regarding enactment of parental role

		Carrier $n=20$	Non-carrier $n=18$	At-risk $n=15$
Perceived impact of carrier status on future plans relating to parental role				
Do you think knowing your carrier status for FXS (at-risk: knowing that you could be a carrier) may affect your plans or goals for marriage (serious partner relationship)?	Would have an impact	25% (5/20)	11% (2/18)	33% (5/15)
	No impact	70% (14/20)	83% (15/18)	66% (10/15)
	Do not plan on getting married	5% (1/20)	6% (1/18)	0
Do you think knowing your carrier status for FXS (at-risk: knowing that you could be a carrier) may affect your plans or goals for having children?	Would have an impact	65% (13/20)	50% (9/18)	60% (9/15)
	No impact	35% (7/20)	39% (7/18)	27% (4/15)
	Uncertain if they want children	0	11% (2/18)	13% (2/15)
Perceptions about personal use of reproductive options				
What do you think about ways other than having biological children to become a parent (for example, adoption)?	Positive	75% (15/20)	94% (17/18)	53% (8/15)
	Negative	10% (2/20)	0	6% (1/15)
	Uncertain	20% (4/20)	11% (2/18)	53% (8/15)
What do you think about prenatal testing or other technologies such as egg donation, preimplantation diagnosis?	Positive	40% (8/20)	40% (6/15)	50% (7/14)
	Negative	20% (4/20)	28% (5/18)	20% (3/15)
	Uncertain	45% (9/20)	33% (6/18)	27% (4/15)
What do you think you would do if you knew before your baby was born that he/she has FXS? (Would you end the pregnancy?)	Yes	0	11% (2/18)	6% (1/15)
	No	95% (19/20)	66% (12/18)	66% (10/15)
	Uncertain	5% (1/20)	22% (4/18)	27% (4/15)
Can you describe for me how you feel about the possibility of having a child with FXS? (Parenting a child with FXS?)	Positive	80% (16/20)	[a]	73% (11/15)
	Negative	15% (3/20)	16% (3/18)	6% (1/15)
	Uncertain	5% (1/20)	[a]	20% (3/15)

[a] Participants who were non-carriers typically did not respond to the question about how they felt about parenting a child with fragile X

more choices about reproductive plans or they no longer had to face difficult ethical decisions. This was also the reasoning provided by those who reported a change in their plans:

> "Yes, if I was a carrier, I would not probably have children naturally, unless there was, I don't know all the new technology they have if they can (test). No, I probably wouldn't because I wouldn't want to have to terminate a pregnancy."
>
> Non-carrier, 21 years

At-risk

The participants who were at-risk who did not feel that this information had affected their plans did not typically offer an explanation. For those who felt this information had influenced their reproductive plans, a few had contemplated reproductive options and said they would do so in the future. Others reported feeling cautious or uncertain about how their genetic risk might affect their future plans:

> "Um, like I mean I do, but like I still kind of hesitate on it and everything just, but I guess kind of, yeah."
>
> At-risk, 14 years

Reproductive Options

Adoption

Although the majority of all participants had a positive attitude towards adoption, some qualified their statements, indicating that although they preferred having biological children, they would adopt if it were the only way to become a mother:

> "Adoption, I think, you know, that it's good, but I don't think that I'd ever do it, unless I knew I couldn't have children, then I probably would. But with me being able to have them, I just don't see it."
>
> Carrier, 24 years

Reproductive Technology

Approximately half of the study participants in all groups reported they were supportive of assistive reproductive technologies, such as prenatal testing, pre-implantation genetic diagnosis, or egg donation. Of the participants who were carriers and non-carriers, the major reason given was that they felt they would do whatever they

needed in order to help increase the chances of having a child without FXS:

> "I agree with it. I know my religion might not feel [it] is right, but, I think if, you know, and you can have a child that isn't going, that can be healthy and can be perfectly normal, that you should take whatever means necessary to make sure that happens."
>
> Carrier, 20 years

> "I don't want to have a child with fragile X simply for their, you know, for them. I don't want them to have to go through all that, so yes, I would definitely. It's a hard world sometimes and having fragile X can make it a really hard world. Yes. It's not that I don't, I mean, I love [my cousin]. I wouldn't change him, but if I had [a] chance to not have one, I'd definitely take it."
>
> Carrier, 18 years

A few of the adolescent girls and young women who were carriers or at-risk qualified their positive response with a statement that they would use reproductive technology only if it could occur before an actual pregnancy, such as pre-implantation genetic diagnosis. Others felt prenatal testing was okay for "someone else" but not for them:

> "It is there, and I am open to it I suppose. Other people want to do it, great. I don't know personally if I want to put that much time and energy and effort and possible disappointment into it."
>
> At-risk, 21 years

Positive statements about the availability of this technology also included statements indicative of personal or moral conflict when they considered using these technologies themselves. This conflict was often expressed as a concern about wanting a child without FXS and the love they felt for their affected relative:

> "I think that it is definitely good, and I think that the ability to do that should be out there, but then at the same time, I wonder like…my cousin might not be here at all if…if that was an option or that had been done. It's almost like me saying my cousin's life isn't really valuable, so I feel like that's not right either. So that's why I'd probably just adopt."
>
> Non-carrier, 21 years

> "I don't know that I could say. I guess if you could choose not to have a fragile X child. But as far as like saying 'No, I don't want a fragile X child,' that's like saying 'No, I can't love him because he's got fragile X'."
>
> At-risk, 17 years

Others specifically commented that they would undergo prenatal testing in order to be better prepared for an affected child if the prenatal test were positive:

> "I think you should definitely do it, so that parents can find out as soon as possible and know about it, prepare themselves for it, that they're not just surprised."
>
> Carrier, 17 years

Those who definitively stated that reproductive technologies were not something they endorsed did so for religious, moral, or ethical reasons, or because they considered them to be just too "high tech":

> "My whole religion holds against having anything like that, and I find it rather repugnant to just sort of pick and choose what child you're going to have. I mean, that's just, that's not how it's supposed to work. You're supposed to get what comes to you and there you go."
>
> Carrier, 20 years

Others were uncertain how they felt or that it was a future concern, so they had not yet thought about it.

Termination of an Affected Pregnancy

We asked study participants to tell us what they thought they might do if they learned during the pregnancy that the baby had FXS. We specifically asked for clarification regarding pregnancy termination or "ending pregnancy." Only 3 participants (2 non-carriers and 1 at-risk) reported they believed they would terminate a pregnancy if the baby was found to have FXS. The reasons given were that life can be difficult for a child who is affected and/or that fragile X was part of their childhood and they did not want it in their future:

> "I really don't know what I would do in that situation. Likely, I would probably have an abortion. I hate the idea of abortion, but I mean…I have my brother, and that's enough for me."
>
> Non-carrier, 22 years

Ninety-four percent (51/54) stated they would continue the pregnancy if they found out during the pregnancy that the baby had FXS:

> "If I was pregnant and if I guess they would go ahead and say that he was fragile X, I would, I wouldn't throw him away, no, I would have him, no doubt. I would just have to deal with it."
>
> At-risk, 18 years

Only 5 participants (1 carrier, 3 non-carriers, and 1 at-risk) stated they would not terminate for moral reasons:

> "Have it. I don't believe in abortion, so I'd have the baby and I would care for it too. I'm not going to give it up just because it has fragile X."
>
> Non-carrier, 18 years

The majority of participants gave other reasons to support continuing their pregnancies. Many commented that they would accept a baby with fragile X, and they would love their baby no matter what:

> "I would just be like, 'Ok. I'll love this baby anyway.' I mean… [So you wouldn't end the pregnancy?] No, not a chance."
>
> Carrier, 18 years

There was also recognition that a child with FXS is difficult and/or more work to raise than a typically developing child:

> "I mean, seeing my brother, I mean, like the things that my family goes through and having the things they went through with him. I mean, he's a kid, I'd take care of him. I couldn't [end the pregnancy], it would be okay in my book. I mean, it would be a lot harder, but whatever. It'd be worth it."
>
> Carrier, 16 years

Some (25-30%) felt learning in advance the baby was affected would allow them the opportunity to plan for how to best meet their child's needs or to be better prepared:

> "Maybe prepare for it. Like help my family or people like my friends or whatever understand it and know that this kid's going to be different. (Would you end the pregnancy?) No."
>
> Carrier, 18 years

A few participants were conflicted or uncertain about what they would do. This was expressed as either conflict between the positive feelings they had for their affected sibling or relative and difficulty in considering pregnancy termination:

> "Looking at my aunt's life, she really, she loves her son so much, and I know that she would never…I mean…but I just…(long pause). Like I've always been like very pro choice, but then when I really think about that situation…but I just don't…I don't know. Especially if you want to have a baby, and you want, I feel like it's wrong to just pick out the perfect ones, you know, the ones that have totally nothing wrong with them.
>
> Non-carrier, 21 years

Or they did not know exactly what they would do:

> "I don't know. Because, I mean my brother has fragile X, and he is not that severely affected at all. In fact he is useful to have around because he remembers things and makes me a great assistant. I don't want to think that it would affect how much I love my baby or how much I wanted my baby."
>
> Carrier, 20 years

Parenting a Child with Fragile X Syndrome

The majority of participants who were carriers (80%) and those who were at-risk (73%), as well as 50% of those who were non-carriers responded with acceptance about the possibility of having their own child with FXS. Of those, the majority included statements that FXS was not a barrier to having biological children:

> "I think again, it is not an issue. I think that it would be a challenge because I haven't always dealt well with it. I guess that it is just growing up as a child, but maybe as I get older it won't be such a, it won't seem like such a challenge. But either way, it's not an issue having children."
>
> At-risk, 16 years

And for some, this acceptance was influenced by the love they felt for their affected relatives and the love they would feel for their own child:

> "I accept it. It doesn't bother me. I mean, I look at my niece, I love her to death. I wouldn't, you know, trade her for a healthy child for nothing, you know."
>
> Carrier, 22 years

Many of the participants in all groups expressed that, as a parent, you need to be able to handle it and were optimistic that they would be able to successfully parent a child with FXS:

> "It will be difficult definitely, but I would get through it day by day."
>
> Carrier, 18 years

Some expressed a spiritual view that they would be given the strength to parent a child with FXS, or that having a child with FXS was their destiny:

> "I believe that God has a path for you. I know He'll help me with, you know, emotional strength, and He would give me the strength to do all that."
>
> Carrier, 18 years

A few of the participants in each group expressed negative emotions at the concept of parenting a child with FXS:

> "My mom always said that she wants me to, but I say that she has never grown up with it, had a sibling. I wouldn't want um my child to have to deal with it, too. I don't know, I just don't want to have to deal with it my whole life. I had it in my childhood."
>
> Carrier 15 years

Only a few participants had never considered the possibility of having a child with FXS and were uncertain about how they felt or what they might do. The non-carriers typically had not thought about it because they knew they were not carriers, or they responded by trying to consider how they might have felt, if they had been a carrier.

Discussion

This study explored self-concept related to parental role in adolescent girls and young women who are members of families where FXS had been diagnosed. To our knowledge, this is the first study to explore parental role identity in adolescents and young adults in the context of genetic risk. We explored both the salience of the role as well as how the parental role would be enacted. We found that genetic risk knowledge appears to have influenced both of these aspects, but the effects are not necessarily negative.

Salience of Parental Role

One developmental task of adolescence and young adulthood is exploring different roles and considering possibilities for the future (Balk 1994; Markus and Nurius 1987). Based on our previous work with adult women going through the carrier testing process for FXS, we have been concerned that an adolescent growing up with knowledge of her genetic risk might abandon exploration of a parent role because of a perceived barrier. We found limited evidence to support this concern in the study participants who had knowledge they were either carriers or non-carriers. There was no statistically significant difference between the participants who were carriers and non-carriers with respect to how they rated the level of importance they assigned to becoming a mother on the FX-VAS. Although 25% of the participants who were carriers reported a negative effect on their desire to have children, the majority of those who were carriers reported wanting biological children. The participants who were non-carriers reported they wanted biological children. They also felt they had more options and were free from the possible moral or ethical decisions about reproduction they would have faced had they been carriers.

A few of the adolescent girls and young women commented that they were uncertain if they wanted children at all, regardless of their genetic risk status. Being childless by choice is a reproductive decision that some women make. Estimates of the proportion of women who choose to be childless by choice vary, from approximately 6-10% in a Western culture (Lee and Gramotnev 2006). Thus, findings from this study of 7% who were uncertain about their desire for children is not outside what has been reported as the general frequency of women who choose to be childless by choice. What is of interest is that none of the participants who were carriers made this statement. It is foreseeable that the participants who were carriers would be at greatest risk for early foreclosure or reduced exploration of the parental identity because they know they have a chance of passing an expanded CGG repeat onto their biological children and, thus, a risk for having affected children. However, we found that the participants who are carriers appear to be contemplating their genetic status and exploring the potential effect on future reproductive plans. This process did not appear to be diminishing their desire to become mothers.

A reduced level of individual importance in becoming a mother was found in the participants who knew only that they are at-risk. Compared to carriers and non-carriers, the participants who were at-risk were also, as a group, less likely to report that their carrier status had not had an effect on their desire for children. They also were more likely to report knowledge of their genetic risk status made them want children less and/or that they were more uncertain about how this information made them feel about having children in the future. Additionally, these girls and young women were also less likely to provide a rationale or a qualifier for their responses. This difference in complexity of responses suggests less exploration of a future role as a mother may be occurring in this group.

It may be that the participants who are carriers and non-carriers, because of knowledge of their actual genetic carrier status, have simply thought about the future and children more and are thus better able to convey their opinions. Additionally, we have previously reported that parents of girls whose positive carrier status was confirmed communicate more frequently, offer more support for the future, and provide an optimistic perspective to their daughters (McConkie-Rosell et al. 2011; McConkie-Rosell et al. 2009). These girls may have had greater opportunity to process the information than girls who are at-risk. The frequent and optimistic communication may have also influenced their feeling FXS is not a barrier to their future plans for having children. It is also possible that some of the girls who are at-risk simply suspend thinking about the

implications of their possible carrier status until this information has more immediate need or until carrier testing is done.

Enactment of the Parental Role

The majority of participants in this study reported wanting a biological child. While there was general support of options such as adoption, the personal primary goal was to have a biological child(ren). Similarly, while there was support for the availability of prenatal diagnosis, it was often viewed as an option for someone else or something they would only consider if testing occurred prior to a pregnancy. Only 3 participants, none of them carriers, felt that they would consider pregnancy termination of an affected child. This finding sharply contrasts with those described by Kay and Kingston in their study of women who were carriers of an X-linked disorder (Duchene Muscular dystrophy, Lesch-Nyhan, Menkes, and Fabry). They found that 13/14 women planned to avoid having an affected son through prenatal diagnosis and pregnancy termination (Kay and Kingston 2002). However, unlike the X-linked disorders in Kay and Kingston's study, FXS is a serious disorder that leads to cognitive impairment, but it is not lethal or life-threatening. Additionaly, our sample included adolescent and young women while Kay and Kingston's study included adult women. There could be developmental differences that influence these perceptions. These differences may warrant further investigation.

Although some study participants were considering using reproductive technologies in the future, it would be to either prevent a pregnancy with an affected child through pre-implantation genetic diagnosis or to allow them to be better prepared to parent an affected child. Interestingly, while the participants who were carriers were typically more certain about their feelings regarding all other aspects of enactment of the parental role, they expressed the most uncertainty about how they felt about prenatal testing. This uncertainly seemed to be the result of conflicting emotions based on their love for their affected sibling and/or relative(s), perceptions that life can be difficult for individuals with FXS, and, for some, wanting to prevent FXS from occurring in their own children. This emotional conflict was found in girls from all three genetic risk groups. For many this conflict was a greater barrier to considering prenatal diagnosis and possible pregnancy termination than religious or moral beliefs.

For some, this conflict seems to have been resolved through acceptance of the possibility of parenting an affected child. Many of the participants expressed that the love they felt for their affected sibling or other relative helped them to accept this possibility in their own child. In addition to acceptance, there was recognition that raising a child with FXS was more difficult than raising a typically developing child, and they expressed confidence that growing up with FXS had helped to prepare them to parent an affected child. In a review of published research of siblings of individuals with developmental disabilities, Heller and Arnold (2010) found that unaffected siblings were knowledgeable about the special needs of their siblings and that they typically had a positive relationship which tended to become stronger with time. This finding does not appear to be threat minimization, as the participants acknowledged the problems and challenges of raising a child with FXS. Contrasting with this acceptance were those who felt FXS had been part of their childhood and they did not want it in their future, if it could be avoided. Thus, enactment was altered by acceptance of an affected child, as well as for some a plan to use reproductive technology to reduce the chances of having a child with FXS.

Enactment of the parental role was also altered through reframing relationships and responsibilities. Participants in all three groups felt they would need to marry or choose a long-term partner who would accept and help take responsibility for the affected sibling(s), co-parent a child with special needs, and/or go through the decision-making process regarding reproductive options and prenatal testing, should that be a choice they make. The participants who were carriers often felt their positive carrier test and the resulting implication for their future children was a type of "litmus test" of a relationship. More specifically, if the relationship could not withstand these possibilities, it was not the right one for them. While the participants who are at-risk expressed some of the same thoughts, they also appeared to have greater concerns about finding this special person than the participants who were carriers.

Reappraisal and Redefinition of Parental Role

In our conceptual framework, we proposed that if parental role salience was high, paired with a strong desire for biological children and a belief against prenatal testing including pregnancy termination, this combination could lead to altered perceptions of becoming a parent. Additionally, if becoming a mother seemed unattainable, then those individuals would be at greatest risk of early foreclosure of the parental role. While there is a suggestion this combination did indeed result in conflicting emotions for some of the study participants, for others an adaptive response may also have occurred. According to Brandtstadter and Rothermund (2002; Rothermund, 2011), if a life goal is deemed unattainable, the goal may be abandoned (disengagement), acceptance of the situation as it is, or modified through redefinition with re-engagement with a newly defined goal. This process of redefining and acceptance of either the situation or a new

definition is an important adaptive response to a blocked goal (Wrosch et al. 2003).

Many of the participants in this study appear to have balanced their strong desire to have biological children and their similarly strong opposition to pregnancy termination through acceptance, re-definition of "parent" or "mother" to include parenting a child with FXS, and identification of the type of person they felt could partner with them to face the challenges ahead. Study findings suggest this process was especially prominent in the participants who are carriers and not as frequent in the participants who are at-risk. Findings regarding the participants who are carriers suggest some may have initially disengaged, expressing they will not have children, but then subsequently actively reassessed and reengaged with a definition of parenting that is not in conflict with their personal beliefs and goals. The process of redefinition and reengagement may be one that the participants who are carriers are more likely to have employed because, for them, the chance of having an affected child has been confirmed by their positive carrier test. The participants who are at-risk do not know if they face a risk of having an affected child or not and thus may not engage in this process until their genetic risk is clarified through testing. The participants who are non-carriers did not typically go through this process as they did not perceive a barrier to becoming a mother.

Study Limitations and Research Recommendations

This study is a cross-sectional exploratory study. The process of developing identities in adolescents into adulthood is complex; this study focused only on one aspect, that of genetic risk and the parental role. The adolescent girls and young women who participated in this study provided their opinions at the time of the interview. The process of identity development is fluid, with movement through the different stages of exploration, commitment, and achievement (Meeus 2011). It will be important to determine longitudinally how perceptions, definitions of the parental role, role salience, and enactment may change over time and to determine if the current views are carried over to actions as adults. Additionally, this study focused on adolescents and young women from families with FXS, an X-linked disorder. We attempted to enroll participants from multiple different parts of the United States in order to reduce a regional effect. However, the girls and young women who participated in this study all had personal experience with FXS. These personal, lived experiences with FXS would be expected to have influenced their responses and limits generalizabity to those who might learn carrier status through other mechanisms, such as population screening. We also did not specifically explore the possibility of a shortened reproductive timeline related to FX-POI. The potential effect of this timeline on girls and young women who are carriers of the premutation should be explored in a future study.

Practice Implications

Knowledge of genetic risk for many of the participants in this study led to reappraisal and redefinition of the parental role. However, findings suggest the participants who knew only that they were at-risk, who had not had genetic testing, were more likely to report a reduced role salience with less exploration of possible options related to the enactment of the parental role. Findings from this study suggest that knowing a disorder is inherited and that there is potential to have an affected child may result in an altered perception of the parental role without carrier testing being done. Parents, genetic counselors, and other health professionals need to consider that this process may be occurring so that the needs of adolescents in all three risk categories are being met. It is important for adolescent girls and young women in all risk categories, not just those who are carriers, to consider different reproductive options and to explore their own perceptions about the possible choices. Genetic counselors can play an important role in focusing attention on the broader concepts of the parental role beyond considering only if genetic testing is appropriate. It is also critically important that those involved with these young women recognize the importance and help facilitate the process of reappraisal and redefinition of "becoming a mother" in light of risk for having an affected child, with an objective of preventing early foreclosure of the parental role and increasing the positive adaptation to whatever option and definition meets the individual girl's personal life goals.

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