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Living with Genetic Risk: Effect on Adolescent Self-Concept

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Abstract

The purpose of this study is to describe the interplay of adolescent girls' self-concept, coping behaviors, and adjustment associated with knowledge of genetic risk for fragile X syndrome. We will report here findings on self concept. Using a multi-group cross-sectional design this study focused on girls ages 14–25 years from families previously diagnosed with fragile X syndrome, who knew they were 1) carriers (n = 20; mean age 18.35 years s.d. 2.5), or 2) noncarriers (n =18; mean age 17.78 years s.d. 2.69), or 3) at-risk to be carriers (n = 15; mean age 17.87 s.d. 3.18). The girls completed the Tennessee Self Concept Scale (TSCS:2), a visual analog scale, and a guided interview. Total and all subscale scores on the TSCS:2 were in the normal range for all three groups. However, threats to self concept were found in personal self (physical self, genetic identity, and parental role), social self, and family self (family genetic identity) as they specifically related to the meaning of genetic information and varied based on risk status. Our findings suggest that risk information itself is threatening and for some girls, may be *as* threatening as learning one is a carrier. Certainty related to genetic risk status appears to make a positive difference for some girls by allowing them the opportunity to face the challenge of their genetic risk status and to begin to consider the meaning of this information.

Keywords

Fragile X syndrome; self concept; genetic testing in children; genetic counseling; carrier testing; parental role; adolescents

INTRODUCTION

A concern often associated with genetic testing in children is harm to self-concept [American Society Human Genetics/American College of Medical Genetics 1995; Clarke 1994; Fanos 1997] and current practice guidelines propose providing minor children with risk information and delaying testing unless there is a clear benefit to the child [Borry et. al. 2006]. Four possible mechanisms by which self concept might be altered based on genetic information have been identified: genetic identity, social self, altered perception of health, and threat to the parental role [McConkie-Rosell and DeVellis 2000].

Despite this concern the existing research is limited and it is often unclear how self-concept is defined and measured. In their pivotal review, Shavelson and colleagues [1976] highlighted

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the need for clarity in the conceptual and operational definition of self-concept when used in research. We identified only one study of minors who had genetic testing where a domain of self concept was assessed. Michie et. al. [2001] found self esteem, the evaluative component of self concept, to be a key predictor of adjustment to presymptomatic testing for familial adenomatous polyposis in 60 children (10–16 yrs). Although they did not formally measure self concept, Mitchell et al. (1993) reported that 9 cystic fibrosis (CF) carriers (ages 15–17 yrs) did not feel that being a carrier had changed their self-image and Clow and Scriver [1977] reported that there was a small chance of altered self-perception of high school students undergoing carrier testing for Tay-Sachs.

The majority of the self-concept research related to genetic risk information has focused on adults. Some studies have concluded that knowledge of carrier status has an adverse effect [Childs et. al. 1976; Evers-Kiebooms et. al. 1994; Fanos and Johnson 1992; Marteau et. al. 1992; Wooldridge and Murray 1989] while others have not supported this concern [Axworthy et. al. 1996; Rowley et. al. 1984]. Only one study using a standardized, validated self-concept measure, found any effect. Denayer et. al. [1996] administered the Tennessee Self-Concept Scale (TSCS) to adult siblings of individuals with cystic fibrosis and found diminished family self, on this subscale, when compared to the study measure norms but no difference in the total score.

In our study of adult women going through the carrier testing process for fragile X syndrome, we found that while global self-concept was stable, as measured on the TSCS (total and all subscales) situationally specific feelings about self related to the implications of being a carrier were affected by carrier knowledge [McConkie-Rosell et. al. 2000]. The women identified five areas of concern: implications of a positive carrier test for children, a possible barrier to having biological children or grandchildren, possible expression of clinical features of fragile X in themselves, an altered or heightened awareness of their genetic identity, and regret over not knowing sooner.

Presently, our understanding of the consequences to children of learning genetic risk information is based primarily on data extrapolated from research on adults. Findings based on research directed at adults may have limited applicability to adolescents because of differences in the stability of adult and adolescent self-concept. Unlike adult self-concept that once formed is generally considered stable; the adolescent self-concept is developing and is affected by tasks and challenges [Baumeister 1998]. Adolescents are 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]. Therefore, although there has been much debate about genetic testing in minors, little research exists that can be used to inform genetic counseling interventions designed to enhance positive adaptation of minors to this information.

The purpose of this study is to describe the relationship among adolescent girls' self-concept, coping behaviors, and adjustment associated with knowledge of genetic risk for fragile X syndrome. Fragile X syndrome is a common X-linked disorder with an estimated frequency of the full mutation of 1/4000- 1/6000 [Crawford et. al. 2002; Morton et. al. 1997; Turner et. al. 1996] and a carrier frequency of the premutation in North America of approximately 1 in 250 for females and 1 in 800 for males [Dombrowski et. al. 2002; Rousseau et. al. 1995]. The full mutation causes a range of cognitive disabilities and autistic-like behaviors [Hagerman and Hagerman 2002]. Individuals who carry the premutation are at increased risk for fragile X associated tremor and ataxia (FXTAS) [Hagerman and Hagerman 2004] and fragile X associated primary ovarian insufficiency [Sherman 2000]. We will report here our findings on self concept.

METHODS

Definition of Self-Concept Used in this Study

In this study, we operationalized self-concept in three different ways. First, global self-concept was defined as a multi-dimensional, hierarchical sense of self and self perception related to identity, feelings, thoughts, behavior, appearance, and personal characteristics [Hattie 1992]. Secondly, it was defined as "feelings about self" related to genetic risk and the interpretive meaning the individual places on this information. Finally, using identity theory, we also defined self-concept as "self in the parental role" [McConkie-Rosell and DeVellis 2000]. Our measurement approach utilized both a standardized multi-dimensional measure of self concept as well as a situationally specific measure of feelings about self and genetic risk status, and a guided interview. A mixed methods approach was used because of the complexity of self concept and our previous findings in adults that suggest global self concept is stable but feelings about self related to genetic information may be affected. Using a multi-group cross-sectional design this study focused on girls, from families previously diagnosed with fragile X syndrome, who knew they were 1) carriers, or 2) noncarriers, or 3) at-risk to be carriers.

Sample Recruitment and Data Collection

After review and approval of this study by the Duke University Health System (DUHS) Institutional Review Board, we recruited females ages 14–25 years of age. Those girls ≤ 18 years must have had knowledge of her particular genetic risk status for at least 6 months prior to study participation. Girls 19 to 24 years must have learned their status prior to the age of 19 years. We expected variability in the length of time each has had knowledge of her status and in the manner in which she learned this information. Our prior research suggested that how families manage genetic information varies based on coping strategies employed by the family and developmental stage and maturity of the child(ren) [McConkie-Rosell et. al. 2002; McConkie-Rosell et. al. 1999] and did not want to bias the study towards families using only one method of informing.

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

The study sample was recruited through the Fragile X Clinic at DUHS, the family support groups from the National Fragile X Foundation, and postings on the FRAXA listserv. After completing the prescreening to ensure subjects met enrollment criteria, researchers traveled to the families and subjects. All interviews were conducted by the study's principal investigator, AMR, and the standardized psychological measures were completed with the assistance of the study genetic counselor.

Study Measures

Global Self Concept—Global self concept was measured using the Tennessee Self-Concept Scale (TSCS):2. The TSCS:2 is multidimensional measure of self-concept with good validity and reliability (Cronbach's alpha .73) and it is normed for both adolescents and adults [Fitts and Warren 1996; Kramer and Conoley 1992]. The TSCS:2 includes subscale measures of conflict, physical, moral, personal, family, social, academic/work, identity, satisfaction, and behavior selves. The TSCS:2 is a self-report measure, written at an 8th grade level, and uses a 5 point Likert scale response option. The TSCS:2 yields both global self concept and subscale scores.

Situationally Specific Feelings About Self Related to Genetic Risk Information—Feelings about self related to genetic risk information were explored using a structured interview and a visual analog scale, FraX-VAS. Both the structured interview and the FraX-VAS were adapted for adolescents from those used previously [McConkie-Rosell et. al. 2002; McConkie-Rosell et. al. 2000; McConkie-Rosell et. al. 2001; McConkie-Rosell et. al. 1997]. The measures were also piloted with 10 adolescents who knew they were carriers of fragile X syndrome. We found that the adolescents in the pilot study were very motivated to "tell their story" and offer their insights and views.

Questions exploring self concept on the structured interview asked the participant to describe: her feelings about self related to genetic risk information, how she thinks knowledge of genetic risk influences family and peer relationships, and perceptions about feelings related to stages of knowledge regarding genetic risk information (learning at-risk status, learning actual carrier status, and current feelings). The interviews were audiotaped and transcribed verbatim.

The FraX-VAS was designed to work in tandem with the interview. The FraX-VAS asks the adolescent to rate her feelings about self related to her genetic risk, perception of how serious a problem she viewed fragile X syndrome, effect of genetic knowledge on family and peer relationships and her feelings related to stages of genetic knowledge (at-risk, carrier status known, current feelings). The measure ranges from 0–10 and is scored to 1/10cm. Each response is indicated on a 10 cm line with positive and negative anchors and a midpoint of 5 indicted. A VAS was determined to be the measurement approach of choice because of its demonstrated sensitivity in measuring intensity of feelings [Bond et. al. 1995], the fine discrimination that makes a VAS very sensitive to subtle differences, and the variability of response options, which allows for increased power with smaller sample sizes [DeVellis 1991].

Analysis

Power calculations were based on the TSCS:2, our adolescent pilot data, and longitudinal adult women data. We determined that a minimum sample size of 15 per group provided us with 80% power to detect mean differences among the three groups if the two most extreme means differed by at least 1.2 s.d. For pairwise group comparisons using an overall 5% level for type I error and Bonferroni adjustment we had greater than 95% power to detect differences in the two most extreme means (two-sided test), but with less power to detect an intermediate difference, if one mean was exactly intermediate between the other two.

Statistical Methods—Descriptive statistics were obtained for the three groups defined by carrier status, and group comparisons were performed using chi-square tests using SAS version 9.1. Assessments of group differences associated with carrier status were made using the nonparametric Kruskal-Wallis procedure. This approach was applied to evaluations of age and FraX-VAS scores. Pairwise comparisons among the three groups defined by carrier status were made according to the method given by Conover [1999], using the Tukey adjustment for multiple comparisons in conjunction with an overall 5% level of Type I error. Since the analytic procedure is based upon ranks, medians are provided as appropriate descriptors of responses in the three carrier status groups.

Within each individual, differences in remembered perceptions between different stages of knowledge were calculated; these were evaluated within each of the three carrier status groups using the Wilcoxon Signed Rank procedure. This approach was used to assess whether there was a nonzero median difference in a given remembered perception between the stages of knowledge being considered. In addition, the Kruskal-Wallis test and the multiple comparisons approach outlined above were used to compare these differences among the three carrier status groups.

Qualitative Method—The transcribed interviews were uploaded into ATLAS Ti 5.0. We used directed content analysis, a qualitative method that is guided by theory or prior research [Potter and Levine-Donnerstein 1999] to analyze the interview data. We utilized findings from previous self concept research, self concept theory, and our parental role model [McConkie-Rosell and DeVellis 2000] to develop our initial coding categories. Additional codes were developed following an inductive approach. Responses were then tabulated for each specific question and sorted based on genetic risk status. The interviews were first independently coded by AMR and GAS then jointly reviewed until consensus was reached.

RESULTS

Fifty-three girls from 13 different states in the US were enrolled between 2003 to 2006 (Table I). The majority of the girls were in high school at the time they were in the study and had at least one sibling affected by fragile X syndrome. Half of the girls had multiple relatives affected with fragile X syndrome. Over half of the girls thought they had learned that fragile X was in inherited disorder and knew they could be a carrier before they were 14 years old (Table II). Twenty-three percent of the girls could not remember a specific age that they learned fragile X syndrome was an inherited disorder in their family. Forty-two percent of the girls who are carriers and those who were not carriers knew their actual carrier status before age 14 years.

Global Self Concept

The total mean scores on the Tennessee Self Concept Scale were in the normal range for all three groups [carrier 56.60 (s.d. 10.88); noncarrier 52.17 (s.d. 5.20); at-risk 53.33 (s.d. 8.08)]. No differences were found among the groups (Kruskal Wallis Test; $p > .1$). This was also true for every subscale of the TSCS:2.

Situationally Specific Feelings About Self

FraX-VAS—Means and s.d. as well as median scores for the self concept items on the FraX-VAS are in Table III. There were no group differences with respect to the distributions of age and the seven FraX-VAS scores (Kruskal-Wallis test; $p > .1$). The only significant results were found for two of the FraX-VAS scores: Feelings about Self ($p=0.01$) and Social Self ($p = 0.009$). There was no evidence for group differences for age or any of the other five FraX-VAS measures ($p>0.10$ in all instances). Where the overall test for group differences was significant, the significance of pairwise group differences was assessed.

Pairwise comparisons among the three carrier status groups found feelings about self differed significantly for the at risk and noncarrier groups (medians of 5.0 and 5.7 respectively) after adjustment for multiple comparisons, with the noncarrier group showing more positive feelings about self. The distribution of scores for the carrier group was intermediate (median of 5.1) and could not be said to differ significantly from that of either the noncarrier group or the at risk group. In contrast, on the social self item, the distribution of this outcome in the at risk group was found to differ significantly from that of the other two groups after adjustment for multiple comparisons with both the carrier and noncarrier groups reporting more positive perceptions. There was no evidence of a difference between the carrier and noncarrier groups. This is reflected in the pattern of median scores: median VAS of 5.00 in the at risk group versus the median scores of 5.45 and 5.35 in the noncarrier and carrier groups, respectively.

For all three groups there was no difference in how the girls initially rated feeling about the possibility of being a carrier on the FraX-VAS (Table IV). Significant differences emerged when considering how they remembered feeling when they learned their test result (for the girls who know they are carriers and noncarriers) and their current feelings about their genetic risk status for all three groups.

We found no significant change in perception of emotions of the girls who are carriers from when they first learned they could be a carrier to when they learned their actual carrier status for fragile X syndrome ($p>.14$ in all instances, two-tailed Wilcoxon signed ranks tests). However, the girls who are carriers reported significant improvement in two emotions, upset and scared, comparing when they first learned their carrier status to their current state (less upset $p= .02$; less scared $p = .01$). Girls in the carrier group also reported feeling less upset ($p=0.04$) and scared ($p=0.003$), and a suggestion of a feeling less anger ($p=0.06$) between the time they learned their actual carrier status to their current feelings at the time of interview.

The girls who were found not to be carriers reported significantly improved feelings at the time they learned their noncarrier status; they were less upset, less scared, more relieved, less angry and happier ($p < .001$ in all instances). There was no change reported from when they learned their actual carrier status relative to their current feelings ($p>.15$ in all instances). For the girls who are at-risk there were no significant changes in their reported emotions on the FraX-VAS related to first learning they could be a carrier compared to their current feelings ($p\geq.12$ in all instances).

Interview

Stages of Knowledge—During the interview, study participants were asked to describe their feelings at three stages of knowledge regarding their genetic risk status. All three groups reported similar feelings when learning they *could be* carriers of fragile X syndrome. Threats to the enactment of the parental role were identified in a majority of the responses from the girls in the at-risk and in the noncarrier groups. This finding was also true, but to a lesser extent in the girls who are carriers. Many of the girls cited negative emotions such as sad, scared, upset, disappointing, or worried to describe how they felt learning this information.

> Carrier (17 years)
>
> Um…I…I was a little sad just because I've always wanted to have kids, and it's just going to make it that more difficult because I can't just get pregnant naturally. I have to go through in vitro or whatever…whatever they can do right now, and maybe in the future, they'll have more advances, but otherwise, it doesn't really affect me that much, but just that it was going to make it more difficult.

They also expressed acceptance or resignation that their carrier status could not be changed. Additionally, several of the girls who were carriers expressed that initially they did not understand that the diagnosis of fragile X in family had risk implications for them as well as their affected relatives. This finding was fairly unique to the girls who are carriers.

> Carrier (15 years)
>
> I think that I thought it was kind of, I kind of liked it because I felt like I was kind of like my brother. But later on, I realized that I didn't really like it.

The girls who had been tested and knew their carrier status were then asked to describe how they felt when they learned their actual carrier status. The girls who are carriers continued to express negative emotions such as upset and sadness as well as acceptance or resignation that their carrier status cannot be changed. Fifty percent of the noncarriers expressed positive emotions such as being relieved or happy that they no longer had to be concerned about the reproductive implications of being a carrier.

> Non carrier (16 years)
>
> I was happy, I mean, it's a good thing. I mean not carrying it is a good thing. It has, it's good for when I get married and have kids, but it wouldn't, I mean, it really is, I'm not, wouldn't have been upset if I was.

Finally, the girls were asked to describe their current feelings about their carrier or at-risk status. Threat to the parental role continued to be a concern for the girls who are carriers and to a lesser extent in the girls who are at-risk. The girls also identified barriers to becoming a parent including when in a relationship to tell a potential partner and cost of prenatal testing options, concerns about having an affected child. At the same time, the girls expressed confidence that they would be able to parent a child with fragile X syndrome if that should happen to them.

> Carrier (age 18 years)
>
> I guess I'm more worried. It's like if I were to get married. How do I tell my husband or like my fiancé or boyfriend you know when I'm much older. Like how would I tell him, if we have kids, it might not turn out so good.
>
> Carrier (14 years)
>
> Kind of proud in a way. I might have a kid that's affected, but I would know kind of how to handle them in a way because I have my sister, but still worried. My cousin was lucky. Her daughter, she just had a kid, she wasn't affected.

Many of the girls who were noncarriers expressed relief that they were no longer at risk to have an affected child.

> Noncarrier (19 years)
>
> I'm glad. I always thought that I would be fine with having a fragile X kid, but now that I am older and I am planning my life and I have a steady boyfriend it is like, I am so glad that I am not you know. I mean I love my brother and I love my uncle, but I'm just really glad that it is not me, you know.

Forty-five percent of the girls who were carriers also included statements about either family genetic identity or personal genetic identity. This was also true for the girls who were noncarriers, although to a lesser extent, and were not made at all by the girls who were at-risk.

> Carrier (16 years)
>
> Um, I didn't really feel sad or anything. I mean, to me, it's just the way things happen. I didn't really feel sad or angry or mad. I mean, of course, I wasn't happy, I was just, well, it's genetics, it's just, it's nothing I can do about it, so I just accepted it naturally. …I don't really have any hate or anger towards it, I mean, it's just part of who I am and it makes up part of who I am, so.

In the girls who were at-risk there was a reduction in statements indicating acceptance or resignation from how they described first learning they could be a carrier (67%) to how they feel now (33%) that was also associated with an increase in statements indicating uncertainty about how they felt (from 0% to 33%).

> At risk (17 years)
>
> Um, it could be bad, but again, still, I still don't really know very much about what I'm going to do, so, it's still is kind of information that's waiting to be used, but not being used right now.
>
> At-risk (14 years)
>
> The same way, really. I mean, I want to know, like it's something that I think, like I said, that I wanted to know and I don't really know how I feel about it.
>
> At-risk (18 years)
>
> I've…I really don't know cause I'm kinda iffy, I don't I'm just in-between kind of cause I don't know.

In contrast, the girls who were carriers did not expressed conflict. These girls showed an increase (from 25%) in statements indicating acceptance or resignation from how they remembered feeling when they first learned they could be a carrier for fragile X syndrome (to 80%).

> Carrier (18 years)
>
> I'm fine with it, and I can't even do anything about but learn…learn more about it, so I don't fret on it or worry about it or…
>
> Carrier (18 years)
>
> It's just one of those things (small laugh) I mean it's…it's just something that you know, we'll have to deal with one day, whenever I get married and have kids and all that, but I mean, it's, you know…well maybe you had a kid, there's always that chance that anything could happen to them, you know, they could…I mean, you never know.

Some of the girls recognized a change in their feelings when they considered how they first felt to how they were feeling in the present and for the at-risk and carriers, the possibility that they might feel differently in the future. This was commonly seen in the girls who were carriers and less in the at-risk girls.

> Carrier (16 years)
>
> Yeah, that is when I thought that it was kind of interesting. I thought it was kind of cool that I had it but that I could not be affected by it. But I didn't really realize that my kids could be.
>
> You didn't realize what it meant.
>
> Yeah.
>
> What about how you feel now about it?
>
> Now it is kind of like I wish that I didn't.

Descriptions of Feeling About Self Related to Genetic Knowledge—Study participants were asked to describe in what ways, if any, they thought knowing their carrier status or at-risk status had affected their feelings about themselves. The majority of the girls (65% of the girls who are carriers, and 56% noncarriers, and 73% of the girls who are at-risk) reported that they did not feel knowing about their genetic risk status had affected their feeling about themselves.

> Carrier (17 years)
>
> I don't think it's made me look at myself really differently. "I'm so horrible" or anything like that…(laugh)

Fifteen percent of the carriers and 20% of the girls at-risk reported that learning their genetic risk status had made them feel worse about themselves. None of the girls who are noncarriers made this observation.

> Carrier (22 years)
>
> Yeah. It has affected the way I feel about myself, but…immediately how I felt about myself was completely different than now…you know. I mean it takes time, you know, to accept something. …I felt like something…like immediately I felt like something was wrong with me type thing, you know. Something was wrong, but now, you know, it's just…it's just me. You know. That's the way I am. I mean…I'm more accepting now, and I understand it now, so I'm not as scared of it or anything.

A similar number of the girls who are carriers and noncarriers indicated that learning their genetic status had made them feel better about themselves while none of the girls at-risk reported these positive feelings. Although the majority of the girls did not report a change in self perception, approximately one fourth of all of the girls expressed concern about how their status might affect having children. They also recognized how the possibility of having an affected child might impact their future.

> Carrier (17 years)
>
> Um…kind of the same, but I'm glad that I know. I mean, I'm glad that I'm not just going to…I'm glad that knowing that I wouldn't just have a child and, you know, surprise, like they have Fragile X.

Some of the girls who are carriers and noncarriers also included statements related to their genetic identity. However, this was not true for the girls who were at-risk.

> Carrier (18 years)
>
> It…it hasn't. I mean it's not…like I said, it's not my fault. It's not my dad's fault. Just because of that doesn't meant that, you know…I mean whenever I found out, it's not like it changed me. You know, it didn't have anything to do with me…you know me as a person.

A few of the girls who were not carriers comment that they would have been okay with the outcome of the test had they been found to be a carrier.

> Noncarrier (16 years)
>
> Myself, it really hasn't like, I mean, it hasn't, like I don't look at myself different because I'm not a carrier than I would if I was a carrier. I look at myself pretty much the same. I mean, it's just me and if I was a carrier, it wouldn't be a problem to me.

However, some also stated that they might have felt differently about their status if they had been found to be a carrier.

> Noncarrier (17 years)
>
> Not at all. It didn't, I mean I was very happy that I wasn't a carrier, but it didn't change anything. I think it'd be a lot different if I was a carrier. It would be pretty hard.

Some of the girls who are noncarriers expressed ongoing concern for their affected relatives and a few expressed survivor guilt.

> Noncarrier (23 years)
>
> It just kind of makes me a little bit more grateful for being able to just grow up, find someone that I love, marry him, be able to have a kid, you know, and not have to think about the things that my sister's going to have to think about or that my parents are going to have to think about and things like, well, what if some man takes advantage of her some day or something like that? You know, I guess just being aware that I'm not ever going to have to make those kinds of decisions for my child. It makes me grateful. But, I don't know, not so much about how I view myself. I mean, I can't really think of anything.
>
> Noncarrier (22 years)
>
> Um…it's affected me in different ways. Like, there's the survivor guilt, which kind of is always in the back of my head of like being so relieved that I'm not having someone like my brother, but I don't think he would want it. Like, I know he's…on some level, he realizes that he's different, and I think that bothers him, so I know… I can know that he would be glad if…I… but it's made me happy and like everything.

Physical Self—Study participants were asked if they felt they had features of fragile X syndrome and if so, to describe what they thought those features were. Subjects in all three risk categories sought symptoms of fragile X syndrome including physical and emotional features. Seventy-five percent of girls who knew they were carriers, and 73% of the girls who are at-risk either sought and found symptoms of fragile X syndrome or thought that they might have features but were a little uncertain. Sixty-seven percent of the girls who are noncarriers also sought symptoms and a few of those identified what they thought could be a feature of fragile X syndrome. Only one fourth of the girls in any group definitively stated that they did *not* have features of fragile X syndrome.

> Carrier (22 years)
>
> Well, if you'd a put the features down on paper before I ever found out about, probably I would have said no, but now that I've been able to look at it and everything, and I think I do a little bit. …Let's see. There was something about the ears. I'm trying to re…like, let's see. I'm trying to think physical. (laugh) Probably not as … My eye… I had that lazy eye. .. I had it surgically corrected when I was 12. Let's see. What else was it that I'd remember? I know at one point I thought there was some emotional behaviors that might have been, but I don't remember exactly.
>
> Noncarrier (19 years)
>
> Um, I've heard different things. They have told us that if you are a carrier, which I am not so it makes me wonder but, um…. Maybe there is something wrong with me (laugh). My cousin, who is a carrier, her and I are a lot alike with learning styles. We are very visual learners- it takes a lot to get through to us I think and I have also heard that irregular periods can be a sign of being a carrier which that runs in our family. I don't know if that is a part of our genetic make-up or if it is the fragile X thing.

Social Self—We asked the girls to consider if knowledge of their genetic risk status impacted on their relationships with friends and if they talked with their friends about their genetic risk status. The majority of the girls (77% girls who are carriers, 76% noncarriers, and 87% of girls who are at risk) felt that their genetic risk status had not made a difference. When asked if they had informed friends or other non family members about their genetic risk the majority of the girls who are carriers (85%) and noncarriers (77%) reported being able to openly discuss their carrier status with their friends and that theirs was a positive experience. Only 27% of the girls at-risk made this observation.

> Noncarrier (15 years)
>
> Well like they (my friends) ask me, they're like, "well, do you have it?" and I tell them no and that I don't have to worry about it cause I'm not a carrier cause I got tested for it and stuff like that.
>
> Carrier (20 years)
>
> Yeah, my best friend has known about it since I've known about it. We've been friends since elementary school, so she's always kept up on it. And I think that's good to have somebody out of your family that you can talk to about it that's not so affected by it, and my boyfriend from the get go…he's…I've never kept it a secret because if I think he's the one, which I think he is, I don't want to have to tell him what's going on, so…I think especially with him, it's been good that he's known everything about it, and he knows my brother and, you know. So I…I think it's good to have told, you know, people that you're close to.
>
> Carrier (17 years)

Yeah, I've told…I've told like my best friend about Fragile X, and I told her that I was a carrier of it, and…

How did she react?

Um, she…I think that she was…I think she was surprised too, but like I explained to her that…that this was why I have it, and I think she understood.

Girls who are at-risk were more likely to "keep it all in the family" than either girls who were carriers or noncarriers.

At-risk (14 years)

They don't know anything about it.

You've never told them?

No. I don't talk about it at all to them.

The girls who are at-risk were also more likely to report that did not feel supported by their friends when they did talk about their risk status than either the girls who were carriers or noncarriers.

At-risk (18 years)

Like that day in biology, we're talking about fragile X, and you know, then I told them, I have a nephew that has fragile X. They're all like how do you get it? I'm like well it just happened that my sister was a carrier and she had somebody with fragile X. Saying how the X chromosome is not stable enough and it was like broke off you could say.

What was their response?

They're like oh well, I don't really care.

A few of the girls also reported that sometimes it can be difficult to explain fragile X syndrome and how it affects their siblings to their friends.

Carrier (17 years)

Sometimes they'll ask me do I have any other brothers or sisters and I'll have to tell them about my brother and sometimes they'll ask, "well, what's fragile X?" and I explain it to them a little bit.

What do you say when they say, "what's fragile X?"

It's hard to explain…I just tell them sometimes how they can turn out or … It's just hard to explain to somebody.

Family Self—When asked if they had informed any of their own family about their carrier status the majority of the girls reported that there was no need to tell as their relatives (grandparents and/or aunts and uncles), had already been told, usually by one of their parents. We asked the girls whether and how knowing carrier status had affected the way they view their family. The majority of the girls who are carriers and noncarriers, but only one of the girls at-risk reported that they felt their family was closer and/or knowing their status had a positive effect on their family. The majority of the girls at-risk reported that their carrier status had no effect on how they viewed their family.

Carrier (20 years)

Um…well, I guess, in a way yes, because I know that they have this disease, and I mean, I know that…that like it happens, you know, and there's something that they

all have to live with. But I think it kind of helps that we're such a close family, so we can all…we all kind of deal with it together, and we, you know, there's a lot of support in our family.

Noncarrier (16 years)

I think it's made me put a lot of things into perspective with my family. It's made me want to grow closer to my family because I want to learn all that I can about fragile X and how to help.

Over half of the girls who were carriers and noncarriers and a few of the girls at-risk also reported an awareness of their family genetic identity.

Noncarrier (16 years)

Um, it makes it easier, it's more structured when I know like, I know everyone's status. Like.. I know my mom is a carrier and I know my brothers have it and my sister and I aren't a carrier and it's easier to look at my family and say like, it's not like oh she could be a carrier. It's just…. You're like wow, or you see something and go maybe that's a Fragile X carrier characteristic. It makes things easier and it's kind of like, like it's all there and you know exactly who has what and what it is and it's easier to understand people when you know what they have… about their status.

At-risk (18 years)

Different…..I have an unusual amount of family members that have some kind of handicap. When you look at most families and they don't have that many of handicapped people.

DISCUSSION

Despite mean scores on the TSCS for all three groups suggesting that global self concept is not affected by knowledge of carrier status for fragile X syndrome, we found differences in feelings about self specifically related to the interpretive meaning of genetic risk knowledge in these girls. The difference in self perception appears to be related to certainty of genetic information. Girls who only know that they are at risk tend to report reduced feelings about self, and a significantly negative effect on their relationships with friends. There were no differences among the groups in how serious a problem fragile X was viewed or level of family upset.

Threats to self concept were found in personal self, social self, and family self as they specifically related to the meaning of genetic information and varied based on risk status.

Personal Self

Personal threats to self concept identified in this study were: physical self, genetic identity, and threat to the parental role.

Physical Self (Symptom Seeking)—Interestingly, symptom seeking occurred in all three groups including girls who know they are not carriers. Symptoms sought included emotional (shyness), academic (problems with math in school), and physical characteristics (large ears, long face). Symptom seeking has been previously reported in a study of adult siblings of individuals affected by cystic fibrosis who were raised with the possibility of being a carrier for cystic fibrosis [Fanos 1999; Fanos and Johnson 1995] and in a similar study of siblings of individuals with ataxia-telangiectasia [Fanos and Puck 2001]. Fanos and colleagues found that misunderstandings, formulated as children, persisted into adulthood regarding the

identification of possible symptoms of the disorder and those symptoms were used to reinforce a believed carrier status.

In our previous study of adult women going through the carrier testing process for fragile X syndrome, we also identified symptom seeking as a common phenomenon [McConkie-Rosell et. al. 2000]. The adult women in our longitudinal study sought symptoms of fragile X syndrome in themselves to support a believed carrier status when they were at-risk. Similar to the adolescent girls, the women in that study cited their poor math skills and shyness as possible symptoms of fragile X syndrome. However, once they learned carrier status, the women who were found not to be carriers no longer sought symptoms of fragile X syndrome. In this study, however, the girls who were not carriers continued to seek symptoms and a few believed they were successful in identifying mild characteristics. This finding suggests that it can be very difficult for some adolescents, regardless of their actual carrier status, to definitively state that they do not have features associated with a genetic disorder that has been diagnosed in the family.

For these adolescents, symptom seeking may have begun in early childhood and may have been incorporated into the developing self concept. These long standing beliefs about self may be challenged as inaccurate assessments or appear to persist even though refuting information is available. It is important to recognize that symptom seeking is common and it is not an indication of outcome of the genetic test (for those who have not been tested) and it is not necessarily confirmation of an expression of the disorder in those that test positive.

Threat to Parental role—We have broadly defined threat to parental role to include both positive and negative perceptions and have proposed that the parental role is a domain of self concept that may be affected by genetic risk information [McConkie-Rosell and DeVellis 2000]. This study, as well as our previous study of adult women [McConkie-Rosell et. al 2000] lends support for this concern. Threat to the parental role was found in the descriptions of feelings about self and in all three groups when they were describing how they felt about first finding out they could be carriers. However, there was also an acceptance as these responses were frequently coupled with expressions of love for their affected relative, as well as confidence that if they did have an affected child they could competently care for that child.

These findings suggest that it is genetic risk information that is threatening, not the learning of actual carrier status. Importantly, unlike the girls who are carriers, in whom this concern was often paired with thinking about the future and considering possibilities, or the relief felt by the girls who are not carriers, the girls who are at-risk are uncertain about what they think and often have not considered future possibilities. Certain knowledge of genetic status appears to allow the girls to move forward in thinking about their future plans, especially those related to becoming a mother.

Genetic Identity—The term, genetic identity was first used by Schild [1981] to describe a shattered self-adequacy syndrome caused by the knowledge that one posses a defective gene. Findings from this study did not support the concept that knowledge of an altered gene was experienced as a threat to the total person resulting in a compromised sense of self. We did, however, find support for the concept that “genetic identity” exists.

The girls who are carriers as well as the noncarriers do not appear to be experiencing their genetic identity as “I’m defective”. Rather, they are able to compartmentalize it as only a small part of their total self. Linville [1985] has highlighted the important protective mechanisms of many different domains of self. Our findings suggest that girls who are carriers and noncarriers are able to incorporate their genetic identity in a positive manner. These girls also acknowledge that their genetic status is uncontrollable. The recognition that genetic status is not controllable

and thus not a personally accountable action, seems, for some of the girls, to be protective. This incorporation of the genetic identity and strategy of compartmentalization was not found in the girls who are at-risk.

Social Self

A significant effect on the perception of genetic risk information on friendships was identified. The girls who are carriers and noncarriers discuss their carrier status with their friends and found it to be a positive supportive experience. These girls seem to choose who to tell, such as a good friend, or they talk about their own status as part of educating people in general about fragile X syndrome. Disclosure appears to be part of developing intimacy, an important component of the adolescent developing self-concept [Berndt and Perry 1990]. The girls who are at-risk reported that knowledge of their genetic risk had a negative effect on their friendships, that they are less likely to inform and seek the support of friends about fragile X syndrome, and typically do not discuss concerns about their possible carrier status. In addition, the girls at-risk indicated that if they did talk about their risk status with their friends, it was sometimes a negative experience. The girls who are at-risk have some uncertainty about what to disclose and what level of concern to express to their friends which may be a barrier to the development of intimate friendship relationships and support as it relates to genetic risk status.

Family self

We found an interesting difference in the girls' perceptions of their families. The girls who are at-risk were more likely to report that fragile X syndrome had no effect on their family. In contrast, the girls who are carriers and noncarriers report that fragile X syndrome has had a positive effect by making their family closer. For the girls who are noncarriers we also found that the relief and reduced concern felt by them regarding their own carrier status was often paralleled by concern for their family and for relatives who were carriers, at-risk, or affected. Family support and adolescent self concept are positively interrelated [Hoelter and Harper 1987]. Demo et. al. [1987] also found that the greater the perceived level of support, participation, and communication, the higher the level of self esteem in adolescents and their parents. It is possible that the open communication regarding genetic status that may occur as part of the process of learning status for girls who know they are either carriers or noncarriers has had a positive effect on aspects of family communication regarding fragile X syndrome leading to this perception.

Family Genetic Identity—For the many of the girls in this study family genetic identity is intrinsic to their family. They had an awareness of a difference in their family because of fragile X and sometimes found comfort in knowing others in their family also faced these concerns. They also expressed concern for those same relatives, both affected and those who could be carriers. They looked to other family members to infer their status, if it were unknown, and felt that they could relate to those whose status was the same as theirs, if it were known. They openly speculated about who and who is not a carrier or affected. We have used the term "family" genetic identity to describe this observation because, for the girls in this study, the knowledge about an altered gene "in the family" has become an integral part of the shared family story.

Stages of Knowledge

Differences were identified in how the girls both ranked and described their feelings about three different stages of knowledge about their risk status. While there were no differences seen in how the girls recalled and rated feeling when they first learned their "at-risk" status; significant differences were found in ratings regarding learning actual carrier status and current feelings. The girls who are confirmed carriers and noncarriers reported improvement in feelings

about their status from when they first learned they *could be* a carrier. The girls who are carriers rated learning they were "at risk" the same as learning they were carriers suggesting that learning at risk status was just as emotionally charged as learning they were carriers. In addition, the girls who are carriers reported significant improvement when considering their current feelings. Improvements in feelings were seen in the girls who were not carriers at the time they learned their genetic status. However, the girls who are at-risk did not report any change in their feelings over time.

On the interview questions, the girls who are carriers demonstrated an acceptance or resignation that genetic status cannot be changed. They reported fewer negative emotions when describing their current feelings about their positive carrier status versus how they reported remembering feeling when they first learned they could be a carrier and when they first learned their actual carrier status. There appears to be a developing maturity in their perception. Although, threat to parental role remains a major concern, this concern is paired with thinking about the future and considering possibilities. There is an acceptance of the possibility of having an affected child as well as consideration of reproductive options such as adoption.

In the girls who are at-risk there was an increasing uncertainty coupled with a reduction in acceptance or resignation in their descriptions of the different stages of feelings on the interview. Unlike the girls who are carriers who have begun to face the certainty of their genetic risk some of the girls who are risk have become more uncertain about how they feel. This may be occuring because carrier status is still unknown.

Study Limitations

There are several limitations to this study. Fragile X syndrome is an X-linked disorder and the findings may not be generalizable to either autosomal recessive or dominant disorders including presymptomatic testing for an adult onset disorder. Study participants also self selected for the study. We attempted to obtain representative sample through recruitment from multiple sources. This study is limited by cross-sectional design. Carrier testing was not done as part of this study and we could not control how or when study participants learned their status. Therefore, the perceptions reported are retrospective. However, each of the three groups provides a unique perspective on adolescent self concept related to genetic knowledge.

CONCLUSION

An important finding in this study was that threats to self concept were found in girls in all three risk categories suggesting that the risk information itself, is threatening, and for some girls, may be as threatening as learning one is a carrier. Our findings also suggest that actual knowledge of carrier status is not universally damaging to the developing self concept. The developing adolescent self concept is influenced by facing adversity, resolving crises and decision making related to life events and is positively influenced by overcoming difficult life situations [Balk 1995]. Certainty related to genetic risk status appears to make a positive difference for some girls by allowing them the opportunity to face the challenge of their genetic risk status and to begin to consider the meaning of this information. For the girls who are carriers, this process leads to less uncertainty about how they feel about being a carrier as well as being able to consider what they might do in the future related to enactment of the parental role. Knowledge that they are not carriers has allowed the noncarriers to feel relief for themselves in parallel with elevated concern for other family members. The girls in this study, who know their carrier status, viewed their families as closer and were also able to positively incorporate and compartmentalize their genetic identity. In these domains of self concept the girls with knowledge of actual carrier status appear to be more similar than the girls who know only that they are at-risk. Because the at-risk girls do not have knowledge of their actual carrier

status they appear to have greater uncertainty about how they feel about themselves, the possibility of being a carrier, as well as experiencing a barrier in their relationships with friends.

Genetic counseling is important for the adolescent and his/her family in offering support, accurate information, and anticipatory guidance. Additional research is needed to develop strategies for families to help positively manage the information and evaluate the effectiveness of these strategies. These strategies need to consider that threat to self concept may be occurring based on informing adolescents about their genetic risk and that they are not spared these threats by not offering testing. The objective of the genetic counseling should be to focus on the resolution of these threats to self concept to enable adolescent to move forward regardless of whether and when testing takes place.

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Table I

Sample Demographics N =53

Ethnicity	**n (%)**	**n (%)**
	Caucasian	50 (94)
	African American	2 (4)
	Hispanic	1 (2)
Religion		
	Baptist	8 (15)
	Protestant/Christian non-denominational	27 (52)
	Catholic	9 (17)
	Jewish	2 (4)
	No formal affiliation	6 (11)
	No religious beliefs	1 (2)
Year completed in school at the time of the interview		
	7–8th grade (Middle School)	6 (11)
	High School (9–12th)	31 (58)
	High School Graduate	2 (4)
	College	16 (30)
Closest relative affected by fragile X syndrome		
	Sibling	35 (66)
	1st Cousin	9 (17)
	niece/nephew	5 (9)
	Uncle/aunt	1 (2)
	Greater than 3rd degree	2 (4)
Multiple relatives affected		29 (55)

There are no significant differences among the three groups (p> .1 Pearson Chi-Square)

Table II

Age of Study Participants and Ages They Remember Learning Their Genetic Risk

	Carrier (N =20)	Noncarrier (N= 18)	At-risk (N = 15)
Age	mean 18.45 years median 18 years (s.d. 2.5)	mean 17.8 years median 18.5 years (s.d. 2.7)	mean 17.9 years median 17 years (s.d. 3.2)
Age learned that fragile X was inherited "runs in the family"*			
	n (%)	n (%)	n (%)
< 5 years	0 (0)	0 (0)	1 (7)
6–10 years	3 (15)	7 (39)	6 (40)
11–13 years	6 (30)	2 (11)	5(33)
14–18 years	7 (35)	2 (11)	0
>18 years	1 (5)	0	1 (7)
Unable to remember a specific age/ always known	3 (15)	7 (39)	2 (13)
Age learned *could be* a carrier*			
< 5 years	0	0	0
6–10 years	1 (5)	7 (39)	6 (40)
11–13 years	7 (35)	5 (28)	5 (33)
14–18 years	10 (50)	3 (17)	0
>18 years	1 (5)	0	1 (7)
Unable to remember a specific age/always known	1 (5)	3 (17)	2 (13)
Age learned actual carrier status (carrier/noncarrier)*			
< 5 years	0	0	
6–10 years	2 (10)	3 (17)	
11–13 years	5 (25)	6 (33)	
14–18 years	8 (40)	4 (22)	
>18 years	3 (15)	1 (6)	
Unable to remember a specific age/ Always known	2 (10)	4 (22)	

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

Table III

FraX-VAS (Feelings About Self Related to Genetic Risk Information) Means (s.d) and Median.

	Carrier n = 20	**Noncarrier n= 18**	**At-risk n = 15**	**p value***
Perception about how serious a problem fragile X syndrome is viewed n± S.d. [median]				
How do you view fragile X, in general? 0 = very serious problem 10 = not a problem	4.9 + 3.0 [5.0]	4.96 + 3.4 [5.0]	3.5 + 2.3 [3.60]	0.3
Feeling about self				
How has knowing your carrier status affected the way you feel about yourself (at-risk: knowing you could be a carrier)?	6.0 + 1.9 [5.1]	6.8 + 2.1 [5.7]	5.2 + 0.5 [5.0]	0.01**
Social Self				
How has knowing your carrier status affected your relationship with your friends (at-risk: knowing you could be a carrier)?	6.5 + 2.0 [5.4]	6.8 + 2.1 [5.5]	4.5 + 1.8 [5.0]	0.01***
Family Self				
Has knowing your carrier status affected your relationship with your parents (at-risk: knowing you could be a carrier)?	6.8 + 2.1 [5.5]	6.7 + 2.1 [5.3]	5.6 + 1.7 [5.5]	0.11
How has knowing your carrier status affected your relationship with your siblings (brothers/sisters)	6.6 + 2.0 [5.4]	6.7 + 2.4 [5.6]	5.8 + 1.6 [5.0]	0.4
How has knowing your carrier status affected your relationship with your extended family (aunts, uncles, cousins)	6.4 + 1.9 [5.5]	6.5 + 2.1 [5.1]	5.6 + 2.0 [5.0]	0.56
How has your family reacted to finding out that fragile X syndrome runs in your family?	7.2 + 2.3 [7.6]	7.1 + 2.0 [7.1]	7.4 + 1.9 [7.7]	0.92

Anchors are negative (0) to positive (10). The midpoint of 5 is intermediate indicating no effect/no difference.

* Significance probability associated with the Kruskal-Wallis Test of the null hypothesis that there is no difference in the distribution of the outcome of interest among the three carrier status groups.

** Data provided evidence that there were differences among the three carrier status groups with respect to the distribution of the VAS reflecting Feelings about Self. After adjustment for three pairwise multiple comparisons among the groups, the at-risk and noncarrier groups were found to differ significantly; the carrier group could not be said to differ significantly from either the noncarrier group or the at-risk group.

*** Data provided evidence that there were differences among the three carrier status groups with respect to the distribution of the VAS reflecting Social Self. The distribution of this outcome in the at-risk group was found to differ significantly from that of the other two groups after adjustment for multiple comparisons; however there was no evidence of a difference between the carrier and noncarrier groups.

Table IV

Stages of Knowledge FraX-VAS Means (s.d.) and Median

How did you feel when you first found out you could be a carrier?				
	Carrier n =16* N+ s.d. [median]	**Noncarrier n = 17* N+ s.d. [median]**	**At-risk n = 15 N+ s.d. [median]**	**p value+**
Upset	5.9 + 2.7 [5.5]	5.5 + 2.2 [5.0]	6.9 + 2.7 [6.8]	0.38
Scared	6.6 + 3.1 [7.5]	5.7 + 2.9 [5.0]	6.1 + 2.3 [5.0]	0.65
Relieved	6.2 + 3.1 [6.1]	6.1 + 2.6 [5.1]	5.5 + 1.9 [5.1]	0.77
Angry	8.5 + 2.1 [9.7]	7.2 + 2.4 [7.9]	7.7 + 2.7 [9.4]	0.18
Happy	6.9 + 2.3 [5.4]	6.9 + 2.4 [5.8]	5.5 + .88 [5.0]	0.23
How did you feel when you first found out your carrier status? (omitted for at-risk group)				
	n = 18**	n = 17**		
Upset	5.4 + 2.9 [5.0]	8.4 + 2.0 [9.5]		0.002
Scared	5.9 + 3.2 [5.4]	9.0 + 2.0 [10.0]		0.003
Relieved	5.8 + 2.6 [4.9]	1.3 + 2.0 [0.2]		0.003
Angry	7.7 + 2.3 [8.6]	9.0 + 1.7 [10.0]		0.04
Happy	6.1 + 2.5 [5.0]	1.7 + 2.3 [0.1]		<.0001
How do you feel now about your carrier status?				
	n = 20	n = 18	n = 15	
Upset	7.4 + 2.8 [9.0]	8.7 + 2.3 [9.7]	8.0 + 2.5 [9.4]	0.19
Scared	8.4 + 2.3 [9.5]	9.1 + 1.8 [10]	7.5 + 3.0 [9.4]	0.09
Relieved	5.8 + 2.9 [5.0]	1.4 + 2.3 [0.35]	5.3 +1.7 [5.0]	<.0001++
Angry	8.8 + 2.1 [9.6]	9.1 + 2.0 [10.0]	8.5 + 2.3 [9.7]	0.31
Happy	5.5 + 2.4 [5.0]	1.8 + 2.4 [0.3]	5.3 + 1.0 [5.0]	<.0001++

Anchors are very (0) to not (10). The midpoint of 5 is intermediate.

*4 girls who are carriers and 1 noncarrier reported that they had no memory of not knowing so could not answer the question.

**Two carriers and one noncarrier reported they had no memory of not knowing their carrier

+Significance probability associated with the Kruskal-Wallis Test of the null hypothesis that there is no difference in the distribution of the outcome of interest among the carrier status groups; exact significance probabilities are given for comparisons of carriers and noncarriers at the time they first found out their carrier status.

++Data provided evidence that there were differences among the three carrier status groups with respect to the distribution of the VAS. After adjustment for pairwise multiple comparisons among the groups, the noncarrier group was found to differ significantly from the at-risk and carrier groups; the latter two groups could not be said to differ significantly from each other.