Published in final edited form as:

J Genet Couns. 2012 December ; 21(6): 845–853. doi:10.1007/s10897-011-9454-x.

Diagnosis of Fragile X Syndrome: A Qualitative Study of African American Families

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Abstract

Fragile X syndrome (FXS) is an inherited genetic condition with critical consequences to the proband and family members at all levels in the generations. Although evidence demonstrates that the rates of diagnosis for FXS are the same in all racial groups, age of diagnosis in African American children has been reported to occur later than in Caucasian children. Additionally, African American families are seriously under-represented in existing FXS research studies. As such, it is important to understand the possible disparities in the underlying factors to receiving a diagnosis in African American families with FXS. Herein, a qualitative approach was adopted to describe the overall FXS diagnosis experiences (*pre-diagnosis, diagnosis, and post-diagnosis* stages) of a convenience sample of 10 African American mothers. We identified three major findings among our participants: (1) FXS testing is not ordered immediately once a parent expresses concerns of developmental delays to the pediatricians, (2) the diagnosis is sometimes delivered in an insensitive manner with information often being outdated and unbalanced towards negative aspects, (3) communication issues among family members exists once the diagnosis is discovered. Although these qualitative data may not be representative of the whole group, these findings have significant implications for genetic counseling and our understanding in providing support and advocacy for African American families with FXS.

Keywords

Fragile X syndrome; African American; Diagnosis; Medical support

Introduction

Fragile X syndrome (FXS, MIM 300624) is the most common inherited form of intellectual disabilities and the most common single gene cause of autism. The vast majority of cases is caused by expansion of CGG repeat in the 5' untranslated region of the *FMR1* X-linked gene. This expansion of over 200 repeats leads to decreased or absent levels of fragile X mental retardation protein (FMRP) (Sutcliffe et al. 1992) and is termed the full mutation. Premutation carriers have unstable alleles with 55 to 200 repeats. These alleles can expand to the full mutation during transmission from mother to child, leading to FXS. The premutation form is common in the general population with a prevalence estimate of approximately 1 in 130–259 females and 1 in 250–813 males (Cronister et al. 2005; Dombrowski et al. 2002; Hantash et al. 2011; Rousseau et al. 1995). FXS is more severe in males because the mutation is located on the X chromosome. This leads to a prevalence of the FXS in males of about 1 in 4,000 and, in females, about 1 in 8,000 (Coffee et al. 2009; Crawford et al. 2001; 2002; P. J. Hagerman 2008; Turner et al. 1996).

As an inherited genetic condition, the fragile X mutation has critical consequences to the child, parents, and extended family members at all levels in the generations. The term “fragile X associated disorders” is now applied to the clinical outcomes of the expansion mutation as it leads to at least three well established disorders, each with a broad spectrum of involvement and these can all occur in a single family (Chonchaiya et al. 2009). For instance, premutation carrier women are at risk for primary ovarian insufficiency (FXPOI) and possibly other conditions such as depression and anxiety, hypothyroidism, fibromyalgia, and neuropathy (Coffey et al. 2008; Hunter et al. 2010; Roberts et al. 2009). Premutation carrier men, and less frequently women, are at risk for developing fragile X-associated tremor ataxia syndrome (FXTAS), a late onset neurodegenerative disorder (Bourgeois et al. 2009; R. J. Hagerman et al. 2001). Thus, in one family that is segregating the fragile X mutation, young children may have FXS, their mother may suffer from ovarian insufficiency and early estrogen-deficiency disorders, and their grandfather or older uncle may have significant medical problems associated with FXTAS. For these reasons, early diagnosis has important implications not only for the child with FXS, but also for extended family members. Thus, once an individual receives the diagnosis, it is important to inform immediate and extended family members about the possibility that they may be carriers of the fragile X mutation.

Fragile X syndrome has no distinct physical features at birth to prompt the diagnosis, and only becomes evident when the child presents with developmental delays. Because of the subtlety of the somatic features and the variability of the intellectual and behavioral characteristics, the identification of a causal mutation is necessary for the diagnosis of FXS (Garber et al. 2008). The majority of the literature discussing barriers to receiving a diagnosis of FXS has primarily involved Caucasian participants. For instance, a study conducted by Bailey et al. in 2003 to determine family experiences and perceptions of FXS in 274 families (94.2% Whites, 2.1% African Americans, 1.8% Hispanics) indicated that males born in 1990 – 1999 received the diagnosis of FXS at approximately 32 months, on average 18 months after the family first identified concerns (Bailey et al. 2003). With patient advocacy, professional recommendations regarding prompt diagnosis for genetic testing, and increased exposure to information about FXS in the pediatric literature, Bailey et al. surveyed 1,250 additional families (85% Whites, 7% Hispanics, and 2% African Americans) in 2009 (Bailey et al. 2009). No change was detected in the age of diagnosis of FXS from 2003 to 2009 despite enhanced efforts to increase FXS awareness in the medical community and advocacy. The average age of FXS diagnosis remained relatively stable across the 7-year period at ~35 – 37 months. Notably, although both of these large effort FXS studies recruited participants nationally, 85–94% respondents were Caucasians compared to a

disproportionately low representation of 2% African-Americans families. Since FXS occurs in all ethnic groups, socioeconomic levels, and geographic region, the experiences and results described in the above national studies may not be representative of all families with a child with FXS as the participants were not ethnically diverse (Bailey et al. 2009; 2003). A study by Crawford et al. conducted in the Atlanta metropolitan area suggested that one-fourth to one-third of children identified with FXS attending public schools were not diagnosed before the age of 10 years, and that African American males were less likely to be diagnosed before the age of 10 than Caucasian males (Crawford et al. 2002). More studies were recommended to determine whether the lack of an earlier diagnosis in African Americans results from social issues, such as a lack of resources in the family or in the associated health-care professional, or to cultural differences related to the motivation to obtain a diagnosis.

Disparities exist in medical care and diagnosis for many children with developmental disabilities including children with autism (Gourdine et al. 2011). The diagnosis of autism in African American children occurs much later than in Caucasian children. As a result, African American children may require longer and more intensive intervention. (Hilton et al. 2010) As such, we are interested in understanding the diagnosis experiences of African American families with FXS. If the diagnosis is not timely, this also delays appropriate genetic counseling for the families on the recurrence risk of FXS, which can be important for family planning. Approximately one-third of the families in the Bailey et al. study gave birth to a second child before a diagnosis of FXS was made in their family, leading to additional affected offspring in many cases (Bailey et al. 2003). Furthermore, it may delay diagnosis of the premutation-associated disorders of FXPOI and FXTAS in older members of the family. For these reasons, it is important to appreciate the disparities in ethnic representation in genetic research and possible underlying factors to obtaining the diagnosis in African American families with FXS.

There are currently no studies evaluating the experiences in discovering FXS in African American families. Such studies are important in enhancing our knowledge of potential benefits and barriers associated with obtaining the diagnosis and its implications. These findings are the crucial initial steps in designing a culturally sensitive plan to promote awareness and earlier diagnosis of FXS. Herein, we examine the diagnostic experiences of African American families with FXS and the consequences after the diagnosis is received. For the purpose of this study, we define the diagnostic experiences beginning from the time the parent first suspected concerns in their child's development. Finally, we describe the potential implications for families once the FXS diagnosis is known.

Methods

Design and Participant Recruitment

A semi-structured, in-depth qualitative interview as well as a brief demographic questionnaire were used to learn more about the FXS diagnosis experiences in African Americans. After receiving approval from the Emory University internal review board, we recruited African American women who had a child with a diagnosis of FXS. Informed consent was obtained from all participants prior to participation. Participants in this study were recruited through the Emory Fragile X Center research studies or through their attendance at the Fragile X Clinic at Emory University. The Emory Fragile X Center includes studies investigating phenotype consequences of the premutation including neuropsychological function, reproduction, and movement disorders throughout the United States. Twenty-seven participants met the eligibility criteria of self-identifying as African American and having a child diagnosed with FXS. The Emory Fragile X Clinic has seen 94 families, and 15 were eligible for this study. However, five of these families overlapped with

the eligible families from the Emory Fragile X Center studies, resulting in a total of 37 eligible participants.

Each family received a letter explaining the details of the study along with the information regarding who they should contact if they were interested in participating. A follow-up phone call was placed two weeks after the letter was sent to ensure that the contact information was correct and that families received the letter. Of the 37 letters that were sent and contact made, ten women agreed to participate. After receiving informed consent, an interview time was scheduled by the participant.

A trained interviewer conducted all interviews over the phone and recorded them with permission. The interviews took approximately 30 min to complete. Each participant also completed a quantitative questionnaire covering topics such as education level, income, child's age at diagnosis, insurance status, and distance from a major medical center. At the conclusion of the interview, each participant received a \$20 gift card.

Interview Guide

The interview guide was created specifically for this study and consisted of semi-structured questions (Table 1). The questions focused on the following areas: (1) first signs that prompted the parent to be concerned; (2) interactions with physicians or other medical professionals; (3) benefits and barriers to receiving a FXS diagnosis; and (4) sources of support and information after receiving the diagnosis. Probing questions were used to gather more information in these areas. All interviews were conducted by the same interviewer who has a Master's degree Behavioral Science and Health Education in Public Health (KC).

Data Analysis

The data from the demographic questionnaire were analyzed using descriptive statistics. The qualitative data were transcribed verbatim from the interviews. The transcribed interviews were uploaded into Atlas Ti 6.2 and analyzed using directed content analysis. Two of the authors independently coded the interviews for themes. They then collaborated to discuss all differences until a consensus was reached.

Results

Study Sample

A convenience sample of ten mothers of children with FXS, who self-identified as African American, enrolled in the study (Table 2). These mothers were from six different states and ranged in age from 27 years to 51 years with a median of 44.5 years. Three families had Medicaid and an annual household income of \$21,000–\$51,000, while five families earned \$51,000–\$100,000 a year, and two families over \$100,000 annually. In terms of maternal education level, there were two high school graduates, six with bachelor's degrees, and two of the mothers had advanced degrees. The median age at diagnosis for each participant's child was two and a half years with a range from 1 to 7 years. At the time of this study, the median years since receiving an FXS diagnosis was 8.5 years with a range of 2 to 17 years. All of FXS testing were done after 1991 when DNA testing became available for FXS. Nine of the children with FXS were male, and one child was female. Table 2

Interview Results

The results have been divided into three stages: *pre-diagnosis*, *diagnosis*, and *post-diagnosis*. The *pre-diagnosis stage* includes the time period beginning when a parent first had concerns about her child (e.g., developmental delay) and consists of all experiences prior to her child receiving a diagnosis of FXS. The *diagnosis stage* is when the child was given a result of

FXS and includes such experiences as the manner in which the result was delivered and recommendations that followed. The *post-diagnosis stage* captures the experiences of where families received support and information about FXS after obtaining the diagnosis.

Pre-diagnosis—The majority of the mothers reported that the first signs of a possible problem were delayed developmental milestones, such as walking, crawling, and talking. Behavioral issues included hyperactivity, averting eye contact, and hand flapping.

“He was not crawling. He was not sitting up...It took him a while to do the things that...his sister did right on cue...She was right on time with her milestones, whereas, he was anywhere from 6 months to a year behind in everything.”
(Interview 6)

“First he wasn’t following objects and then he had the recurring ear infections, and then when it came time for his speech to develop, his speech wasn’t developing on target.” (Interview 7)

In response to parents expressing concerns, many pediatricians tried to treat the symptoms with medication and/or therapies and did not initially pursue chromosomal and/or fragile X testing. Some of the children were given diagnoses, such as attention deficit hyperactivity disorder (ADHD), Autism, Cerebral Palsy, and Developmentally Delayed prior to the diagnosis of FXS.

“I told him that he was very hyper. The school was complaining, and he was just like, ‘put him on meds.’ He didn’t do any tests. He didn’t mention fragile X.”
(Interview 1)

“They were testing him to maybe find out if he could hear. They were testing to make sure that his vocal cords were okay. Like I said, we went to speech therapy and those type of things, and I don’t think they were actually testing for a diagnosis.” (Interview 4)

Differing reports were given about dysmorphic features. Some mothers reported a large forehead and long ears. However, other mothers said that neither they nor their doctor felt that their child had any of the “typical” FXS physical features.

“But he doesn’t have long ears. He doesn’t have a very long, slender face. Particularly at that time, he had a fat little face.” (Interview 2)

Lack of awareness regarding FXS by both physicians and family members was cited as a barrier to receiving a diagnosis of FXS. Three of the participants revealed that they had never heard of FXS until their child received the diagnosis. Four mothers first heard of FXS from other family members and three individuals were diagnosed by a Geneticist and one by the Pediatrician. The other three mothers first found information about FXS in a variety of ways. One was a Biology teacher who taught an annual lesson on genetics that mentioned FXS. Another participant had a friend mention it to her, and the third heard a health report on the news, which prompted her to mention FXS to her doctor.

“He was 6 years old when we finally got a diagnosis for him and it was only because I saw a show on TV, and it was a medical blip that came on the news every night. The lady was describing what her son did and she talked about flapping the arms and eating a lot and ear infections, and it caught my attention. I said this sounds exactly like what (name of child) does... I mentioned it to the doctor when he went the next time.” (Interview 4)

Five of the ten participants first brought FXS to their doctor’s attention and requested testing. In three of these cases, there was a family history of FXS, and the doctor proceeded

to test without any question. However, the two participants without a family history of FXS reported a hesitance to test on the doctor's part.

"I...mentioned the fragile X. He didn't think he looked like a fragile X child, but I insisted as a matter of fact. We were leaving his office, and I said, 'Here, you didn't do the blood test. Aren't you're supposed to do something?' He reluctantly did the blood test, and when we got the results, he did say that it was fragile X." (Interview 1)

Some participants had a prior diagnosis of FXS in their extended family, but there were issues with communication between family members and misinformation about who was at risk for FXS. In fact, the median diagnosis age only differs by 1 year between those who had a previous diagnosis of FXS in their families and those who did not.

"It (FXS) was in the family but we really only thought that it was like a one man deal. You know we weren't really educated on it." (Interview 7)

A common theme found in all participants was the personality trait of being assertive. Some mothers demanded FXS testing when a doctor was hesitant or visited a developmental pediatrician when the pediatrician wanted to wait. Others advocated for their children to receive services in schools.

"The pediatrician said: 'here's a number for a developmental pediatrician, but I think that you should just wait until he is like 14 months old because sometimes boys are late.'... So at the 1 year visit, he is saying, 'wait until he's 14 months old'. So I go home, and I say, 'no', I am going to call this developmental pediatrician now and see what they think and I'm going to call for the early intervention evaluation now." (Interview 2)

"He made the comment that I was basically an overzealous parent and you know that I shouldn't compare both of my children and just you know the lack of support from him identifying issues that I thought were critical. Even on the referral that he gave me, he wrote on the referral, "per mom's request" so indicating that I'm doing this because of the mother not because I think that anything is wrong. And I still have that. I still have that referral though because I'm glad that I pushed but it was very offensive to me that he would put that." (Interview 10)

Diagnosis—The experience of receiving the diagnosis of FXS varied among the participants. Some of the mothers received the result in the doctor's office and credited their doctor as a great source of support and information. Some were referred to a genetic counselor where they reported positive interactions. Others described a more terse exchange with the doctor, and two of the mothers received the diagnosis over the telephone.

"He did the blood draw and um I will never forget this. I was 9 months pregnant. I was driving home from work, and he called me on my cell phone...and said, 'oh yeah, we got the test results. Um, you know (name of child) does have fragile X and basically that just means that he will be mentally retarded for the rest of his life. If you have any questions, give me a call' and that was pretty much it." (Interview 6)

Upon receiving the diagnosis of FXS, most mothers recounted being on a "huge emotional rollercoaster" consisting of being devastated and relieved that their child had a diagnosis.

"It's like okay; you have cancer, now go home and die. That's how I felt. Where are the answers now? There's no cure. That was a big blow. There was not an injection or vaccine that you could take." (Interview 1) "It was bittersweet. I was excited. I felt like I looked and I searched, and I searched, and I searched, and I

found an answer. You know because sometimes in life, you search and you never find the answer. So, the fact that there was an answer and not only for my son, for me.” (Interview 1)

“I was devastated. I felt like I was grieving for a child that was still living.” (Interview 10)

Post-diagnosis—Once the participants received a diagnosis of FXS for their child, they sought information and support. Frequently mentioned sources of information included the National Fragile X Syndrome Foundation website and conferences, support groups, listserv, and academic centers. Family also played a large role regarding support. Five of the participants cited their family as a positive source of support, and two of these participants had a family history of FXS.

“I guess my biggest support was my sister. You know, she had already been through all of this and so she was my biggest support.” (Interview 3)

“My family was always accepting. You know that was never a problem.” (Interview 4)

The other five participants, with one having a family history of FXS, explained negative reactions from family members. These five mothers told stories of family members in denial, and others felt unsupported when family members would not be tested to see if they carried the premutation form of the FXS gene.

“I mean my family, my in-laws did not want to hear this, did not want to think that anything was wrong with (name of child).” (Interview 2)

“The family that I had that is affected um for whatever reason, she doesn’t give me a lot of information.” (Interview 9)

Sharing the diagnosis of FXS with family members proved to be challenging for some participants. They felt the responsibility to recommend testing for their family members, but it was not always well-received.

“I just felt, wow, I would have appreciated somebody telling me this. And you know, knowing this, I felt like I had a social responsibility, a moral responsibility, to tell my cousins.” (Interview 2)

“I just tried to go through my family...and so my family really refused to be tested. Nobody wanted to be tested.” (Interview 6)

Other participants shared experiences of family members being open to testing.

“When I was tested and found out that I was a carrier and found out that he was affected, it kind of prompted my whole family to get tested... Because I have other cousins and other people who were approaching reproductive age who just needed to know.”

As participants reflected on their diagnosis experiences, they also mentioned what they would change about the process if they were given the opportunity to do so (Interview Item #8). Three of the participants stated that the appropriate time for carrier testing and education regarding FXS should occur during the preconception or prenatal period. Two participants state that they would not change the current testing process, and the other participants’ answers varied from better educating doctors to shortening the amount of time it takes to receive a diagnosis.

“I think I would like pediatricians or OB’s who really deal with the mother before... give us that kind of information. ‘There’s a possibility of this existing and

here's some info.' I think pediatricians and OB's should be more aware of it so that they can tell mothers or prospective mothers this exists." (Interview 1)

Finally, participants were given the opportunity to share anything else that they thought might help other families (Interview Item #9). Four of the mothers shared that parents should be proactive and bold when seeking information. Two participants advised other mothers to seek help or therapy for themselves if they felt they might need it. Additional responses included: "trust your instincts," "love your child," and "don't be ashamed of your child." Some of the responses included a combination of the preceding answers.

"Be persistent. Just because a doctor is a doctor, it does not mean that they know... you are the best authority for your child... I will tell educators, 'you may have the book knowledge, but I know my child better than you or anyone else so you need to listen to me.' And don't be shy, speak up. If something in your gut is telling you that this isn't right. Go with your gut. Don't let anybody intimidate you... You are your child's best advocate." (Interview 6)

Discussion and Implications

This study describes the diagnosis experiences of 10 African American mothers with a child with FXS. Our findings are divided into three stages: *pre-diagnosis*, *diagnosis*, and *post-diagnosis* with common themes (Table 3). In the *pre-diagnosis stage*, we noted a common theme of diagnosis odyssey including delayed diagnosis, lack of FXS awareness among the medical community, and issues with communication among family members. Similar to previous national studies, the median age of diagnosis is approximately 30 months of age (Bailey et al. 2009; 2003). When parents first became concerned, the pediatrician is the immediate point of contact. Despite expressing their worries about their child's development and/or behavior, the pediatricians typically reassured the parents that their child may catch up with their developmental skills or the parents needed to "wait and see" as boys tend to develop at a slower pace or they may provide a symptoms-based treatment approach, and did not attempt to pursue a diagnosis for the cause of their child's developmental delay, learning and behavioral problems.

Indeed, parents who are assertive and proactive continue to raise concerns to the pediatricians and requested testing and/or referral to a specialist (e.g., developmental-behavioral pediatrician, neurology) for further evaluation. Being "persistent" and continually requesting further testing and/or referrals to specialists were essential in receiving a diagnosis of FXS in many cases.

Communication barriers among family members were noted in many participants. Families may not discuss FXS as some participants were not aware that their extended family members have a prior diagnosis of premutation and full mutation FXS. Additionally, when families discussed FXS, there was often misinformation about who was at risk for FXS among family members.

The *diagnosis stage* includes the experiences of receiving the diagnosis from medical professionals, and for many, reaching the end of the diagnosis odyssey. The diagnosis of FXS often completely changes the lives of many families who suddenly find themselves faced with the challenges of understanding the genetics, medical, developmental, and psychiatric issues related to FXS.

Participants also reported that pediatricians typically did not have sufficient knowledge of FXS and provided insensitive information with emphasis on the negative aspects of FXS.

The pediatricians may not be familiar with FXS, and is therefore likely to provide limited information to family. This may lead a family to express the lack of sensitivity and thoughtfulness by the pediatrician. Many mothers found valuable resources and information through the National Fragile X Foundation Web site (www.nfxf.org) with a specific section for newly diagnosed families. Genetic counselors could easily download information from the website for parents, and offer to connect parents to a FXS local support group, if the mother is interested. A list of local support group can be found on the National Fragile X Foundation website. Additionally, genetic counselors should refer the family to a local Fragile X Syndrome Clinic. The genetic counselor can play an important role in helping the family deal with the diagnosis by obtaining a detailed family history, providing a comprehensive genetic counseling and balanced information regarding the medical, psychological, educational, and social potential for the child, mother, and extended family. Furthermore, genetic counselors can be a valuable resource to extended family members who have questions about FXS and/or interested in seeking FXS testing.

The *post-diagnosis* stage captures the experiences of where families received support and information about FXS after obtaining the diagnosis. Once the participants received a diagnosis of FXS for their child, they sought information and support through a variety of sources often independently. The resources from medical professionals may be outdated and/or insufficient. The majority obtained information and resources through the National Fragile X Syndrome Foundation website. Others joined their local FXS support groups and listserv, and scheduled an appointment at a FXS clinics to receive specialized evaluation and recommendations.

Possibly overwhelmed by an unexpected diagnosis, parents may struggle to understand what the future holds for their family, while also being expected to coordinate the necessary services for their child. As a result, they may lack the time and effective method to inform other family members that their child has received the diagnosis of FXS and potential implications to other family members. The National Fragile X Foundation Web site (http://www.nfxf.org/html/new_family.htm) has a sample letter to help with informing and explaining FXS and its inheritance to family members. It is important for families to communicate as they can play an important role in providing support.

Interestingly, our participants reported the barrier of communication issues among family members occurring in the *pre-diagnosis* and *post-diagnosis* stages. These communication issues are two-fold: 1) families may not share the diagnosis of FXS with immediate and extended family members, and 2) families who share the diagnosis may not have sufficient knowledge to fully explain every aspect of FXS. They may also encounter unwillingness of family members to accept the diagnosis and to obtain testing. Furthermore, emotions of defensiveness, denial, anger, and disinterest may emerge. Since FXS is an inherited condition, it is important that families inform each other about the diagnosis as other family members may need to be tested or be informed of their risk for future medical and/or reproductive issues as it relates to the fragile X mutation

Study Limitations

Although our sample size is small and may not be generalized to all African American families with FXS, this is the first study to examine the FXS diagnosis experiences in African American families. All participants were recruited through the Emory Fragile X Center studies or the Fragile X Clinic at Emory University. However, their state residence, socioeconomic, and educational level were diverse (Table 2). We were unable to compare the demographics of those who participated and those who did not due to lack of consent from the latter group. It is possible that the experiences of mothers who participated differ

from those mothers who are unlikely to participate in research and/or attend a fragile X specialty clinic. Thus, our sample may be biased as it represents a group of parents who are active in the fragile X community and proactive in seeking specialty clinic care for their child. Nonetheless, our results are similar to reported experiences of families in previous national studies who were predominately reporting as Caucasian (Bailey et al. 2009; 2003). Importantly, within our sample, all encountered similar barriers to the diagnosis and communication of information within the family, in spite of their high level of education (all but one had at least some college or post high-school education). Again, this is similar to that reported in previous FXS studies and other developmental disabilities disorders (Bailey et al. 2009; 2003).

Practice Implications

The diagnosis experiences of African Americans are relatively similar to the studies by Bailey et al. which included predominantly Caucasian families (Bailey et al. 2009; 2003). The results of this study, combined with the previous studies on family experiences and perceptions in FXS, reinforce that FXS may take a long time to recognize and diagnose in all ethnic groups. As such, we have a long way to go to ensure that all children receive the diagnosis of FXS earlier, regardless of race or ethnicity. Similar to Bailey et al. studies, we identified the presence of a communication gap in families once the diagnosis of FXS is known. This lack of communication or inaccurate information may exacerbate the diagnostic dilemma, sometimes even limiting effective cascade testing that is so important in an inherited disorder. We recognize that this is a challenging and complicated process. Health care professionals can serve as advocates by connecting families to resources or providing those resources that are easy to navigate and sensitive to their needs.

Research Recommendations

This is the first study to examine the overall experiences of African American families with FXS through three stages: *pre-diagnosis*, *diagnosis*, and *post-diagnosis*. Additional studies among families from ethnically and culturally diverse backgrounds are important to further understand the experiences of FXS families. Furthermore, the father's perspective should be studied in a similar study design to describe the similarities and differences in their perceptions compared to mothers. Future studies should also evaluate physicians' reflections and correlate these findings with the perceptions of the patients. This may help bridge the gap in the communication barriers that often exist between physicians and parents when seeking a diagnosis for the child's developmental delay.

Our participants indicated the lack of communication, support, and potential denial among family members. These families need an immediately accessible link that is easy to navigate and culturally-sensitive to their needs. Future studies should focus on designing innovative tools to enhance the process of connecting family members together once they receive the FXS diagnosis.

Acknowledgments

We would like to thank the parents and children for their participation in this study. Financial support for this study was provided by the D. J. Shockley Foundation, Holly Lane Foundation and the Fragile X Association of Georgia.

Grant sponsors: NIH/NICHD; Grant number: 1K23HD058043-01A1 (JV).

Grant sponsor: NIH/NHCHD; Grant number: R01 HD29909 (SS).

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Table 1**Semi-structured Interview Guide**

Interview Questions	Probing Questions
1. Tell us about your diagnosis experience starting from when you first recognized any problems to when you got the diagnosis of fragile X syndrome for your child.	<ul style="list-style-type: none"> • What were some of the first symptoms that you noticed? • What doctors did you see? • What doctor gave you the diagnosis of FXS?
2. What part of the whole process of receiving a diagnosis did you think went the smoothest?	
3. What were the barriers in the process?	
4. What part of the process was the most challenging?	
5. How did you feel when your child was given a diagnosis of fragile X syndrome?	
6. Tell us about any support you received when you first got the diagnosis?	<ul style="list-style-type: none"> • What role did you family play in the support you received? • What role did your friends play in the support you received? • Did you receive support from other types of organized groups?
7. Describe any experience you may have gone through to get information about fragile X syndrome.	
8. If you could change anything about the process of getting a diagnosis of fragile X syndrome, what would you change?	
9. Is there anything else you would like to tell us about your experience that might help us assist other families in the future?	

Table 2

Participant Descriptions

	Child's Age at Time of FXS Diagnosis	Doctor Who Gave the FXS Diagnosis	Insurance at Time of FXS Diagnosis	Mother's Highest Level of Education	Annual Household Income	Years of Diagnosis (Years Since Diagnosis at Time of Interview)	Known Family History of FXS Prior to Diagnosis
Interview 1	5	Neurologist	Medicaid	Bachelor's Degree	\$21,000–50,000	1997 (11)	No
Interview 2	1	Pediatrician	Private	Bachelor's Degree	\$51,000–100,000	1999 (10)	No
Interview 3	7	Geneticist	Private	Some Graduate Classes	>\$100,000	2004 (5)	Yes
Interview 4	6	Geneticist	Private	Advanced Degree	\$50,000–100,000	1992 (17)	No
Interview 5	3	Geneticist	Medicaid	High School Degree	\$21,000–50,000	2005 (4)	Yes
Interview 6	3	Neurologist	Private	Some Graduate Classes	>100,000	1998 (11)	No
Interview 7	1	Geneticist	Private	Bachelor's Degree	\$51,000–100,000	2001 (8)	Yes
Interview 8	2	Pediatrician	Private	Bachelor's Degree	\$51,000–100,000	2000 (9)	No
Interview 9	2	Pediatrician	Medicaid	Some College	\$21,000–50,000	2006 (4)	Yes
Interview 10	1	Neurologist	Private	Advanced Degree	\$51,000–100,000	2008 (2)	No

Table 3**Common Themes in Pre-diagnosis, Diagnosis, and Post-diagnosis Stages**

Stages	Common Themes
Pre-diagnosis	<ul style="list-style-type: none"> • First signs of concern included delayed developmental milestones (e.g., sitting, crawling, walking, and talking), and behavioral issues (e.g., hyperactivity, averting eye contact, and hand flapping). • Many doctors did not initially pursue chromosomal and/or fragile X testing. Instead, they treated symptoms with medications or intervention therapies. • Some participants reported their child lacking the “typical” dysmorphic features associated with FXS. • A common reported barrier to receiving a diagnosis of FXS was lack of FXS awareness by both physicians and family members. • Half of the participants had to bring FXS to the attention of the physician. • Participants who had a prior family history of FXS reported issues of miscommunication and misinformation about FXS among family members. • All participants demonstrated the personality trait of being assertive and proactive about finding the cause for their child’s delays.
Diagnosis	<ul style="list-style-type: none"> • The experience of receiving a FXS diagnosis varied among participants. Some described their physicians as supportive and informative while others described the interaction as terse and insensitive. • A common response to the diagnosis of FXS was both devastation and relief in discovering the diagnosis.
Post-diagnosis	<ul style="list-style-type: none"> • Sources of support included family, the internet, support groups, and the National Fragile X Foundation. • Communication gap included challenges in sharing the diagnosis of FXS with other family members.