Journey of Fathers Following their Child’s Fragile X Diagnosis: Support Seeking Behaviors of Fathers

Master’s Thesis

Presented to

The Faculty of the Graduate School of Arts and Sciences
Brandeis University
Graduate Program in Genetic Counseling
Sharyn Lincoln, MS, CGC, Advisor

In Partial Fulfillment
of the Requirements for the Degree

Master of Science
in
Genetic Counseling

by
Aman Kaur Mann

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ABSTRACT

Journey of Fathers Following their Child’s Fragile X Diagnosis:
Support Seeking Behaviors of Fathers

A thesis presented to the Graduate Program in Genetic Counseling

Graduate School of Arts and Sciences
Brandeis University
Waltham, Massachusetts

By Aman Kaur Mann

Studies show that fathers of children with a disability or chronic illness experience significant psychological impact. Appropriate support has shown to improve their well-being and that of their child. Despite these studies, the needs of these fathers are continually overlooked in healthcare settings. The purpose of this study was to understand the father’s experience receiving a fragile X syndrome (FXS) diagnosis for their child, focusing on their support seeking behaviors and their satisfaction with the supports available. We used an anonymous online survey and received 32 responses. The majority of fathers were satisfied with their experience with medical professionals. However, 15.7% of fathers were dissatisfied with their experience. We found that fathers want (1) more access to FXS information, specialists and local resources, and (2) specialized support from family, friends, other FXS families and FXS-related organizations. Healthcare providers can meet the needs of the fathers by (1) increasing awareness and education of FXS and FXS-related resources, (2) assessing both parents level of understanding, addressing their questions and concerns, and providing psychosocial support, and (3) acknowledging the
fathers role, discussing the impact on them when receiving a diagnosis for their child and providing referrals to appropriate support resources. Support organizations can also meet fathers’ needs by (1) creating literature tailored to fathers, (2) encouraging more father-related activities in support groups, and (3) creating a network to connect fathers to other fathers. With the support of this data, we recommend implementation of these suggestions in order to address and meet the individual needs of fathers of children diagnosed with FXS, and support them through their unique journey as fathers.

Keywords: Fragile X syndrome; paternal needs; paternal support-seeking behavior; paternal involvement; paternal and/or family support; genetic counseling
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INTRODUCTION

The needs of fathers who have a child with a disability or chronic illness have been historically overlooked in healthcare settings, despite studies showing that these fathers experience significant psychological impact, similar to mothers (Shin et al., 2006; Cummings, 1976). Providing fathers with the appropriate support is not only important for their well-being but also for their children as it has been shown to improve child outcomes (Alio et al., 2009; Anding et al., 2016).

Fathers who receive any clinical diagnosis for their child show changes in their levels of pessimism, depression, coping styles and anxiety (Hartley et al., 2012; Cummings, 1976; Oers et al., 2014). Without taking appropriate steps, such as seeking support and creating appropriate coping mechanisms, emotional, physical and cognitive burnout is possible (Lindström et al., 2010). A study published in 2010 found that significantly more parents of children with chronic diseases scored higher for clinical burnout (persistent stress and psychological reactions) on the Shirom-Melamed Burnout Questionnaire compared with parents of healthy children (Lindström et al., 2010). Another study found that fathers of children with learning disabilities have higher avoidant attachment, indicating they are emotionally unavailable or unresponsive to the child, and have lower use of active coping strategies compared to mothers of children with learning disabilities and fathers of children with no disability (Al-Yagon, 2005). These changes are associated with paternal age, child’s behavioral problems, risk of having additional children with disability, maternal depressive symptoms and/or perception of the maternal experience, economic status and satisfaction with social support network (Hartley et al., 2012; Shin et al.,
Studies have shown that fathers’ initial needs after their child receives a diagnosis were related to resources, type of information desired, parenting skills, sources of support and time flexibility (Rosebrough, 2011). Support in particular was a recurring theme. Fathers lacked actual support and information regarding support. They wanted more access to other parents of children with similar diagnoses, support geared towards males, and support from family and friends (Rosebrough, 2011). Providing fathers with family-centered resources has shown to promote effective coping mechanisms that can increase the level of involvement in their child’s life, leading to better outcomes for both parent and child (Broger & Zeni, 2009; Anding et al., 2016). A literature review published by Hornby found that the satisfaction fathers received from social support is just as important, if not more so, than the existence of the support system itself (Hornby, 1995).

Previous studies have found that fathers of chronically ill children used work habits, avoidance and emotion-, problem solving- and active-coping styles with religious dimensions as ways to cope with the diagnosis and receive support (Broger & Zeni, 2009; Hovey, 2005; Allen, 2005). Primarily, fathers’ main source of support and information is their spouse (Lewis, 1988; Rosebrough, 2011). Overall, the support seeking behaviors of fathers is multifaceted and influenced by complex social ideals, such as the perception of medical providers, partners, gender identity and stereotypes (Rosebrough, 2011; Allen, 2005).

Although these studies discussed types of support and coping mechanisms the fathers used and the importance of the fathers’ satisfaction from their supports, they did not examine the factors that contributed to the fathers’ satisfaction or dissatisfaction. In addition, studies have assessed the impact of a child’s diagnosis on the father, but fathers continue to be neglected and
overlooked by service providers. In a study conducted in 2011, Chinese fathers of children with disabilities experienced no supportive communications from providers, missed critical time for disability management and were excluded from medical decision making when interacting with health care professionals (Huang et al., 2011). Another study conducted in England and Wales had similar findings, in which only a third of fathers of children with disabilities surveyed felt included in sessions with health care practitioners (Carpenter & Towers, 2008).

The purpose of this study was to understand fathers’ experience with receiving a fragile X syndrome (FXS) diagnosis for their child, focusing on their support seeking behaviors and their satisfaction with the supports available. FXS is a rare genetic condition that may cause intellectual disability, and learning and behavioral challenges in both genders (FRAXA, 2017; NFXF, 2017). However, males are more frequently affected and with greater severity (FRAXA, 2017; NFXF, 2017). The FXS population was chosen because there are intellectual disabilities and behavioral issues associated with the condition, and, as noted above, these issues appear to be more significant when considering how fathers cope with their child’s diagnosis. There are also large FXS-related organizations and support networks available, allowing us to better assess fathers’ knowledge, access, use and satisfaction with these support resources. We examined (1) the father’s experience during the diagnostic odyssey, including their satisfaction with the healthcare providers, and (2) factors that influenced the father’s decision to seek out support and their satisfaction with the support they received. Understanding the experience of these fathers can provide healthcare providers and organizations insight on how to improve their services in order to better meet the needs of this overlooked population.
METHODS

This study was reviewed and approved by the Brandeis University Institutional Review Board.

Sample and recruitment

Fathers of at least one child diagnosed with FXS within the past 10 years (≥ 2006) were invited to participate in a confidential and anonymous online survey. We recruited participants through the National Fragile X Foundation (NFXF), the FRAXA Research Foundation, the “I love someone with Fragile X Syndrome” Facebook page and the “Fragile X Syndrome” Facebook page. A reminder recruitment notice was sent four weeks after the initial posting date. We offered all participants who completed the survey a chance to enter a draw for one of three $50 Amazon.com gift cards.

Data Collection

We designed an anonymous survey using Qualtrics®, an online survey tool. The survey had 47 questions: questions 1-6 was the father’s demographics, 7-11 was the first affected child’s demographics, 12-29 was regarding the father’s interaction and satisfaction with healthcare providers, 30-42 was regarding the father’s experience after receiving the FXS diagnosis for their child, and 43-47 was regarding the fathers’ thoughts on the overall journey and their suggestions for providers on how to better meet their needs. The survey was open for approximately six weeks. Data was collected using multiple-choice questions in the form of single or multiple selections, Likert scale questions, and open-ended questions. Participants were also asked what they thought healthcare providers and organizations can change to better meet their needs.
The survey was voluntary and anonymous. Participants had the option to not answer any of the survey questions and to exit the survey at any time. All raw and analyzed data was securely stored, and only the principal investigator and the student researcher had access to these documents and materials.

**Data Analysis**

Descriptive statistics on quantitative data was performed using SPSS, a statistical software package. Comparisons between variables were made using univariate and bivariate statistical analysis. Microsoft Excel was used to manually analyze open-ended questions to identify themes.
RESULTS

**Demographics of fathers**

There were 46 respondents. Incomplete responses and those that did not meet inclusion criteria were removed, and data from 32 participants were used. All the fathers were married with a majority of them being 31 to 40 years of age (58.0%). The majority of fathers had two or more biological children (84.4%, N=27). Of these, 78.1% had one child diagnosed with FXS and 21.9% had two children diagnosed with FXS. This study focused on the experience of their first child’s diagnosis with FXS. Table 1 contains a summary of study participants’ demographic data.

**Table 1. Demographic information of the fathers**

<table>
<thead>
<tr>
<th>Frequency</th>
<th>Percent (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Father’s age at first child's FXS diagnosis (Years) (N=31)</td>
<td></td>
</tr>
<tr>
<td>≤ 20</td>
<td>2</td>
</tr>
<tr>
<td>26-30</td>
<td>4</td>
</tr>
<tr>
<td>31-35</td>
<td>9</td>
</tr>
<tr>
<td>36-40</td>
<td>9</td>
</tr>
<tr>
<td>41-45</td>
<td>7</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Marital status (N=32)</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Married</td>
<td>32</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Number of biological children (N=32)</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>5</td>
</tr>
<tr>
<td>2</td>
<td>19</td>
</tr>
<tr>
<td>3</td>
<td>4</td>
</tr>
<tr>
<td>4</td>
<td>3</td>
</tr>
<tr>
<td>5</td>
<td>1</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Number of children diagnosed with FXS (N=32)</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>25</td>
</tr>
<tr>
<td>2</td>
<td>7</td>
</tr>
</tbody>
</table>

**Demographics of first child diagnosed with FXS**
81.3% of fathers had a male child diagnosed with FXS and 18.8% had a female child diagnosed with FXS. The average age at FXS diagnosis was 2.9 years, with the diagnostic odyssey beginning on average at 2.2 years and lasting approximately 1.0 years. Table 2 contains a summary of the study participants’ child’s demographic data.

**Table 2. Demographic information of the first child diagnosed with FXS**

<table>
<thead>
<tr>
<th></th>
<th>Frequency</th>
<th>Percent (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Sex (N=32)</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>26</td>
<td>81.3</td>
</tr>
<tr>
<td>Female</td>
<td>6</td>
<td>18.8</td>
</tr>
<tr>
<td><strong>Age at diagnosis (Years) (N=32)</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Minimum</td>
<td>0</td>
<td></td>
</tr>
<tr>
<td>Maximum</td>
<td>7</td>
<td></td>
</tr>
<tr>
<td>Average</td>
<td>2.94</td>
<td></td>
</tr>
<tr>
<td><strong>Age when diagnostic odyssey began (Years) (N=32)</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Minimum</td>
<td>0</td>
<td></td>
</tr>
<tr>
<td>Maximum</td>
<td>6</td>
<td></td>
</tr>
<tr>
<td>Average</td>
<td>2.19</td>
<td></td>
</tr>
<tr>
<td><strong>Length of diagnostic odyssey (Years) (N=32)</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Minimum</td>
<td>0</td>
<td></td>
</tr>
<tr>
<td>Maximum</td>
<td>6.5</td>
<td></td>
</tr>
<tr>
<td>Average</td>
<td>1.03</td>
<td></td>
</tr>
</tbody>
</table>

Fathers were asked the clinical symptoms of FXS their children expressed, and could choose more than one. Fathers reported their child having mild to moderate severity of developmental delay (75.1%), intellectual disability (68.8%), hyperactivity (62.5%), hand flapping (68.8%), temper tantrums (71.9%), autism (56.2%), lack of impulse control (75.0%), hyperarousal or sensory issues (59.4%) and sleep problems (68.8%). Hand biting and seizures were generally reported to be not applicable (53.1% and 75.0%).

**Fathers’ satisfaction with medical professionals**

43.8% of respondents reported attending 100% of their child’s medical appointments, whereas 56.2% reported attending 90% or less. Of those who did not attend 100% of their child’s
medical appointments, the majority stated work as their primary reason (88.9%). Other reasons included care for other children (5.6%) and location of the appointment (11.1%).

Fathers initially met with a developmental pediatrician (31.2%), geneticist (25.0%), genetic counselor (21.9%), neurologist (18.8%) or psychologist (3.1%) to discuss the FXS diagnosis for their child. Fathers were asked to rate their level of agreement with statements addressing different aspects of a clinic visit, such as decision-making, information giving and referrals to resources.

Fathers generally agreed that they were included in the decision to do molecular genetic testing on their child (71.9%). 21.9% neither agreed nor disagreed and 6.2% disagreed that they were included. Of those who disagreed, one stated FXS was never explained to him and neither was the testing. Another stated he advocated for the test to be done.

Fathers generally agreed that their questions and concerns were addressed after receiving the diagnosis (65.7%). However, 15.7% disagreed with this statement. These fathers stated the medical professional was not knowledgeable about FXS and, therefore, did not provide useful information about prognosis and next steps. Some fathers found detailed information from “downstream specialists and families” or from doing their own research online.

The majority of the fathers did not feel that the medical professional addressed their needs as a father after receiving the diagnosis (53.1%). 34.4% were neutral on this point and 12.5% agreed their needs were addressed. Of those that felt their needs were not addressed, 73.3% stated that their needs were never assessed or addressed and 13.3% felt their needs were dismissed or ignored. Three fathers in particular described how their needs were dismissed or ignored.

“Everything that was discussed through the diagnosis they communicated to my wife. If I said anything it was quickly dismissed or redirected or given a short answer. We assumed that it was
because my wife was the carrier. But still was shocked that opinions an[d] questions I had ever not as important to them.”

“I’ve rarely felt like any medical professional have so much considered my needs as a father. I’d say a more fitting question would be, “The ____ acknowledged my presence as a father...” That feeling started the day my first son was born, when I wasn’t able to sign the birth certificate without my wife’s written permission.”

“They kept looking at and speaking to my wife. Hey I am the parent too. I felt like the only time they did speak to me it was patronizing like I am not involved with or take any care of my child”

One father pointed out that little father-centric information on FXS exists.

Fathers generally agreed that the medical professional provided them with information on FXS diagnosis (68.8%) and information they needed to care for their child (40.7%). However, 12.5% and 31.3%, respectively, did not feel information was provided, and 18.8% and 28.1%, respectively, were neutral on this point. Of those that did not feel information was provided, 75.0% stated that the medical professional left the information-giving to the counselor or subsequent medical professionals and 50.0% were told to research online. 89.9% explained that the medical professional was just not knowledgeable about FXS.

The majority of fathers disagreed with the statement that they were provided information on family centered or male-focused support groups (65.7%). 15.6% were neutral on this point and 18.8% agreed they were provided this information. Of those that disagreed, 89.5% stated support was just not offered, 21.0% stated the medical professional had limited knowledge, and 21.0% thought a subsequent medical professional may offer it. One father explained,

“It was not discussed and I was too “shell-shocked” with the diagnosis to think about the impact on myself.”

Approximately half of the fathers also disagreed with the statement that they were provided referrals to support organizations such as the National Fragile X Foundation (NFXF) (50.1%). 15.6% neither agreed nor disagreed and 34.4% agreed they were provided these
referrals. Of those that disagreed, 78.6% stated this information was just not provided, 14.3% stated that subsequent medical professionals provided this information, and 14.3% were told to research online.

The majority of fathers disagreed with the statement that they were connected with another family with a FXS diagnosis (84.4%). 15.6% neither agreed nor disagreed. Of those that disagreed, 100% stated this information was just not provided, 24.0% indicated the medical professional was not aware of another FXS family, 12.0% indicated a subsequent medical professional may provide this information, and 16.0% were told to research online. One father pointed out that,

“...privacy requests from other FX[S] parents, or privacy policies in general, seem to hinder medical professionals ability to make such connections.”

Overall, half of the fathers were satisfied with their experience with the medical professional because the medical professional was knowledgeable and provided the information they needed clearly (75.0%), they received a diagnosis (25.0%), the medical professional was caring (18.8%), the medical professional provided referrals and resources (12.5%), and the medical professional provided good counseling (6.2%). For some fathers the end to the diagnostic odyssey is what provided them with the most satisfaction:

“We went through a lot of other medical searching for what was wrong with my kid, but she was the first [to] figure it out.”

Nearly 16.0% of fathers were not satisfied with their experience with the medical professionals. Their reasons included the medical professional not being knowledgeable about FXS (80.0%) and the medical professional not providing any referrals (i.e. NFXF or other families) (80.0%). One father stated that the medical professional,

“...had absolutely no knowledge of FXS nor resources to support our family.”

10
Another father explained,

“Very good from a medical perspective. Not so good in terms of how to help my son or how to cope with diagnosis.”

Fathers’ satisfaction with Genetic Counselors

Twenty-nine fathers met with a genetic counselor at some point during their diagnostic odyssey. Of these, twenty-eight provided details regarding their experience. Themes included the session being positive, informative, clear, overall helpful, and professional. Information they found helpful was regarding the genetics of FXS, the clinical picture including issues that may arise later in life, and implications for other children and extended family. One father described his experience as,

“Emotionally rough, but ultimately helpful and enlightening.”

However, some of the fathers described the genetic counseling experience as redundant, overly optimistic or pessimistic about the diagnosis or uninformative. Some felt the genetic counselor had limited knowledge. Others felt there was a lack of counseling or information overload. One father found the genetic counselor,

“Didn’t present any new information we hadn’t read about on the [I]nternet.”

Another father found that,

“One was very direct stating that our son would need to be reside with us for his entire life with little independence, while the other was overly empathic to the point of appearing insincere.”

Fathers’ emotional response to the diagnosis

Fathers were asked if they experienced any or all of the following after receiving a FXS diagnosis for their child: depression, anger, anxiety, stress, relief or other, and they could choose more than one. This data is summarized in Figure 1.
Figure 1. Fathers’ emotional experience after receiving a FXS diagnosis for their child

<table>
<thead>
<tr>
<th>Emotional response</th>
<th>Percentage of fathers (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Stress</td>
<td>80.0</td>
</tr>
<tr>
<td>Anxiety</td>
<td>56.7</td>
</tr>
<tr>
<td>Depression</td>
<td>53.3</td>
</tr>
<tr>
<td>Anger</td>
<td>40.0</td>
</tr>
<tr>
<td>Relief</td>
<td>33.3</td>
</tr>
<tr>
<td>Grief and Despair</td>
<td>7.7</td>
</tr>
<tr>
<td>Sadness</td>
<td>7.7</td>
</tr>
<tr>
<td>Hopelessness</td>
<td>3.3</td>
</tr>
<tr>
<td>Fear for the future</td>
<td>3.3</td>
</tr>
<tr>
<td>None of the above</td>
<td>7.7</td>
</tr>
</tbody>
</table>

Fathers’ experience seeking support

Fathers were asked whether they sought out support after receiving a FXS diagnosis for their child. Of the thirty respondents, 53.3% said no and 46.7% said yes. Of those that did not seek out support, 53.8% said they did not feel they needed it and/or were focused on educating themselves and finding resources to support their partner and child. Other reasons for not seeking out support included previous exposure to individuals with disability (7.7%) and that it was never offered (15.4%). One father stated,

“...because it wasn’t really given that they cared. [M]ore focused on my wife as she was carrier. Felt was my position to stay strong and support her and my daughter.”

Another father stated,

“I will be fine.”

And a third father questioned,

“What kind of support do you ask for?”
Of the fourteen fathers that said they did seek out support, the majority received it from their partner (71.4%), FXS-related organizations (64.3%), and FXS support groups (50.0%). Other common sources were family members (42.9%), internet resources (42.9%), and other FXS families (28.6%). Only two (14.3%) sought support from religious/spiritual communities. Other sources of support included licensed counselors, family doctors and autism spectrum disorder support groups.

The majority of the fathers were either somewhat or extremely satisfied with the support they received from their partner (N=10). Many fathers were also satisfied with FXS related organizations (N=9), internet resources (N=6), FXS support groups (N=7), and other FXS families (N=4). One of the six fathers that sought out support from their family members was somewhat dissatisfied. He explains they,

“...took more of a defensive stance when we first got the diagnosis, e.g. “he’ll be fine...just let te kid be a kid...” It was only until symptoms started to become undeniable at about age 4 that others began to be more silent about the diagnosis. Family does not proactively assist.”

Of the two fathers that sought out support from their religious or spiritual community, one was extremely dissatisfied as the support they desired was not offered or provided, and if so, it was not adequate. Of the five fathers that sought out support from other sources that weren’t listed, one was somewhat dissatisfied due to the inability to find an appropriate counselor. The father explained he

“Requested a referral for counseling to help emotionally deal with the diagnosis but they weren’t able to find a suitable counselor to refer me to, so nothing was provided.”

Factors influencing fathers support seeking behavior

Using a chi-squared test for independence, we aimed to determine whether there was a significant association between various factors and the fathers’ decision to seek out support. No association was found between the father’s age at diagnosis (N=29, p=0.370), medical
appointment attendance (N=30, p=0.543), or number of children affected by FXS (N=30, p=0.134) and whether or not the father sought out support. No association was found between the severity of the child’s clinical features (N=30) and whether or not the father sought out support: developmental delay (p=0.897), intellectual disability (p=0.224), hyperactivity (p=0.953), hand flapping (p=0.222), hand biting (p=0.131), temper tantrums (p=0.845), autism (p=0.985), lack of impulse control (p=0.555), hyperarousal or sensory issues (p=0.932) and seizures (p=0.462). Figure 2 displays the association between the severity of each clinical feature and whether or not the father sought out support. No association was found between the fathers’ emotional response and their decision to seek out support or not, except for those that experienced relief. There was a significant negative association between relief and support seeking behavior (N=30, p=0.038) where fathers who experienced relief did not seek out support. Figure 3 displays the association between the fathers’ emotional response and whether or not the father sought out support.

Using independent samples t-Test, we analyzed whether the fathers’ satisfaction with their health care provider while receiving their child’s FXS diagnosis was associated with their decision to seek out support. The only association found was the fathers’ agreement with the statement that the medical professional provided referrals to support organization such as NFXF. There was a significant association between providing referrals to support organizations and support seeking behavior (N=30, p=0.026). Figure 4 displays the association between the fathers’ satisfaction with the medical professional and whether or not the father sought out support.
Figure 2. Association between severity of the child's clinical features and whether the father sought support (N=30)

*Severity of child’s clinical feature ranges from mild to severe; 1=Mild, 2=Moderate, 3=Severe, 4=N/A.*
Figure 3. Association between fathers’ emotional response to diagnosis and whether the father sought support (N=30)

***Significant at p<0.05.
**Figure 4. Association between the fathers’ level of agreement with statements assessing interaction with the medical professional and whether or not the father sought out support (N=30)**

<table>
<thead>
<tr>
<th>Statement</th>
<th>No</th>
<th>Yes</th>
<th>Average Level of Agreement</th>
</tr>
</thead>
<tbody>
<tr>
<td>I am overall satisfied with my experience with the medical professional.</td>
<td>1.50</td>
<td>3.31</td>
<td>3.50</td>
</tr>
<tr>
<td>The medical professional connected me with another family with FXS diagnosis.</td>
<td>1.44</td>
<td>2.25</td>
<td>*</td>
</tr>
<tr>
<td>The medical professional provided referrals to support organizations such as NFXF.</td>
<td>2.13</td>
<td>2.88</td>
<td>*</td>
</tr>
<tr>
<td>The medical professional provided me with information on family centered or male-focused support groups.</td>
<td>2.43</td>
<td>3.75</td>
<td>3.79</td>
</tr>
<tr>
<td>Received the information I needed to care for my child.</td>
<td>1.50</td>
<td>2.75</td>
<td>*</td>
</tr>
<tr>
<td>The medical professional provided me with information on FXS.</td>
<td>2.13</td>
<td>3.75</td>
<td>3.79</td>
</tr>
<tr>
<td>The medical professional addressed my needs as a father after receiving the diagnosis.</td>
<td>2.07</td>
<td>3.81</td>
<td>3.79</td>
</tr>
<tr>
<td>The medical professional addressed my questions and concerns after receiving the diagnosis.</td>
<td>2.43</td>
<td>3.81</td>
<td>3.79</td>
</tr>
<tr>
<td>I was included in the decision to do molecular genetic testing on my child for FXS.</td>
<td>2.25</td>
<td>4.19</td>
<td>4.29</td>
</tr>
</tbody>
</table>

* Level of agreement with statements assessing interaction with medical professional ranges from strongly disagree to strongly agree; 1=Strongly disagree, 2=Somewhat disagree, 3=Neither agree nor disagree, 4=Somewhat agree, 5=Strongly agree.

**Experience of fathers with multiple children diagnosed with FXS**

The seven fathers who had two children diagnosed with FXS were asked whether the experience receiving the diagnosis and coping with the second diagnosis was any different. Four fathers said their experience was different. 50.0% stated this was due to the fact that it was a
bigger shock the second time. 50.0% also stated the second child had a different phenotype and 25.0% indicated they experienced greater depression and anxiety. One father further explained, “I suspect it produced greater depression and anxiety knowing that both of my children had this bad luck in the genetic lottery of their lives.”

Three fathers said their experience was not different because they all had already suspected it and thus were not surprised.

*Fathers’ final comments on how to better meet their needs*

Fathers were asked what they found most helpful when they were going through the process of receiving a FXS diagnosis for their child. Of the twenty-seven fathers that responded, 37.0% stated access to information on FXS (i.e. what it means, what to expect, research, etc.), 18.5% stated support from their partner and family and 18.5% stated support from FXS-related organizations were the most helpful. Fathers also found access to FXS specialists (11.1%), support from other FXS families (14.8%), praying (7.4%), previous exposure to individuals with disabilities (7.4%), access to local resources (3.7%) and having an answer (7.4%) helpful.

Fathers were also asked how they think health care providers and support organization can better meet their needs after receiving a diagnosis. Of the twenty-nine that responded, many stated increasing awareness and education of FXS for providers (41.4%) and increasing awareness of resources to organizations like NFXF and support groups (20.7%) would help to better meet their needs. Other suggestions included mandating or recommending therapy (13.8%), providing appropriate referrals and access to health care and service providers (17.2%), recognizing and discussing the impact on parents (10.3%), acknowledging the presence of fathers (10.3%), creating more literature for fathers (6.9%), creating inclusive support groups (6.9%) and connecting them to other fathers (6.9%).
DISCUSSION

The study aimed to understand the father’s experience with receiving a FXS diagnosis for their child, focusing on their support seeking behaviors and their satisfaction with the support available. By understanding the father’s experience and satisfaction with the healthcare providers, and by examining factors that influenced their decision to seek out support, our hope is that healthcare providers and organizations can develop ways to better meet the needs of fathers.

The majority of the fathers that participated in the study were young, <40 years of age (77.4%), and they were all married. This was unexpected given that there is a significantly higher risk of divorce in small families with a child with disabilities (Namkung et al., 2015). Most had one child diagnosed with FXS (78.3%), but there were a few that had more than one child diagnosed (21.9%). The prevalence of FXS is higher in males than females, consistent with the findings of this study. The average age of diagnosis, 33 months, in this study is slightly lower than the reported age at diagnosis, 35-37 months for males and 42 months for females (Bailey et al., 2009). The average age at which the process to diagnose a child began is 2.2 years, or 26 months, which is roughly consistent with the general population: 20 months for males and 26 months for females (Bailey et al., 2009). Finally, the average length of the diagnostic odyssey, from the first clinic visit to the time of diagnosis, is 12 months, which is slightly lower than the 16 months quoted in the Bailey et al. paper (2009). The features of FXS reported by fathers include developmental delays and intellectual disability, two of the hallmark features of FXS. Other common features reported included hyperactivity, hand flapping, temper tantrums, and
lack of impulse control, all of which are commonly reported in the FXS population. 25% of fathers reported their children have seizures, which is relatively consistent with the reported frequency of seizures in the FXS population.

In general, fathers were satisfied with their experience with the medical professional they met with regarding their child’s FXS diagnosis. The fathers felt this was because the medical professional was knowledgeable and provided them with the information they needed. A common reason for their satisfaction was the fact that they finally received a diagnosis for their child. Bailey et al. found that more than one third of families report greater than 10 visits with medical professionals before receiving a FXS diagnosis for their child (Bailey et al., 2003). This was suggested at by one father who commented that they had seen a number of providers before a diagnosis was made. Several fathers reported feeling relief after receiving the FXS diagnosis for their child, which may also relate to the satisfaction of finally receiving a diagnosis. Several fathers also pointed out that not only receiving a diagnosis was important, so was the way it was delivered. Although they were generally satisfied with their experience, they felt that the results disclosure was handled poorly. Some found the healthcare provider to be overly optimistic or empathic to the point of appearing insincere, whereas others found them to be overly pessimistic, depressing and direct. There are different ways to break the news to families regarding a genetic diagnosis, and balancing the hopes and desires of the parents, while also providing realistic expectations, is difficult. Healthcare providers have to be cautious when delivering bad news, keeping in mind the impact of both overly pessimistic and overly optimistic information.

Two major themes appeared when the journeys of the fathers were examined: (1) fathers want access to FXS information, specialists and local resources, and (2) fathers want support from family, friends, other FXS families and FXS-related organizations.
Access to FXS information, specialists and local resources

One of the main sources of dissatisfaction for fathers was associated with the lack of FXS information, specialists and local resources. Studies have shown that fathers of children with chronic illnesses often take on the role of advocating for their child’s medical needs, and their immediate concerns are usually regarding the child’s long-term future (Yogman & Garfield, 2016; Carpenter & Towers, 2008). It is therefore not surprising that fathers are information seekers once a diagnosis is made and are satisfied only when they are provided the information they need to care and advocate for their child.

A common reason why fathers felt they did not receive the information they needed was because their presence and role in childcare was not acknowledged. Some fathers pointed out that the focus at medical appointments was primarily the mother of the child. They felt that their questions and concerns were quickly dismissed, their opinions were not considered important, and if they were spoken to, it was in a patronizing manner. Healthcare providers can improve the father’s experience by acknowledging their presence and listening to them. Directly speaking to the father along with the mother, answering their questions, asking them how they feel and then addressing those feelings and concerns are important. It is also important to keep in mind that a father may need different information than the mother, and that this often involves information regarding immediate and long-term care for their child.

In cases where the father cannot attend the child’s medical appointment, they often receive information secondhand from the mother (Carpenter & Towers, 2008). This may result in fathers receiving more general information and reduce their engagement in their child’s care. It also puts different stressors on the family as both parents are trying to cope with a new diagnosis, and if only one parent is able to attend the results session, the other may not be receiving the
information they need. In this study, work-related commitments were the most common reason for not attending medical appointments. Fathers traditionally fill the economic provider role in families, and, therefore, their lack of attendance due to work-related reasons is not surprising (Lewis & Pleck, 1979). It is important healthcare providers are aware of the possible work-related commitments of fathers and try to be accommodating of the fact. When scheduling an appointment to discuss results, providers should try to ensure that both parents are available for the visit.

Another reason fathers felt the information they desired was not provided to them might be that FXS is a rare genetic condition and it is difficult to find healthcare providers that are familiar with it. This is often the case with rare genetic conditions, but healthcare providers should have the ability to provide appropriate referrals to specialists to bridge this gap in knowledge when needed. For example, there may be dedicated clinics or specialists for a particular rare disease. Much of this information can be found on the Internet, making it easier for providers to find and provide to families.

An important step in improving access to FXS information, specialists and local resources is to increase awareness and education of FXS for healthcare and service providers. Awareness has increased as testing for FXS has increased, but there is still the perception from families that healthcare providers have limited knowledge of FXS. Many fathers indicated doing their own research on FXS prior to meeting with medical professionals. This is becoming more and more common as there may be a delay between giving the results to a family and seeing them in clinic to discuss the results in detail. Healthcare providers should keep this in mind when meeting with a family to discuss a new diagnosis, especially if they previously gave the results to the family or if the family had access to the results via a patient portal. As families, especially
fathers, are likely to have done research prior to their visit, it is important for healthcare providers to assess the level of understanding of the parents and address the questions they have, rather than fulfilling their own agenda. Fathers stated that information should be presented in a clear, helpful and professional manner without being redundant and overloading on information. Therefore, the discussion regarding the diagnosis should not be a regurgitation of facts, but rather a conversation addressing their questions and concerns and providing psychosocial support.

Despite increased awareness of FXS, fathers noted that healthcare providers did not provide them with resources and many had to seek these resources out themselves. Healthcare providers should make an effort to better educate themselves about resources that are available for fathers, and families in general, and provide appropriate referrals. If the healthcare provider is not familiar with FXS or FXS-related resources, they should make the appropriate referral to specialists, if possible, or make the effort to find these resources. Providers may want to vet some of these resources before referring families as fathers who did use online resources for information gathering found that the literature was complex in medical terminology and did not address them directly.

Support from family, friends, other FXS families and FXS-related organizations

Another source of dissatisfaction for fathers was the lack of support from family and friends and lack of referrals to other FXS families and FXS-related organizations. Although some of their comments indicated some dissatisfaction, overall fathers were satisfied with the support they sought out. However, they did note that they were dissatisfied with healthcare providers for not providing referrals to other FXS families and FXS-related organizations. We found that fathers did not show a preference for one source of support over another. Healthcare providers should, therefore, be familiar with and provide fathers with a wide range of options
when discussing support resources. The Internet has made it substantially easier for providers to look up support networks, Facebook pages for rare diseases, and other sources of support. Therefore, it should take relatively little time to find possible support organizations and should not be an added burden to healthcare providers.

It is important to note that not all fathers sought out support even when presented with options. The main reasons fathers stated for not seeking out support were that they felt they did not need the support and/or they were focused on educating themselves and finding resources to support their partner and child. One father explained that they felt their role was to stay strong and support their partner and child, as the focus was mainly on the carrier mother and the father was not acknowledged or cared about. Studies have shown that although fathers have a difficult time receiving the news of their child’s diagnosis, many decided to “get on with it” and focus on supporting their families instead (Carpenter & Towers, 2008). One father decided not to seek out support because he felt he will be fine, and another father even questioned what kind of support one even asks for. These reactions may be rooted in the gender and parental role stereotypes enforced by society and accepted by healthcare professionals. Healthcare providers need to be careful about assuming the fathers’ reaction and behaviors based on stereotypes. They should instead engage, understand and address the fathers’ individual experience. The father’s comments indicated that healthcare providers might need to check in with them at follow-up visits to discuss how they are doing and if they feel they require support at that time.

It is interesting to note that although greater than 50% of the fathers did have an emotional response to receiving a FXS diagnosis for their child, the majority of them did not seek out support. Studies have found that the expression of feelings like depression is different in men and women (Yogman & Garfield, 2016). Men are likely to avoid emotional expression,
deny vulnerability, and not seek help, consistent with the results of this study (Yogman & Garfield, 2016). Healthcare providers should be cognizant that the way men show emotion is different than women, but it does exist, and thus needs to be addressed. Studies have shown that fathers are typically reluctant in engaging with healthcare providers and services, but they are willing to engage where they feel this leads to improved outcomes for their children (Carpenter & Towers, 2008). A way healthcare providers can engage fathers and encourage support-seeking behaviors is by emphasizing that the benefit is not only for themselves, but also their child.

Generally, it seems as though the fathers felt that the healthcare providers were good at providing medical information, but were lacking in terms of assessing their needs as a child care provider and as a parent of a child who was given a diagnosis of FXS. Healthcare providers should assess the father’s emotional response to the child’s diagnosis and address these feelings with appropriate counseling and/or referrals. It is also important that healthcare providers not assume that fathers are not significantly impacted if more than one child is diagnosed with FXS. One father said he had greater depression and anxiety because both of his children had “bad luck in the genetic lottery of their lives.”

We found there to be a significant association between healthcare providers providing referrals to support organizations and whether or not the father sought out support. This may be because this raises the father’s awareness of such an organization but may also be because a healthcare provider specifically referred them to a support organization. If the father is not aware of the resources available to them, their ability to make a decision of whether or not to seek out support may be limited and uninformed. Once again, healthcare providers must be aware of the resources available in order to provide the appropriate referrals to fathers as these types of referrals can improve the outcome for both fathers and their children.
The fathers’ experiences with seeking support provided several areas that healthcare professionals can address to ensure fathers’ needs are met. Some suggestions for healthcare providers include acknowledging the father’s role and discussing the impact on them when receiving a FXS diagnosis for their child. Some fathers suggested mandating or recommending therapy to help them cope with the diagnosis. One father’s experience requesting counseling was difficult as their healthcare provider was not able to provide them with a suitable counselor. However, finding the “right” counselor depends on multiple factors that are outside providers’ control. Therefore, providers can recommend counseling, but families will need to take an active role in finding the right counselor for them.

FXS organizations can also better meet the needs of fathers by creating more inclusive support groups. One father acknowledged that fathers-only support groups may not work like mothers-only support groups do, but if there were a parents’ support group where mothers and fathers can both attend, that would be more inclusive and accessible to fathers. Further work can also be done to create literature that is accessible and more specific to fathers and their needs. Although FXS-related organizations are generally able to connect parents to other FXS families, they might consider creating a network through which fathers can be connected to other fathers.

Limitations

This pilot study was limited by a small sample size, consisting of only thirty-two participants. Our analysis, therefore, represents a limited population, and may not be generalizable to all fathers of children with FXS, or any other genetic condition. Recruitment was done through organizations such as NFXF and FRAXA and Facebook pages, which may have resulted in a sampling bias. Fathers who are members of these organizations or Facebook pages may be more inclined to share their thoughts and experiences, and are more likely to have
received support and be satisfied with it. As our overall goal was to look at fathers’ support seeking behaviors and better understand their needs, by sampling through support organizations it is inherent that the father had sought out some support. This study was not limited to a particular geographical location. Therefore, we may have had respondents from different parts of the world that have a different standard of care for FXS. Recall bias is another potential limitation to this study as any father of a child diagnosed with FXS after 2006 was included. Finally, qualitative analysis was done by one researcher, which may have led to subjectivity in the identification of themes.

**Future directions**

Further studies on a larger sample size should be conducted. This would allow us to determine if the results from this study are consistent with the larger population of fathers of children with FXS. It would be interesting to see whether the results of this study are consistent with the experiences of fathers of children with other genetic conditions as well. This study only had fathers that were married participate; it would be interesting to see whether marital status influenced their experience. Interviewing fathers along with their partners, if applicable, might be interesting to see how their experiences are similar and different, as previous research has always done studies on the two populations separately. Additional studies could develop father-centric literature for various diagnoses and compare this to traditional literature to see if there is a difference in satisfaction between the information provided.
CONCLUSION

This study aimed to understand fathers’ experiences during the diagnostic process, examine factors that influenced their decision to seek out support and determine ways that healthcare providers and organizations can improve their services to better meet the needs of fathers. We found that generally the fathers were satisfied with their experience going through the diagnostic odyssey. Two major themes appeared when the journeys of the fathers were examined; (1) fathers want access to FXS information, specialists and local resources, and (2) fathers want support from family, friends, other FXS families and FXS-related organizations. By understanding the fathers’ experiences, healthcare and service providers can improve their services to better meet their needs. Healthcare providers can better meet the needs of fathers by (1) increasing awareness and education of FXS and FXS-related resources, (2) improving the discussion of FXS by assessing the parents level of understanding, addressing their questions and concerns, and providing psychosocial support, and (3) acknowledging the fathers role and discussing the impact on them when receiving a diagnosis for their child. Support organizations such as NFXF can also better meet the needs of fathers by (1) creating literature tailored to fathers, (2) encouraging more father-related activities in support groups, and (3) creating a network to connect fathers to other fathers.

The results of this study support the anecdotal clinical experience. With the support of this data, and suggestions from the fathers, we can hopefully implement these changes and suggestions in order to address and meet the individual needs of fathers of children diagnosed with FXS, and support them through their unique journey.
REFERENCES


APPENDICES

Appendix A: Recruitment notice

National Fragile X Foundation and FRAXA Research Foundation Listserv E-blast

Subject: Journey of Fathers Following their Child’s Fragile X Diagnosis

Seeking Fathers of Children with Fragile X Syndrome to Participate in a Research Study

You are invited to participate in an online research survey to investigate the journey of fathers following their child’s fragile X diagnosis, focusing on their support seeking behaviours. As a part of my Master’s thesis at Brandeis University, I am looking to understand the unique experience of fathers in order to determine what improvements can be made to services provided by healthcare providers and organizations to better meet their needs.

This study is open to fathers of at least one child diagnosed with fragile X syndrome within the past 10 years (≥ 2006).

The specific goals of this research study are to:

1. understand the fathers experience during the diagnostic odyssey, including their satisfaction with the healthcare providers
2. identify factors that influenced the fathers decision to seek out support and their satisfaction with the support they received
3. determine what improvements can be made to the services provided by healthcare providers and organizations to better meet their needs

This anonymous survey will take an average of 10 minutes of your time. All participants who complete the survey will have the opportunity to enter a draw for one of three $50 Amazon gift cards. Your survey responses will not be connected to your email address.

This study was reviewed and approved by the Brandeis University Institutional Review Board. If you wish to be a part of this study please click the link provided below.

If you have any questions, concerns or comments, please feel free to contact me by email at amann5@brandeis.edu, or the Brandeis University faculty sponsor, Sharyn Lincoln, at Sharyn.Lincoln@childrens.harvard.edu or (617) 355-4688.

Thank you in advance for your time and participation.

Click here to take the survey!

Sincerely,

Aman Mann
Master’s Degree Candidate, Class of 2017

Sharyn Lincoln, MS, CGC
Program Coordinator, Fragile X Program,
Are you a father of a child diagnosed with fragile X syndrome?

Was your child diagnosed within the past 10 years?

If you answered yes to the questions above, you are invited to participate in an online research survey to investigate the journey of fathers following their child’s fragile X diagnosis, focusing on their support seeking behaviours. As a part of my Master’s thesis at Brandeis University, I am looking to understand the unique experience of fathers in order to determine what improvements can be made to services provided by healthcare providers and organizations to better meet their needs.

This anonymous survey will take an average of 10 minutes of your time. All participants who complete the survey will have the opportunity to enter a draw for one of three $50 Amazon gift cards. Your survey responses will not be connected to your email address.

This study was reviewed and approved by the Brandeis University Institutional Review Board. If you wish to be a part of this study please click the link provided below.

If you have any questions, concerns or comments, please feel free to contact me by email at amann5@brandeis.edu, or the Brandeis University faculty sponsor, Sharyn Lincoln, at Sharyn.Lincoln@childrens.harvard.edu or (617) 355-4688.

Thank you in advance for your time and participation.

Click here to take the survey!

Sincerely,

Aman Mann
Master’s Degree Candidate, Class of 2017
Genetic Counseling Program
Brandeis University
Appendix B: Study instrument

Welcome!

You are being asked to participate in this study because you are a father of at least one child diagnosed with fragile X syndrome within the past 10 years. The purpose of this study is to understand the unique experiences of fathers after their child is diagnosed with fragile X syndrome, focusing on their support seeking behaviors. This information will be used to determine what improvement can be made to services provided by healthcare providers and organizations to better meet the needs of fathers.

The specific goals of this research study are to:
4. understand the fathers experience during the diagnostic odyssey, including their satisfaction with the healthcare providers
5. identify factors that influenced the fathers decision to seek out support and their satisfaction with the support they received
6. determine what improvements can be made to the services provided by healthcare providers and organizations to better meet their needs

Your responses to this online survey will be anonymous. This survey is expected to take an average of 10 minutes of your time. Participation of this survey is voluntary. We ask that you respond to all survey questions, however, you may skip any questions that you are not comfortable answering or end your participation at any point.

This study involves minimal risk to participants. Some participants may experience some distress when answering questions regarding their experience receiving their child’s diagnosis as it may result in them re-living the experience. Participants may benefit from the feeling that their experience, feelings and opinions are being heard and that they are contributing useful information to the field of genetic counseling.

All participants who complete the survey will have the opportunity to enter a draw for one of three $50 Amazon gift cards. If you are interested, please enter your email address on a separate link as directed at the end of the survey. Your survey responses will not be connected to your email address.

This study was reviewed and approved by the Brandeis University Institutional Review Board. If you have any questions about your rights as a research subject please contact the Brandeis Institutional Review Board at irb@brandeis.edu or (781) 736-8133.

If you have any questions, concerns, or comments about this study, please feel free to contact me, Aman Mann, at amann5@brandeis.edu, or the Brandeis University faculty sponsor, Sharyn Lincoln, at Sharyn.Lincoln@childrens.harvard.edu or (617) 355-4688.

By clicking Next button (>>, you acknowledge that you have read the information above and consent to participate in this survey.
Thank you in advance for your time and participation.

**Survey Questions**

**I. Demographics - Father**

Q1 Are you the father of at least one child diagnosed with fragile X syndrome?
- Yes
- No

*If No Is Selected, Then Skip To End of Survey*

Q2 Was your child diagnosed with fragile X syndrome in 2006 or later?
- Yes
- No

*If No Is Selected, Then Skip To End of Survey*

Q3 How old were you when your FIRST child was diagnosed with fragile X syndrome? (Years)
- ≤ 20
- 21-25
- 26-30
- 31-35
- 36-40
- 41-45
- ≥ 46
- I do not remember

Q4 What is your current marital status?
- Single, never married
- Married or Domestic Partnership
- Divorced
- Separated
- Widower
- Prefer not to answer

Q5 How many biological children do you have?
- 1
- 2
- 3
- 4
- 5
- >5
Q6 How many of your biological children have been diagnosed with fragile X syndrome?

- 1
- 2
- 3
- 4
- 5
- >5

II. Demographics – First Affected Child

Q7 Please answer the following questions ONLY for your FIRST child diagnosed with fragile X syndrome.

Q8 How old was your child at diagnosis?

- Age (Years) ____________________
- I do not remember

Q9 What year was your child diagnosed?

- Year (YYYY) ____________________
- I do not remember

Q10 Please indicate the clinical features your child exhibits and their severity, from mild to severe. If they do not have that particular clinical feature please select N/A.

<table>
<thead>
<tr>
<th>Clinical Feature</th>
<th>Mild</th>
<th>Moderate</th>
<th>Severe</th>
<th>N/A</th>
</tr>
</thead>
<tbody>
<tr>
<td>Developmental Delay</td>
<td>☑</td>
<td></td>
<td></td>
<td>☑</td>
</tr>
<tr>
<td>Intellectual Disability</td>
<td>☑</td>
<td></td>
<td></td>
<td>☑</td>
</tr>
<tr>
<td>Hyperactivity</td>
<td>☑</td>
<td></td>
<td></td>
<td>☑</td>
</tr>
<tr>
<td>Hand flapping</td>
<td>☑</td>
<td></td>
<td></td>
<td>☑</td>
</tr>
<tr>
<td>Hand biting</td>
<td>☑</td>
<td></td>
<td></td>
<td>☑</td>
</tr>
<tr>
<td>Temper tantrums</td>
<td>☑</td>
<td></td>
<td></td>
<td>☑</td>
</tr>
<tr>
<td>Autism</td>
<td>☑</td>
<td></td>
<td></td>
<td>☑</td>
</tr>
<tr>
<td>Lack of impulse control</td>
<td>☑</td>
<td></td>
<td></td>
<td>☑</td>
</tr>
<tr>
<td>Hyperarousal/sensory issues</td>
<td>☑</td>
<td></td>
<td></td>
<td>☑</td>
</tr>
<tr>
<td>Seizures</td>
<td>☑</td>
<td></td>
<td></td>
<td>☑</td>
</tr>
<tr>
<td>Sleep problems</td>
<td>☑</td>
<td></td>
<td></td>
<td>☑</td>
</tr>
</tbody>
</table>
III. Diagnostic Odyssey – Interaction with Healthcare Providers

Q11 How old was your child when you started the process of getting a diagnosis?
   Years __________
   Months __________

Q12 How long did it take to get a fragile X diagnosis for your child? (From the time of the initial visit to the time of diagnosis)
   Years __________
   Months __________

Q13 What percentage of your child's medical appointments did you attend? (Fragile X related appointments only)
   ○ 0%
   ○ 10%
   ○ 20%
   ○ 30%
   ○ 40%
   ○ 50%
   ○ 60%
   ○ 70%
   ○ 80%
   ○ 90%
   ○ 100%

Answer If Of all the medical appointments with all of the providers, what percentage of appointments did you attend with your child? 100% Is Not Selected

Q14 What was the reason(s) you could not attend the medical appointments?

Q15 Did you meet with a genetic counselor?
   ○ Yes
   ○ No

Answer If Did you meet with a genetic counselor? Yes Is Selected

Q16 What was your experience like meeting with the genetic counselor?

Q17 Who was the FIRST medical professional you met with to discuss the fragile X diagnosis?
   ○ Neurologist
   ○ Geneticist
   ○ Genetic Counselor
   ○ Developmental Pediatrician
   ○ Other ____________________
Q18 Please indicate your level of agreement, from strongly disagree to strongly agree, with the following statements in relation to your interactions with the [Answer from Q17].

<table>
<thead>
<tr>
<th>Statement</th>
<th>Strongly disagree</th>
<th>Somewhat disagree</th>
<th>Neither agree nor disagree</th>
<th>Somewhat agree</th>
<th>Strongly agree</th>
</tr>
</thead>
<tbody>
<tr>
<td>I was included in the decision to do molecular genetic testing on my child for fragile X syndrome.</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>The [Answer from Q17] addressed my questions and concerns after receiving the diagnosis.</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>The [Answer from Q17] addressed my needs as a father after receiving a diagnosis.</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>The [Answer from Q17] provided me with information on the fragile X diagnosis.</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>I received the information I needed to care for my child.</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>The [Answer from Q17] provided me information on family centered or male-focused support groups.</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>The [Answer from Q17] provided referrals to support organizations such as the National Fragile X Foundation.</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>The [Answer from Q17] connected me with another family with fragile X diagnosis.</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>I am overall satisfied with my experience with the [Answer from Q17].</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
</tbody>
</table>

Follow-Up Questions for Q18

Q19 You selected strongly disagree or somewhat disagree to the following statement: I was included in the decision to do testing on my child for fragile X syndrome. Please explain why.

Q20 You selected strongly disagree or somewhat disagree to the following statement: The [Answer from Q17] addressed my questions and concerns after receiving the diagnosis. Please explain why.

Q21 You selected strongly disagree or somewhat disagree to the following statement: The [Answer from Q17] addressed my needs as a father after receiving a diagnosis. Please explain why.
Q22 You selected strongly disagree or somewhat disagree to the following statement: The [Answer from Q17] provided me with information on the fragile X diagnosis. Please explain why.

Q23 You selected strongly disagree or somewhat disagree to the following statement: I received the information I needed to care for my child. Please explain why.

Q24 You selected strongly disagree or somewhat disagree to the following statement: The [Answer from Q17] provided me information on family centered or male-focused support groups. Please explain why.

Q25 You selected strongly disagree or somewhat disagree to the following statement: The [Answer from Q17] provided referrals to support organizations such as the National Fragile X Foundation. Please explain why.

Q26 You selected strongly disagree or somewhat disagree to the following statement: The [Answer from Q17] connected me with another family with fragile X diagnosis. Please explain why.

Q27 You selected strongly disagree or somewhat disagree to the following statement: I am overall satisfied with my experience with the [Answer from Q17]. Please explain why.

Q28 You selected somewhat agree or strongly agree to the following statement: I am overall satisfied with my experience with the [Answer from Q17]. Please explain what made the experience satisfying.

IV. Post-fragile X Diagnosis – Father’s Experience

Q29 Did you experience any of the following after your child received a fragile X diagnosis? Check all that apply.
- Depression
- Anger
- Anxiety
- Stress
- Relief
- Other _______________________

Q30 Did you seek out support after receiving your child's fragile X diagnosis?
- Yes
- No
Answer: If Did you seek out support after receiving your child's fragile X diagnosis? No Is Selected

Q31 Please explain why you chose not to seek out support after receiving your child's fragile X diagnosis.

Answer: If Did you seek out support after receiving your child's fragile X diagnosis? Yes Is Selected

Q32 Who did you reach out to for support? Check all that apply.
- Your partner
- Family members (i.e. siblings, parents, cousins, aunts/uncles, etc.)
- Fragile X related organizations (i.e. National Fragile X Foundation)
- Internet resources (i.e. Facebook groups and blogs)
- Fragile X support groups
- Other families with a child diagnosed with fragile X syndrome
- Religious/spiritual community
- Other ____________________

Q33 Please rate the level of satisfaction you felt from these sources of support.

*The options selected in Q32 will appear in a matrix using a satisfaction scale. For example, if “Your partner” is selected, the table would look like this:*

<table>
<thead>
<tr>
<th>Source</th>
<th>Extremely dissatisfied</th>
<th>Somewhat dissatisfied</th>
<th>Neither satisfied nor dissatisfied</th>
<th>Somewhat satisfied</th>
<th>Extremely satisfied</th>
</tr>
</thead>
<tbody>
<tr>
<td>Your partner</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**Follow-up Questions for Q33**

Q34 You indicated you were extremely dissatisfied or somewhat dissatisfied from the support you received from your partner. Please explain why.

Q35 You indicated you were extremely dissatisfied or somewhat dissatisfied from the support you received from family members (i.e. siblings, parents, cousins, aunts/uncles, etc.). Please explain why.

Q36 You indicated you were extremely dissatisfied or somewhat dissatisfied from the support you received from fragile X related organizations (i.e. National Fragile X Foundation). Please explain why.

Q37 You indicated you were extremely dissatisfied or somewhat dissatisfied from the support you received from Internet resources (i.e. Facebook groups and blogs). Please explain why.

Q38 You indicated you were extremely dissatisfied or somewhat dissatisfied from the support you received from fragile X support groups. Please explain why.
Q39 You indicated you were extremely dissatisfied or somewhat dissatisfied from the support you received from other families with a child diagnosed with fragile X syndrome. Please explain why.

Q40 You indicated you were extremely dissatisfied or somewhat dissatisfied from the support you received from religious/spiritual community. Please explain why.

Q41 You indicated you were extremely dissatisfied or somewhat dissatisfied from the support you received from [Other from Q32]. Please explain why.

V. Final Thoughts

Answer If Given you have had multiple children diagnosed with fragile X syndrome, was your experience receiving the diagnosis and coping with it different in any way? Yes Is Selected

Q42 Given that you have multiple children diagnosed with fragile X syndrome, was your experience receiving the diagnosis and coping with it any different?

○ Yes
○ No

Q43 In what way(s) was the experience of receiving the fragile X diagnosis for your other child(ren) and coping with it different from your first child?

Q44 In what way(s) was the experience of receiving the fragile X diagnosis for your other child(ren) and coping with it similar to your first child?

Q45 What did you find most helpful when going through the process of receiving the fragile X diagnosis for your child?

Q46 As a father of a child with a fragile X diagnosis, how do you think health care providers and support organizations can better meet your needs after receiving a diagnosis?

VI. Raffle Participation

Q47 Would you like to enter a draw to win one of three $50 Amazon gift cards? Note: Your contact information will not be connected to your survey responses if you select yes.

○ Yes
○ No

If No Is Selected, Then Skip To End of Survey
If Yes Is Selected, Then Go To Separate Survey To Input Contact Information
Appendix C: Permission letters

National Fragile X Foundation

Brandeis University Thesis Project

Linda Sorensen <linda@fragilex.org>
To: "amann5@brandeis.edu" <amann5@brandeis.edu>
Cc: David Salomon <david@fragilex.org>

Hi Aman – Thank you for contacting the NFXF. We will post your survey on our website and on our social media sites within the next 7 to 10 days.

Linda Sorensen
Chief Operating Officer
linda@fragilex.org
Direct 202-747-6202
Mobile 925-283-1897

Together, We're Stronger!
Invest in the Future of Fragile X Research and a Stronger Community
Donate today

National Fragile X Foundation
2100 M St., NW
Ste.170 Box 302
Washington, DC 20037-1233

Main 202-747-6210
Toll-Free 800-688-8765
Fax 202-747-6208
Hi Aman,

Yes, you have our permission to use both facebook and the Listserv. You can just post your survey on our facebook page and the instructions are below to join the listserv and do the same. If you need anything further let me know!

Good luck!
Melissa

FRAXA Listserv:

To Join: Send email from your email address to LISTSERV@LISTSERV.CC.EMORY.EDU  
In the body of your message type: SUBSCRIBE FRAGILE-X-mynname (“mynname” is whatever name you want to appear as the “from name” on your Listserv postings. Type it in the way you want it to appear in upper/lower case letters.)  
[Quoted text hidden]

--
Melissa Budek  
Office Manager  
FRAXA Research Foundation  
10 Prince Pl., Newburyport, MA 01950  
tel: (978) 462-1866  
Sign up for email updates (free) at FRAXA.org

Please consider the environment before printing this email.
Fragile X syndrome

Hi Aman. https://fragilex.org/ is probably the best resource. Is it your father?

Aman M

Hi,
It is not my father. I am conducting a study to see what the father's experience is like receiving a Fragile X diagnosis for their child. I wanted to know if I can post my survey and recruit fathers that are a part of your Facebook group for my study?

December 22, 2016

Hi Aman, I will post something on the page for you. Have you contacted admins of any Fragile X groups on Facebook?

December 25, 2016

Aman M

Hi. Yes I have contacted some admins, such as the NFXF to use their facebook page and listserv and have received approval.

If you could post something on the page for me that would be excellent. I have not opened up my survey at this time, as I would like to send it out after the New Year. Would it be okay if I sent you my recruitment notice and survey link next week, in order to post it on the page for January 9th?

I appreciate all your help.

Thank you,