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Assessing Parental Satisfaction of Current Fragile X Syndrome Resources Provided at Diagnosis

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ASSESSING PARENTAL SATISFACTION OF CURRENT FRAGILE X SYNDROME RESOURCES PROVIDED AT DIAGNOSIS

By

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ABSTRACT

Diagnosis of a child with fragile X syndrome (FXS) is associated with parental anxiety. Existing literature recommends FXS resources, but there has not been a formal assessment of when parents learn about these resources, or their satisfaction with them. Studies have demonstrated negative feelings surrounding the diagnostic experience of FXS. This exploratory analysis assessed the gap in knowledge on parental satisfaction of the recommended resources from healthcare providers during the time of diagnosis. We predicted parents would not be satisfied with the way resources were presented to them. We discussed parental satisfaction or dissatisfaction, utility of the recommended resources, and determined what resources parents independently identified as valuable for healthcare professionals and genetic counselors to recommend to families in the future.

Seven participants took an online questionnaire containing inclusion criteria and demographic type questions. Four of these participants partook in further focus group discussion (n = 3) or individual interview (n = 1) and discussed FXS resource satisfaction and preferences. The results were analyzed descriptively.

Overall, participants were not satisfied with the information and advice they received from providers at diagnosis. All participants expressed a need for more positive and balanced information about FXS. Parents primarily relied on information and support from national organizations like NFXF and FRAXA, Facebook groups, and national FXS conferences. Two participants were not given any informational or support resources at the time of diagnosis. Participants recommended that professionals educate families

about specific resources at the time of diagnosis and provide actionable steps forward in an age-appropriate manner.

This study suggests parents would like the names of specific resources at diagnosis like national organizations, Facebook group names, local resources, and a way to connect with a parent who has already experienced a FXS diagnosis. Parents feel isolation and anxiety during a new diagnosis of their child, so it is important to emphasize positivity and that a group of providers, therapists, and the entire FXS community is there to support the family. Genetic counselors and providers should give age-appropriate recommendations so parents can continue to adjust and adapt to their child's FXS diagnosis.

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CHAPTER 1: BACKGROUND

1.1 Fragile X Syndrome Genetics

Fragile X syndrome (FXS) is a genetic condition characterized by intellectual disability, developmental delay, and behavioral issues. FXS is one of the most common forms of inherited intellectual disability and is estimated to affect approximately 1 in 4,000 males and 1 in 8,000 females (Hunter et al., 2019; Van Remmerden et al., 2020). The Fragile X Mental Retardation 1 (*FMR1)* gene, located at chromosome Xq27.3, encodes a protein known as Fragile X Mental Retardation Protein, FMRP. FXS is caused when the *FMR1* gene is not functioning within nerve cells properly (Hunter et al., 2019). A lack of this protein can lead to the behavioral and cognitive features seen in individuals with FXS.

Fragile X syndrome is a trinucleotide repeat expansion disorder. The *FMR1* gene has a 5' untranslated region containing a trinucleotide repeat sequence (Garber et al., 2008). This region is made up of a cytosine-guanine-guanine (CGG) nucleotide triplet that repeats a variable amount in all individuals. The typical number of CGG repeats in *FMR1* is 6 to 44 repeats, which is referred to as a normal allele (Finucane et al., 2012). Males typically have one normal allele and women have two normal alleles. A normal allele is unmethylated, so the gene is able to be transcribed and translated to make FMRP (Hunter et al., 2019). The repeating region is unstable, and an expansion in the repetitive sequence is possible, which can lead to premutation or full mutation of the gene

(Sherman et al., 2005). If the CGG repeats expand past a certain threshold, it can become hypermethylated, effectively turning the gene off by halting transcription of *FMR1* (Hunter et al., 2019).

Individuals with 45 to 54 CGG repeats have an intermediate genotype, have a small risk for expansion, and may have mild symptoms (Finucane et al., 2012). Some mutations in this intermediate range are stable, while others are not and are at greater risk of expanding to a premutation (Nolin et al., 1996). Someone with 55 to 200 CGG repeats is known to have a *FMR1* premutation and there is a risk for expansion in subsequent generations to a full mutation due to the *FMR1* instability (Nolin et al., 1996; Sherman et al., 2005). The premutation allele is associated with the risk of developing a *FMR1*-related disorder but does not cause classic FXS. An individual with more than 200 repeats has a full mutation, so methylation of *FMR1* occurs, so no FMRP is made and that individual has FXS (Garber et al., 2008; Hunter et al., 2019). Approximately 99% of individuals with FXS have the expanded CGG repeat size, but 1% of individuals have either a deletion or a point mutation in the *FMR1* gene (McConkie-Rosell et al., 2005).

As mentioned earlier, the normal allele (less than 44 repeats) is generally considered stable, so it is unlikely to expand as it gets transmitted to the next generation (Monaghan et al., 2013). As the repeat size increases into the intermediate zone or to the premutation allele, there is a greater chance of an expansion to occur (Hunter et al., 2019). Expansion risk depends on CGG repeat size, maternal or paternal transmission, and if there are AGG triplets interrupting the sequence (Eichler et al., 1996; McConkie-Rosell et al., 2005). In the general population, around 95% of individuals have one or two AGG interruptions that stabilizes the *FMR1* gene during transmission, which prevents

DNA polymerase slippage during DNA replication, thereby preventing expansion. Some individuals with the premutation can still have one or more AGG interruptions, which greatly increases the stability (Villate et al., 2020). In the presence of an interrupting AGG repeat, premutation carriers have a decreased risk of expansion of CGG repeats to the full *FMR1* mutation (Nolin et al., 2015).

Fragile X syndrome and *FMR1*-related disorders are inherited in an X-linked manner (Sherman et al., 2005). Every person has 23 pairs of chromosomes, 22 pairs are autosomes and are the same between males and females, and there is one pair of sex chromosomes (typically XX or XY). *FMR1* resides on the X chromosome, so males have one copy of *FMR1* since their sex chromosomes are "XY" and females have two copies of the gene because their sex chromosomes are "XX" (Hunter et al., 2019). A mother always passes one of her two X chromosomes to her child, while a father will pass on his Y chromosome to his sons, and his X chromosome to his daughters. If a female is a premutation carrier, she has a 50% chance of passing on the premutation, which resides on one of her X chromosomes, to each of her children. When a female premutation carrier transmits the premutation allele to the next generation, there is almost always an expansion (Nolin et al., 2003). A male with the premutation will pass this allele to all of his daughters, but none of his sons. The premutation in males can undergo a small expansion, a contraction, or remain stable. The premutation very rarely expands to a full mutation due to male transmission (Nolin et al., 2003).

1.2 Fragile X Syndrome Features and Management

There is a variable clinical phenotype observed in FXS. Individuals can range from having learning difficulties but a normal IQ, to severe intellectual disability and/or

autism (Garber et al., 2008). FXS is one of the most common forms of inherited intellectual disability and is the most common inherited form of autism spectrum disorder (Hunter et al., 2019; Van Remmerden et al., 2020). Each individual with FXS is unique, but there are characteristic features of this condition. These features may affect males and females, but males are typically more severely affected (Finucane et al., 2012). Individuals can have physical features such as hypotonia, large ears, long face, macroorchidism (in males), hypermobility of the joints, increased susceptibility to seizures, mitral valve prolapse, and increased frequency of ear infections (Garber et al., 2008). Common cognitive and behavioral features include various learning or intellectual disabilities, autism spectrum disorder (ASD), motor or speech delays, behavior issues (e.g. attention deficit hyperactivity disorder), shyness or difficulty with transitions, echolalia, coprolalia, and/or stereotypies (Hunter et al., 2019; McConkie-Rosell et al., 2005).

Females with a full mutation allele may have similar features as an affected male; however, it is more likely that a female will have a more mild and subtle presentation (McConkie-Rosell et al., 2005). Less than 50% of females with a full mutation allele have intellectual disability (Spector et al., 2021). Females may have a normal IQ or only slight learning disabilities, and they are also more likely to have minor physical characteristics and psychological concerns in comparison to males with FXS (McConkie-Rosell et al., 2005). The most common manifestations are behavioral, such as: avoidant personality, mood disorders, and stereotypic disorders (Spector et al., 2021). Non-random X inactivation is thought to explain the wide spectrum of features in females and can explain the milder phenotype seen in some females with the full mutation (Spector et al.,

2021). Typically, the somatic cells in females undergo random X chromosome inactivation where one X chromosome is transcriptionally silenced in each cell (Hall et al., 2016). This results in roughly a 50/50 split of which X chromosome is active within regions of the body. Variation in the activation ratio away from a typical 50/50 ratio can cause the affected *FMR1* premutation or full mutation allele to be disproportionately active or inactive within the body, contributing to heterogeneity of FXS and *FMR1*–related disorder features and severity (Hall et al., 2016).

FMR1-related disorders are associated with having a premutation allele and include Fragile X Primary Ovarian Insufficiency (FXPOI) and Fragile X-Associated Tremors/Ataxia syndrome (FXTAS) (Monaghan et al., 2013). FXPOI causes ovarian dysfunction, and symptoms include early onset menopause, irregular menses, and infertility. About 20% of women with a premutation can have features of FXPOI (Finucane et al., 2012; Spector et al., 2021). More rarely, premutation carriers can develop FXTAS. FXTAS more commonly affects males starting in their 50s and is associated with tremors and progressive nerve degeneration, leading to balance and coordination difficulty, and occasionally slurred speech (Finucane et al., 2012). Cognitive decline is also a feature of FXTAS and can include memory loss, dementia, and a loss of decision-making and executive functioning skills (Finucane et al., 2012).

There is currently no cure or standard treatment for Fragile X syndrome. The management of FXS is based on treating an individual's symptoms that present and following the recommended surveillance guidelines (Hunter et al., 2019). Various therapies are valuable to help an individual achieve their highest neurodevelopmental potential. These include applied behavior analysis therapy (ABA therapy), speech

therapy, occupational therapy, and early intervention (Garber et al., 2008). Seizures, psychological, and behavioral features can be treated via medication(s). It is optimal for a patient to receive a referral and have their care managed through a Fragile X clinic that is part of the Fragile X Clinical and Research Consortium (FXCRC) (National Fragile X Foundation). These clinics provide multidisciplinary care and can provide medications, genetic counseling, consultations with appropriate specialists, referrals to other providers, and help coordinate various therapies (Finucane et al., 2012). Many clinics partake in research efforts and can help patients get involved in ongoing FXS research and clinical trials.

1.3 Genetic Testing of Fragile X Syndrome

Due to varied clinical phenotypes, a FXS diagnosis is made based on genetic testing of the *FMR1* gene to assess the number of CGG repeats (Garber et al., 2008). The American College of Medical Genetics (ACMG) Standards and Guidelines for Fragile X testing recommends genetic testing for:

Fragile X syndrome:

1. Individuals of either sex with intellectual disability, developmental delay, or autism, especially if they have (a) any physical or behavioral characteristics of fragile X syndrome, (b) a family history of fragile X syndrome, or (c) male or female relatives with undiagnosed mental retardation.
2. Individuals seeking reproductive counseling who have (a) a family history of fragile X syndrome or (b) a family history of undiagnosed mental retardation.
3. Fetuses of known carrier mothers.

4. Affected individuals or their relatives in the context of a positive cytogenetic fragile X test result who are seeking further counseling related to the risk of carrier status among themselves or their relatives. The cytogenetic test was used prior to the identification of the FMR1 gene and is significantly less accurate than the current DNA test. DNA testing on such individuals is warranted to accurately identify premutation carriers and to distinguish premutation from full mutation carrier women.

Ovarian Dysfunction:

1. Women who are experiencing reproductive or fertility problems associated with elevated follicle stimulating hormone (FSH) levels, especially if they have (a) a family history of premature ovarian failure, (b) a family history of fragile X syndrome, or (c) male or female relatives with undiagnosed mental retardation.

Tremor/Ataxias:

1. Men and women who are experiencing late onset intention tremor and cerebellar ataxia of unknown origin, especially if they have (a) a family history of movement disorders, (b) a family history of fragile X syndrome, or (c) male or female relatives with undiagnosed mental retardation. (Sherman et al., 2005, p. 586)

Trinucleotide repeat size and methylation status needs to be determined on the *FMR1* gene through polymerase chain reaction (PCR) and Southern blot analysis (Spector et al., 2021). PCR is able to amplify the repeated region on *FMR1* to allow the number of repeats to be determined. PCR decreases in effectiveness as the number of

repeats increases and it is able to better determine repeat size up to a premutation allele. Southern blot analysis can determine methylation status as well as approximate allele size. ACMG recommends that laboratories combine the usage of traditional PCR with Southern blot analysis (Monaghan et al., 2013). If repeat-primed PCR or methylation PCR is used, this can decrease the use of Southern blot analysis. Laboratories can use various tests depending on presenting clinical features (Monaghan et al., 2013). Monaghan et al. (2013) explains that, since full mutations of over 200 CCG repeats account for over 99% of FXS cases, the tests are over 99% sensitive and 100% specific. If testing identifies a full *FMR1* mutation allele in a male, this is considered diagnostic because FXS is 100% penetrant. In females, these results can be diagnostic if her clinical features are consistent with FXS, but it should be noted that less than 50% of females with a full mutation allele will be affected with intellectual disability (Spector et al., 2021). Less than 1% of FXS cases are caused due to a *FMR1* sequence variant or a partial or full gene deletion (Sitzmann et al., 2018). These patients would usually be detected with a multi-gene sequencing panel or deletion/duplication analysis that includes *FMR1* (Hunter et al., 2019).

1.4 Fragile X Syndrome Results Counseling and Resources

It is known that patients primarily rely on their healthcare professionals to provide them with credible information about their condition (Genetic Alliance, 2010). Genetic counselors are valuable and serve as a resource about genetic conditions for patients, as well as other healthcare professionals (Genetic Alliance, 2010). Education that usually occurs during a results disclosure of FXS includes the inheritance pattern, the genetics of the condition, risk assessment for other family members, potential prognosis,

management options, and patient resources (McConkie-Rosell et al., 2005). In a FXS results disclosure session, the parent or guardian can be overwhelmed emotionally due to their child's diagnosis and the large amount of information provided during the disclosure session(s). Parents may feel in shock at the time of diagnosis, feel uncertainty about their child's long-term development, and are often coping with many new feelings and fears (Lebmann and Sarimski, 2013). Genetic counselors can help support the family by gathering relevant local, national, and online support groups and resources (Goodwin et al., 2015).

A study looking at the diagnostic experience of parents of children affected with 22q11.2 deletion syndrome, Fragile X syndrome, and Down syndrome found that the diagnostic experience is generally negative because healthcare providers show a lack of support and provide unsatisfactory quantity and quality of information on the condition (Goodwin et al., 2015). A parent experiences a new diagnosis more positively when information is presented in a calm and supportive manner (Goodwin et al., 2015). Parents expect the healthcare professional to understand their emotional distress at the time of diagnosis and deliver advice and provide information about support, treatment, and next steps (Lebmann & Sarimski, 2013). Lebmann and Sarimski (2013) studied parental experiences of FXS diagnosis and found that some parents overall felt grateful for understanding the reason for their children's delays, but 58.4% stated they desired more information about the disability, and 52.8% wanted more emotional support from the professional during the diagnosis. Support groups and parents' groups are known to benefit parents and help them cope with a child's diagnosis (Finnegan et al., 2014). One study found that 81.3% of parents found support groups to be important following a FXS

diagnosis (Lebmann & Sarimski, 2013). Parents may cope and manage their reactions to a child's diagnosis through information seeking behavior about the condition as a way to "restore order in a chaotic existence" (Starke & Moller, 2002). When parents have a child with a disability, it is common that a mother will seek out advice from the healthcare provider to help her understand her child's needs, while a father is more likely to manage using resources such as medical literature or books (Starke & Moller, 2002).

It is important that the healthcare professional is well informed about the best resources available. Resources can include printed information pamphlets, websites, information about patient advocacy groups, books, or even national organizations (Genetic Alliance, 2010; Uhlmann et al., 2010). Patients have reported a preference for written materials such as brochures, pamphlets, and books over websites (Genetic Alliance, 2010). Mac et al. (2020) states that online resources are often written above the recommended reading level and can be perceived as more difficult to understand than printed materials, which highlights the importance of finding patient-friendly resources. Recently, social media is allowing patients and their families to be more involved in their care and advocacy. Such platforms are becoming a medium to empower patients, increase a sense of community and participation, and increase research recruitment (Pogue et al., 2018). The complexity of the many resources makes it important that providers are comfortable with the resources that they recommend to patients. Uhlmann et al. (2010) warns that not all advocacy and support groups benefit each family in the same way, so healthcare providers must assess every family individually to determine which group(s) to introduce a family to.

There are many Fragile X syndrome resources that exist, but families often do not know of these resources without appropriate guidance. McConkie-Rosell et al. (2005) recommended resources such as National Fragile X Foundation, FRAXA Research Foundation, family-oriented books and articles, and other online resources. The National Fragile X Foundation provides informational resources about FXS and *FMR1*-related disorders. They also maintain support groups and a Facebook page for affected families (Finucane et al., 2012). The National Fragile X Foundation provides additional information about current research studies, the Fragile X Online Registry with Accessible Research Database (FORWARD) natural history study, and the location of Fragile X Clinics. The FRAXA Research Foundation is also a good resource dedicated to FXS research. FRAXA also has information surrounding medical treatment and education strategies to use for children with FXS and contains a for-parents-by-parents guide relating to challenges of daily life with FXS. In addition to websites, there is a parent-written physician-reviewed book called "Children with Fragile X Syndrome: A Parent's Guide" by Jayne Dixon Weber including information about FXS concerns, daily and family life, child development and education, as well as family advocacy. Resources like the ones listed, along with a strong healthcare/social team and personal coping strategies help families make a successful adjustment to a new diagnosis (Lebmann & Sarimski, 2013).

1.5 Rationale of Study

There has been little research conducted to assess the feelings and satisfaction of current resources that exist for FXS. There is no literature to our knowledge that assesses how parents of children with FXS feel about the resources they are provided when their

child is newly diagnosed with the condition. Existing literature recommends resources, but there has not been an assessment of when parents learn about these resources or their satisfaction with the resources that healthcare professionals provide (i.e. Finucane et al., 2012; McConkie-Rosell et al., 2005). Goodwin et al. (2012) reported a negative outlook and feelings of lack of support surrounding the diagnostic experience for various conditions: 22q11.2, Down syndrome, and related conditions, including Fragile X syndrome. Lebmann and Sarimski (2013) also found negative feelings surrounding the diagnostic experience of FXS. This demonstrates that parents need support when their child is diagnosed with a chronic condition, so it is important that the best resources are being provided to families.

Genetic counselors often play a direct and important role in disclosing a new genetic diagnosis and providing the appropriate resources to families with a genetic condition. A genetic counseling session is often recommended to increase understanding about the condition, management, and resources, with the goal of reducing family stress and anxiety (Genetic Alliance, 2010). Genetic counselors are not the only healthcare providers to explain a diagnosis of FXS, and not all providers can dedicate the same amount of time to a result disclosure.

1.6 Purpose of Study

The purpose of this study was to assess parental satisfaction with the recommended resources they received at the time of a FXS diagnosis of their child. Parents with children who were diagnosed with a full *FMR1* mutation in the past 10 years were targeted to take an initial survey to meet study criteria. A focus group and one-on-one interviews were used to gather information about parental satisfaction with current

FXS resources and their satisfaction with the resources healthcare professionals are recommending at the time of diagnosis. The study identified common resources that are provided to parents as they receive a FXS diagnosis. We hypothesized that parents would not be fully satisfied with the way resources were presented to them at the time of diagnosis. This study aimed to improve the diagnostic experience for families receiving a FXS diagnosis by identifying the resources that parents find most informative, resources they dislike for any reason, and those they wish they could have learned about sooner.

Providing helpful resources improves the support a family feels from their health provider, thereby promoting a more positive diagnostic experience. This information will benefit the genetic counseling profession and other healthcare providers by identifying the resources that parents find most informative and helpful to improve the FXS diagnostic experience, as well as patient-provider relationships. Ultimately, this study was an exploration of how parents view resources at diagnosis. It can be used as a point of reference for genetic counselors and other healthcare professionals to begin developing a more comprehensive list of resources to introduce parents to at the time their child is diagnosed with FXS.

CHAPTER 2

ASSESSING PARENTAL SATISFACTION OF CURRENT FRAGILE X SYNDROME RESOURCES PROVIDED AT DIAGNOSIS[1]

[1]Michalski, M., Ferrante, R., McConkie-Rosell, A., & Drazba, K. To be submitted to *Journal of Neurodevelopmental Disorders*.

2.1 Abstract

Diagnosis of a child with fragile X syndrome (FXS) is associated with parental anxiety. Existing literature recommends FXS resources, but there has not been a formal assessment of when parents learn about these resources, or their satisfaction with them. Studies have demonstrated negative feelings surrounding the diagnostic experience of FXS. This exploratory analysis assessed the gap in knowledge on parental satisfaction of the recommended resources from healthcare providers during the time of diagnosis. We predicted parents would not be satisfied with the way resources were presented to them. We discussed parental satisfaction or dissatisfaction, utility of the recommended resources, and determined what resources parents independently identified as valuable for healthcare professionals and genetic counselors to recommend to families in the future.

Seven participants took an online questionnaire containing inclusion criteria and demographic type questions. Four of these participants partook in further focus group discussion (n = 3) or individual interview (n = 1) and discussed FXS resource satisfaction and preferences. The results were analyzed descriptively.

Overall, participants were not satisfied with the information and advice they received from providers at diagnosis. All participants expressed a need for more positive and balanced information about FXS. Parents primarily relied on information and support from national organizations like NFXF and FRAXA, Facebook groups, and national FXS conferences. Two participants were not given any informational or support resources at the time of diagnosis. Participants recommended that professionals educate families about specific resources at the time of diagnosis and provide actionable steps forward in an age-appropriate manner.

This study suggests parents would like the names of specific resources at diagnosis like national organizations, Facebook group names, local resources, and a way to connect with a parent who has already experienced a FXS diagnosis. Parents feel isolation and anxiety during a new diagnosis of their child, so it is important to emphasize positivity and that a group of providers, therapists, and the entire FXS community is there to support the family. Genetic counselors and providers should give age-appropriate recommendations so parents can continue to adjust and adapt to their child's FXS diagnosis.

2.2 Introduction

Fragile X syndrome (FXS) is an X-linked genetic condition characterized by intellectual disability, developmental delay, behavioral issues, and certain physical features. FXS is one of the most common forms of inherited intellectual disability caused by a trinucleotide repeat expansion in the promoter region of the Fragile X Mental Retardation 1 (*FMR1)* gene located at Xq27.3 (Hunter et al., 2019). Normally, *FMR1* contains a cytosine-guanine-guanine (CGG) triplet that repeats 6 to 44 times. The normal allele is stable, unmethylated, and does not cause FXS or *FMR1*-related disorders (Finucane et al., 2012; Hunter et al., 2019; Monaghan et al., 2013). Occasionally, due to polymerase slippage during DNA replication, the expansion may increase during transmission to the next generation into the intermediate or grey zone (45-54 repeats), premutation (55-200 repeats), or eventually into the full mutation (200+ repeats) (Nolin et al., 1996). The intermediate genotype has a risk for expansion to the premutation, but not the full mutation (Nolin et al., 1996). The premutation allele can expand to the full mutation in subsequent generations and is associated with an increased risk of developing

FMR1-related disorders such as Fragile X Primary Ovarian Insufficiency (FXPOI) and Fragile X-Associated Tremor/Ataxia syndrome (FXTAS) (Monaghan et al., 2013). A female premutation carrier almost always transmits an expansion to the next generation, whereas males rarely experience expansion at transmission (Nolin et al., 2003). The *FMR1* gene becomes methylated at the full mutation causing FXS (Hunter et al., 2019).

FXS has a variable clinical phenotype and individuals can range from having learning difficulties to severe intellectual disability and/or autism (Garber et al., 2008). Typically, males experience more severe features while females are more likely to have minor physical characteristics and more mild psychological concerns (Finucane et al., 2012; McConkie-Rosell et al., 2005). Less than 50% of females with a full mutation allele have intellectual disability (Spector et al., 2021). Common physical features include hypotonia, large ears, long face, macroorchidism (in males), hypermobility of joints, as well as cognitive and behavioral features like intellectual disabilities, autism spectrum disorder (ASD), and motor or speech delays (Garber et al., 2008; Hunter et al., 2019; McConkie-Rosell et al., 2005). Ideally, after diagnosis, a patient is referred to a Fragile X clinic that is part of the Fragile X Clinical and Research Consortium (FXCRC) to ensure appropriate multidisciplinary care (Finucane et al., 2012).

Patients primarily rely on healthcare professionals to provide them with credible information about their condition (Genetic Alliance, 2010). Genetic counselors serve as resources about genetic conditions for patients as well as other healthcare professionals (Genetic Alliance, 2010). Education that usually occurs during a results disclosure of FXS includes inheritance pattern, the genetics of the condition, risk assessment for other family members, potential prognosis, management options, and patient resources

(McConkie-Rosell et al., 2005). In a result disclosure of FXS, the parent can be emotionally overwhelmed due to their child's diagnosis and the large amount of information introduced and reviewed (Lebmann & Sarimski, 2013). Parents expect the healthcare professional to understand their emotional distress at the time of diagnosis and deliver advice and information about support, treatment, and next steps (Lebmann & Sarimski, 2013). Genetic counselors can help support the family by gathering relevant local, national, and online support groups and resources (Goodwin et al., 2015).

A study found that parents of children affected with 22q11.2 deletion syndrome, FXS, and Down syndrome generally report having a negative experience when receiving the diagnosis because healthcare providers show a lack of support and provide unsatisfactory quantity and quality of information on the condition (Goodwin et al., 2015). A parent experiences a diagnosis more positively when information is presented in a calm and supportive manner (Goodwin et al., 2015). Lebmann and Sarimski (2013) found that 58.4% of parents stated they desired more information about the disability, and 52.8% wanted more emotional support from the professional during the diagnosis of Fragile X Syndrome. These researchers also found that 81.3% of parents consider support groups to be important following a FXS diagnosis. Parents may cope and manage their reactions to a child's diagnosis through information seeking behavior about the condition as a way to "restore order in a chaotic existence" (Starke & Moller, 2002). When parents have a child with a disability, it is common that a mother will seek out advice from the healthcare provider to help her understand her child's needs, while a father is more likely to manage using resources such as medical literature or books (Starke & Moller, 2002).

It is important that the healthcare professional is well informed on the best resources available. Resources can be printed information pamphlets, electronic websites, information about patient advocacy groups, books, or even national organizations (Genetic Alliance, 2010; Uhlmann et al., 2010). Patients generally prefer written materials such as brochures, pamphlets, and books over websites (Genetic Alliance, 2010). Mac et al. (2020) stated that online resources are often written above the recommended reading level and can be perceived as more difficult to understand than printed materials. However, recently, social media has become a medium to empower patients, increase a sense of community and participation, and increase research recruitment (Pogue et al., 2018). The complexity of the many resources makes it important that providers are comfortable with the resources they recommend to patients. Uhlmann et al. (2010) warns that not all advocacy and support groups benefit each family in the same way, so healthcare providers must assess every family individually to determine which group(s) to introduce a family to.

McConkie-Rosell et al. (2005) recommended resources such as National Fragile X Foundation, FRAXA Research Foundation, family-oriented books and articles, and other online resources. The National Fragile X Foundation provides informational resources, current research studies, and locations of Fragile X clinics. They also maintain support groups and Facebook pages (Finucane et al., 2012). FRAXA Research Foundation is dedicated to FXS research and informing about medical treatment and education strategies for children with FXS. Additionally, the book "Children with Fragile X Syndrome: A Parent's Guide" by Jayne Dixon Weber can be useful for families. A combination of resources, a strong healthcare/social team, and personal coping strategies

help make a successful adjustment to a new diagnosis in the family (Lebmann & Sarimski, 2013).

There has been little research conducted to assess the feelings and satisfaction parents have of resources that exist for FXS. There is no literature to our knowledge that assesses how parents of children with FXS feel about the resources they are provided when their child is newly diagnosed with the condition. Existing literature (i.e. Finucane et al., 2012; McConkie-Rossel et al., 2005) recommends resources but there has not been an assessment of when parents learn about these resources or their satisfaction with the resources that healthcare professionals provide. Lebmann and Sarimski (2013) found historically negative feelings surrounding the diagnostic experience of FXS. It is important that parents find support when their child is diagnosed with a chronic condition, so providing good resources is beneficial to everyone involved. While genetic counselors are not the only professionals involved in a results disclosure, they do play a role in disclosing a new diagnosis and discussing the condition, management, and resources, with the goal of reducing family stress and anxiety (Genetic Alliance, 2010).

The purpose of this exploratory analysis was to assess parental satisfaction with the recommended resources they received at the time of a FXS diagnosis of their child. Providing helpful resources improves the support a family feels from their provider, thereby promoting a more positive diagnostic experience. This information will benefit the genetic counseling profession and other providers by identifying the resources that parents find most informative and helpful to improve the Fragile X diagnostic experience as well as patient-provider relationships.

We predicted that parents would not be fully satisfied with the way resources were presented to them at the time of diagnosis. The parents provided recommendations for the kind of information that is most beneficial to them and explained why certain information is more valued at the time of diagnosis. This study aimed to assess the types of resources healthcare professionals recommend parents review when their child is diagnosed with FXS. The researchers then discussed with parents their satisfaction or dissatisfaction and the utility of the recommended resources. Another goal of this study was to learn which resources parents discovered individually to fill gaps in desired knowledge about FXS. Ultimately, this exploratory analysis studied how parents are exposed to resources at diagnosis. This study can be used as a point of reference for genetic counselors and other healthcare professionals to begin developing a more comprehensive list of resources to introduce parents to at the time their child is diagnosed with FXS.

2.3 Materials and Methods

2.3.1 Participants and Recruitment

This study was approved by the Institutional Review Board at the University of South Carolina in July 2021 (Pro00113410). Participants were over the age of 18 years old and were parents or caretakers of children under the age of 18 years old with a full Fragile X mutation, diagnosed within the past 10 years. Only English-speaking participants were included in this study due to insufficient resources for translation and interpretation. Survey exclusion criteria is detailed in Figure 2.1.

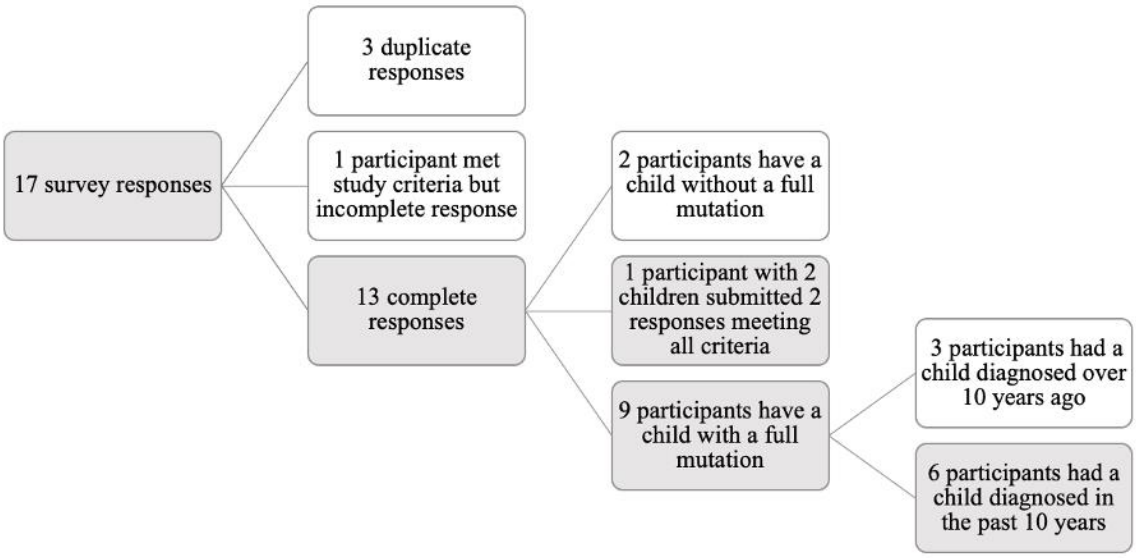

Figure 2.1 Methods by which participant eligibility was stratified. Participants included in the white boxes were excluded from the study due to unmet inclusion criteria and their responses were not incorporated into data analysis.

Of the seven participants who were identified to qualify for a focus group, one did not provide contact information and one respondent was unable to be contacted by email or phone. Both of their survey responses were included in the quantitative portion of data analysis. One participant was identified to not meet full criteria during the focus group, as the diagnosis of FXS was made in adulthood, so her responses were not included in qualitative or quantitative analysis. Ultimately, three qualified participants were scheduled for a focus group discussion and one participant was interviewed individually.

Recruitment for this study was done at both a national level through the National Fragile X Foundation (NFXF) and a local level through the Greenwood Genetic Center (GGC). Both organizations were involved in social media postings alerting their followers about the study (Appendices A and B). The social media posts included a brief description of the study as well as a link to a pre-focus group survey for GGC or to the research portal and pre-focus group survey for NFXF. NFXF posted on their research portal about the study as well as emailed an advertisement for the study in a research

round up to parents of children with FXS (Appendix C). GGC additionally posted advertisement flyers in their offices (Appendix D). Participation was voluntary, and those who participated in the online survey had the option to opt in to a further focus group discussion. The participants also had an opportunity to be entered into a raffle at the end of the focus group and interview for a $25 Amazon gift card.

2.3.2 Materials and Measures

This exploratory analysis consisted of a self-applied online questionnaire and a subsequent focus group or interview conducted by the primary researcher via a phone call or video conference. Both the online questionnaire and the focus group discussion questions were created by the primary researcher and reviewed by the research committee for content, clarity, and accuracy. The focus group discussion questions were created with the intent to answer study objectives. Demographic questions were developed by researching other surveys and modeling the questions in a similar fashion. The online questionnaire was developed through Qualtrics and incorporated skip logic to tailor the questions to each participant to ensure they met study requirements. The questionnaire contained items about inclusion criteria, when and where parents received their child's diagnosis, *FMR1* mutation status, contact information, and demographics (Appendix E). Answers were structured in multiple choice, select all that apply, and open-ended text entry. Participants were contacted via telephone to obtain an email, or via email with a Doodle Poll questionnaire to select multiple times that fit in their schedule to participate in the focus group. If a participant was unable to participate in the scheduled focus group, they were offered a phone interview option.

The focus group was conducted over a video conference meeting protected by a passcode. The individual interview was conducted via telephone. Both the focus group and interview used the same set of questions and were conducted by the primary researcher. See Appendix F for the focus group script and questions and Appendix G for the interview script and questions. Participants were asked nine overarching questions, with supplementary questions asked when appropriate to elicit further responses about participant background, types of resources they were exposed to at the time of diagnosis, satisfaction or dissatisfaction with these resources, the kind of information and resources they looked for near their child's diagnosis, and if there were gaps in the information parents desired.

2.3.3 Procedures

From the initial recruitment flyer or research posting, participants were provided the link to the survey or could directly click the link for the survey on the electronic post. The first page of the survey outlined the specific details of the project and provided information about the survey and subsequent focus group. Participants needed to consent to proceed to the rest of the survey. Participants were able to leave the questionnaire at any time and could skip any question. If a participant did not answer a question that determined their eligibility, they were prompted to answer, but were still able to skip the question. Completing the survey served as consent to be contacted about participating in the focus group.

Due to an error in the survey logic, some participants were unable to complete the full survey, but were still able to leave a telephone number after completing the eligibility questions. In the instances where these participants met criteria, the researcher called

these individuals to gauge their interest and requested a second completion of the survey, then proceeded with the interview/focus group scheduling. This resulted in three partially complete and duplicated responses which were eliminated from data analysis. The researcher was unable to contact one qualified participant, so the incomplete response was not included in the analysis.

The primary investigator contacted volunteers via email with a DoodlePoll questionnaire to schedule the focus group. Those who could not schedule a focus group participated in a one-on-one interview. A week before the focus group and interview, participants were provided the discussion questions. At the beginning of the focus group and interview, verbal consent was obtained to record the meeting. The phone interview was recorded with Apple Simple Recorder and the Zoom meeting was recorded via Zoom. Both were saved to the primary investigator's password-protected laptop and were transcribed verbatim. All identifying information was removed from transcriptions to protect participant identity. Audio and video recordings were destroyed upon completion of transcription. Participants were emailed after the interview or focus group with links to resources that were discussed for their future reference as well as information about other FXS resources that were compiled by the researcher (Appendix H). Data was collected from July 2021 to December 2021. One participant was selected by lottery for the Amazon gift card, and was emailed the redemption code.

2.3.4 Data Analysis

In order to meet the objectives of the study, the data collected required quantitative and qualitative analysis. The pre-focus group survey was analyzed quantitatively in Excel. Much of the data from this pre-focus group survey was

categorical, so percentages and frequencies were calculated. Duplicated or partially completed survey responses were discarded from final analysis. The focus group and interview responses were jointly analyzed through a descriptive model for each discussion question. Each transcript was analyzed and used to describe what participants agreed on and where participants had contradictory viewpoints.

2.4 Results

2.4.1 Demographics

Of the initial responses to the survey, seven responses submitted by six eligible individuals were analyzed. The sample population consisted of all mothers (n = 6, 100%) who identified as females and as the primary caregiver of their child with FXS. The most common racial and ethnic identity was Caucasian/White (n = 5, 83.33%) and one participant (n = 1, 16.6%) identified as Hispanic/Latino. There were three participants (50%) who had completed a Master's or Doctoral degree, two participants (33.33%) with a Bachelor's degree, and one participant (16.67%) who had some college education. All six participants (100%) had a son with FXS, and two participants (33.33%) also had a daughter with a full fragile X mutation who was symptomatic. As mentioned earlier, one participant took the survey twice because she has two affected children, a son and a daughter, and during the focus group, one participant shared that she also has an affected son and daughter.

2.4.2 Fragile X Syndrome Diagnosis

Every participant (n = 6, 100%) responded that they were present at the time their child was diagnosed with FXS. Based on survey responses, the average age at diagnosis was approximately 1.67 years, and participants learned the diagnosis of their child an

average of 2.67 years ago. The age range of the affected children was between four months old and seven years old, and the age at diagnosis ranged from the first few weeks of life to about two and a half years old. The majority of participants (n = 4, 66.67%) had never visited a specialized fragile X clinic, and two participants (33.33%) reported that they had been seen at a fragile X clinic.

The details of the type of provider who gave the participants their child's diagnosis, as well as which healthcare professional discussed resources with the family, is presented in Figure 2.2. The survey did not distinguish if the resources needed to be discussed in any specific time frame, but asked which healthcare professionals had ever discussed resources with the parents. All six participants had spoken with a healthcare professional about FXS resources. No participant selected that a psychologist, special educator, or educator provided them with a diagnosis or resources for their child. One participant selected that they had received resources additionally from a board-certified behavioral analyst (BCBA).

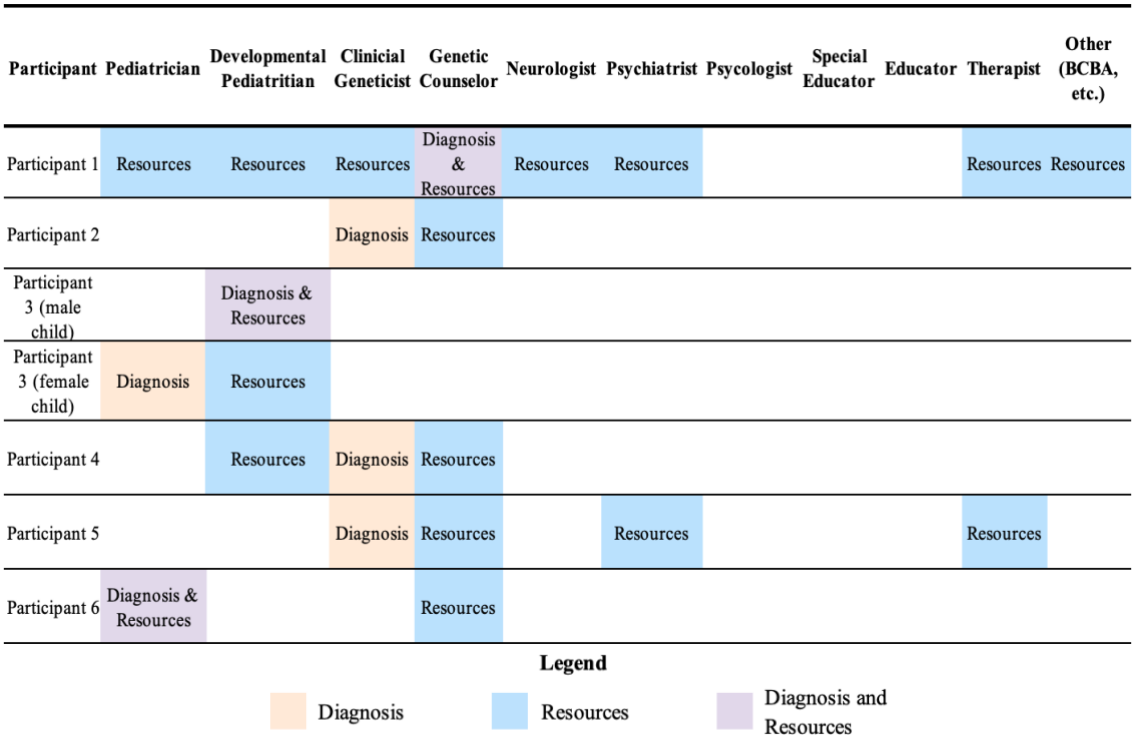

Figure 2.2 Highlights which healthcare provider discussed resources with the participant, provided a diagnosis, or discussed resources and provided a diagnosis.

2.4.3 Qualitative Analysis

For reference, Participant 3 was interviewed one-on-one, and Participant 2, Participant 5, and Participant 6 were in a focus group discussion together. Participants 1 and 4 are not included in the following analysis as they completed the survey but did not participate in an interview or focus group.

2.4.3.1 Introductions and discussion of how a diagnosis was received. The four interviewed participants were the first in their family to learn about FXS and did not have prior experience with FXS. There were two participants with two affected children, three of the four saw a genetic counselor after diagnosis, and there was a range of ages at diagnosis, as mentioned previously. Participant 3 had two affected children, an older son who was diagnosed first, and younger daughter who also was symptomatic and has a full

mutation. Participant 3 was the only participant to not see a genetic counselor after diagnosis of her children.

Participant 2 had two affected children, a younger son and older daughter. Her son was diagnosed first at eight months of age after failing to meet appropriate milestones; he was referred to genetic counseling by an early intervention specialist. Testing found a mosaic normal allele and full mutation allele in the son, and a geneticist recommended FXS testing for the daughter who was about two and a half years old at the time. Participant 2's daughter was diagnosed with a full FXS mutation but was not mosaic. Regarding her son's diagnosis, she stated: "luckily, it was very early on, on so we had the ability to have lots of resources."

Participant 6 reported that she has an affected son who is four months old, who was diagnosed at six weeks after postnatal testing following her diagnosis as a premutation carrier during pregnancy. Due to concern for early onset ovarian insufficiency following struggles with infertility, the participant was tested for FXS during her pregnancy. She received prenatal genetic counseling about the 50% risk of having an affected son. Her son was tested for FXS at birth, and the pediatrician who disclosed the test results confirming the FXS diagnosis referred her to a genetics team for more information.

- "It was such a devastating appointment. My husband ended up in the hospital with suicidal thoughts and um I had to go on antidepressants because of the things this uh genetic counseling team told us about our child…It was very much like this was something that I uh did wrong as a parent, and that I should of chose.

Basically, told me I should have terminated my pregnancy once I found out I was a carrier." – Participant 6

Participant 5 found out her son had FXS when he was three weeks old from a state newborn screening research initiative which offered screening for spinal muscular atrophy and FXS in a research setting. The state contacted the family through a phone call to disclose the positive result and then they followed up with a genetic counselor and research team at a university.

2.4.3.2 Resources on FXS that healthcare professionals recommended parents at diagnosis. Participants were recommended a few different methods of learning more about FXS and how to care for their child. Two participants were not given much information and told to do their own research online. Of those two individuals, one mother was informed about medical assistance programs.

- "We didn't get emails. We didn't get pamphlets; we didn't get resources like we got literally not it was like just go out and figure it out." – Participant 6
- "Okay basically they just said look into the local groups and look online on Facebook and that was it." – Participant 3

Participant 2 was given multiple handouts, pamphlets, and national organization names and referred to a FXS clinic.

- "A lot of like, business cards and pamphlets. It may be. I think at that point, the fragile X Foundation had been like the beginner's guide to fragile X. Possibly was like maybe 30-page handout kind of document that I was handed or maybe emailed the PDF." – Participant 2

- "There's a fragile X clinic in Cleveland that's less than an hour from me. So we did, I would say meet with that position within probably six weeks to two months." – Participant 2

Participant 5 was told to only research through reputable sites like university pages and national organizations. No participant had been given any books or support group contacts at the time of their child's diagnosis.

- "We were told by the genetic counselor, to actually stay off Google and not Google it and to only reference that specific website that I was talking about and use those documents and use that website to learn more." Participant 5

Participant 5 also stated that her son is being followed in a three-year long research initiative, but did not disclose the university or the specific website she mentioned in the above quote, but described it:

- "…they had like different documents like PDFs where it is. They were very specific as to here's a document if you want to talk to your child's pediatrician or doctor about their diagnosis, and then there was another one that was specifically like, here's how to tell your family members about your child and their diagnosis, and like how it could also potentially affect them…" – Participant 5

Participant 6 met with a genetics team after diagnosis to learn more about FXS and future implications. A genetic counselor discussed FXS with her.

- "The only thing we actually, saw was this PowerPoint that was written like with a medical angle and not being in a medical profession. Like it was basically just like not, it was just not helpful" – Participant 6

Participants were also asked to expand on their satisfaction or dissatisfaction and thoughts on the resources they were recommended at diagnosis. Some responses were:

- "Totally unsatisfied, ya I had to do a lot of research myself. It was by talking to other people, and letting uh them know of my son's diagnosis was, that I was connected to other parents in the area who had kids with Fragile X" - Participant 3
- "It wasn't very personalized, it was just like take this and read it, take this and read it" – Participant 2

2.4.3.3 Initial information parents most wanted to learn about after diagnosis. Half of the participants in this study stated they wanted more information about their child's developmental potential and prognosis, and what they should do moving forward from a diagnosis. Two participants also discussed the importance of learning about medical implications, features of FXS, and their varied abilities, while some were concerned about long term care and how they should prepare for the future.

- "Like what to expect for the long haul, like was you know, are there kids who can live on their own like [son's name], specifically [son's name], because boys are more affected than girls. What is the trajectory in terms of his like potential and like what resources can I get, and hit the ground running to have him reach you know the potential that he's meant to reach." – Participant 3
- "Your son or child has this diagnosis. Here's what you should do. These are the steps that you should take going forward." – Participant 5
- "I was hysterical and I just said, you know, should I be saving money for medical funds? Because that's what it sounds like. Because right now we have a college fund." – Participant 6

- "What do I need to do for his future, especially at the point where I'm no longer here, and my husband is no longer here to care for him. What happens to him, then?" – Participant 5
- "One thing I'm going to add but maybe a few of the medical things that show up along the way." – Participant 2
- "I remember asking for like a template, like if it's truly is a spectrum, like what are the phases of the spectrum" – Participant 6
- "I want to know hey, if they have verbal communication, or if they have moderate like, fine motor skills, like what are those different layers to what that spectrum means."– Participant 6

Participant 3 expressed that the information she wanted to learn, including long-term prognosis, necessary therapies, and future concerns, were the same in regards to her affected son and daughter.

2.4.3.4 Resources on FXS that parents found after their child's diagnosis. The participants found information on national organization webpages, like NFXF and FRAXA, through internet-based searches. Two participants found information about ongoing research on the NFXF website as well as through other organizations. All four participants found active Facebook groups to join which allowed them to connect with other parents online. Two participants also found more resources relating to therapies and intervention services for their children.

- "…getting information from the Fragile X Foundation about studies that are available for your children. And for yourself, like, if obviously, you know, if your

children have it, someone is a carrier. So having the resources for the carrier also, like, I was able to educate myself through those web pages." – Participant 3

- "I found a lot of helpful information in the National fragile X website…" – Participant 5
- "I dig and connected with a social worker, who did help us get into the birth to three program. But locally, there's there's no real community of fragile X parents here in Wisconsin. There's a branch in Milwaukee through the National Fragile X Foundation, but the woman who runs it, this is her first year, so the community is she she's trying to build a community now … like to me, the only thing I've really been able to use is the Facebook page." – Participant 6
- "The Facebook groups, the specialists, the clinics we have done a lot of research, like I mentioned. Stanford has a girl band study that we're a part of. Cincinnati Children's, Dr. Erickson is one of the biggest, and so everything is under his care, so we get in a lot of research with that." – Participant 2
- "…how to adapt ABA for fragile X kids that I was able to send to, like our therapist … I Googled it and it came up. But also, I called the fragile X Foundation" – Participant 3

Participant 3 expressed that being able to communicate with other parents taught her what kinds of behaviors to expect and how to manage various behavioral differences or challenges that her children may have.

- "I found like just talking to people who've been where I am or people like in the area… who are going through the same things and their kids are the same age as mine …I think that like, we've just been a resource for each other." – Participant 3

One mother had mixed feelings about Facebook groups:

- "The great thing about it is that obviously there's a lot of parents and everyone's really loving and positive. The bad thing about is you see everything… like because it's a spectrum, you don't know where your kid's going to fall, and some of it is really overwhelming." – Participant 6

Participant 3 found specific Facebook groups related to clinical phenotype:

- "There are one's I'm a member of. There's a, there's a fragile X group. There's a fragile X syndrome group. There's fragile X female carrier symptoms group. Ummm, there's just a lot like, a lot of groups actually start with fragile X. There's a fragile X advocacy group" – Participant 3

Participant 2 expressed the importance of finding the FXS resources by stating:

- "Once you get to know the people that are big in the fragile X world, that feel like you have a lot more hope, and a lot more of the positive resources" - Participant 2

2.4.3.5 FXS resources parents wished they discovered or had access to sooner following FXS diagnosis. Participants described the importance of having support groups, Facebook groups, and information on current research from the time of diagnosis, along with more information on medical complications that children with FXS may be at-risk for.

- "Maybe a few of the medical things that show up along the way. So, like with my son was when he was feeding as a young kid, he threw up a lot… the tonsils and adenoids are often an issue in fragile X kids… so that would be something if it was in the research would be very helpful." – Participant 2

Additionally, Participant 6 described wanting more balanced and positive information “Just highlighting the positives. So, like I'm, I'm desperate for someone telling me something good.” Participant 3 discussed that she would have liked specific suggestions to join certain Facebook groups or make connections with local FXS groups. She added:

- “My husband and I walked out of that appointment and being like, okay, we don't know what fragile X is and now we're just supposed to look on Facebook groups online? And like where's the benefit in that? And like what can we do? And what studies are there? And I just felt like we were lost.” – Participant 3

2.4.3.6 Topics parents identified as lacking resources or information.

Participants expressed a desire for more resources related to educational needs of children with FXS, girls with FXS transitioning to adulthood, possible medical complications, as well as managing the behaviors of a child with FXS. Additionally, parents wanted more ways to connect with parents of older children to gain guidance from them and specific information on current research progress.

- “I would love to know more about the best learning programs every year. We're looking at what's the best kind of math to teach them? What's the best kind of reading program around to teach them?” – Participant 2
- “So yes, as much information, as the be put together for school districts, especially related to writing IEPs” – Participant 2
- “For people who have fragile X, and who are, you know, functioning enough to have relationships like, for my daughter. I'm very like hopeful that when she gets older, like what are the resources for her to have kids?” – Participant 3

- “I think just like, where are we in finding a cure for this? Is that like even a possibility in my kid's lifetime?” – Participant 3
- “I don't think anything gave me any usable tools and how to deal with a child for fragile X…doesn't talk about stimming or like, how you deal with a kid who's stimming or having food issues…” – Participant 3
- “So yeah, more of maybe an advocacy be helpful if they could have parent mentors or something to be available to answer specific questions” – Participant 2
- “Like is there a way you're connecting all the parents so that you know, we can all like, you know, talk and you know, share ideas and like others have mentioned, you know, maybe they're further along, you know, their children are older, you know, maybe they can shed some light and experience” – Participant 5

2.4.3.7 Outdated, inaccurate, or confusing resources. No participant specifically mentioned any inaccurate or otherwise confusing or outdated resources that they would not recommend. Participant 3 discussed that she thinks that resources need to be more “easily searchable” but she specifically said “No I haven’t found anything that’s fairly outdated or anything.” Participant 5 said “Not really” when asked the same question. Additionally, Participant 2 said:

- “Some of them are just long. So if we make abstracts out of some of them, like…I can't hand a teacher a 30 page PDF and be like, read about fragile X, like, possibly, maybe a power point, or some sort of slide deck will be more timely for a teacher.” – Participant 2

2.4.3.8 Additional information parents wanted to discuss about resources.

Two participants discussed the frequency that they have to educate medical professionals and teachers about FXS.

- "I know that there're obviously are a lot of genetic diseases, but I was just very um surprised with the lack of knowledge that I found from general practitioners." - Participant 3
- "Every time we need a teacher, like 'I think I read that in a textbook.' Like it's just it's not that common and I don't expect people to know it, but we're constantly teaching and learning" – Participant 2

The researcher asked participants at this point about two specific resources to gauge familiarity: a website called Positive Exposure (positiveexposure.org), and the book "Children with Fragile X Syndrome: A Parent's Guide" by Jayne Dixon Weber. Both were briefly overviewed when describing them to participants. All four participants expressed interest in these resources but had never come across them. They requested the primary researcher send the website link and title of the book to them after each meeting.

2.4.3.9 Types of information, readings, and resources parents would provide a newly diagnosed family. This discussion point elicited many detailed responses from participants. Participants emphasized the importance of discussing how FXS is a spectrum, connecting parents to support groups and Facebook groups, and providing information to national organizations to improve diagnostic experience.

- "I want to know that it's a spectrum and that there are, you know, parent networks available" – Participant 3

- "The National fragile X website, and then also that State University, that was doing the research that initially diagnosed my son" – Participant 5
- "Have written down in the paper that they give you after your appointment like the local groups or the other, you know, fragile X Foundation websites..." – Participant 3
- "I would have liked her to give me more resources, said join this Facebook group …or we have this is the person in [city] that you should talk to." – Participant 3

Participant 2 explained the value of educating new parents about fragile X conferences, and Participant 6 explained how creating a supportive team decreases feelings of isolation.

- "My husband and I were able to go to one of the about the national conferences…that was kind of in a way an eye-opening experience because literally were put in a room where you saw the severity and saw the positive…we got to sit in the room and talk with individuals that had been diagnosed, and maybe their sibling been diagnosed, and I think that was probably one of the best experiences" – Participant 2
- "A lot of people from what I've heard have like, really found some amazing resources and friendships from going to those conferences" – Participant 3
- "It's yes, you feel alone now, and yes, you're unbelievably sad. And we're going to give you time to grieve, but here's like a team of people waiting for you to like help you pick yourself up and get going again" – Participant 6

2.4.3.10 Positivity and Empathy. Throughout these discussions, three of the participants said they would like more positivity and balanced information at the time of

diagnosis. Additionally, the manner in which providers discuss FXS with patients is important; in short: empathy is valued.

- "Can you just explain and just tell us what this means for his future and eventually they just told us the best we can hope for was that he could be a bagger at a grocery store? So, I just I wish they would get more sensitivity training when delivering the news and what that means for their futures rather than giving such a doom and gloom type of answer" – Participant 5
- "There's not a lot of positive stories out there. So, it's kind of It's very daunting to look ahead" – Participant 6
- "…once you turn into a teenager, he's going to revolt he's going to be so aggressive. You should probably start thinking about where his future placement would be. Yeah. I definitely agree that they need to spend more on the positive in the potentials instead of the what you're not going to reach…" – Participant 2

One participant highlighted how she appreciated people showing an interest in her child and stated the people who work at NFXF are good people.

- "The foundation for fragile X like get back to you right away and they really care, um and they take it personally" – Participant 3

Participant 2, who had children which were older than the other participants, expressed:

- "There is a lot to look forward to. They certainly the personalities are so funny. They're so loving." – Participant 2

2.4.3.11 FXS Age-Related Information. Participants 2 and 6 shared that information that was stratified by age group, and developmental levels should be

more accessible to parents to help families plan and adjust to their child as they grow.

- "Some of the Facebook you look at the general fragile X, you have some people talking about 55-year-old some people talking about 22 year olds and some of brand new infants" – Participant 2
- "Let's do a fragile x like one to three group, a fragile x three to eight group, like, kind of break it down into like let's put us into groups." – Participant 2

Additionally, this kind of information can help assess a child's progress and help create new benchmarks.

- "I feel like if I had a better sense of fragile X kids typically where learned to read by 5, and then you're like you're making benchmarks based on the actual peer set at versus, just the kids who happen to have been born around the same time as him." – Participant 6
- "So, if we could kind of chunk it out into developmental stages, whatever that might look like. I think that might help our planning personalities a little bit to just think in life stages" – Participant 2
- "I think that would be really helpful for me to understand because then I know what to advocate for and what to push back against when people tell me like, hey, he's not doing well or he is" – Participant 6

2.5 Discussion

This exploratory analysis assessed the satisfaction of parents on the types of resources they were recommended by various healthcare professionals when their child was diagnosed with FXS. There have already been studies which demonstrate the

importance of informational and support resources to families in the time of a new diagnosis (Goodwin et al., 2015; Lebmann & Sarimski, 2013; Stark & Moller, 2002). Although some research promotes various resources for families during a diagnostic experience, and some studies recommend FXS specific resources, there has not been a study which assessed parental opinions of the existing and recommended resources for FXS (Finucane et al., 2012; Genetic Alliance, 2010; McConkie-Rosell et al., 2005). The purpose of this study was to begin to understand the types of resources parents desire at the time of diagnosis to improve patient-provider relationships and overall diagnostic experience of families affected by FXS. We predicted that the parents would not be satisfied with the resources received at the time of diagnosis and that parents are seeking additional resources independently after a diagnosis to fill a need for more information and support.

The information revealed in this study supports previous literature discussing how parents are often unsatisfied with the initial diagnostic appointment at least in part because they did not receive appropriate provider support or resources (Goodwin et al., 2015). The information and resources that were most strongly desired at the time of diagnosis were support resources like local parent groups and Facebook groups, as well as actionable steps for parents to take with their child moving forward. Another common sentiment was that more information needs to be provided to participants in an age appropriate, supportive, and empathetic manner, and more emphasis needs to be placed on balanced information. These findings align with studies like Lebmann and Sarimski (2013) which reported that parents expect a supportive approach from their healthcare

professional during a diagnosis and anticipate gaining informational resources and advice for next steps in the appointment.

The participants experienced differing initial exposure to resources at the time of diagnosis. Participant 2 who was provided with the most informational materials and a referral to a fragile X clinic after diagnosis overall expressed more positivity about the time of diagnosis than those participants who were directed to search online for resources or provided with limited guidance on future directions. Participant 2 received information that other studies have proposed as valuable resources within the FXS community such as the NFXF and family-oriented articles (McConkie-Roselle et al., 2005). Additionally, the parents who were told to search online for more information then also expressed in later discussion that they wished the healthcare professionals would have provided the names of the best resources on FXS during the diagnosis. This finding supports other literature which demonstrates that patients generally rely on their healthcare professionals to provide information and resources which promotes a more positive diagnostic experience (Genetic Alliance, 2010; Goodwin et al., 2015; Lebmann & Sarimski, 2013). Genetic counselors are trained in providing information and community and support resources to patients and utilize these skills in aiding patients through a genetic diagnosis (Genetic Alliance, 2009).

The participants in this study were newly diagnosed families with little to no prior knowledge of FXS. Therefore, it is important to remember that the informational needs of these parents may be different from those where there was a previous family history of FXS. Families without prior knowledge of FXS may require more detailed information and basic resources at the time of their child's diagnosis compared to patients who are

aware of their risk for years ahead of bearing children. A study which assessed the communication of genetic risk in adolescent girls with FXS found that many participants prefer genetic counselors and other healthcare professionals to communicate information about FXS and genetic risk in an open and honest discussion that is presented in a positive manner (McConkie-Rosell et al., 2009). Although our participants were not as prepared for a diagnosis as the girls and women in the McConkie-Rosell et al. (2009) study, participants in our study agreed that a method of providing positive information and reassurance to the family is important.

The suggestions from participants of this study support the findings of previous research that organizations like NFXF, FRAXA, as well as Facebook groups have been identified as the most beneficial informational and support resources for families (Finucane et al., 2012; McConkie-Rosell et al., 2005). The mothers who were in this study also relayed that the national organizations are critical resources at the time of diagnosis, and two mothers expressed the importance of educating future families about ongoing research and national conferences. These various resources were often discovered by participants in their own research. Two of the participants also discussed how learning about the possible medical complications of FXS or how to adapt therapies like applied behavior analysis to children with FXS is something they had to search for on their own. Additionally, emphasis needs to be placed by genetic counselors and other healthcare professionals that FXS is a spectrum, and some children will be more affected than others, but there are still resources available to meet your child's developmental needs.

All of the participants recommended Facebook groups as a method of joining the FXS community and emphasized the groups are a principal component of their resource and support network. This supports the findings of Lebmann and Sarimski (2013) stating that 81.3% of parents consider support groups to be important following a FXS diagnosis. One of the participants in this study specifically identified the various groups she has found to be beneficial and would want providers to introduce to newly diagnosed families. Pogue et al. (2018) similarly found that social media has been increasing the sense of community for patients and families affected by various conditions. One of the participants explained how social media as a resource can occasionally be overwhelming. This sentiment demonstrates how genetic counselors and other providers should assess the needs of the patient when providing resources and may need to provide qualifications about resources or choose to not disclose certain resources until a later appointment (Uhlmann et al., 2010).

This study identified areas where parents were unable to locate the resources they desired. Three of the participants explained that they would like more accessibility to connect and talk with parents who have older children who had already gone through the diagnosis; these parents could act as mentors for newly diagnosed parents. One mother wanted more information on her female daughter transitioning into adulthood and available resources for females addressing sexuality, relationships, and possible resources for having biological children. The desires of the parents when viewed holistically show that parents are looking for age-specific resources for their children. Two participants specifically mentioned having resources broken up to address the needs of different age groups would be helpful. It can be noted that there are already resources that exist for

these topics of interest on the NFXF website and in books such as “Children with Fragile X Syndrome: A Parent’s Guide” by Jayne Dixon Weber; however, none of the participants were familiar with the book. It was not explored if participants are not aware of the resources on the NFXF website about age related guidance, transitioning to adulthood, education, and methods to connect with other parents, or if they were not finding these resources satisfactory for their needs. The responses of the participants support prior research showing that parents require the help of their healthcare professional to guide them down appropriate avenues of information and to help families establish strong support networks (Genetic Alliance, 2010; Goodwin et al., 2015; Lebmann & Sarimski, 2013).

Multiple participants described the time of diagnosis as “shocking”, “devastating”, or “unbelievably sad” which led to a large discussion in the interview and focus group about the importance of provider empathy and positivity. Genetic counselors and healthcare providers should strive to present information about a diagnosis in a supportive manner to enhance the diagnostic experience (Goodwin et al., 2015). Many participants in this study expressed they wanted more hope to come across from the providers, more positive language, balanced negative aspects of the condition with strengths of people with FXS, and support. One study showed that parents expect healthcare professionals to deliver advice and information about a diagnosis with an understanding of the great emotional distress that parents are feeling (Lebmann & Sarimski, 2013).

The results of this study support previous guidance on the factors that make informational resources patient appropriate. Davis et al. (2010) at the Center of Disease

Control (CDC) created a document titled “Simply Put” about creating patient education materials. It highlights the importance of encouraging and supporting your audience, providing the audience with action items or next steps to take, and limiting scientific jargon (Davis et al., 2010). The respondents in this study reflected these principals in their answers. Parents stressed positivity and empathy from the healthcare provider, the importance of providing direction after diagnosis, and how parent-friendly materials aid in comprehension and resource satisfaction. Other resources on developing patient education materials also stress the importance of focusing on actions the patient can take, using “plain language”, personalizing educational materials, and the value of providers organizing their educational materials so they are easily accessible to provide patients with resources at the time of diagnosis (Guidelines for developing patient education materials, 2017; Use Health Education material effectively: Tool #12, 2020). This study supports these previous guidelines on resources; it found that parents feel the provision of resources needs to be more personalized, comprehensive, and accessible.

2.5.1 Practice Implications

Participants expressed that it is most beneficial for genetic counselors and other healthcare professionals involved in giving a diagnosis to provide balanced and supportive information. Parents feel isolation and anxiety during a new diagnosis of their child and it is the role of the genetic counselor and other providers to appropriately educate a family about FXS, and also help the family establish a strong resource network. Genetic counselors are uniquely trained in providing information in an easy-to-understand manner and often are the healthcare professionals who offer and provide families with resources. It is important to emphasize that there is a group of providers,

therapists, and an entire FXS community that is there to support the family. Based on the results of this study, it seems that parents would like the names of specific resources like organization names, specific Facebook group names, information about local support groups, or providers with expertise in FXS, and if possible, a way to connect with a parent who has already experienced a FXS diagnosis. Multiple parents also expressed the need-to-know action steps on how to move forward from a diagnosis, so genetic counselors and providers can remember to give age-appropriate recommendations so parents can adjust to the diagnosis. Genetic counselors and providers should be prepared to provide access to resources like the ones listed above, as well as books on FXS or other regional resources, for future diagnoses of FXS. Additionally, participants did request information that can be found on web pages such as the NFXF, so it might be necessary to reassess how accessible these resources are in order to ease the usability of these sites for parents/guardians.

2.5.2 Limitations and Future Investigation

The findings from this study are limited due to the small sample size (n = 4). The sample was not representative of all FXS parents as it was primarily composed of Caucasian females; only one participant identified as part of a minority group, and no males participated in an interview or focus group discussion. Findings from this exploratory study can be used in a future study inclusive of a larger, more diverse population. Study participation could be enhanced by extending time for recruitment and expanding recruitment avenues possibly through FXS Facebook groups.

Recall bias could have influenced the results of this study as participants were asked to reflect on a time in their life that was both stressful and shocking. Specific

questions asked parents to recall resources provided at diagnosis versus ones they discovered individually which could lead to some participants unintentionally confusing the resources they were provided at diagnosis with ones they identified online themselves and vice versa. Additionally, participants self-selected for this study so it is possible that the participants experienced a more positive or negative diagnosis experience compared to the typical parent.

Another limitation of this study was the error in the logic of the survey. Future work should aim to correct this error thereby allowing more participants to initially reach the end of the survey and prevent complications in contacting participants to begin the survey again ahead of scheduling them for a focus group or interview.

Future research could also focus on expanding this study to collect more parent experiences and opinions on exposure to resources at the time of diagnosis. If a larger sample is obtained, it would be beneficial to stratify participants in focus groups by sex of the parent or child, age of child at diagnosis, time since diagnosis, prior family history of FXS, or the type of healthcare provider present at diagnosis. Although the sample size was small, each of the mothers who participated in the study did not have experience with FXS prior to their children's diagnosis, so their reflections begin to highlight the needs of newly diagnosed families with no prior family history of FXS. The openness of these families to share their experiences will allow future research to continue to explore parent experiences with resources and provide more guidance on how genetic counselors and other healthcare professionals can work to improve the support parents feel during a new diagnosis. After more information is collected with a broader population, it could be

possible to utilize parent recommendations to develop a comprehensive guide of patient friendly resources about FXS to provide parents when their child is diagnosed with FXS.

CHAPTER 3: CONCLUSIONS

Parents of children recently diagnosed with FXS require more direction and support from the healthcare professionals involved in providing the diagnosis. Participants in this study did not feel satisfied with the information and advice they received from providers and all expressed a need for more positive and balanced information about FXS. The present study also found that parents are primarily relying on information and support from national organizations like NFXF and FRAXA, Facebook groups, and national conferences. These resources are ones that participants would also recommend providers and genetic counselors educate families about when their child is diagnosed with FXS. The study identified that parents would like more opportunities to connect with previously diagnosed families who can serve as friends and mentors during the adjustment to the new diagnosis. If genetic counselors and providers know of other parents or local FXS groups who would be willing to speak with the families, referral or contact information is critical to decrease feelings of isolation. Interestingly, many participants expressed a desire for information to be presented in an age-appropriate manner, but some of this can be found on organization pages like NFXF, so it may be interesting to explore this in further research.

Genetic counselors play a vital role in the diagnosis of children with FXS as they are uniquely trained to provide complex medical and genetic information in a patient-friendly manner and support the patient by providing appropriate resources. It is important that genetic counselors and other healthcare professionals remember that the

time of diagnosis is stressful and parents value balanced information on FXS as well as further guidance about best resources, local support groups, and age-appropriate next steps. This research can be used as an initial exploration of how we can continue to improve future patient experiences, and if expanded to a broader population, can be the background to developing a comprehensive guide for healthcare professionals on FXS resources.

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APPENDIX A: NATIONAL FRAGILE X FOUNDATION SOCIAL MEDIA ADVERTISEMENT

Caption for the below images:

Attention parents of children with Fragile X syndrome: Did you receive the info you needed when your child was diagnosed? What other resources would have been helpful? @USCGCP Genetic counseling student, Megan Michalski's thesis project will help us better meet the needs of families. You input is critical! Click here to participate! @NatlFragileX @fragileXsyndrome

Figure A.1 National Fragile X Foundation social medial photo

APPENDIX B: GREENWOOD GENETIC CENTER SOCIAL MEDIA ADVERTISEMENT

Caption for the below images:

Attention parents of children with Fragile X syndrome: Did you receive the info you needed when your child was diagnosed? What other resources would have been helpful? @USCGCP Genetic counseling student, Megan Michalski's thesis project will help us better meet the needs of families. You input is critical! Click here to participate! @NatlFragileX @fragileXsyndrome

Figure B.1 Greenwood Genetic Center social media photo

APPENDIX C: NATIONAL FRAGILE X FOUNDATION RESEARCH PORTAL POSTING

Focus Group: Parental Satisfaction with Fragile X Resources

Assessing Parental Satisfaction of Current Fragile X Syndrome Resources Provided at Diagnosis

A research study at the University of South Carolina

Why are we doing this research?

University of South Carolina Genetic Counseling Master's Program is conducting a research study to learn about parents' satisfaction with informational resources provided at the time of their child's Fragile X syndrome diagnosis. This will help make recommendations to healthcare professionals of resources parents want to receive at the time their child is diagnosed.

Who can participate?

Parents over 18, with a child diagnosed with Fragile X Syndrome in the past 10 years may be eligible to participate. Parents must read and speak English fluently.

What will happen in the study?

If the individual qualifies and decides to be in this research study, they will complete a less than 5-minute online survey and later participate in a single 60-minute to 90-minute virtual focus group discussion via zoom. The survey may be taken immediately while the focus group discussion might occur up to a couple months later.

The following is a list of some of the study procedures that will happen during the study:

- Complete intake survey to verify you meet the inclusion criteria of the study
- Participate in one 60-to-90-minute virtual focus group discussion to provide opinions about resources you received at the time of your child's diagnosis

What are the good things that can happen from this research?

Participants will receive a single page summary of the discussion they participated in with mentioned resources highlighted for easy reference. It is possible you learn of new resources from other parents that can help you or your child. Each participant will additionally be able to access the final research study to see the results.

What are the bad things that can happen from this research?

During the study, participants will reflect on the time their child was diagnosed with Fragile X syndrome. This can bring up difficult emotions for some. Due to the nature of a focus group, participants do not remain anonymous. We will ask all participants be respectful and not share anyone's identifying information outside of the study.

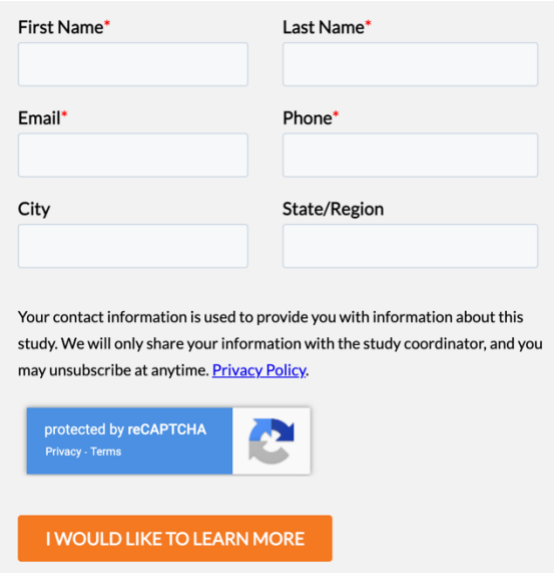
Will I/my child be paid to complete this survey?

At the end of each focus group, a $25 Amazon gift card will be raffled.

How can I participate?

Please complete the form below for access to the intake survey. You must complete the intake survey to be considered for a focus group.

If you have questions please contact Megan Michalski at (843)-345-3071 or Megan.michalski@uscmed.sc.edu



When someone completed the contact form, they saw this message.

Thank you for your interest in this project!

Here is the link to the intake survey. You must complete the intake survey to be considered for a focus group.

Your information has been sent to Megan Michalski. She will contact you. If you have questions or want to reach out directly, you can contact her at (843)-345-3071 or Megan.michalski@uscmed.sc.edu

APPENDIX D: GREENWOOD GENETIC CENTER OFFICE ADVERTISEMET

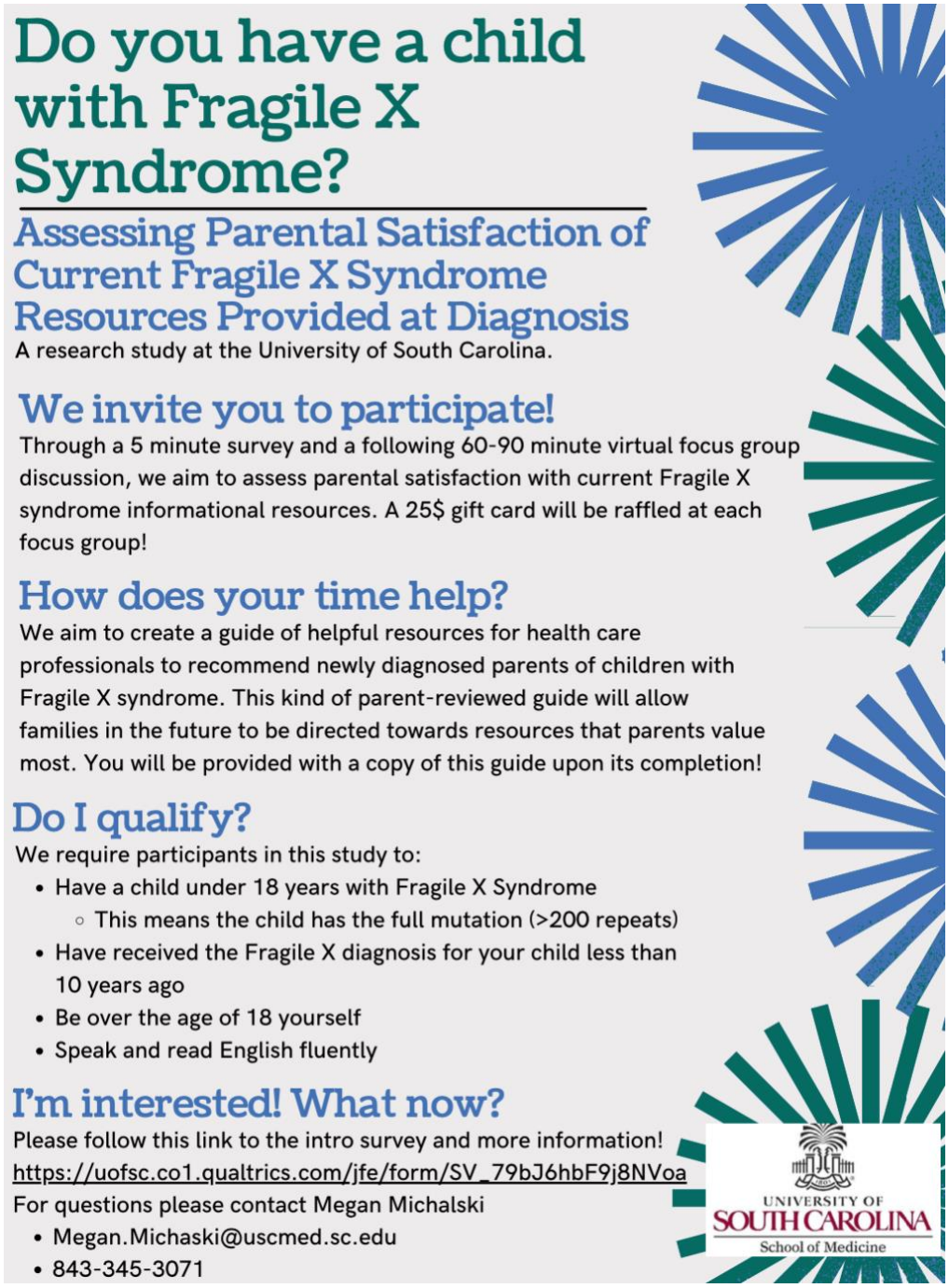


Figure D.1 Recruitment flyer for Greenwood Genetic Center

APPENDIX E: STUDY QUESTIONNAIRE

Start of Block: Default Question Block

Q1 Dear Potential Participant,

You are invited to participate in a graduate research study focusing on parent satisfaction with informational resources provided near the time of a Fragile X diagnosis in your child. I am a graduate student in the genetic counseling program at the University of South Carolina School of Medicine. Through an intro survey and following focus group discussion, we hope to obtain feedback about the needs of parents at time of diagnosis of their child with Fragile X in order to create a guide for healthcare professionals.

This kind of parent-reviewed guide will allow families in the future to be directed towards resources that parents value most when their child is diagnosed with Fragile X syndrome. This virtual focus group will allow providers to take into consideration the preferences of parents when receiving a Fragile X diagnosis. We hope this will improve diagnostic experiences for parents.

This less than 5-minute survey gathers information to determine if you meet the inclusion criteria for the focus group and information about your child's diagnosis of Fragile X syndrome and resources you were directed to. Questions marked with "requested answer" specifically relate to inclusion criteria. We kindly request you answer these questions to be a part of the focus group. If there is any other question you would prefer not to answer just press "Next" at the bottom of the screen.

If it is determined that you qualify for the focus group portion of this study, you will be invited to participate in a 60–90-minute virtual focus group discussion over Zoom. This discussion will focus on your satisfaction and any recommendations or opinions you have about the resources you received when your child received a Fragile X syndrome diagnosis.

While some contact information must be obtained in order to reach you to coordinate the focus group discussion, your contact information will be kept confidential. The results of this study might be published or presented at academic meetings; however, participants will be deidentified.

Your participation in this research is voluntary. By clicking "I consent" below and completing the survey, you are consenting that you have read and understand this information and are consenting to being contacted to participate in a virtual focus group. At any time, you may withdraw from the study by not completing the survey or joining the focus group. Thank you for your time and consideration to participate in this research study. If you have any questions regarding the research, you may contact either myself or my faculty advisor, Richard Ferrante, using the contact information below. If you have any questions about your rights as a research participant, you may contact the Office of Research Compliance at the University of South Carolina at (803) 777-7095.

Megan Michalski, B.S.
Genetic Counselor Candidate
Megan.michalski@uscmed.sc.edu
(843)-345-3071

Dr. Richard Ferrante
Faculty Advisor
Richard.Ferrante@uscmed.sc.edu

- ○ I consent (4)
- ○ I do not consent (5)

Skip To: End of Survey If Dear Potential Participant,You are invited to participate in a graduate research study focusing = I do not consent

Page Break

Q2 Has your child been diagnosed with a Fragile X

- ○ Full Mutation (Has Fragile X Syndrome) (>200 repeats) (6)
- ○ Premutation (7)
- ○ My child does not have Fragile X syndrome (8)
- ○ Other (please specify) (9) _____

Skip To: End of Survey If Has your child been diagnosed with a Fragile X != Full Mutation (Has Fragile X Syndrome) (>200 repeats)

Page Break

Display This Question:

If Has your child been diagnosed with a Fragile X = Full Mutation (Has Fragile X Syndrome) (>200 repeats)

Q3 Are you your child's primary caregiver?

- ○ Yes (1)
- ○ No (2)

Display This Question:

If Are you your child's primary caregiver? = Yes

Or Are you your child's primary caregiver? = No

Q4 Were you the parent/caregiver who was present at the time of diagnosis?

- ○ Yes (1)
- ○ No (2)

Skip To: End of Survey If Were you the parent/caregiver who was present at the time of diagnosis? = No

Display This Question:

If Were you the parent/caregiver who was present at the time of diagnosis? = Yes

Q5 What is your relationship to your child?

- ○ Mother (1)
- ○ Father (2)
- ○ Other (3) _____

Page Break

Display This Question:

If Were you the parent/caregiver who was present at the time of diagnosis? = Yes

Q6 How many years ago did you child receive their Fragile X diagnosis? (in years)

_____

Skip To: End of Survey If Condition: How many years ago did you ... Is Greater Than or Equal to 11. Skip To: End of Survey.

Display This Question:

If If How many years ago did you child receive their Fragile X diagnosis? (in years) Text Response Is Less Than or Equal to 10

Q7 How old is your child currently (in years)

_____

Page Break

Display This Question:

If If How many years ago did you child receive their Fragile X diagnosis? (in years) Text Response Is Less Than or Equal to 10

Q8 What kind of healthcare professional diagnosed your child with Fragile X syndrome?

- ○ Pediatrician, MD (1)
- ○ Developmental Pediatrician, MD, NP, or PA (2)
- ○ Clinical Geneticist, MD (3)
- ○ Genetic Counselor (4)
- ○ Neurologist, MD (5)
- ○ Psychiatrist, MD (6)
- ○ My child has not been diagnosed by a healthcare professional (7)
- ○ Other (please specify) (8)

_____

Page Break

Display This Question:

If If How many years ago did you child receive their Fragile X diagnosis? (in years) Text Response Is Less Than or Equal to 10

Q9 Have you spoken to a healthcare professional about resources?

Resources can include informational books or pamphlets, links to videos, handouts, reference to patient advocacy groups and informational sites like the National Fragile X Foundation, packets about management and prognosis, or even how to access support groups or Facebook groups.

- ○ Yes (1)
- ○ No (2)

Page Break

Display This Question:

If If How many years ago did you child receive their Fragile X diagnosis? (in years) Text Response Is Less Than or Equal to 10

Q10 Which of these health care professionals have provided you with resources? (select all that apply)

Resources can include informational books or pamphlets, links to videos, handouts, reference to patient advocacy groups and informational sites like the National Fragile X Foundation, packets about management and prognosis, or even how to access support groups or Facebook groups.

- ☐ Pediatrician, MD (1)
- ☐ Developmental Pediatrician, MD, NP, or PA (2)
- ☐ Clinical Geneticist, MD (3)
- ☐ Genetic Counselor (4)
- ☐ Neurologist, MD (5)
- ☐ Psychiatrist, MD (6)
- ☐ Psychologist (7)
- ☐ Special Educator (8)
- ☐ Educator (9)
- ☐ Therapist (10)
- ☐ Other (please specify) (11) _____

Page Break

Display This Question:

If If How many years ago did you child receive their Fragile X diagnosis? (in years) Text Response Is Less Than or Equal to 10

Q11 Have you ever visited a specialized Fragile X Clinic?

- ○ Yes (1)
- ○ No (2)
- ○ Unsure (3)

Page Break

Display This Question:

If If How many years ago did you child receive their Fragile X diagnosis? (in years) Text Response Is Less Than or Equal to 10

Q12 If you are willing to be contacted to participate in a focus group discussion about resources you received from your healthcare professional when your child was diagnosed please fill out some basic contact information.

Preferred first and last name:

_____

Display This Question:

If If How many years ago did you child receive their Fragile X diagnosis? (in years) Text Response Is Less Than or Equal to 10

Q13 Email address:

_____

Q19 Phone number:

_____

Page Break

Q14 Does your child with Fragile X syndrome identify as a

- Male (1)
- Female (2)
- Non-binary / third gender (3)
- Prefer not to say (4)

Q15 What gender do you identify as?

- Male (1)
- Female (2)
- Non-binary / third gender (3)
- Prefer not to say (4)

Page Break

Q16 What race/ethnicity do you identify as? (choose all that apply)

- White or Caucasian (1)
- Hispanic or Latino (2)
- Black or African American (3)
- Native American or American Indian (4)
- Asian or Pacific Islander (5)
- A race not listed here (please specify) (6)

_____

- Prefer not to specify (7)

Page Break

Q17 What is the highest level of education you have completed?

- Some high school (1)
- High school degree (2)
- Some college (3)
- Technical degree (8)
- Associate's degree (4)
- Bachelor's degree (5)
- Some graduate school (6)
- Master's or Doctoral degree (7)

Page Break

Q18 Please click "Next" to submit this survey.

You qualify to participate in the virtual focus group! Thank you for your willingness to participate and your time. Your participation in this study is valuable to all healthcare professionals involved in distributing information and resources to Fragile X families.

You will be contacted via email within a couple of weeks to determine your general availability for a focus group meeting to assist with scheduling the virtual focus group.

We invite you to share this online survey with other parents of children with Fragile X Syndrome.

If you have any questions, please contact Megan Michalski at:
megan.michalski@uscmed.sc.edu
(843)-345-3071

End of Block: Default Question Block

APPENDIX F: FOCUS GROUP SCRIPT AND QUESTIONS

Focus Group Script Introduction

Thank you everyone for participating in today's focus group! I really appreciate that you are willing to share your valuable time with us. My name is Megan, I am a genetic counseling graduate student at the University of South Carolina. I will be guiding today's discussion. I would like to remind all of you that participation in today's focus group is completely voluntary and you can choose to not respond to any questions. You may decide to leave this virtual meeting at any point. Please let me know at any time if there are any technical difficulties on my or your end so we can try to resolve them to make this discussion flow smoothly.

As was explained on the initial recruitment letter, the purpose of today's focus group is to learn more about your opinion about Fragile X resources. Participants in this group are parents of a child with a full Fragile X mutation. Using these focus groups, the research team will work to develop reference materials (like a document or brochure) for healthcare providers to use to suggest resources to parents about Fragile X Syndrome that are most beneficial.

I really want to hear all of your thoughts and opinions during the discussion today, so I will be recording the session. Please know that we have strategies to protect your confidentiality and anonymity. Only researchers will have access to today's recording or the transcript from the recording. When we are summarizing results from this discussion all identifying information will be excluded from your comments or

quotes to protect your identity. You are not required to share personal identifying information, including names of yourself or your children.

Due to these measures, I hope you feel comfortable to speak your mind. Remember you don't have to answer any question you are not comfortable answering. I want to ensure everyone today has a chance to speak, so please do not be offended if I limit any of your comments to allow someone else to speak. I ask that we are conscious of others and do not talk over someone if they are speaking. I do have specific questions which you all received ahead of time, but the conversation may take a natural flow. We do have a random raffle to receive a 25$ Amazon gift card after the completion of this focus group. Lastly, I ask that you please keep your microphone muted when you are not speaking if possible, and feel free to utilize the chat function. Does anyone have any questions before we begin?

Focus Group Questions

1. To start could you please introduce yourself, state how old your child is and when you got their diagnosis? (You may choose to share your child's name if you feel comfortable)
    a. What kind of healthcare professional gave you the diagnosis?
    b. General pediatrician, Geneticist, Genetic counselor, other
2. Now thinking back to when your child was first diagnosed, what kinds of resources about Fragile X did that healthcare professional recommend so that you could learn how to care for your child?
    a. (Resources here can include informational books or pamphlets, links to videos, handouts, reference to patient advocacy groups and

informational sites like the National Fragile X Foundation, packets about management and prognosis, or even how to access support groups or Facebook groups.)

   b. Did you look on the web, Facebook, book, brochure, etc.
   c. Could you give a specific example of resources that you first read when your child was diagnosed with Fragile X.
   d. your satisfaction or dissatisfaction with them?
   e. Topics these references covered

3. When your child was first diagnosed what did you most want to learn about?
   a. Did the recommended resources cover this information to the extent that you wanted?
      i. Please expand on this
   b. Were you satisfied with the information you learned and how it was presented from these sources?
4. What other resources about Fragile X did you find on your own?
   a. Provide examples
   b. Why did you like about these resources, topics covered, etc?
   c. What aspects were most useful in these resources?
      i. Give me an example of what was useful?
   d. What gaps in information did these fill?
   e. How did you use the information?
5. Which resources do you wish you knew about earlier than when you found them?

    a. Provide examples.
    b. Explain what made these resources the most beneficial
6. What areas do you wish there were more information on that you cannot find?
    a. Is there any information you would like to learn that you cannot find information on?
    b. How could these resources have been made more accessible to you?
7. Are there resources that you have come across or were recommended that were outdated, overly confusing, inaccurate, or for another reason ones you did not like?
    a. If yes, provide examples please
    b. What made you dislike these resources?
    c. How could these resources be improved?
8. Have we missed anything about resources that you would like to still discuss?
9. If you were creating resources for parents who have a child with Fragile X Syndrome, what information do you think is necessary to be included, where would you direct them for further readings, what do you want them to know?
    a. What resources were most useful when learning about the condition?
    b. What resources were most useful when learning how to care for your child?

Focus Group Conclusion

We have reached the end of this focus group. Thank you again for taking the time to participate and share your thoughts. As I mentioned the goal of this discussion is to aid in the creation of a reference for healthcare professionals to know what resources

to recommend to families affected by Fragile X. You should receive a 1 page summary of today's focus group notes within a couple weeks, and as my thesis is completed you will be updated of the results of this study. Do you have any final questions or thoughts you would like to share?

The 25$ Amazon gift card can be raffled to this group. If you are selected as the winner, you will receive an email containing details to the Amazon gift card tonight! Thank you again for your time this afternoon! Take care.

APPENDIX G: INTERVIEW SCRIPT AND QUESTIONS

Interview Script Introduction

Thank you for participating in this interview! I really appreciate that you are willing to share your valuable time for this study. My name is Megan, I am a genetic counseling graduate student at the University of South Carolina. I would like to remind you that participation in today's interview is completely voluntary and you can choose to not respond to any questions. You may decide to leave this phone call at any point. Please let me know at any time if there are any technical difficulties on my or your end so we can try to resolve them to make this interview flow smoothly.

As was explained on the initial recruitment letter, the purpose of today's discussion is to learn more about your opinion about Fragile X resources. Participants in this study are parents of a child with a full Fragile X mutation. Using focus groups and interviews, the research team will work to develop reference materials (like a document or brochure) for healthcare providers to use to suggest resources to parents about Fragile X Syndrome that are most beneficial.

I really want to hear all of your thoughts and opinions during the interview, so I will be recording the call. Please know that we have strategies to protect your confidentiality and anonymity. Only I will have access to today's recording or the transcript from the recording. When we are summarizing results from this interview all identifying information will be excluded from your comments or quotes to protect your

identity. You are not required to share personal identifying information, including names of yourself or your children.

Due to these measures, I hope you feel comfortable to speak your mind. Remember you don't have to answer any question you are not comfortable answering. I do have specific questions which you received ahead of time, but the interview may take a natural flow. I also have a random raffle to receive a 25$ Amazon gift card after the completion of yours and a couple other interviews. Do you have any questions before we begin?

Focus Group Questions

1. To start could you please introduce yourself, state how old your child is and when you got their diagnosis? (You may choose to share your child's name if you feel comfortable)
    a. What kind of healthcare professional gave you the diagnosis?
    b. General pediatrician, Geneticist, Genetic counselor, other
2. Now thinking back to when your child was first diagnosed, what kinds of resources about Fragile X did that healthcare professional recommend so that you could learn how to care for your child?
    a. (Resources here can include informational books or pamphlets, links to videos, handouts, reference to patient advocacy groups and informational sites like the National Fragile X Foundation, packets about management and prognosis, or even how to access support groups or Facebook groups.)
    b. Did you look on the web, Facebook, book, brochure, etc.

    c. Could you give a specific example of resources that you first read when your child was diagnosed with Fragile X.
    d. your satisfaction or dissatisfaction with them?
    e. Topics these references covered
3. When your child was first diagnosed what did you most want to learn about?
    a. Did the recommended resources cover this information to the extent that you wanted?
        i. Please expand on this
    b. Were you satisfied with the information you learned and how it was presented from these sources?
4. What other resources about Fragile X did you find on your own?
    a. Provide examples
    b. Why did you like about these resources, topics covered, etc?
    c. What aspects were most useful in these resources?
        i. Give me an example of what was useful?
    d. What gaps in information did these fill?
    e. How did you use the information?
5. Which resources do you wish you knew about earlier than when you found them?
    a. Provide examples.
    b. Explain what made these resources the most beneficial
6. What areas do you wish there were more information on that you cannot find?

    c. Is there any information you would like to learn that you cannot find information on?
    d. How could these resources have been made more accessible to you?
7. Are there resources that you have come across or were recommended that were outdated, overly confusing, inaccurate, or for another reason ones you did not like?
    a. If yes, provide examples please
    b. What made you dislike these resources?
    c. How could these resources be improved?
8. Have we missed anything about resources that you would like to still discuss?
9. If you were creating resources for parents who have a child with Fragile X Syndrome, what information do you think is necessary to be included, where would you direct them for further readings, what do you want them to know?
    a. What resources were most useful when learning about the condition?
    b. What resources were most useful when learning how to care for your child?

Focus Group Conclusion

We have reached the end of this interview. Thank you again for taking the time to participate and share your thoughts. As I mentioned the goal of this discussion is to aid in the creation of a reference for healthcare professionals to know what resources to recommend to families affected by Fragile X. You should receive a 1 page summary of recommendations of resources from other participants, and as my thesis is completed

you will be updated of the results of this study. Do you have any final questions or thoughts you would like to share?

The 25$ Amazon gift card can be raffled between those who completed an interview and focus group. If you are selected as the winner, you will receive an email containing details to the Amazon gift card in next couple of weeks.

Thank you again for your time this afternoon! Take care.

APPENDIX H: RESEARCHER COMPILED RESOURCE LIST: PARTICIPANT RECOMMENDATIONS AND OTHER FRAGILE X RESOURCES

RESOURCES

Thank you everyone for participating in my thesis research highlighting parent satisfaction related to fragile X syndrome resources provided by healthcare professionals at the time of diagnosis. Based off of the discussions that we had, I complied a short list of resources that you as parents seemed to gravitate towards, along with a couple of resources that I have found in my own research, these are in *italics*. Maybe some of these are new to you and you find some new information. Thank you again for your time!

Websites:

- National Fragile X Foundation
  - https://fragilex.org/
  - https://fragilex.org/living-with-fragile-x/newly-diagnosed/ - information for a newly diagnosed family
- National Fragile X Foundation – Community Support Networks
  - https://fragilex.org/living-with-fragile-x/community-support/
- *Positive Exposure*
  - https://positiveexposure.org/frame/fragile-x/

Facebook Groups:

- Fragile X Female Carrier Symptoms
- Fragile X Syndrome
- NFXF Fragile X Advocacy
- Fragile X
- Girls with Fragile X Syndrome
- *Fantastically Fragile X* – a Fragile X "brag room" for families

Current Research Studies:

- FRAXA Research Foundation
  - https://www.fraxa.org/
- *UC Davis MIND Institute*
  - https://health.ucdavis.edu/mindinstitute/research/fragilex/fragilex-current-research.html

- *ClinicalTrials.gov*
    - https://clinicaltrials.gov/ct2/results?cond=Fragile+X+Syndrome&term=&cntry=&state=&city=&dist=&Search=Search

Books:

- *Children with Fragile X Syndrome: A Parent's Guide* – by Jayne Dixon Weber
- *X Stories: The Personal Side of Fragile X Syndrome* – edited by: Charles W. Luckmann and Paul S. Piper