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Parental Experience Of Divulging a Diagnosis Of Fragile X Syndrome To Their Affected Child

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Parental Experience of Divulging a Diagnosis of Fragile X Syndrome
to their Affected Child

by

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Bachelor of Science
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Abstract

Fragile X syndrome (FXS) is a genetic condition with varied presentation that may include intellectual and learning disabilities, behavioral and learning challenges, and certain physical characteristics. When an individual is affected with FXS, it often leads to complex discussions within the family, including parental disclosure of a FXS diagnosis to a child. This study explored how a FXS diagnosis is communicated between a parent and their child diagnosed with FXS. An online questionnaire was disseminated through FXS organizations and support groups to parents who have at least one child diagnosed with FXS. The online questionnaire was supplemented by eight brief telephone interviews. Qualitative analysis was conducted on eighty-three responses. Parents commonly practiced resilient communication while providing age and developmentally appropriate information at a level their child was able to understand. Though parents would focus on how their child was different from others to explain FXS, they also used affected family members and other FXS families as examples to prevent their child from feeling isolated by the diagnosis. Parents worried that their child would not understand or that the information provided would have a negative effect on the child's emotional being. Resources were not often used, but parents expressed a desire for a children's book written specifically for children with FXS as well as easier access to input from other FXS families and healthcare professionals. Awareness and understanding of this experience will allow genetic counselors to provide appropriate education, support, resources, and referrals to ensure successful communication between a parent and child.

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Chapter 1. Background

1.1 Fragile X Syndrome

The *FMRI* gene is located on Xq27.3 and when functioning properly, makes a protein product called FMRP (Nolin et al., 1996). This protein is expressed throughout the body but is most commonly found in neurons (Devys, Lutz, Rouyer, Bellocq, Mandel 1993). In the 5' untranslated region of *FMRI* there is a region of repeating nucleotides, specifically repetitive sequences of CGG nucleotides (Verkerk et al., 1991). This region is unstable and expansion of the repetitive sequence can occur, leading to various related disorders that are determined by the size of the expanded CGG repeat and the degree of methylation that occurs (Sherman, Pletcher, & Driscoll, 2005). Within the general population, most people have a “normal” allele that ranges from 6 to 44 repeats and an individual is considered to have a “premutation” allele if 55-200 CGG repeats are present. Some overlap exists between the “normal” and “premutation” allele definition at about 45-54 repeats, known as the “grey zone” or “intermediate” mutation (Nolin et al., 1996). While some intermediate alleles are stable, other intermediate alleles are not. When over 200 CGG repeats are present, an individual is considered to have a full mutation and a diagnosis of Fragile X syndrome (McConkie-Rosell et al., 2005). Most individuals affected by FXS have an increased repeat size, but 1% of FXS diagnoses are caused by either a point mutation or deletion within the *FMRI* gene (McConkie-Rosell et al., 2005). A point mutation is present when there is a single base pair change in the DNA

sequencing, while a deletion is present when a piece of the DNA sequence is missing (Mahdiah & Rabbani, 2013).

Generally if an individual has less than 40 CGG repeats per allele, his or her allele is stable, meaning the number of repeats will likely stay the same through transmission to his or her child (Macpherson et al., 1995). However, as an individual's number of CGG repeats increases, his or her chance to pass on an expanded allele increases. This can be dependent on numerous factors including CGG size, the presence of AGG interruptions, and maternal age as it is thought that the process of DNA replication is interrupted by these factors, leading to expansion of the allele (McConkie-Rosell et al., 2005; Yrigollen et al., 2014).

It is estimated that about 1 in 259 females and 1 in 755 males carry the premutation (McConkie-Rosell et al., 2005), while about 1 in 4000 males and 1 in 8000 females are affected by FXS (de Vries et al., 1997; Finucane et al., 2012; Turner, Webb, Wake, & Robinson, 1996). FXS and the associated phenotypes are inherited in an X-linked manner. Females have a genetic make-up of two X-chromosomes, while males have an X-chromosome and a Y-chromosome. Because of this, a father will only pass his X-chromosome to his daughter and his Y chromosome to his son. A mother will always pass on one of her two X-chromosomes to each of her children. When a male carries the premutation, he has a 100% chance of passing the mutation to all of his daughters and a 0% chance of passing the mutation on to his son. When a female carries the premutation, she has a 50% chance of passing the allele with the mutation on to each child and a 50% chance of passing her normal allele on to each child, as each child will inherit one or the other allele.

The number of CGG repeats are categorized by the phenotype with which they are associated. For individuals with a normal or intermediate allele, no associated phenotype of FXS is expected. When individuals have a premutation, their mutated allele is unstable and can expand during the replication process, potentially causing FXS in their children or grandchildren if the number of repeats increases into the full mutation range (Nolin et al., 1996). Individuals with a premutation are also at risk for Fragile X-associated Tremor/Ataxia Syndrome (FXTAS) and Fragile X-associated primary ovarian insufficiency (FXPOI; Finucane et al., 2012). FXTAS is a type of neurodegenerative disorder that is more likely to affect premutation males than females. Affected individuals can present with progressive tremor, cerebellar dysfunction, Parkinson-like symptoms, and cognitive decline that can include memory loss, dementia, and loss of executive functioning (Hagerman et al., 2001). FXPOI affects female premutation carriers and causes a decline in ovarian function (Murray, Webb, Grimley, Conway, & Jacobs 1998). Finucane et al. (2012) describes that approximately 15 to 20% of women with a premutation allele are affected by FXPOI, accounting for about 2 to 7% of women with sporadic ovarian insufficiency. Allen et al. (2007) confirmed an increased risk of ovarian insufficiency with a 13-fold higher frequency in premutation carriers than in non-mutation carriers, with menopause occurring five years earlier than average. Women with one premutation allele also had short, irregular, and missed cycles as well as reduced fertility (Allen et al., 2007).

When an allele expands to over 200 repeats, an individual is considered to have a full mutation and a diagnosis of FXS. McConkie-Rosell et al. (2005) described that when an individual has a full mutation, a process called methylation occurs in a region of the

FMRI gene, causing the gene to become silenced. When this occurs, the gene is unable to make the FMRP protein. Though FXS affects both sexes, males often have a more severe presentation than females. Affected males frequently present on a spectrum ranging from mild learning disabilities to severe intellectual disabilities. Males may also show behavioral challenges that can include Attention Deficit Hyperactivity Disorder (ADHD), Autism Spectrum Disorders (ASD), anxiety, hand-biting, hand-flapping, echolalia, and coprolalia. Physical characteristics may include large ears, a long face, soft velvet-like skin, prominent forehead, macroorchidism, flat feet, double-jointed fingers, and hyper-flexible joints. Females who are affected will often have a similar presentation in all areas when compared to an affected male, including intellectual disability, behavioral challenges, and characteristic physical features. Though the presentation is similar, females are more likely to have a subtler phenotype, often having normal IQ and only slight physical characteristics along with potentially milder learning disabilities and/or behavioral/psychological conditions (McConkie-Rosell et al., 2005).

1.2 Genetic Testing of Fragile X Syndrome

The American College of Medical Genetics (ACMG) Professional Practice and Guidelines Committee provided FXS testing guidelines (Sherman et al., 2005) that recommends genetic testing for:

1. Individuals of either sex with mental retardation, developmental delay, or autism, especially if they have (a) any physical or behavioral characteristics of FXS, (b) a family history of FXS, or (c) male or female relatives with undiagnosed mental retardation.

2. Individuals seeking reproductive counseling who have (a) a family history of FXS 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 *FMRI* 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.
5. 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 FXS, or (c) male or female relatives with undiagnosed mental retardation.
6. 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 FXS, or (c) male or female relatives with undiagnosed mental retardation. (p. 586)

Sherman et al. (2005) explained that when genetic testing is recommended, the size of the trinucleotide repeat as well as methylation status is determined for the *FMRI* gene on the X chromosome through Polymerase Chain Reaction (PCR) and Southern blot analysis. PCR amplifies the region of the *FRM1* gene with the trinucleotide repeat, which allows the approximate number of repeats to be determined for each allele. As the

number of repeats increases, this test becomes less effective as it is more difficult to amplify larger repeats. Because of this, PCR is adequate for identifying normal, grey zone, and premutation alleles. A Southern blot analysis will provide an approximate measurement of all allele sizes and can distinguish between methylated and unmethylated *FMRI* alleles, but is more labor intensive than PCR. Labs typically maintain both of these tests and use them as appropriate, either in combination or individually based upon the clinical conditions. For the individuals with a point mutation or deletion, other testing strategies should be implemented such as sequencing, assays, linkage studies, or cytogenetic studies (Sherman et al., 2005).

1.3 Communication within the Family Structure

According to Rolland and Walsh (2008), a stressful event not only has an effect on a primary individual, but it affects the whole family as a functional unit. This can be seen as a ripple effect throughout the family, as each member has a response that may affect relationships within the family or outside of it (Rolland & Walsh, 2008). The ripple effect explains how an individual's actions can either consciously or unconsciously affect others through direct or indirect means and is observed across all genetic counseling specialties. For example, in a pediatric session, the focus would often be on the child who has a genetic diagnosis but others possibly affected by this diagnosis also are considered. Unaffected siblings highlight this issue, as they may feel overlooked if they do not receive as much attention as their sibling with a diagnosis. Additionally, an unaffected sibling may experience anxiety if they do not understand their own personal risk for the condition (Plumridge, Metcalfe, Coad, & Gill, 2011).

According to Rolland and Walsh (2008), a family's belief systems, organizational patterns, and their communication processes largely comprise a framework that dictates family function and can influence the family's and/or an individual's ability to positively adapt when situations arise. A family's belief system is important in determining the meaning a situation could have on a family, how the situation came about, and what can be done to improve upon the situation if necessary. Family organization patterns dictate how a family is able to maintain themselves as a unit through adaptability, connectedness, family boundaries, and community networks. Communication within families is essential to the framework as it establishes an ability to have positive family functioning. Within communication, families should recognize that the ability to verbalize their thoughts openly and clearly, along with trust and problem solving, are key to successfully navigating through problems when they arise. This framework and the ripple effect cause the family's response to a stressful event to contribute greatly to how both the family and individuals outside of the family adapt and cope with the event (Rolland & Walsh, 2008).

Harris et al. (2010) found that familial coping, adaptation, and cohesion along with familial health beliefs were associated with an open style of communication, where family members were willing to share information with others and were supportive in these endeavors. Families that experience higher levels of open communication reported more active coping styles as well as higher levels of adaptation and cohesion within their family. At the same time, negative coping mechanisms such as denial were negatively associated with an open communication style (Harris et al., 2010). Peer and Hillman (2012) found that when an individual does not successfully cope with stress, the build-up

of stress may have adverse implications not only for him or herself but also for those in his or her family structure. For example, if stress prevents parents from meeting their own needs, it could limit them from providing a full level of care to their child. This could inhibit the child from reaching his or her maximum developmental potential (Peer & Hillman, 2012). Altogether, this provides a basis for the importance of open communication within a family to ensure adequate coping mechanisms and family function.

The family systems theory recognizes multiple facets that interplay within the family as well as outside of it; together these influences help shape an individual's role within his or her family, school, the workplace, and within health care systems. This is a shift from a previous theory that the family system is a linear, deterministic view of causality in a traditional psychoanalytic theory (Rolland & Walsh, 2008). Over time and experiences, family systems tend to develop themes that are unique to their structure and will influence how they deal with change and stress. Certain themes may also influence the development of patterns that regulate family communication as the patterns become reciprocal and repetitive over time (Bylund, Galvin, & Gaff, 2010). Interventions to explore such themes and patterns help families recognize what might be influencing their family structure and communication, allowing families to learn from the past to make a difference in communication for current relationships. While patterns of communication are important, it is vital that conveyed messages are clear and congruent. Otherwise, the risk of anxiety and depression increases for the receiving individual in the presence of uncertainty or confusion (Rolland & Walsh, 2008). Clarity of messages can be related to an individual's ability to communicate and can influence whether or not s/he is

understood. Due to the complexity of communication, there must be consideration of many factors when examining a family's communication patterns, as it is possible that certain influences may provide insight on how a family communicates health information (Harris et al., 2010).

1.4 Communication of Genetic Risk to Children

Sullivan & McConkie-Rosell (2010) reported that when a child has a genetic disorder, the parents are often responsible for discussing that diagnosis with their child and answering any questions that may arise. Additionally, Metcalfe, Coad, Plumridge, Gill, and Farndon (2008) discovered that individuals often feel protected by keeping what they consider problems to themselves, but the inability to openly discuss such issues can lead to tense relationships and poor communication within families. While such communication does not lessen the psychological and emotional pain of living with a genetic disability or risk, it does allow for open discussion when concerns arise and an increase in support and care within the family (Metcalfe et al., 2008). When parents choose not to communicate genetic information to their child, it creates considerable strain for both parties as well as on the relationship between them (Metcalfe, Plumridge, Coad, Shanks, Gill, 2011). When a parent is considering disclosure to their child about a genetic diagnosis, many factors may influence such a discussion. Parents may consider whether the child is affected or at an increased risk, how the family perceives the diagnosis, what might the personalized meaning to the information be, and what are the implications of the diagnosis or risk upon their child (McConkie-Rosell, Del Giorno, & Heise, 2011).

Metcalfe et al. (2011) described how parents often report that discussing genetic risk information is emotionally painful and a difficult subject for them to broach with their child, often requiring a particular event to occur to prompt a discussion. Dennis, Jowell, Cordeiro, and Tartaglia (2014) reflected these findings as parents frequently reported that they withheld explaining to their child about a diagnosis until the child needed to see a specialist or had numerous medical appointments related to their diagnosis. This would particularly occur if the child began to question the utility of the appointments. Parents also reported that if their child began to experience problems related to their diagnosis then those experiences would prompt discussion of the diagnosis (Dennis et al., 2014). Parents may shy away from such a conversation due to shock they felt at the time of diagnosis, feelings of guilt and fear, an increased burden due to emotional and physical caregiving to others, and feelings of grief and bereavement (Metcalfe et al., 2011). Gallo, Angst, Knafl, Hadley, & Smith (2005) found that some parents choose nondisclosure of a condition as they feel it could negatively affect a child's will to live or the parent is afraid their child may inadvertently disclose about the condition to others. Parents may consider themselves open for discussion of their child's diagnosis, but often times this is just in reference to answering questions posed by the child instead of initiating a discussion with their child (Metcalfe et al., 2011). Though the focus should likely be on the needs of the child affected with a genetic disorder, this attention will be shifted by individuals who are interconnected with the child, such as a parent or sibling.

When a parent considers disclosure of a diagnosis to their child, a common concern is the timing and content of the conversation. Studies have found that parents

withholding information until their child become an adult could have an impact on the young adult's coping mechanisms, self esteem, reproductive decision-making, and family cohesion (Metcalf et al., 2011). Individuals at-risk for genetic conditions have reported they feel it is important to learn about their risk early on at a developmentally appropriate level (Wehbe, Spiridigliozzi, Heise, Dawson, & McConkie-Rosell, 2009). Parents and children alike recommended honesty, early disclosure, and continuing the discussion over time (Dennis et al., 2014). This allows the child to adapt and cope, provides an open stream of communication, and ensures no misinformation. Parents also should encourage their child to ask questions and should be supportive and nonjudgmental throughout discussions (Metcalf et al., 2011).

Parents recognize the need to adapt discussions with their child to a developmentally appropriate level but find themselves unable to access advice and support for different developmental stages (Metcalf et al., 2011). Parents often feel uncertain as to what they should say, what vocabulary they should use, and how to prevent the conversation from exceeding their child's level of understanding (Dennis et al., 2014). For example, Metcalf et al. (2011) explained that when a child is seven years old or younger, s/he will be able to notice physical differences between individuals and wonder if they could have the same condition. A child at that age may be able to understand that the condition could be related to both the parent and child, but would not be able to grasp how it could be passed down through the family. This understanding is something that would continue to evolve over time and is crucial to how information should be presented to a child about his or her diagnosis (Metcalf et al., 2011). To ease this concern, parents consult healthcare providers with the hopes of gaining assistance in

risk assessment as well as how to explain a diagnosis to their child (Sullivan & McConkie-Rosell, 2010). When asked about their experiences, parents reported that they felt they had a lack of support from healthcare providers in regards to discussing a diagnosis with their child (Metcalf et al., 2008). Parents feel that more advice and assistance on how to give their child developmentally appropriate information should be provided (Metcalf et al., 2011). This lack of support often leads to feelings of anxiety, worry, and concern as parents are relying on their own understanding and experiences with a genetic condition to help inform their child (Metcalf et al., 2008). If a healthcare provider were able to evaluate a family through the family systems theory, it would allow the provider to explore and understand the impact that a genetic diagnosis could have on a family and determine the needs of the family (Galvin & Young, 2010). Along with parents, young individuals at risk for a genetic condition felt a need to meet with a healthcare professional with whom they could discuss their risk in order to become further informed (Metcalf et al., 2011). This highlights the importance of a relationship between a family at-risk for a genetic condition and their healthcare provider to ensure that a foundation is built upon which a child will be supported and adequately educated about their risk and/or diagnosis (Sullivan & McConkie-Rosell, 2010). Thus, during this continuous process, healthcare providers should help parents set appropriate goals and utilize resources to develop a plan for communication with their child.

1.5 Family Structure Involving Individuals with an Intellectual Disability

When parents have a child with an intellectual disability, they find difficulty in determining when they will move into a more supportive role as the child matures instead of remaining in the role of primary caregiver (Peer & Hillman, 2012). This may alter the

course of communication through their child's life, specifically in regards to genetic disability. Stark and Moller (2002) describe how mothers of a child with disabilities often seek out advice from healthcare providers until they began to develop a better sense of their child's wants and needs. When a mother finds herself dissatisfied with her child's healthcare, she would often seek a relationship with the provider to maintain control in that relationship. When the mother found more satisfaction in the relationship, she used it to gain information to share with others about her child's condition. Fathers, on the other hand, felt that their responsibility is to manage their child's developmental growth due to available resources, such as medical literature and reference books, which fathers can investigate (Starke & Moller, 2002).

Reilly, Murtagh, and Senior suggest that having a child with intellectual disability can introduce numerous challenges into a family, depending on the child's specific disabilities. It has been found that the presence of behavior or psychiatric conditions as well as decreased receptive language abilities have been associated with more severe restrictions on family and parental activities in daily life. This includes activities such as going shopping, going on vacation, and taking public transportation. The authors found that the presence of a behavioral or psychiatric condition is associated with a decrease in positive aspects, as reported by the parent. Positive aspects include items such as happiness brought by the child, helping the family become more tolerant/patient/spiritual/compassionate, creating friendships with other parents, and providing opportunities to learn and develop. There is a higher threat to family functioning when there are comorbid conditions present, along with intellectual disability, that need to be adequately identified and managed. For these families,

awareness is crucial to ensure correct diagnosis so that proper resources, support, and interventions can be accessed that will supplement positive family functioning (Reilly et al., 2015).

1.6 Communication with Individuals with an Intellectual Disability

Communication of a diagnosis is already a challenging process and becomes further complicated when the child has an intellectual disability. Parental experiences in a communicating a diagnosis to a child with an intellectual disability will elicit differing discussion strategies and parental concerns. In regards to communication, Shearn and Todd (1997) reported that when a child has an intellectual disability, parents purposefully avoid conversation about the disability with their child due to a fear it will cause the child emotional distress. To achieve this, parents will avoid use of any terminology that may lead to awareness in their child about his or her disability, such as ‘handicapped’ (Shearn & Todd, 1997). Jones, Oseland, Morris, and Larzeler (2014) found that some parents will avoid talking to their child about his or her disability to emphasize the child’s similarities to other children and minimize the disability. When parents chose to discuss their child’s disability with him or her, they are more likely to use words such as ‘difference’ and may use that to focus their conversation on how their child is different from other children his or her own age (Jones et al., 2014). Parents may try to conceal their child’s disability by creating an illusion of a normal life and helping their child maintain that vision even if it does not parallel reality (Shearn & Todd, 1997). Jones et al. (2014) found that often times, parents choose not to explain to a child about their intellectual disability with the hopes of shielding their child from the truth and occasionally because the parent considers the disability as a negative. If a child notices such a belief, it could trigger him

or her to have a negative self image and influence future beliefs and interactions (Jones et al., 2014).

In their study of parents who had a child diagnosed with Down syndrome, Cunningham, Glenn, and Fitzpatrick (2000) described parents who did not communicate with their child about the diagnosis. These parents cited reasons such as they worried their child would not understand, they felt there was “no point” in communicating the diagnosis with their child, or they felt uncomfortable broaching the subject. Similar to general communication of genetic risk to children, parents who communicate with their child about his or her diagnosis reported doing so after an event triggered the conversation. The most common reasons for initiating discussion were that their child began asking why they looked like someone else with Down syndrome, their child began asking why people were staring at him or her, their child began asking why they could not participate in activities his or her siblings could, or other individuals began asking questions about the child. Parents emphasized that discussion should focus on details relevant to a child’s life at that point in time and should be explained at a level the child can comprehend. This supports that a parent’s awareness of his or her child’s ability to understand and open communication within the family are key elements for successful discussion about Down syndrome with an affected child (Cunningham et al., 2000). Talking with children about their diagnosis is important to help facilitate understanding on how it affects them as they transition through life.

Similar to typically developing children, those with an intellectual or any other disability will eventually grow up and have their own wants and needs. This may involve having a family, pursuing a specific career, choosing medical treatment, or any other

decision in life that may require the support from family, friends, or professionals. When a couple affected by an intellectual disability is seen for counseling, there are numerous challenges that may arise within the session (de Vries, van den Boer-van den Berg, Niermeijer, & Tibben, 1999). Depending upon the comprehension of the couple, they may not fully understand the risk being discussed, their options, and possible outcomes. It is important to assess understanding throughout the session as well as the couple's ability to cope with the ramifications of the chosen decision. It may be more difficult to facilitate informed decision making with the couple if there is limited skill in comprehension and reflection, which could lead the counselor to take on a more directive role. When that occurs, it is important to utilize other healthcare professionals to create a multidisciplinary care team for the couple. This will help prevent the counselor from directly leading the couple towards one decision or another based on his or her personal opinions about the situation. Some sort of support, such as a family member or friend, will often accompany individuals with an intellectual disability. This can become problematic when the views of the patient differ from that of his or her support system and the counselor has to attempt to successfully navigate through both sides' needs in order to facilitate a decision (de Vries et al., 1999).

When an individual has an intellectual disability, effective counseling will require a substantial amount of time invested by the genetic counselor as well as individuals within the individual's family structure and support network. Time spent together will often be more conversational and be conducted at a slower speed to ensure the patient is able to follow along (Finucane, 2010).

While prior studies have focused on the sharing of genetic information with daughters at risk for carrying an unstable *FMRI* allele (McConkie-Rosell et al., 2011), there is little information available to parents who wish to discuss genetic information with their child diagnosed with FXS. This study explored parental experiences and attitudes of communicating a diagnosis of FXS to their affected child to provide insight into the challenges and successes parents have during these conversations. The study explored if a parent had communicated the diagnosis to their child, when and how did such a conversation arise, what information was provided to the child, what resources were used during the discussion, what were some of the parent's thoughts and feelings throughout this process, and did the presence of autism or an intellectual disability influence the experience. The information gained through this study is expected to benefit genetic counselors and their patients with a diagnosis of FXS, as it will allow genetic counselors to better identify what these families need to successfully communicate about a diagnosis. Once a family's needs are identified, genetic counselors can provide appropriate education, support, resources, and referrals to help facilitate more successful communication between a parent and their child.

Chapter 2. Parental Experience of Divulging a Diagnosis of Fragile X Syndrome to their Affected Child¹

2.1 Abstract

Fragile X syndrome (FXS) is a genetic condition with varied presentation that may include intellectual and learning disabilities, behavioral and learning challenges, and certain physical characteristics. When an individual is affected with FXS, it often leads to complex discussions within the family, including parental disclosure of a FXS diagnosis to a child. This study explored how a FXS diagnosis is communicated between a parent and their child diagnosed with FXS. An online questionnaire was disseminated through FXS organizations and support groups to parents who have at least one child diagnosed with FXS. The online questionnaire was supplemented by eight brief telephone interviews. Qualitative analysis was conducted on eighty-three responses. Parents commonly practiced resilient communication while providing age and developmentally appropriate information at a level their child was able to understand. Though parents would focus on how their child was different from others to explain FXS, they also used affected family members and other FXS families as examples to prevent their child from feeling isolated by the diagnosis. Parents worried that their child would not understand or that the information provided would have a negative effect on the child's emotional being. Resources were not often used, but parents expressed a desire for a children's book written specifically for children with FXS as well as easier access to input from

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other FXS families and healthcare professionals. Awareness and understanding of this experience will allow genetic counselors to provide appropriate education, support, resources, and referrals to ensure successful communication between a parent and child.

2.2 Introduction

Fragile X syndrome (FXS) is a genetic condition that causes intellectual disability, behavioral and learning challenges, and certain physical characteristics. FXS is caused by a mutation in the *FMR1* gene on Xq27.3 (Nolin et al., 1996). Both males and females affected with FXS present on a spectrum that can range from mild to severe features, but females are more likely than males to present with a subtler phenotype (McConkie-Rosell et al., 2005). When FXS is present in a family, individuals may be unaffected, carry a premutation, or have a diagnosis of FXS. This leads to complex discussion within families that may affect not only the primary individual, but the whole family as a functional unit (Rolland & Walshe, 2008). When a parent is considering disclosing to his or her child a genetic diagnosis, many factors may influence such a discussion. Parents may consider whether the child is affected or at an increased risk, how the family perceives the diagnosis, what might the personalized meaning to the information be, and what are the implications of the diagnosis or risk upon their child (McConkie-Rosell et al., 2011). For some parents, discussing genetic risk information may be emotionally painful and a difficult subject to broach with children, often requiring a related event to occur to prompt a discussion (Metcalf et al., 2011). This could include the child needing to see a specialist, having numerous medical appointments, or the child experiencing problems related to his or her diagnosis (Dennis et al., 2014).

Past studies have shown that parents withholding information until their child become an adult might impact his or her coping mechanisms, self-esteem, reproductive decision-making, a family cohesion (Metcalf et al., 2011). Individuals that are at-risk for genetic conditions have reported that they feel it is important to learn about their risk early on and at a developmentally appropriate level (Wehbe et al., 2009). Parents and children alike have recommended honesty, early disclosure, and continuing the discussion over time (Dennis et al., 2014). This allows the child to adapt and cope, provides an open stream of communication, and ensures the child is not receiving misinformation. Parents should also encourage their child to ask questions and be supported and nonjudgmental throughout any discussions (Metcalf et al., 2011).

Parents recognize the need to adapt discussions with their child to a developmentally appropriate level but often find they are unable to access advice and support for different developmental stages (Metcalf et al., 2011). Parents often feel uncertain as to what they should say, what vocabulary they should use, and how to prevent the conversation from exceeding their child's level of understanding (Dennis et al., 2014). When asked about their experiences, parents reported that they felt that they had a lack of support from healthcare providers in regards to discussing a diagnosis with their child (Metcalf et al., 2008). Along with parents, young individuals at risk for a genetic condition felt a need to meet with a healthcare professional with whom they could discuss their risk to become further informed (Metcalf et al., 2011). This highlights the importance of a relationship between a family at-risk for a genetic condition and their healthcare provider to ensure that a foundation is built upon which a child will be

supported and adequately educated about their risk and/or diagnosis (Sullivan & McConkie-Rosell, 2010).

Communication of a diagnosis is already a challenging process and becomes further complicated when the child has an intellectual disability. Parental experiences in a communicating a diagnosis to a child with an intellectual disability will elicit differing discussion strategies and parental concerns. Shearn and Todd (1997) reported that when a child has an intellectual disability, parents purposefully avoid conversation about the disability with their child due to a fear it will cause the child emotional distress. When parents chose to discuss their child's disability with him or her, they are more likely to use words such as 'difference' and may use that to focus their conversation on how their child is different from other children his or her own age (Jones et al., 2014). Parents emphasized that discussion should focus on details relevant to a child's life at that point in time and should be explained at a level the child can comprehend. This supports that a parent's awareness of his or her child's ability to understand and open communication within the family are key elements for successful discussion of a genetic condition with an affected child (Cunningham et al., 2000).

In this study, we explored parental experiences and attitudes of communicating a diagnosis of FXS to their affected child. Specifically, our aim was to gain an understanding of the challenges and successes parents have had as they discussed such a diagnosis with their child. Our main goal was to gather baseline information about the experiences parents had when communicating a diagnosis to their child with FXS.

Specific questions asked were as follows:

- 1) Had the parents pursued a conversation with their child about the FXS diagnosis?
- 2) How old was the child when parents chose to engage in that discussion?
- 3) Which aspects of the condition did parents find easier to explain?
- 4) What challenges did parents face during the discussion?
- 5) Why do parents choose not to discuss the FXS diagnosis with their child?
- 6) Did the presence of autism or an intellectual disability influence the experience?

The information gained through this study is expected to benefit genetic counselors and their patients with a diagnosis of FXS, as it will allow genetic counselors to better identify what these families need to successfully communicate about a diagnosis.

2.3 Materials and Methods

2.3.1 Participants. This study targeted parents of children diagnosed with FXS. Individuals were invited to participate if they met the following criteria: have a child who has been diagnosed by a medical professional with FXS; the diagnosis has been confirmed with genetic testing; participants are over the age of 18; participants have access to the Internet; and participants speak fluent English. Individuals were eligible to participate whether or not they have discussed their child's diagnosis of FXS with him or her. It was requested that participants with more than one child diagnosed with FXS focus on only one child throughout the questionnaire. If these participants were interested, they could fill out a second questionnaire for each child.

Invitations to possible participants were offered through posting of an announcement about the research study, shown in Appendix B, which provided a brief introduction to the study and a link to the standardized invitation and questionnaire on

SurveyMonkey. The announcement was posted on the National Fragile X Foundation Facebook, a FXS Facebook group, and the FRAXA research foundation website, electronic mailing lists, and social media outlets. Permission was obtained through each of the groups separately before posting of the announcement. The standardized invitation, shown in Appendix A, invited and explained the study to participants, provided contact information for the investigators, and allowed participants to begin the study. Participants were able to choose whether they wanted to continue with the survey upon reading the research description and requirements. Data collection was performed from July, 2015 through October, 2015.

2.3.2 Study methods. Potential participants were able to view the invitation letter to gain an understanding of the study and determine if they met eligibility and were interested in participating. A respondent was considered as a participant once he/she began the questionnaire, which was expected to take approximately 15-20 minutes for participants to complete. If a participant did not wish to answer a specific question, s/he could skip it and continue to the next question. At any time, a participant could withdraw from the study by not continuing the questionnaire.

At the end of the questionnaire, participants who had discussed a diagnosis of FXS with their child were invited to participate in an optional 15-minute telephone interview, which was used to collect qualitative data on parental experiences that may complement the findings from the questionnaire. If parents had not yet discussed their child's diagnosis of FXS with the child, they were excluded for participation in the interview section of the study. A sub-sample of parents who were eligible and agreed to be interviewed was randomly chosen for participation. To achieve this, a number between

1 through 10 was randomly drawn from a hat and possible interview candidates were chosen using that number. For example, the number seven was drawn so every seventh person who was eligible for an interview was selected until each of the eight interview slots was filled. Interviewees were contacted initially and a time was agreed upon to conduct the interview. If an interviewee could not be contacted after two attempts, a new candidate was randomly selected to fill the slot by determining who was the next seventh eligible individual.

Each parent was provided an explanation of the interview process on the phone and then asked four open-ended questions (see Appendix D). Additional questions were asked as necessary during interviews to ensure a thorough explanation of a participant's response.

Eight interviews were completed and each was recorded and transcribed by the principal investigator. The interviews were conducted from a private room and the audio files were stored on a password-protected computer. Once transcribed, the audio file was destroyed and any identifying information was removed from the transcripts. Other than contact information for the optional phone interview, no identifying information was collected from participants. Contact information was kept in a password-protected computer and destroyed after the completion of the study. This study was approved by the Institutional Research Board of the University of South Carolina, Office of Research Compliance, Columbia, SC, in June 2015.

2.3.3 Study measures. Demographic information was collected on participants including data about gender, ethnicity, highest level of education completed, and job title. An original questionnaire was created to assess parental experiences on communicating

about the diagnosis of FXS with his or her child, including why these conversations happen, when do parents choose to start these discussions, what resources are utilized, what thoughts and emotions are experienced, as well as what challenges and successes these conversations may bring to a family.

The questionnaire, (see Appendix C), contained multiple choice, select all that apply, and open-ended questions. The questionnaire utilized branched logic, with particular responses leading to a specific path of questions. Development of the questionnaire and interview guide was driven by interest in specific factors of parental communication to their child by the research team. A literature review focused on family communication, communication of genetic risk, and FXS was utilized to help inform and refine questions posed to parents. A team comprised of two genetic counselors and one licensed psychologist was consulted throughout development and helped refine the questionnaire based on their clinical and research experience. Four parents piloted the questionnaire for comprehension and readability, as well as to determine approximate questionnaire completion time. These results were not included in data analysis. The questionnaire was then revised based on their feedback and received further review by the research team for finalization.

2.3.4 Data analysis. Incomplete questionnaires of less than 80% were not included in analysis. Quantitative analysis was performed using SPSS Version 23, Microsoft Excel, and SurveyMonkey. Descriptive statistics were used to analyze demographic information and to calculate frequencies, proportions, and averages. Descriptive statistics were calculated based on the number of participants who answered a specific question. A chi-square test for independence was used to determine if there was an association

between the presence of autism or an intellectual disability in a child and the experience the family had with communicating a diagnosis. Qualitative data was analyzed using a directed content analysis to identify emerging and recurring themes. The development of the initial codes by the principle investigator (BAA) was guided by the initial literature review that focused on family communication, communication of genetic risk, and FXS. Following preliminary coding, the data was independently coded by a genetic counselor (AMR) and then reviewed with the primary investigator for clarification. BAA then systematically re-coded the data, with AMR assessing for consistency throughout. Upon completion of qualitative coding, the data was analyzed for themes providing insight to the parental experience of discussing a diagnosis of FXS with his or her child.

2.4 Results

2.4.1 Participant demographics. One hundred participants began the questionnaire, with 83 of those meeting eligibility requirements and completing greater than 80% of the survey. 17 responses were excluded. Respondent demographics are displayed in Table 2.1. The majority of participants were Caucasian ($n = 76, 97\%$), female ($n = 71, 90\%$), and completed at least some college ($n = 75, 95\%$). Five participants completed the questionnaire twice, while one participant completed the questionnaire three times, presumably for numerous affected children within the same family.

The majority of participants' children were reported to be males ($n = 65, 81\%$) under the age of 18 at the time the survey was completed ($n = 47, 64\%$), with an average of 16.05 years ($SD = 8.58, \text{range} = 2 \text{ to } 37$). Two participants reported their child as deceased. Age of diagnosis of FXS averaged at 4.89 years ($SD = 5.30, \text{range} = 0 \text{ to } 36$), with the most common response being children diagnosed 6 years or younger ($n = 65,$

Table 2.1 Participant Demographics

	Frequency (N = 83)	Percentage (%)
Gender	<i>n</i> = 79	
Female	71	90
Male	8	10
Ethnicity	<i>n</i> = 78	
Caucasian	76	97
Black or African American	2	3
Education	<i>n</i> = 79	
Did not graduate high school	0	0
High school	4	5
Some college	17	22
Associates degree	5	6
Bachelor's degree	26	33
Some graduate school	7	9
Masters/Doctoral degree	20	25
Primary Caregiver	<i>n</i> = 83	
Yes	81	98
No	2	2
Relationship	<i>n</i> = 83	
Mother	75	90
Father	8	10

81%). Three parents reported they had a prenatal diagnosis. The most common mental health diagnosis was intellectual disabilities ($n = 48, 59\%$) and when the option “other” was chosen, anxiety was the most commonly reported response ($n = 10, 59\%$). Parents often selected multiple comorbidities diagnosed in their child. The healthcare provider making these diagnoses most frequently was a developmental pediatrician ($n = 28, 35\%$). This data is displayed in Table 2.2. When questioned about use of a genetic counselor ($n = 82$), most parents reported seeing either a general genetic counselor ($n = 38, 46\%$) or a pediatric genetic counselor ($n = 21, 26\%$). Few had not seen a genetic counselor ($n = 14, 17\%$).

Table 2.2 Children Demographics

	Frequency (N = 83)	Percentage (%)
Gender	<i>n</i> = 79	
Male	64	81
Female	15	19
Current Age	<i>n</i> = 74	
6 years or younger	11	15
7 to 9 years	7	10
10 to 12 years	11	15
13 to 15 years	12	16
16 to 18 years	6	8
18 years or older	27	36
Age at Diagnosis	<i>n</i> = 83	
6 years or younger	68	82
7 to 9 years	6	7
10 to 12 years	3	4
13 to 15 years	3	4
16 to 18 years	1	1
18 years or older	2	2
Mental Health	<i>n</i> = 82	
Autism	43	52
Intellectual disability	48	59
Learning disabilities	37	45
ADD/ADHD	40	48
None of these	5	6
Other	17	21
Healthcare Professional	<i>n</i> = 80	
Pediatrician	24	30
Psychologist	23	29
Psychiatrist	16	20
Developmental Pediatrician	28	35
Neurologist	20	25
Not Applicable	1	1
Other	9	11

Parents reported most often that one of their child’s greatest strengths is his or her sociability (*n* = 25, 32%) with determination (*n* = 20, 25%) being the second most common out of all of the responses (*n* = 79). In regards to what parent’s felt their child excels at, it seemed that the choices provided did not fit well and almost half of the 76

respondents chose “other” ($n = 33$, 43%). Some of the more common answers provided when “other” was selected included using technology and having a great memory. The second most common response selected by respondents was music ($n = 18$, 24%), followed by reading ($n = 12$, 16%).

2.4.2 Initial communication of diagnosis. Participants were questioned about various aspects of their experiences with discussing FXS with their child. When parents were asked if they had shared with their child that s/he had been diagnosed with FXS, 57% ($n = 47$) responded yes. Forty-one of those 47 parents (87%), have discussed with their child what that diagnosis means for him or her. The majority of those parents ($n = 35$, 85%) had the first discussion about FXS with their child by the time s/he was 15, with the age ranges 7-9 and 13-15 being equally chosen and the most common ($n = 11$, 27%). Nineteen parents (49%) indicated that the discussion most often arose because parents wanted their child to have an understanding of his or her diagnosis ($n = 39$). Forty respondents reported that discussions typically involved the child’s mother ($n = 38$, 95%) and less often the father ($n = 14$, 35%).

When asked how they decided what information to include in their initial discussion with their child, 39 parents provided a response. Most parents seemed to base their discussion on helping their child understand his or her diagnosis ($n = 16$, 41%). Primarily, parents provided basic information at a level developmentally appropriate for their child. Additionally, parents had decided to discuss how their child was different from others in an attempt to help him or her better understand FXS ($n = 13$, 33%). These two approaches were intertwined by one mother who stated, “*Basically it was dependent*

on what he could understand, which was simply some things might take him longer to learn, might seem difficult.”

In their initial discussion of FXS with their child, parents most often included information about their child’s intellectual disability or learning difficulties ($n = 30$, 86%) and emotional or mental health challenges ($n = 18$, 51%). Parents felt it was easiest to discuss their child’s intellectual disability or learning difficulties ($n = 18$, 46%) and hardest to discuss potential medical interventions ($n = 12$, 32%). Figure 2.1 shows how often topics were included in discussion ($n = 35$) and which topics parents considered to be easier ($n = 39$) or more difficult ($n = 37$) to discuss.

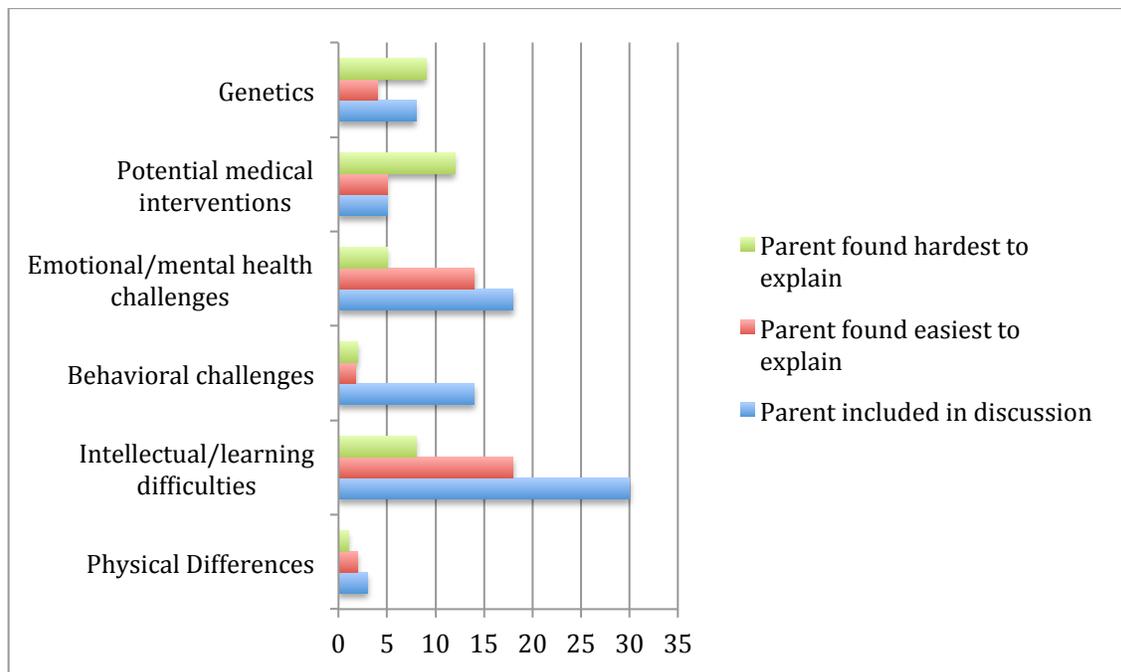


Figure 2.1 Topics Included During Initial Discussion and Topics Found Easiest and Hardest to Explain

Thirty-eight respondents provided insight into resources used in their initial discussion. Parents commonly reported that they did not utilize any resources during the initial discussion with their child ($n = 29$, 76%). When resources were used, healthcare

professionals ($n = 5$, 13%) and websites ($n = 4$, 11%) were selected most often. When asked about what resources would be useful for their discussion, the most common suggestion out of 26 responses was for an illustrated children’s book, written specifically for children with FXS at a level they would be able to understand ($n = 8$, 31%).

In an attempt to understand how these experiences affected parental emotions, we asked how parents were feeling before and after the discussion with their child. These responses, from 40 participants, can be seen in Figure 2.2 Prior to the conversation, parents most commonly reported feeling sad ($n = 17$, 43%) and calm ($n = 16$, 40%). This was compared to after the conversation, where parents still commonly reported feeling calm ($n = 23$, 58%) and more parents felt relieved afterwards ($n = 14$, 35%).

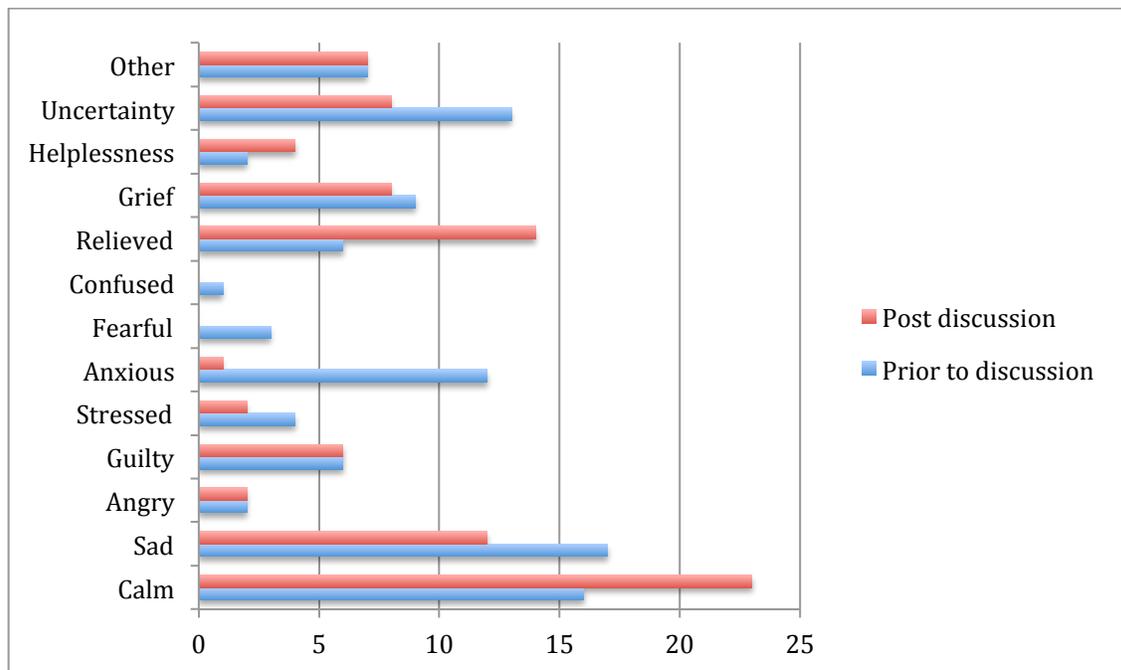


Figure 2.2 Parental Emotions Prior to and After Discussing FXS with Child

Parents considered what their initial worries and concerns were prior to talking with their child ($n = 40$). Parents most often shared about their worry that discussing FXS with their child will have a negative effect on the child’s emotional status or feelings

about him/herself ($n = 17, 43\%$). Parents shared that their worry focused on “...his feelings. That he’d feel worse about himself, feel less than others” and “...how she would react to the information.” Parents were also commonly concerned that their child would not understand what they were trying to explain ($n = 10, 25\%$). One mother shared that, “I was most worried that she wouldn’t ‘hear’ or understand what I was saying because she was already so emotional.” In an interview, a mother shared about her son’s abilities, stating, “I wouldn’t say profoundly affected, he was able to function and do everything but as far as his understanding – I don’t think he really understood a whole lot”

When asked about their concerns and worries after the initial conversation with their child, the majority of parents reported that their original concerns were eased through the discussion ($n = 22, 67\%$). However, some parents found their original concerns were replaced by new worries ($n = 8, 24\%$), while a minority felt their original concerns were intensified by the discussion ($n = 3, 9\%$). Figure 2.3 shows the spread of these 33 responses.

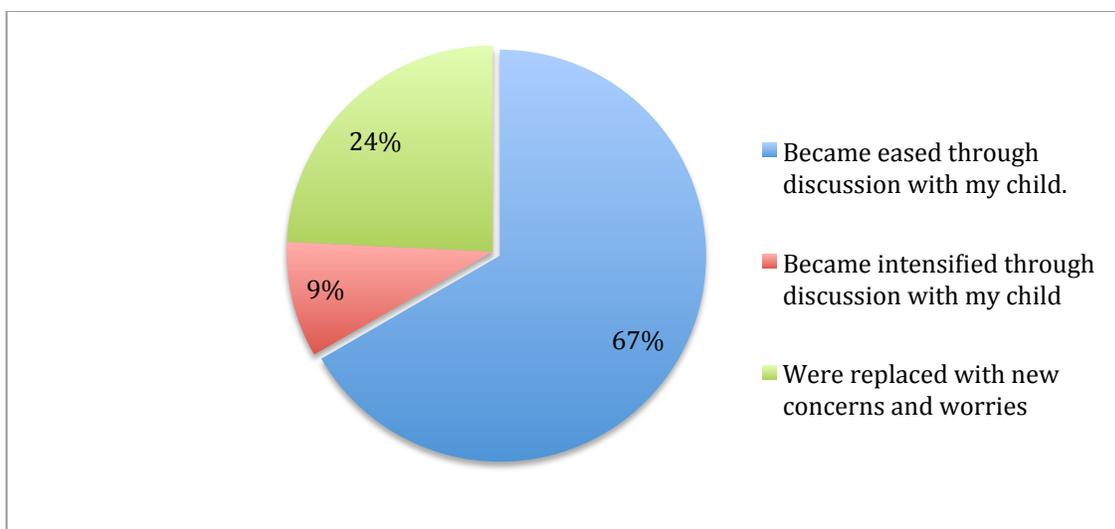


Figure 2.3 Discussion Effect on Initial Parental Concerns and Worries

For parents whose concerns were eased by the discussion, there commonly seemed to be a neutral or positive response from their child after the discussion or the parent felt comfortable with how the conversation went and the information provided. In an interview, a parent described her post-discussion experience as “...*a weight off my back*” and that she was “...*glad that I finally told her.*” When original concerns were replaced by new worries, it seemed to be driven by parental uncertainty of how the child would react to the information, how much was understood, and what might happen in the future. After talking with her daughter, one mother was “*Not sure what she would do with the information. Not sure if she would obsess over it*” while another worried “*What should I do now? Who would help us?*” For those that became intensified, one mother shared that her daughter “...*was upset about the diagnosis*” while another mother was concerned because her son “...*did and still does feel defective and ashamed*” after discussing his diagnosis of FXS.

Parents shared what they found most surprising during the initial discussion with their child about his or her FXS diagnosis ($n = 36$). Parents were commonly surprised at how well their son or daughter received the information s/he was provided ($n = 16, 44\%$). Parents indicated their surprise at “*How well she accepted and understood what we talked about*” and “*That he seemed to embrace it*” as well as with “*How easily he took the diagnosis.*” One father even shared that after his daughter found out about her diagnosis, she accepted it, and “...*when she would meet new people she was [sic] introduce herself and explain that she had fragile X.*” Overall, 29 parents (83%) did not identify a specific area where they lacked information to successfully discuss FXS with their child ($n = 35$).

2.4.3 Successive discussions. Out of the 39 respondents, the majority of parents reported that the diagnosis of FXS has been an ongoing conversation ($n = 27$, 69%) with their child. Twenty-six of these parents shared insight into what prompted successive discussions. Fifty-four percent of the time ($n = 14$), a child had a question or wanted to discuss something related to his or her diagnosis. For one parent, their child's questions "...would often result from a test of some new medication or dosage change" while another child "...wanted to know where he got fragile X." Less often, a significant event occurred related to a child's diagnosis ($n = 7$, 27%), and one mother shared, "*We try to use events that occur that relate to her condition to bring the subject up for discussion.*" In some families, a parent approached their child wanting to discuss his or her diagnosis ($n = 7$, 27%). In regards to her aging daughter, one parent discussed that "*As she got older we were able to talk about the genetics and how that would impact her decisions later in life.*" In another family, they "...talk about FX all the time. Especially when we are going to attend support group events or see other FX families, which we do often."

Similar to the initial conversation, most parents ($n = 13$, 54%) did not report using resources in successive conversations with their child ($n = 24$). The most commonly selected resources that were utilized included other parents whose children have FXS ($n = 4$, 17%) and websites ($n = 3$, 13%).

2.4.4 Advice from parents. Thirty-four parents shared what they found most helpful in facilitating discussion of a FXS diagnosis with their child. It was most commonly suggested to normalize the situation by using affected family members and other families as examples to show that the child is not alone with his or her diagnosis ($n = 9$, 26%). Parents achieve this by "*Making sure that she had interaction with others with*

fragile X of varying degrees of affectedness” and others have “...made it clear that fragile X affects many members of our family, not just my daughter so she doesn’t feel singled out and different.” A parent in an interview shared that what worked for their family was that they have:

Always kept it open, that we’ve kept it ‘this affects the whole family and we’re all in it together and it’s just part of who we are it’s not something bad it’s just a part of who we are. It’s not everything about us, it’s just a part of who we are. It’s something that we deal with and we go on with who we are and our daily life.

Parents were asked to share advice with other parents who are planning to discuss their child’s diagnosis of FXS with him or her ($n = 34$). A common piece of advice given was that parents should focus on strategies to help their child understand the information they will be providing ($n = 15, 44\%$). This included providing information in a basic, developmentally appropriate manner that is tailored to their child’s specific needs. One parent suggested *“If they have questions, answer them honestly at a level appropriate to the child’s understanding”* while another pointed out that it *“Depends on the cognitive function of the child, also the maturity.”* Another common piece of advice suggested was to maintain resilient communication ($n = 15, 44\%$). This involved the parent initiating conversation, using opportunities that arise to create a discussion with their child, and being open and honest with their child. Specific advice included *“Talk about it early and often...”* and *“Don’t wait, do it early and let the information grow with your child.”* Others recommended parents *“Phrase things in the most in the most [sic] positive manner possible and be factual like not judgmental”* and *“Try to use everyday impacts of*

FX as opportunities to discuss the condition, rather than having ‘the big talk’” and lastly “Be open and honest, give age appropriate information at a level they understand” because “There’s never going to be a perfect time or an easy time for some people to do this.” One parent shared:

Keep it simple, stress that everyone has something that makes them unique and special and fragile X is one of their unique qualities. That being aware of how it affects them is a tool to use in helping them overcome any obstacles. Make sure your child knows that you are approachable about this subject anytime. I liked to use specific examples, ‘you know you are a really good reader and I am not,’ or ‘I am good at cooking and my friend is not.’ We all have different skills. That is what makes the world interesting. Above all, be honest!

While it was not commonly brought up, one mother felt that parents should not put their child into a FXS box because of his or her diagnosis. Instead, parents should let them live to their full potential. This was echoed in an interview, where a mother said, “*Don’t put restrictions on them, you know?*” and that she explained to her son, “*You know, you learn differently but you can learn whatever you want.*”

There were no strong themes across parental responses when asked if they had any thoughts, feelings, or emotions that they would like to share that had not been previously addressed by our questionnaire ($n = 27$). A powerful quote arose from a mother whose daughter is affected that she wanted to share:

I know of many families with fragile X that want [sic] to keep it a secret, especially from their girls. I feel that this is not fair to the girls who grow

up thinking what is wrong with me or I can't wait to have kids and then wham [sic] they are told that they have a genetic disorder that could result in having a seriously disabled child. Additionally when parents act as if fragile X is something to be depressed about or unhappy about or something that should be kept secret, this can often affect how a child sees himself.

2.4.5 Parents have not discussed. Fifty-three percent of parents ($n = 44$) reported that they have not discussed with their child what a diagnosis of FXS means for him or her ($n = 83$). These parents commonly expressed that they have not pursued such a discussion because of the child's ability to understand ($n = 25, 57\%$) or the child's age ($n = 18, 41\%$) by sharing, "...his understanding is too low level" and "...my child is too young to understand." Approximately 77% of these parents feel that they will discuss FXS with their child in the future ($n = 34$), while 23% feel that they will not ($n = 10$).

2.4.6 Parents who will not discuss. Twenty-three percent of parents ($n = 10$) who have not discussed their child's diagnosis of FXS with him or her felt that they would never pursue that discussion ($n = 44$). When asked to share how they feel when considering talking to their child about his or her FXS diagnosis, parents most often reported feeling calm ($n = 4, 40\%$) and other ($n = 4, 40\%$). Those that selected other did not provide an alternative feeling, but instead expressed that their child did not have the cognitive abilities to understand and thus they did not foresee such a discussion occurring in the future. Sad ($n = 3, 30\%$) and uncertainty ($n = 3, 30\%$) followed as the next most common selected feelings. All parents who have no plans to discuss their child's diagnosis with him or her felt that their child's level of disability or understanding

contributed most to their decision ($n = 10$, 100%). There were no commonalities across responses when parents were asked if there were any thoughts, feelings, or emotions that they would like to share ($n = 10$). It was great to see positivity expressed by one parent, who responded and shared *“I am so very proud of my son(s) with fragile X. They are wonderful, compassionate, funny, loving human beings who prove to me every day that worth is not held in your abilities or lack thereof.”*

2.4.7 Parents who will discuss. Seventy-seven percent of the parents ($n = 34$) who have not discussed their child’s diagnosis of FXS with him or her felt they would broach the topic in the future ($n = 44$). When asked to consider at what age parents hoped to begin a discussion about their child’s diagnosis of FXS, 32 parents provided input. The age range of 7-9 years was most commonly selected ($n = 8$, 25%) followed by 10-12 years ($n = 7$, 22%). Out of 33 respondents, the majority of parents felt that they would be prompted to discuss with their child by their interest in their child having an understanding of his or her diagnosis ($n = 16$, 48%), with a significant event occurring that will warrant discussion as the second most common prompt for the discussion to occur ($n = 8$, 24%). The majority of participants feel that they would like for the child’s mother ($n = 29$, 88%) and father ($n = 25$, 76%) to be included ($n = 33$).

The majority of parents feel that in their initial discussion, they would like to discuss their child’s intellectual disability or learning difficulties ($n = 28$, 85%) followed by their child’s emotional or mental health challenges ($n = 22$, 67%). Parents indicated that it would be easiest to explain to their child about emotional or mental health challenges ($n = 16$, 52%) and hardest to explain to their child about genetics ($n = 19$, 61%). Figure 2.4 displays how often each topic was selected by parents to include in their

initial discussion ($n = 33$) and how often each topic was selected as being the easiest ($n = 31$) or hardest ($n = 31$) to explain.

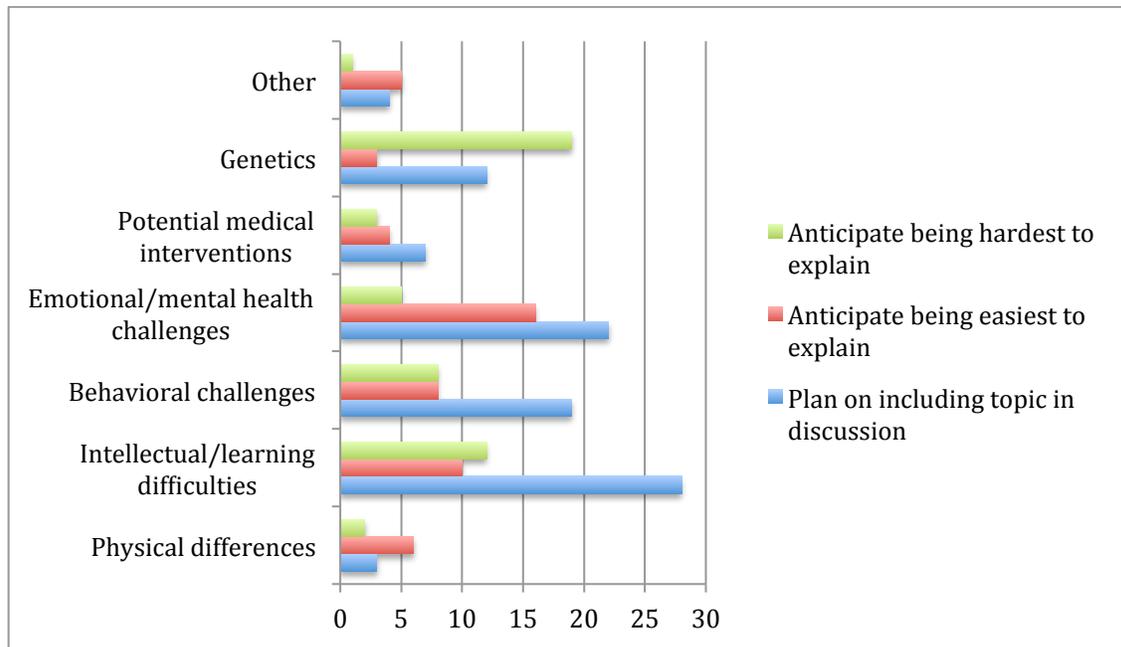


Figure 2.4 Interest in Including Topics in Discussion and Topics Anticipated to be Easiest and Hardest to Explain

When asked to consider how they will choose what information to share with their child, 25 parents shared their thoughts on the subject. Almost half of parents feel that they will practice resilient communication with their child through maintaining open conversation and discussing what is relevant at the time ($n = 11$, 44%), while others will base it on what they feel their child is capable of understanding ($n = 4$, 16%). When asked what resources they hope to use when discussing FXS with their child, pictures ($n = 13$, 41%) was selected most often. Figure 2.5 indicates how often each resource was selected by 32 parents hoping to use it during their discussion. When asked if there were any resources that would be useful in facilitating discussion with their child that parents did not feel they had adequate access to, 15 parents provided suggestions. Similar to parents who have discussed their child’s diagnosis with him or her, parents often thought

that it would be useful to talk with another family or healthcare professionals to learn about their experience with explaining FXS ($n = 5$, 31%) as well as to have a simple children’s book that explained FXS at a level written for their child ($n = 4$, 25%).

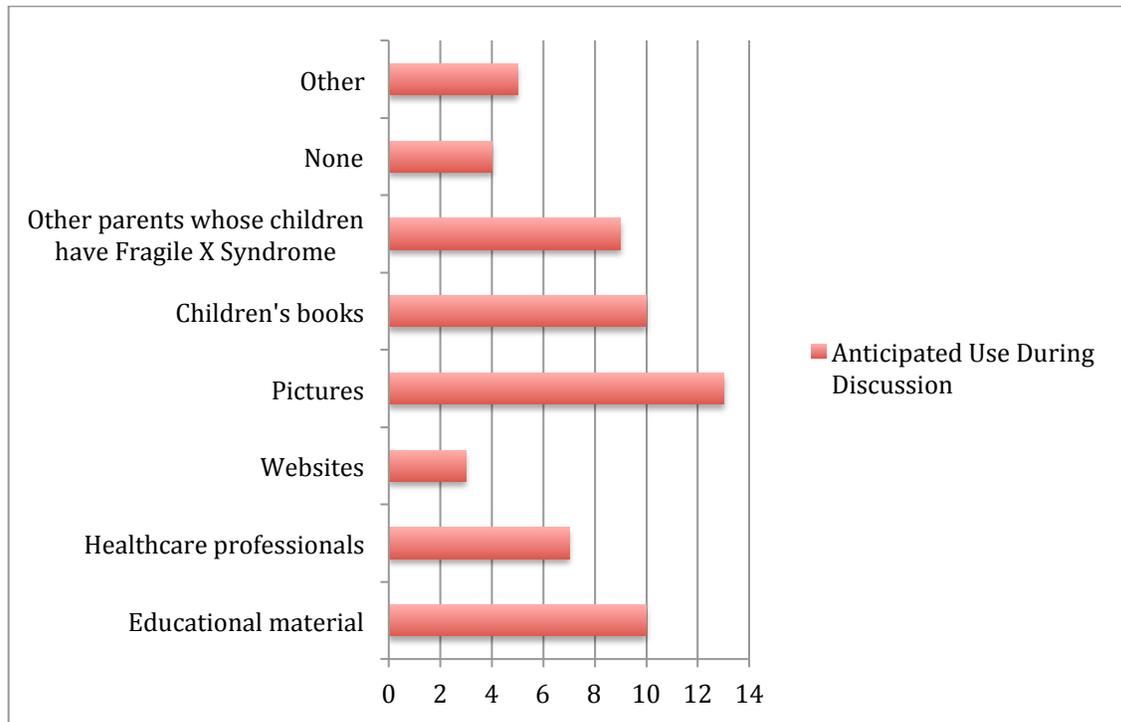


Figure 2.5 Anticipated Use of Resources by Parents in Discussion

Forty-seven percent of parents ($n = 15$) reported that they feel sad when considering discussing their child’s FXS diagnosis with him or her ($n = 32$). Anxiety ($n = 14$, 44%) and uncertainty ($n = 14$, 44%) were the next most common emotions selected by parents when thinking about such a discussion. Parents were asked to consider what they were most worried or concerned about in regards to talking with their child about FXS ($n = 28$). Parents frequently described that their major concerns included their child’s ability to understand ($n = 14$, 50%) as well as the conversation having a negative effect on the child’s emotional status or feelings of self worth ($n = 13$, 46%). Parent’s wonder “*How will he react to the fact that he is going to be different his whole life?*” and

worry *“That it will upset him.”* They also express concern as to *“If he would understand it”* and *“That he won’t understand what we are saying or that he won’t be able to tell us he understands or properly voice any questions that he may have.”*

Parents were asked if there was anything that they felt they did not have adequate information about or experience with to discuss with their child and the majority of the 22 respondents answered “No” with no further explanation ($n = 14$, 64%). Two parents suggested that it would be helpful to talk with other families affected by FXS to help aid in their discussion. Additionally, two other parents wished they had more information about their child’s long-term outlook, as they felt a lot of uncertainty of what to expect in the future in regards to their child’s diagnosis. There were no common themes when asked if there were any thoughts, feelings, or emotions that parents wished to share, but one mother expressed frustration because she needs *“...resources that I don’t believe exist. It is very difficult to get help and buy-in from providers with my daughter who has fragile x but is nearly typical.”*

2.4.8 Autism and/or intellectual disability present. When asked about comorbidity with FXS, 82 parents provided information on additional diagnoses. Seventy-five percent of parents selected their child had a diagnosis of autism, intellectual disability, or both ($n = 62$). Nineteen percent of parents indicated that their child had a diagnosis of learning difficulties, ADD/ADHD, or both of these ($n = 16$), while six percent indicated that their child had no additional comorbidities ($n = 5$) with their diagnosis of FXS. For all participants, the presence of autism and/or intellectual disability as reported by participants was not statistically associated with a child’s gender, use of genetic counseling by families, whether or not the parent told their child that he or she

had a diagnosis of FXS, or whether or not the parent discussed what FXS means with their child. For participants who have discussed a diagnosis of FXS with their child, the presence of autism and/or intellectual delay was not associated with what age the discussion occurred, how the discussion arose, or what effect the conversation had on parent’s initial worries and concerns. For participants who have not discussed, the presence of autism and/or intellectual disability was not statistically associated with whether or not parents who have not discussed will do so in the future, nor was it statistically significant at what age they would like to discuss and what will prompt discussion in the future for parents who will discuss (Table 2.3).

Table 2.3 Pearson Chi-square Test of Independence of the Presence of Autism and/or Intellectual Disability Compared to Numerous Variables

Variable	Chi-square value	Degree of freedom	<i>p</i>
All respondents			
Gender	1.49	1	.22
Genetic counselor use	8.15	5	.15
Shared diagnosis	1.25	1	.26
Discussed diagnosis	.02	1	.89
Parents have discussed			
Age at discussion	7.01	7	.43
How did topic of FXS arise	9.23	4	.06
Effect on initial worry	.31	2	.86
Parents have not discussed			
Will parents discuss	.159	2	.92
Parents will discuss			
Age at discussion	3.78	6	.71
Prompt for discussion	4.16	5	.53

2.5 Discussion

There has been minimal research completed that focuses on the communication of a diagnosis of FXS between a parent and their affected child. Most of the previous research focuses on parental communication with their child regarding other specific

genetic conditions and parental communication of genetic risk information to daughters about their FXS carrier status (McConkie-Rosell et al., 2011). Our study, which was comprised of eighty-three participants who have at least one child diagnosed with FXS, explored parental experiences with communicating information to their child about his or her diagnosis of FXS. The results from this study provide a valuable understanding of the process which families go through when communicating genetic risk information to their affected children in the presence or absence of autism and/or an intellectual disability.

Across all areas explored in our study, numerous themes were consistently seen, even when parents had not yet had a discussion with their child about FXS. A major commonality was that parents tended to support a resilient communication style, which has been described in previous literature on communicating information about a genetic condition. According to McConkie-Rosell et al. (2005), “Resilient communication was defined as any aspect of the conversation that emphasized the importance of honest, truthful, open communication between parent and daughter, an effort at reassurance, optimism, or an attempt to normalize the situation” (p. 60). We found that the parents in our study tended to initiate the conversation with their child, be open and honest, use opportunities as they arose to direct the conversation, continue the conversation over its natural progression, and respect the child’s right to know about his or her diagnosis. Dennis et al. (2014) further supports the idea of resilient communication, reporting that both parents and children alike recommend honesty, early disclosure, and continuing the discussion over time. We found that this occurred whether or not there was a presence of autism and/or intellectual disability in a child. In contrast, Shearn and Todd (1997) reported parents of children with intellectual disabilities purposefully avoided

conversation with their child about the disability and some even tried to conceal it from their child in an attempt to normalize life. It seemed that, at least within the FXS community, most parents want their child to have an understanding of their diagnosis instead of hiding it from him or her.

Parents felt it was important to find strategies to help their child understand their diagnosis, which reinforced the theme of resilient communication. In order to facilitate understanding, parents would often report simplifying information to a developmentally and age appropriate level. This aligns with previous studies that report that individuals at-risk for a genetic condition prefer to learn about their risk early on at a developmentally appropriate level (Webbe et al., 2009). Cunningham et al. (2000) asserts that a parent's ability to focus a discussion on an age and developmentally appropriate topic is contingent upon the parent's awareness of his or her child's ability to understand. This seemed to be an area of concern for some parents in our survey. Parents supplemented their explanation of FXS by providing concrete examples of what it means to have FXS to help further their child's understanding of the condition. Parents often identified a specific skill that their child struggled with or a behavior that s/he often exhibited which could be attributed to a diagnosis of FXS. By using concrete examples, parents can acknowledge their child's diagnosis without having a negative perspective. In contrast, Jones et al. (2014) described how some parents would avoid talking about their child's disability to instead focus on how his or her child was similar to other children and minimize the disability.

The strategies used by parents to ensure understanding were similar to strategies used to help the child manage his or her diagnosis and normalize the situation. Parents

would often use affected family members or other FXS families to normalize the diagnosis, allowing their child to feel more like others. Though they are not shielding their child from the diagnosis, as suggested by Shearn and Todd (1997), parents send the message of normalcy rather than shame. Though parents accepted the diagnosis and wanted their child to understand why s/he was different, they did not want the diagnosis to define their child. This is supported by prior research by Jones et al. (2014), which described how parents are more likely to use words such as ‘difference’ and will focus on how their child is different than other children, rather than use words such as ‘handicapped’ (Shearn & Todd, 1997). Instead of focusing on labels, parents chose to focus on differences they could explain to their child and highlighted their child’s strengths. A mother clearly demonstrated this practice through one of her responses where she shared, *“My son is so much more than his diagnosis. It’s always been a part of him, but never has it been all of him. Fragile X doesn’t define him.”*

When considering the repercussions of discussing their child’s diagnosis of FXS with him or her, parents were often worried about their child being unable to understand/accept the information provided. They were also concerned that the information they were providing would have a negative effect on the child’s emotional status or feelings about their self. We found that a child’s ability to understand was the primary reason a parent had not yet discussed FXS with him or her, which supported by the study by Cunningham et al. (2000) where parents felt there was “no point” in talking to their child about Down syndrome because of an inability to understand. Previous studies have suggested that when there is limited skill in comprehension by an individual, it could be helpful to utilize healthcare professionals to help ensure that necessary

information is being communicated properly (de Vries et al., 1999). When considering a present intellectual disability, prior research typically mentions a negative emotional status or negative feelings of self when a child's diagnosis has been hidden from him or her and the child eventually establishes an understanding without direct communication from his or her parents (Jones et al., 2014). In our study, we found that parents were concerned about their child having a negative self-image because of the discussion, not because it was avoided. Prior research tends to support the idea that when a family has a more open communication pattern, a child will often have a greater ability to cope in contrast to families with less open communication, which can lead to a child becoming stressed, withdrawn, and easily upset (Metcalf et al., 2011).

Our study found that approximately forty-eight percent of parents had discussed FXS with their affected child, while a recent study by Raspa, Edwards, Wheeler, Bishop, and Bailey Jr. (2016) found that seventy-nine percent of their cohort discussed FXS. This is likely because our study focused on children affected with FXS, while the study by Raspa et al. (2016) included premutation carriers as well as children who tested negative. When considering why parents have chosen not to tell their child about his or her test results, Raspa et al. (2016) found that the majority of participants felt that their child would not understand the results, which was commonly reported by parents in our study. Additionally, most parents in our study who had not already shared their child's diagnosis with him or her felt that they would share the diagnosis in the future when s/he is old enough to understand. This was supported by forty-six percent of participants in the study by Raspa et al. (2016) who had not yet told their child about his or her test results.

It was interesting to note overlapping characteristics between parents who have discussed FXS with their child and parents who have not but plan on discussing with their child. Both groups indicated that they felt it was most appropriate to begin the conversation with their affected child when he or she was between the ages of seven and nine. They also felt that this discussion should focus on information that is relevant at the time at a developmentally appropriate level. Children aged seven to nine are typically able to notice visible physical differences, recognize that a genetic condition may be a familial trait, and understand the specifics of a condition that they can see or related to their daily lives (Metcalf et al., 2011). It is difficult to determine how an intellectual disability affects a child's ability to understand, as it often depends on the severity of the disability. As numerous parents in our study pointed out, it is important to evaluate a child's cognitive abilities to determine at what level information should be provided a task that can be difficult for parents to navigate alone.

Mothers were much more likely to be involved in the conversation than fathers across both groups, but this could be due to the fact that 90% of the respondents identified as a child's mother. A study by Metcalfe et al. (2011) found that mothers are more likely to be involved in the sharing of genetic information, which could also be a likely cause of our findings. Most often, parents initiated conversation because they wanted their child to have an understanding of a FXS diagnosis, but even parents who had not discussed also felt the discussion might arise if a significant event occurred that required an explanation. The utilization of an event supports findings from Metcalfe et al. (2011), which described how discussing genetic risk information can be emotionally painful and difficult to bring up, and so the presence of a particular event that can prompt

discussion is a strategy often utilized by parents for discussion. Jowell et al. (2014) had similar findings, where parents would withhold information about a diagnosis until the child needed to see a specialist or had numerous medical appointments that needed an explanation.

Parents who have previously discussed FXS with their affected child most commonly discussed their child's intellectual disability or learning disability along with their emotional or mental health challenges. Similarly, parents who plan to discuss FXS with their affected child in the future also felt they would focus on these aspects of the diagnosis. This makes sense, as these are difficulties that a child with FXS might notice they are experiencing and question their parents about, which is a commonly described way that initial communication occurs (Dennis et al., 2014). These are also more concrete examples of what it means to have FXS, which was commonly reported by parents as a useful strategy to help their child understand his or her diagnosis. These topics were also commonly selected as easier to include in a parent's discussion with his or her child. This is unsurprising, as Cunningham et al. (2000) has reported that parents feel discussion should focus on details relevant to a child's life at the point in time of the discussion and both of these topics would be relevant if they are problematic for a child. Genetics was considered the most difficult topic to explain by both groups, which is likely due to both the generally complex nature of genetics, and the even more complicated nuances of the *FMR1* gene and the variability of presentation with FXS.

For parents who have previously discussed FXS syndrome with their child, they commonly reported using no resources during their discussion. This contrasts with parents who have not yet had the discussion, where only a small percentage anticipated

using no resources during their discussion. Instead, parents hoped they would be able to use pictures, children's books, educational material, and other FXS families to facilitate their discussion. Parents who did not use resources shared that an illustrated children's book written for their child with FXS may have been a helpful resource. Parents who have not discussed also suggested a children's book would have been useful, but they also thought it would be more beneficial to have access to another FXS family or healthcare professional to help with the experience. Previous studies have shown that parents feel they have a lack of support from healthcare providers in regards to discussing a diagnosis with their child (Metcalf et al., 2008). This sentiment was echoed by one of our parents, who shared, "*I specifically asked her regular pediatrician about how and when to explain it. He said I should wait to tell her. I do not believe this is good advice but don't know how to address it with my daughter.*" This highlights how important it is for healthcare professionals to be knowledgeable about various conditions and be willing and able to help parents navigate through the process of discussing the condition with his or her child. This is especially significant for the small group of parents who expressed concern that they could not perceive how much their child was able to understand, as that makes these discussions much more difficult and confusing. Metcalfe et al. (2011) describes how parents recognize the need to adapt discussions to their child's specific abilities, but find themselves unable to access advice and support for different developmental stages. By having a knowledgeable healthcare provider who is familiar with typical developmental milestones and a child's particular abilities, parents can develop a partnership to ensure that a solid foundation of information exists to build upon with further discussion of genetic information (Sullivan & McConkie-Rosell, 2010).

Approximately 13% of our participating parents who have discussed their child's diagnosis have utilized healthcare professionals. However, it is difficult to know what the outcome of those conversations were as we did not specifically ask.

Both groups commonly felt sad prior to having a discussion about FXS with their child. For parents who have not discussed their child's diagnosis, anxiety and uncertainty were the next most commonly selected feelings, which is not unusual considering the thought that must go into such a conversation. Parents who have previously discussed the diagnosis reported feeling calm and sad prior to the discussion, and afterwards felt calm and relieved. It is curious that parents who discussed the diagnosis felt calm before and after, which does not typically align with prior research on the subject. One explanation for this finding may be attributed to research bias. These findings suggest that not only does the discussion benefit the child by providing information but that it could also be beneficial for the parents. Many reported that they were surprised at how well their child took the information, which leads to the parents also having a more positive experience.

Based on prior studies, we thought that the presence of autism and/or an intellectual disability might influence whether or not a parent chose to discuss FXS with their child. Interestingly, there seemed to be no statistically significant association between the presence of autism and/or intellectual disability and whether or not the conversation was previously initiated or whether or not it would be discussed in the future. Previous reports illustrate how parents often choose to avoid conversation and conceal a disability from their child for various reasons, which is not what we found in our study (Cunningham et al., (2000); Jones et al., 2014; Shearn & Todd (1997)

2.5.1 Study limitations. Dissemination of the questionnaire through online methods limited the population to individuals with internet access, while utilization of FXS-based websites and social media sites, along with the parent's ability to self-select to participate may have biased the participant population. The majority of respondents identified as Caucasian and female, which is likely not representative of the general population and prevents this study from being generalizable. Though no systematic study has been conducted for an accurate estimation, previous studies have suggested the prevalence of FXS is overall similar across various ethnicities/races (Crawford et al., 1999; McConkie-Rosell et al., 2005). The information collected from parents who have had a discussion with their child about his or her diagnosis is retrospective, and may not accurately represent what actually occurred in the past. Our study focused on children who were diagnosed with FXS and participation was based on parent report that their child has an actual diagnosis of FXS with a full *FMRI* mutation. Some areas of interest, such as experience with healthcare professionals, were difficult to explore due to the nature of the original question or due to lack of follow up questions for more information.

2.5.2 Future research. Additional studies on this topic addressing the previously mentioned limitations would likely provide more generalizable data. The questionnaire could also be restructured to allow for a more detailed quantitative analysis on the subject. Furthermore, more interviews could be completed and then an in-depth analysis conducted to determine if additional themes arise when parents are able to share more detailed, verbal responses to questions about their experiences with discussing FXS with their child. Specifically, it would be interesting to query parents on outcomes when they attempted to utilize resources that they did not feel they had easy access to, such as

healthcare providers. While the majority of parents reported seeing a genetic counselor, it would be interesting to determine how many parents had follow up visits and investigate how a genetic counselor could aid in the discussion. With parents consent, brief interviews with children diagnosed with FXS would shed insight into what these children are gleaned from conversations with their parents about a FXS diagnosis.

Numerous parents suggested that a developmentally appropriate illustrated book for their child with FXS would be helpful in facilitating discussions. Future research can help determine the content of such a book so that a team of experts can create such a resource for the FX community. Additionally, it could be interesting to focus on healthcare professional's contributions and what they feel their role is in facilitating parental communication with a child about a genetic diagnosis, but this may have to be across general genetic disorders and not so narrow as to be about FXS in specific.

2.6 Conclusions

This study has implications for the field of pediatric genetic counseling, specifically in the presence of a FXS diagnosis, as it was conducted to help provide insight on how parents communicate a diagnosis of FXS to their child that has been diagnosed with the condition. Approximately half of the respondents reported that they discussed their child's diagnosis with him or her while the other half have not. Parents often report that is important to initiate and maintain open and honest communication with their child, along with utilizing strategies to help their child understand and cope with their FXS diagnosis. This allows for the child to be knowledgeable about their diagnosis and also leads to a more positive experience for both parent and child.

Parents often found it easiest to discuss intellectual disability or learning problems and emotional or mental health challenges with their child. These topics are often relevant to the child and can be explained by the parent with examples. Parents seemed to consistently struggle with discussing the genetics of FXS with their child, likely due to the complex nature of inheritance of the *FMRI* gene and the variability seen with the syndrome. Parents commonly reported no use of resources in past discussions, while parents who had yet to discuss hope to utilize written resources or healthcare professionals. Both groups desired an illustrated children's book, written at a developmentally appropriate level for their child to be able to utilize in their discussion to help facilitate understanding of the FXS.

Prior to discussing their child's FXS diagnosis, parents were most often worried that the discussion would lead to their child having reduced self-esteem or a negative self-perception. Parents were also commonly concerned about their child's ability to understand the discussion, which was the leading cause of parents postponing or avoiding discussing FXS with their child. Parents were commonly surprised at how well their child responded to the conversation and generally, the worries that parents initially had were dispelled.

2.6.1 Implications for practice. This study provides insight for genetic counselors to gain an understanding of what parents experience when they discuss or consider discussing a diagnosis of FXS with their affected child. While the primary focus was on parents who have previously discussed with their child, it also gathered data on parents who planned to discuss in the future, as well as those who never planned to discuss. Many facets of discussing a FXS diagnosis were explored with the hopes that genetic

counselors can easily recognize what a family may need in order to begin a discussion with their child. Once these needs are recognized, genetic counselors should be able to provide appropriate education, support, resources, and referrals to ensure successful communication between a parent and child. Essentially, genetic counselors should be willing to create a partnership with parents to help facilitate understanding of a diagnosis and ensure that a child is able to properly understand, cope with, and manage the information.

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Appendix A: Participant Invitation

Dear Potential Participant:

You are invited to participate in a graduate research study focusing on communication by parents who have a child with FXS. I am a graduate student in the Genetic Counseling program at the University of South Carolina School of Medicine. My research investigates the experiences of parents when they communicate to their child a fragile X diagnosis. Your role in the research study includes completing an online questionnaire and participating in an optional telephone interview.

We are asking parents to tell us about their experiences in discussing their child's diagnosis with him or her. A parent is eligible to participate even if they have not yet discussed their child's diagnosis with him or her. This information will be used to give us an understanding on why these conversations happen, when do parents choose to start these discussions, as well as the challenges and successes that such a conversation may bring. It should take about 15-20 minutes to complete. Please complete only one survey for each child – you may submit additional surveys if you have multiple children diagnosed with FXS. If you do not wish to answer a certain question, please skip it and continue on to the next question.

All responses gathered from the surveys will be kept anonymous and confidential. We only ask for your name and phone number in the event that you are interested in an optional telephone interview at a later date. It is not necessary that you provide this information. The results of this study might be published or presented at academic meetings; however, participants will not be identified. Your participation in this research is voluntary. By completing the survey, you are consenting that you have read and understand this information. At any time, you may withdraw from the study by not completing the survey.

Thank you for your time and consideration to participate in this survey. Your responses will help genetic counselors create more helpful education materials for parents and caregivers of children with FXS. If you have any questions regarding this research, you may contact either myself or my faculty adviser, Crystal Hill-Chapman, PhD, LP, ABPP, NCSP 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.

Aly Athens, BS
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Appendix B: Research Study Announcement

Parents in the fragile X Community!

Researchers at the University of South Carolina are looking for parents who have at least one child with fragile X to participate in a survey regarding their experience with explaining the diagnosis to their child with fragile X.

To participate you must be 18 years or older and have at least one child with FXS. Our goal is to gain an understanding of the successes and challenges parents face when they discuss their child's diagnosis of fragile X with him or her. You are eligible to complete the questionnaire even if you have not yet discussed your child's diagnosis of fragile X with him or her. If you choose to participate in this study, you will be contributing to the fragile X Community and will benefit other families who pursue genetic counseling for a diagnosis of fragile X in the future.

If you have any questions or concerns, please contact Aly Athens at aly.athens@uscmed.sc.edu.

To read more about this study and begin the questionnaire, please click the link below:

<https://www.surveymonkey.com/r/2VZB37T>

Appendix C: Online Questionnaire

1. Are you the child's primary caregiver?
 - Yes
 - No

2. What is your relationship to your child with FXS?
 - Mother
 - Father
 - Grandmother
 - Grandfather
 - Adoptive mother
 - Adoptive father
 - Aunt
 - Uncle
 - Other (please specify)

3. One of my child's greatest strengths is his/her
 - Eagerness to learn new things
 - Determination
 - Sociability
 - Humor
 - Independence
 - Passion
 - Creativity
 - Other (please specify)

4. What does your child excel in? Please select all that apply.
 - Art
 - Music
 - Reading
 - Writing
 - Mathematics
 - Communication
 - Science
 - History
 - Athletics
 - Other (please specify)

5. Please describe how your child speaks.

For example: my child speaks fluently, communicates clearly, speaks in short phrases that are hard to understand, has difficulty expressing his or her thoughts and feelings, does not communicate verbally, etc

6. Please describe how well your child understands.

For example: my child has difficulty understanding general information, has difficulty understanding multiple meaning words, demonstrates literal understanding of language, has difficulty comprehending questions, has difficulty understanding what to do, etc.

7. At what age, in years, did your child receive their fragile X diagnosis?

8. Has a medical professional diagnosed your child with FXS?

- Yes
- No
- Unsure

9. Did your child receive genetic testing to confirm their diagnosis?

- Yes
- No
- Unsure

10. Did you see a genetic counselor?

A genetic counselor is a healthcare professional trained in medical genetics and counseling to provide risk assessment, education, and support to families at risk for or affected by a genetic condition.

- Yes, we saw a prenatal genetic counselor
- Yes, we saw a pediatric genetic counselor
- Yes, we have seen a prenatal and pediatric genetic counselor
- Yes, we saw a general genetic counselor
- No, we have never seen a genetic counselor
- I am unsure if we have seen a genetic counselor

11. Has a healthcare provider ever diagnosed your child with one of the following mental health conditions? Please check all that apply.

- Autism
- Intellectual Disability/Mental Retardation
- Learning Disabilities
- ADD/ADHD
- None of these

- Other (please specify)
12. Who diagnosed your child’s mental health condition in the previous question?
Please check all that apply.
- Pediatrician
 - Psychologist (PhD)
 - Psychiatrist (MD)
 - Developmental Pediatrician
 - Neurologist
 - A healthcare professional has not diagnosed my child
 - Other (please specify)
13. Where did your child receive their mental health diagnosis? (e.g., Texas Children’s Hospital, Houston, TX; Lester Elementary School, Florence, SC)
14. Have you shared with your child that s/he has been diagnosed with FXS?
- Yes
 - No
15. Have you discussed with your child what that diagnosis means for him or her?
- Yes
 - No
16. About how old was your child when you first began the conversation about your child’s fragile X diagnosis with him or her?
- 6 years old or younger
 - 7-9 years old
 - 10-12 years old
 - 13-15 years old
 - 16-18 years old
 - 19 years old or older
 - I do not remember
17. How did the topic of your child’s fragile X diagnosis first come up?
- My child had a question about something related to his or her diagnosis
 - We wanted our child to become familiar with the words “FXS” but did not discuss it further at that time
 - A significant event occurred that warranted discussion
 - We wanted our child to have an understanding of his or her diagnosis
 - Other (please specify)
18. Who was involved in the initial discussion? Please check all that apply, including yourself.
- Mother
 - Father

- Older brother/sister
- Younger brother/sister
- Grandparents
- Primary Care physician
- Genetic Counselor
- Other (please specify)

19. In our initial discussion, I chose to talk to my child about his or her (please select all)

- Physical differences
- Intellectual/learning difficulties
- Behavioral challenges (ADHD, hand flapping, etc.)
- Emotional/mental health challenges (e.g. general anxiety, social anxiety, etc)
- Potential medical interventions (e.g. doctors appointments, clinical trials, etc.)
- Genetics (e.g. X linked inheritance, number of trinucleotide repeats, stability and expansion, etc.)
- Other (please specify)

20. How did you decide what information to provide to your child during that initial discussion?

21. In our initial discussion, I found it easiest to explain to my child about his or her (please select all)

- Physical differences
- Intellectual/learning difficulties
- Behavioral challenges (ADHD, hand flapping, etc.)
- Emotional/mental health challenges (e.g. general anxiety, social anxiety, etc)
- Potential medical interventions (e.g. doctors appointments, clinical trials, etc.)
- Genetics (e.g. X linked inheritance, number of trinucleotide repeats, stability and expansion, etc.)
- Other (please specify)

22. In our initial discussion, I found it hardest to explain to my child about his or her (please select all)

- Physical differences
- Intellectual/learning difficulties
- Behavioral challenges (ADHD, hand flapping, etc.)
- Emotional/mental health challenges (e.g. general anxiety, social anxiety, etc)
- Potential medical interventions (e.g. doctors appointments, clinical trials, etc.)

- Genetics (e.g. X linked inheritance, number of trinucleotide repeats, stability and expansion, etc.)
- Other (please specify)

23. Did you use any resources during your initial discussion? Please choose all that apply.

- Educational books
- Healthcare professionals
- Handouts or pamphlets
- Websites
- Pictures
- Children's books
- Other parents whose children have FXS
- Support groups
- None
- Other
- If possible, please provide specific examples of resources used as selected above

24. Are there any resources you feel would have been useful to have during your discussion but you didn't have access to or feel adequately prepared to use? Please briefly explain.

25. How did you feel prior to talking with your child about his or her fragile X diagnosis? Please choose all that apply.

- Calm
- Sad
- Angry
- Guilty
- Stressed
- Anxious
- Fearful
- Confused
- Relieved
- Grief
- Helplessness
- Uncertainty
- Other (please specify)

26. Prior to beginning the initial conversation with your child, what were you most worried or concerned about?

27. What did you find most surprising during the initial conversation with your child about his or her fragile X diagnosis

28. How did you feel after talking with your child about his or her fragile X diagnosis? Please choose all that apply.
- Calm
 - Sad
 - Angry
 - Guilty
 - Stressed
 - Anxious
 - Fearful
 - Confused
 - Relieved
 - Grief
 - Helplessness
 - Uncertainty
 - Other (please specify)
29. After our initial conversation, I found that my original concerns and worries
- Were eased through discussion with my child
 - Became intensified through discussion with my child
 - Were replaced with new concerns and worries
 - Please briefly explain your response
30. Is there anything you felt you did not have adequate information about or experience with to successfully discuss with your child?
31. Has your child's diagnosis been an ongoing conversation with him or her?
- Yes
 - No
32. What prompted further conversations about your child's diagnosis after the first discussion? Please choose all that apply.
- My child had a question or wanted to discuss something related to his or her diagnosis
 - A significant event occurred related to my child's diagnosis
 - I approached my child wanting to discuss his or her diagnosis
 - Other
 - Please briefly explain your response
33. Did you use any resources in successive discussions with your child that you did not use in the initial discussion? Please choose all that apply.
- Educational books
 - Healthcare professionals
 - Handouts or pamphlets
 - Websites
 - Pictures

- Children's books
 - Other parents whose children have FXS
 - Support groups
 - None
 - Other
 - If possible, please provide specific examples of resources used as selected above
34. What have you found most helpful, either in initial or successive conversations, in facilitating discussion with your child about his or her diagnosis?
35. What advice would you give to other parents planning on discussion their child's diagnosis with that child?
36. Do you have any thoughts/feelings/emotions you would like to share that have not been addressed in this survey?
- Yes
 - No
37. Are you interested in receiving a phone call for a 15-minute follow-up discussion about your experiences?
- Yes
 - No
38. Thank you for your willingness to follow up this survey with a brief interview via the telephone. Interested individuals will be randomly selected for the interview. Please provide the following information so we are able to contact you:
- First name
 - Contact number
 - Time and day that works best for you to receive a telephone call
 - Please list a second time and day to receive a telephone call
39. Please explain what has contributed to your decision not to discuss your child's fragile X diagnosis with him or her at this point in time?
40. Do you feel you will have this discussion at some point in the future with your child?
- Yes
 - No
41. How do you find yourself feeling most often when you consider talking with your child about his or her fragile X diagnosis? Please choose all that apply.
- Calm
 - Sad
 - Angry

- Guilty
- Stressed
- Anxious
- Fearful
- Confused
- Relieved
- Grief
- Helplessness
- Uncertainty
- Other (please specify)

42. What factors have contributed to your decision not to talk to your child? Choose all that apply.

- My child's level of disability or understanding
- We don't talk about FXS in my family
- Fear that my child will think s/he is different
- Guilt about my carrier status
- I don't feel qualified to adequately explain my child's diagnosis to him or her
- Other

43. Do you have any thoughts/feelings/emotions you would like to share that have not been addressed in this survey?

44. At what age, in years, do you hope to begin this conversation with your child?

- 6 years or younger
- 7-9 years
- 10-12 years
- 13-15 years
- 16-18 years
- 19 years or older

45. Which of the following would most likely prompt you to initially discuss your child's fragile X diagnosis with him or her?

- My child has a question about something related to his or her diagnosis
- We want our child to become familiar with the words "FXS" but will not discuss it further at that time
- A significant event occurring that will warrant discussion
- We want our child to have an understanding of his or her diagnosis
- Other (please specify)
- Please briefly explain

46. Who do you plan on involving in the initial discussion? Please check all that apply, including yourself.

- Mother

- Father
- Older brother/sister
- Younger brother/sister
- Grandparents
- Primary Care Physician
- Genetic Counselor
- Other (please specify)

47. In your initial discussion, which of the following do you plan on discussing with your child? Please select all that apply.

- Physical differences
- Intellectual/learning difficulties
- Behavioral challenges (ADHD, hand flapping, etc.)
- Emotional/mental health challenges (e.g. general anxiety, social anxiety, etc)
- Potential medical interventions (e.g. doctors appointments, clinical trials, etc.)
- Genetics (e.g. X linked inheritance, number of trinucleotide repeats, stability and expansion, etc.)
- Other
- Please briefly explain your response

48. How did you decide what information you will provide to your child during the initial discussion?

49. I think it will be easiest to explain to my child about his or her (please select all that apply)

- Physical differences
- Intellectual/learning difficulties
- Behavioral challenges (ADHD, hand flapping, etc.)
- Emotional/mental health challenges (e.g. general anxiety, social anxiety, etc)
- Potential medical interventions (e.g. doctors appointments, clinical trials, etc.)
- Genetics (e.g. X linked inheritance, number of trinucleotide repeats, stability and expansion, etc.)
- Other
- Please briefly explain your response

50. I think it will be hardest to explain to my child about his or her (please select all that apply)

- Physical differences
- Intellectual/learning difficulties
- Behavioral challenges (ADHD, hand flapping, etc.)

- Emotional/mental health challenges (e.g. general anxiety, social anxiety, etc)
- Potential medical interventions (e.g. doctors appointments, clinical trials, etc.)
- Genetics (e.g. X linked inheritance, number of trinucleotide repeats, stability and expansion, etc.)
- Other
- Please briefly explain your response

51. What resources, if any, do you hope to use during your discussion? Select all that apply.

- Educational material
- Healthcare professionals
- Websites
- Pictures
- Children's books
- Other parents whose children have FXS
- None
- Other
- If possible, please provide specific examples of resources as selected above

52. Are there any resources you feel would be useful to have during your discussion but don't have access to or feel adequately prepared to use? Please explain.

53. How do you find yourself feeling most often when you consider discussion your child's fragile X diagnosis with him or her?

- Calm
- Sad
- Angry
- Guilty
- Stressed
- Anxious
- Fearful
- Confused
- Relieved
- Grief
- Helplessness
- Uncertainty
- Other (please specify)

54. Prior to beginning the initial conversation with your child, what are you most worried or concerned about?

55. Is there anything you feel you do not have adequate information about or experience with to successfully discuss with your child?
56. Do you have any thoughts/feelings/emotions you would like to share that have not been addressed in this survey?
57. How old is your child currently, in years?
58. Is your child that has been diagnosed with FXS a
- Male
 - Female
 - I choose not to specify
59. What is your gender?
- Male
 - Female
 - I would prefer not to specify
60. What is your ethnicity?
- White
 - Hispanic or Latino
 - Black or African American
 - Native American or American Indian
 - Asian/Pacific Islander
 - Other
61. What is your highest level of education completed?
- Did not graduate high school
 - High school degree
 - Some college
 - Associates degree
 - Bachelor's degree
 - Some graduate school
 - Masters/Doctoral degree
62. What is your job title?

Appendix D: Interview

Follow-up Interview

Thank you for agreeing to share your experiences with us! Your involvement in this study will help us to create guidelines for genetic counselors to better assist parents with the process of communicating a diagnosis of FXS to their children. To follow up the questionnaire, we have a few questions that we will ask you today. The interview will be recorded and transcribed to ensure accurate recollection. After the recording is transcribed, the sound recording will be destroyed. We appreciate your willingness to join us in this study!

1. Is there anything you wish you would have known prior to the initial conversation with your child?
2. What details stand out to you from the first discussion you had with your child?
3. Please explain what you feel your child's understanding of your discussion was.
4. On the questionnaire you were asked how you felt before and after the discussion with your child. Can you please discuss how you were feeling during the discussion?

If any answers to the questionnaire or interview are unclear, we will follow up on those during the interview.