THE EXPERIENCES AND DECISION MAKING PROCESSES OF COUPLES WITH A CHILD WITH A CHILDHOOD GENETIC DISORDER

By

Kate Szubeczak McKee

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ABSTRACT

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There is a disparity in the research between the individual's and the family as a whole's interactional processes in the collection and dissemination of genetic information. There is a gap in our understanding of how couples deal with genetic information, especially when a child is diagnosed with an inherited genetic condition. Also, there is a lack of understanding of the decision-making processes regarding genetic testing. Furthermore, medical professionals and family therapists are becoming more aware of the effect these tests will have on family systems and on the health beliefs of the family. Once a diagnosis has been received, families now have the task of making sense of the diagnosis and deciding on how the family system needs to change or adapt in order to accommodate the genetic disorder.

The purpose of this qualitative study was to understand the lived experiences of couples who have a child suffering from a genetic disorder (affected child) and to understand the couple's experiences of receiving the diagnosis of an inherited genetic disorder in a child, and how they as a couple, and by extension, their family, adapted to that diagnosis. Nine couples from the Midwestern United States were interviewed using a semi-structured, joint interview process. Using thematic analysis and the couples' own words, two studies emerged from the data. The focus of study 1 was on the decision-making processes about genetic testing and the decision to have future children of couples who have a child with an inherited genetic disorder.

The focus of study 2 was on how the family adapted after receiving the diagnosis for their child. Implications and further considerations for family therapists are addressed.

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CHAPTER 1: INTRODUCTION

Background of the Problem

Genetic testing in the United States started in an experimental laboratory the late 1950s (Feetham & Thomson, 2006). Since the completion of the Human Genome Project in 2003, genetic tests have become widely available, and at relatively low cost, for more than 1,000 genetic disorders (Feetham & Thomson, 2006). As the human genome is more completely understood and new genetic tests are developed to detect both the predisposition to, and the presence of, specific diseases, medical professionals and family therapists are becoming more aware of the effect these tests will have on family systems and on health beliefs of the family. Both the benefits and risks of genetic testing are high (Johnson, Wilkinson, & Taylor-Brown, 1999). Predictive genetic testing benefits couples by allowing them access to numerous preemptive medical care options that they likely would not have otherwise known about. Diagnostic genetic testing can confirm diagnoses and substantiate the appropriateness of targeted therapy (Peterson, 2005).

Prenatal testing for genetic conditions/mutations is also available to individuals and families. Examples of such testing include prenatal diagnosis (PND), performed using amniocentesis during the first 10-18 weeks of pregnancy, and preimplantation genetic diagnosis (PGD), performed before a woman becomes pregnant using in vitro fertilization (Dewanwala et al., 2011). Several private PGD centers report that cystic fibrosis, hemoglobin disorders, and muscular dystrophies are among the most common genetic disorders for which PGD is used (Hershberger et al., 2012). In both cases, depending on the results, the couple can choose whether to continue the pregnancy or whether to implant only unaffected embryos. Over 10,000

babies have been born worldwide after pre-implantation screening using PGD (Kosicka-Slawinska, Clarke, & Lashwood, 2013). With the help of testing procedures such as PND and PGD, parents are able to gain a greater sense of control over the health and wellbeing of their future children (Dewanwala et al., 2011). This sense of control can extend to the screening and implantation of only unaffected embryos through IVF or by selective termination of the embryos during the early weeks of pregnancy (Gotz & Gotz, 2006; Kelly, 2009; Myring et al., 2011).

The primary risk associated with all types of genetic testing is that such testing can reveal information that may not be actionable by the couple. Said differently, even after receiving a prediction or diagnosis, couples may not be able to act to change a likely outcome. This inability to act can increase anxiety, alter family relationships, and result in both perceived and actual discrimination and stigmatization (Street & Soldan, 1998; Van Riper & Gallo, 2006). For couples who give birth to an affected child, this inability to act can take on a different meaning. Parents of a child who has been referred for genetic testing or whose child has a genetic disorder report an elevated level of anxiety due to the concern about the health and future of their child, and upon receiving results indicating an abnormality, may feel worried, hopeless, and guilty (Dinc & Terzioglu, 2006).

Not only must the parents of a child with a genetic disorder consider the time, energy, and costs involved in the medical management of the genetic disorder; they also face other potentially negative effects in multiple other areas including decreased self-esteem and self-worth of the diagnosed child, disruption of parent-affected child relationship, parent-unaffected sibling relationship, or sibling-sibling relationships, along with an increase in parental anxiety and guilt (Knafl, Knafl, Gallo, & Angst, 2007). Several studies have identified a number of sources of stress for families who have a child with a genetic disorder, including a lack of

understanding of risk, lag time between genetic discoveries and available treatments, fear of loss of privacy, stigma, and possible discrimination in employment, insurance reimbursement, or by school systems (Knafl, Knafl, Gallo, & Angst, 2007; Kelly, 2009). Research has demonstrated that once a child has been diagnosed with a genetic disorder there are numerous implications for parental mental health and overall family functioning including parental feelings of denial, anger, guilt, and depression (Jacobs & Deatrick, 1999), lower relationship and sexual satisfaction (Quaid & Wesson, 1995), greater relationship strain (Manne et al., 2004) and the real and perceived losses of child and family functioning with the diagnosis of a genetic disorder (Jacobs and Deatrick, 1999). On average, families with a child with a disability face greater levels of stress and dysfunction than other families (Collins & Williamson, 2003). However, little is known about the role these factors play in parent decision-making about future pregnancies. The proposed study seeks to understand the experiences of these parents with a genetic condition diagnosis in one of their children, and the processes related to a couple's decision-making about future pregnancies.

In a study completed by D'Amico et al. (1992), the authors found that among couples with previous experience of a child affected with genetic disease, a significantly higher percentage (65.5%) of couples avoided further pregnancies as compared to couples who opted for further reproduction (34.5%). Surprisingly, couples with a first affected deceased child were far less likely (13.6%) to refrain from reproduction, and thereby more likely to reproduce. In a similar study, the majority of parents of a child affected with a genetic disorder simply chose to avoid future pregnancies, so as to avoid any decisions about prenatal screening or testing to identify whether that child may be affected (Kelly, 2009). Although prenatal diagnosis and preimplantation diagnosis are made available to families who are at high risk for genetic

abnormalities or who are carriers of genes that cause childhood onset disorders, many couples choose not to test for a number of personal, ethical, or religious reasons. Based on feelings of guilt or disrespect for the first-born, in the case of selective termination, or if the parents are unsure of whether they could make a decision about termination, many parents prefer to choose not to test and thereby wait and see if their child is affected (Gotz & Gotz, 2006). To the contrary, a study done of parents of children diagnosed with cystic fibrosis (CF) (Ormond et al., 2007) found that the majority of parents were likely to pursue (or had already pursued) future pregnancies. In fact, the initial experience of diagnosis and care of a child with CF lessened any anxiety about caring for future children with CF.

An additional study found that when parents perceived a medical test as a genetic test, the condition at issue was perceived as uncontrollable and threatening. This implies that perceptions of a self-regulation model of illness are involved. Said differently, how people think about a disease is determined by the label that the disease is given (Senior, Marteau, & Peters, 1999). Some couples consider genetic testing terrifying. It makes sense then that these couples are less likely to pursue such testing. Many other couples pursue access to genetic tests as those tests become commercially available (Campbell & Ross, 2005). Nevertheless, the methodology used by parents to arrive at decisions regarding future pregnancies is unclear. The proposed study seeks to understand what variables contribute to a couple's decision about pursuing future pregnancies in view of an existing disorder.

There is a disparity in the research between the individual's and the family as a whole's interactional processes in the collection and dissemination of genetic information. The lack of understanding between the psychological processes between the individual versus the family, places families at an increased risk for altered relationships (Feetham & Thomson, 2006). It is

known that potential stigma exists in families regarding genetic illnesses. Many times information is shared within a family based on relationship proximity, and not based genetic risk (Campbell & Ross, 2005). Some studies have begun to address the role that extended family members play in the coping and management of a child with a genetic disorder, as well as in the decision making processes of the couple. With regards to how families and individuals mutually influence each other (Feetham & Thomson, 2006; Bubolz & Sontag, 1993), and can affect an individual's interpretation and response to genetic information (McDaniel, Rolland, Feetham, & Miller, 2006), one study suggested that family functioning depended less on the specific genetic condition identified and more on the patterns of family response to the psychosocial challenges of having a child with a genetic condition (Knafl, Knafl, Gallo, & Angst, 2007).

Once a couple decides to proceed with genetic testing, their primary care physician or other medical specialist can order the test. Before the couple has a genetic test they are typically referred to a genetic counselor. The role of the genetic counselor is to help the couple understand the testing procedure, the benefits and limitations of the genetic test and the possible consequences of the test result. To be more specific, according to the National Society of Genetic Counselors website (www.nsgc.org), "genetic counseling is the process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease". Families meet with a genetic counselor once or twice, discuss family history, discuss options for genetic testing and what the results may mean, and review the test results. The families are then are referred back to their primary care physician or to a specialist for future treatment.

Unfortunately, a quick review with a genetic counselor and primary care physician is typically not sufficient to fully process all of the emotions, facts, and processes related to the

outcome of the testing. In fact, it is well recognized that "as the technology of medical genetics surges forward, the emotional needs of families often get left in the wake. Families are often left on their own to find important information and support, and for many, the Internet is the only place to turn" (p. 454, Wamboldt). Said differently, if families are still seeking information after meeting with the genetic counselor or have not had all of their questions answered, the families are typically left on their own to find it. This can be particularly harmful not only because inaccurate information can be found, but also because accurate information is many times not appropriately decipherable by the family. Findings from one study suggest that a therapy approach to genetic counseling may be key to family and child functioning (Knafl, Knafl, Gallo, & Angst, 2007). This therapy would focus on facilitating communication and problem-solving within the family rather than just disseminating information to other family members. In addition, this therapy would also seek to answer some of the potentially unanswered questions generated by the families after meeting with the genetic counselor and/or primary care physician.

Research is still needed to study the family management of genetic information and subsequent treatment (McDaniel, Rolland, Feetham, & Miller, 2006). The current study seeks to address how couples and families have previously communicated about and currently communicate, about genetic information, how these couples and families cope with negative genetic test results, and what role the medical system has played in the family management of genetic information.

Given that each individual in the couple brings his/her own belief systems and multigenerational history of coping with illness, loss, and other adversity, the purpose of this qualitative study is to understand the lived experiences of couples living in the Midwestern United States who have a child suffering from a genetic disorder (affected child). In addition,

this study explored how the couple views future family planning and related health care decisions. In this study, the affected child will be defined as a child who has been diagnosed with a genetic disorder that "causes significant morbidity and mortality" (p. 549, Dewanwala, 2011).

Theoretical Framework

Family Systems Illness Model

The Family Systems Illness Model (FSI) provides a systemic framework for assessing the interaction of a disease or illness with an individual and the family system (Rolland, 1987b). The model emphasizes the interface between time, relative to disease progression and family developmental phases, illness type, and components of family functioning (Rolland, 1987a). Not unlike Bronfenbrenner's Ecology Theory (Bubolz & Sontag, 1993), the FSI model highlights the interactive nature between the type of illness/disability/loss, the individual, family, and illness life cycles and the family's ability to cope. This model also highlights the family's belief systems, culture, ethnicity, spirituality, and gender, wherein each one is nested in the other (Rolland & Williams, 2006). The FSI model was originally developed to provide a framework for psychosocial types of chronic illness and disability before there was a need to include a genetic component of illnesses. The typologies for genetic disorders that will be used in this study are described in detail below as related to the Family Systems Genetic Illness model. Having an understanding of the family system's reaction, behavior, and adaptation to illness over time and in relationship to different points in the family life cycle, is crucial for the practitioner to help the family to navigate current and future illness crises (Rolland, 1987a; Rolland & Williams, 2006). There are four key elements of the family illness belief system: 1) the family's sense of mastery and control over the illness; 2) the family's multigenerational evolutionary

process with illness, loss, and crisis; 3) the family's ethnic, cultural, and religious beliefs; and 4) the family's assumptions about the etiology of illness (Rolland, 1987b).

Family Systems Genetic Illness Model

Until recently, none of the health behavior models could accurately capture the experience of families and individuals as they went through the process of predictive genetic testing. In 2005, Rolland and Williams developed the Family Systems Genetic Illness Model (FSGI) to describe the course of genetic testing and to be used in conjunction with the FSI model (Rolland & Williams, 2005; Rolland & Williams, 2006). Based on a systemic framework, the FSGI model helps organize thinking about the interaction between genetic testing, important family and social networks, and the professionals involved in providing care (Rolland & Williams, 2006). This model was the first model to emphasize the genetic component of health and uniquely included the nonsymptomatic and symptomatic phases of a disease, as well as the interaction between individual and family life cycles (Rolland & Williams, 2006).

One particularly unique portion of the FSGI model is the creation of a psychosocial typology of genomic disorders. This typology classifies different categories of genomic disorders across several dimensions: 1) likelihood of developing a condition based on genetic mutations; 2) overall clinical severity; 3) timing of clinical onset in the life cycle; and 4) whether effective treatment interventions exist that can alter clinical onset of progression (Rolland & Williams, 2005). These four dimensions play an integral role in how a family or individual decides whether to pursue genetic testing, and how they may cope with the results. Some genetic conditions currently included in this typology are breast cancer, Huntington's disease, Hemophilia, Tay-Sachs disease, Hemochromatosis, and Alzheimer's disease. These diseases are examples of those in which genetic mutations or alterations contribute to the expression of the disease and for

which predictive, pre-symptomatic, or carrier testing is available (Rolland & Williams, 2005; Rolland & Williams, 2006).

Another unique aspect of the FSGI model is the addition of a time element. Rolland and Williams (2005) discuss the importance of addressing time and state that, "we need a model that considers the unfolding of illness-related developmental tasks over the entire course of a disorder" (p. 5). This element of time not only addresses the onset and course of illness throughout a lifetime, but, with the availability of genetic testing, the FSGI model adds another dimension of time, i.e., the time associated with the nonsymptomatic phases of a (potential) genetic illness. The FSGI model explains that the nonsymptomatic phases are the phases before a person decides to pursue genetic testing. The nonsymptomatic phases are awareness, crisis I pretesting, crisis II test/posttesting, and long-term adaptation (Rolland & Williams, 2005; Rolland & Williams 2006). In the awareness phase, families are aware of a possible genetic risk or mutation associated with having a child, but are not interested in any kind of testing at the time. In the crisis I pretesting phase, the family is actively considering genetic testing. The decision to test can coincide with major life transitions (e.g. the decision to get married or to have children). The decision to test can also coincide with a life assessment, such as the realization that a person is the same age as a close relative that suffered, or died, from a particular genetic disease. These types of decisions can be revisited several times. It is during the crisis II test/post-testing phase that families have decided to test, and must adapt and grieve depending on the results of the tests. The final phase, long-term adaptation, is the time span between receiving a positive test result and the time in which the condition manifests.

Research Questions

Based on the literature reviewed above, the following research questions were used to guide this study.

1.) What were participant experiences with the medical system before, during, and after the genetic testing process for their affected child?
2.) How did the diagnosis and related medical system interfaces affect individuals in their roles as spouses/parents?
3.) How did the diagnosis in their child affect medical decision-making processes of the couple, especially around having additional children?
4.) What types of supports and resources did the couple utilize as they moved through the diagnosis and related decision making processes?

Rationale and Significance

The rationale for this study is derived from the researcher's desire to assist couples as they navigate the decision making processes associated with genetic testing of any embryo after a couple already has one child with a genetic disorder, and recognize what information may be needed to make decisions about future family planning in relation to the embryo. These couples already have a child who has received a diagnosis of a genetic disorder and the couple may be unsure whether to have additional children based on their experiences with their diagnosed child.

Family therapists and medical professionals will benefit from an increased understanding of how couples communicate about genetic testing, the ramifications of receiving a positive test result, and the role of extended family in the process. Therapists and professionals will also benefit from better understanding how couples communicate about future family planning decisions in the view of a positive test result. Furthermore, couples who are considering genetic

testing and family planning will also benefit from a greater understanding of the aforementioned concepts as related to informed decision-making and family planning guidance.

CHAPTER 2: REVIEW OF LITERATURE

Decision Making in the Family

The family strongly influences health behaviors (Peterson, 2005; Rolland & Williams, 2005). For this reason, it is important to understand the process of decision making within the couple related to their health behaviors. At a familial level, decision-making is a process of reducing dissonance between and among family members (Bubolz & Sontag, 1993) and is the central control system of family organization. This reduction of dissonance leads to less stress and conflict around critical decisions. At its most basic, decision-making involves: 1) the recognition of need for decision, 2) identifying and weighing acceptable alternatives, and 3) selecting or mediating an alternative and facilitating its action (Paolucci, Hall, & Axinn, 1977). From a different perspective, Hastie (2001) describes decision-making as involving three components: 1) choice options and courses of actions; 2) beliefs about objective states, processes, and events in the world, including outcomes states and means to achieve them; and 3) desires, values or utilities that describe the consequences associated with the outcomes of each action event combination. In addition, decisions can also carry meaning for individuals and families in ways that may not make sense to others outside the family (Patel, Kaufman, & Arocha, 2002). In short, the mutual decision making required by families, especially concerning stressful issues, is complex, emotional, and often stressful.

In families, decision making can involve a seeming unlimited number of possible outcomes (Bubolz & Sontag, 1993) and can be a complex process. Not only are families faced with making many types of decisions (Bubolz & Sontag, 1993), but family decisions are also influenced by the environment in which the family functions, which can either restrict decision

making or offer opportunities (Paolucci, Hall, & Axinn, 1977). In many instances, family decisions involve group decisions (Bubolz & Sontag, 1993) and group decision-making processes. Since family decisions are typically made by more than one family member, it is necessary to analyze decision making from an ecological perspective, because each family member is different, with a different psychological make-up. Moreover, each member brings their unique experiences into the decision-making process, (Paolucci, Hall, & Axinn, 1977) uses their own personal style of thinking to make their way through the decision-making process, with different meanings for their experiences, values, expectations, and uses of information (Anderson, 2007).

Risk, Family Decisions, and Genetics

There are two types of risk. A first type of risk is viewed negatively. Knowledge of this type of risk constitutes a heightened awareness of potential unknown dangers or hazards that then becomes a major source of anxiety within the family. A second type of risk is viewed positively. Knowledge of this type of risk helps families predict the likelihood of events occurring in the future and allows the family to feel empowered and more in control of their actions and medical destiny (Hallowell et al., 2004). Both of these types of risk are evident in families considering genetic testing. An understanding of genetic risks, both positive and negative, is considered crucial for informed decision-making (Henneman, Marteau, & Timmermans, 2008).

In addition, many family decisions are made under conditions of uncertainty and risk (Bubolz & Sontag, 1993). The concept of risk is especially highlighted relative to predictive genetic testing, where decisions of risk are paramount because the information gleaned from such testing can drastically alter an individual's sense of self, can change personal identities, and

can cause extreme anxiety and stress (Hallowell, Foster, Eeles, Ardern-Jones, & Watson, 2004). Risk embodies aspects of ambiguity and uncertainty. When linked with genetic inheritance, risk can be very difficult to deal with from a personal and familial perspective (Peters, Djurdjinovic, & Baker, 1999).

Decision-making in families does not rely solely on one family member. Rather, the decision is shared on some level by all family members (Paolucci, Hall, & Axinn, 1977). In making decisions about whether, when, or how genetic information should be evaluated and shared, the interests of both the individual being tested and those with whom they are genetically linked must be considered (Van Riper & Gallo, 2006).

It is important to elicit each family member's understanding of his or her genetic risk and any previous health beliefs prior to beginning to convey risk information to the family (Peters, Djurdjinovic, & Baker, 1999). Genetic testing and risk knowledge can disrupt core beliefs (Rolland, 2006). Bartle-Haring and Gregory (2003) emphasize that the genetic testing decision-making process has a psychological impact on everyone who is genetically linked, and not just on the proband (i.e., person of genetic interest). Several steps can be taken to communicate genetic risk information within the family. First, it is important to construct an accurate family history across generations (McCann et al., 2009). Second, genetic risk information given to one member of the family should be circulated to the rest of the family members (Bowen, Bourcier, Press, Lewis, & Burke, 2004). Third, decisions about what future genetic testing or healthcare decisions should be discussed among family members (Bowen et al., 2004). Shared health beliefs help family members find meaning in their illness experience, whether current or future, and help guide decisions and actions (Rolland, 2006). Research indicates that families who discuss illness and genetic risks as a family, are more likely to participate in genetic testing (McCann et al.,

2009). These discussions may also serve as social support functions to build relational bonds and to facilitate coping among family members (Peterson, 2005).

Risk perception correlates with decision making about genetic testing (Lerman, Croyle, Tercyak, & Hamann, 2002), and perceived risk can be more influential than objective risk (Hiraki, Chen, Roberts, Cupples, & Green, 2009). Understanding perceived risks for complex genetically based diseases can improve risk communication, optimize coping strategies, and encourage the uptake of preventive health behaviors (Hiraki et al., 2009). Decision-making is not merely dependent on obtaining accurate risk information for weighing pros and cons or to resolve or mitigate risk (Anderson, 2007). Decision-making is a complex process that can involve emotion based- and/or rational- modes of perceiving and responding to information (Hopwood, 2000), and using these modes in the context of personal relationships (e.g. within couples and families) and professional relationships (e.g. with doctors and genetic counselors) (Anderson, 2007).

Reproductive Decision Making Related to Genetic Testing

Decisions to have children are usually not made solely by one individual, but typically as part of a couple. When considering the potential genetic risks, couples often make these reproductive decisions not in isolation, but with input from family members, health care workers and others (Klitzman, Thorne, Williamson, Chung, & Marder, 2007). One study showed that 67% of the women made the decision to genetically test prenatally with their partners. However, no women reported making the decision alone (Humphreys, Cappelli, Aronovitch, Allanson, & Hunter 2008). Another study confirms that decision making concerning genetic testing is primarily a joint enterprise within the couple (Kenen, Smith, Watkins, & Zuber-Pittore, 2000). Genetic testing has made it possible to clarify uncertainties before or when making reproductive

decisions (Downing, 2005). However, the decisions that couples make in situations of genetic risk are not based solely on risk information. Instead, these decisions are multifaceted and depend on factors such as desired family size, perceived risk, social context, and personal experiences with the genetic condition (Myring et al., 2011). Beeson and Golbus (1985) and Myring et al. (2011) found that couples with personal experiences of a genetic condition (i.e., those who already had an affected child) had a better understanding of the issues regarding further reproductive decisions, as compared to those couples with little experience of a genetic condition were unclear in their decision making about future children.

Prenatal genetic counseling and testing emphasize couple decision-making. This counseling is not only a potential stressor, particularly when spousal conflict or disagreement exists regarding testing, but can also act as a source of comfort and support (Humphreys et al., 2008). Factors considered by the couple in reproductive decision making include interests of self, interests of current offspring, interests of future offspring, preferences of spouse, preferences of family of origin, obligations to broader society (Klitzman et al., 2007). Religious, ethical, and intellectual values have also been shown to influence the decision making process (Pivetti & Melotti, 2013). In one study, race/ethnicity was associated with the degree of influence on prenatal genetic testing (Learman et al., 2003). For example, African Americans and Latinas were more likely influenced by their faith/religion, while Caucasian women were most likely to report that their health care provider would influence their decision-making. Couples may discover that making decisions about subsequent children involves finding a balance between wanting to have another child, the future needs of existing children, and the at-risk parent (Downing, 2005).

Decisions About Genetic Testing in Children

Many common genetic conditions occur in families with no previous family history or experience of the disorder (Collins & Williamson, 2003). In their study on prenatal genetic testing, Pivetti and Melotti (2013) found that people who had a good understanding of genetic testing and who could count on the support of family members and friends showed a positive attitude toward genetic testing. In making the decision about whether to undergo prenatal genetic testing, women and their partners must consider age-related risk information, the physical risks to the fetus associated with the procedure (Verp, 1992), and their own values and attitudes concerning pregnancy termination and the possibility of raising a child with a genetic disorder (Humphreys et al., 2008; Moyer et al., 1999). The most frequent reasons which lead a couple to consider genetic testing include the worry that the baby is at an increased risk of genetic disorder, the need to make a decision about family planning, a desire for relief from uncertainty, and the intention to take better care of their own health (Pivetti & Melotti, 2013; Moyer et al., 1999). One study found that receiving a positive (i.e., confirmatory genetic) prenatal diagnosis may be experienced by couples as a traumatic event with long term consequences (Nazare, Fonesca, Gameiro, Canavarro, & Datilio, 2011).

In dealing with the uncertainty and anxiety that can come with the decision for genetic testing, it is unclear what information is necessary for couples and families to make the decision that is best for their family and situation. Broadstock & Michie (2000) discovered that an informed decision, i.e., one based on all relevant information, may not be straightforward because people differ in how much information they need or want. Moreover, couples may be looking for an effective decision that is both informed and consistent with the decision maker's values. Disclosure and communication about genetic testing appears to be fairly open among

family members for common, treatable conditions such as inherited cancers. However, this may not be true for more severe and stigmatizing disorders such as sex-linked or recessive conditions (e.g., cystic fibrosis) or Huntington's Disease (Peterson, 2005).

The dynamics operating between partners, particularly the extent to which the views of each partner are respected or taken into account by the other partner are relevant to the nature of couples' decision making process, especially with genetic testing (Daniels, Lewis, & Gillett, 1995). Specifically, the more men perceived that their wives valued and accepted their opinions, and who felt more appreciated by their wives, the more women felt that the decision was shared and that couple's agreement about genetic testing was high (Nazare et al., 2011). If genetic testing is a subject that causes tension within the relationship, then it is likely that both men and women will forge alliances with others, such as extended family or medical professionals (Daniels, Lewis, & Gillett, 1995). One study found that couples in a position to consider genetic testing revealed the genuine care and concern they expressed for their future child(ren) and their understanding of the profound significance of their decision, regardless whether the couple tests or not (Hershberger et al., 2012).

In summary, couples have much to consider when faced with a decision about genetic testing. First, couples need to consider what family members may be involved with the testing or who they would want to include in the decision making process (Bubolz & Sontag, 1993), knowing that each person brings their own experiences, values, and way of thinking to the process (Anderson, 2007). Second, couples must understand that inherent in genetic testing is a certain amount of risk (Lerman, Croyle, Tercyak, & Hamann, 2002), and testing has a psychological impact on everyone genetically linked, not just the individual being tested (Bartle-Haring & Gregory, 2003). Third, couples must understand that reproductive decision making is a

joint enterprise by the couple (Kenen, Smith, Watkins, & Zuber-Pittore, 2000) based on numerous factors, including preferences of spouse and self, (Klitzman et al., 2007), perceived risk, (Myring et al., 2011), and religious and ethical values (Pivetti & Melotti, 2013). Finally, it is necessary for couples to gather information and assess their personal values about the possibility of pregnancy termination or raising a child with a genetic disorder when deciding about genetic testing in children (Humphreys et al., 2008).

Family Adaptation After Receiving a Diagnosis

As the human genome is more completely understood and new genetic tests are developed to detect both the predisposition to, and the presence of, specific diseases, medical professionals and family therapists are becoming more aware of the effect these tests will have on family systems and on the health beliefs of the family. Families can know about a genetic diagnosis sooner than ever before and they can also learn about the expected course of a disorder over time. Once a diagnosis has been received, families now have the task of making sense of the diagnosis and deciding on how the family system needs to change or adapt in order to accommodate the disorder. A genetic disorder diagnosis brings an array of challenges to parents of a child with a genetic disorder, and depending on the severity and prognosis, can lead to considerable stress on all in the family system.

Emotional Roller Coaster for Parents

The literature is clear on how stressful the diagnosis of a genetic disorder in a child is for parents. For the majority of parents, receiving the diagnosis was understandably the worst experience of their lives, but for most, it did not change the way they loved their child. Studies describe a wide array of emotions including anguish, bitterness, confusion, despair, devastation, disbelief, distress, fear, grief, guilt, loneliness, numbness, sadness, shock or upset (Strehl &

Middlemiss, 2007, Nusbaum et al., 2008). Upon receiving a diagnosis of Fragile X Syndrome (FXS), most mothers recounted being on a "huge emotional rollercoaster" consisting of being devastated on the one hand about the seriousness of the condition, and relieved on the other that their child had a definitive diagnosis (Visootsak et al., 2012). In a study done by Lord, Wastell, & Ungerer (2005), researchers reported parents of a child diagnosed with Phenylketonuria, a manageable, inherited childhood genetic disorder, experienced a mild trauma reaction when first learning about the diagnosis and learning how to manage the disorder. Fonseca, Nazare, & Canavarro (2014) found that adjustment had improved and there was a reduction in symptoms related to anxiety and depression for parents six months post diagnosis of a chronic illness of a child.

Activities to Manage the Diagnosis

Managing the diagnosis requires many activities that invariably include frequent visits with medical professionals. The literature suggests that the role medical professionals play during the diagnosis and subsequent care of children is crucial. In several studies, it was very common for parents to report that initial difficulties in obtaining clear, balanced, and useful information about their child's condition and questions about future functioning contributed significantly to their feelings of distress, confusion and frustration. These findings support other studies which have concluded that parents benefit most from receiving clear, balanced, up-to-date written and verbal information, with follow-up appointments for further clarification (Helm et al., 1998; Schuth et al., 1994; Statham et al., 2000). Parents report that experience contact with health-care providers is a vital connection to expertise, to advice, to reassurance, to instructions (Grob, 2008). In one study, parents reported that positive encounters with doctors had a significant impact on their coping, while negative encounters had an inverse effect on coping

(Chaplin, Schweitzer, & Perkoulidis, 2005). In another study, participants reported both negative and positive experiences with their medical professionals; however, the negative experiences outnumbered positive experiences 2.5 to 1 and, as a result, this was still a net negative experience for these parents (Goff et al., 2013).

The Long Term Burden of a Genetic Diagnosis of a Child on Families

Several studies suggest that for many genetic disorders, substantial burdens are created for families, and these burdens are not time limited, but often endure for years and decades. Some families report they live in a state of hyper vigilance, heightened anxiety, and emotional exhaustion (Kratz et al., 2009). The psychosocial burden experienced by family members of affected children may begin to wear on families over time. For example, some couples report marital disharmony, limitations on their time, financial distress, and hindrance from enjoyment of life (Panepinto et al. 2005; Tunde-Ayinmode 2007). For example, mothers of children with Sickle Cell Disease (SCD) have commonly reported daily emotional challenges of constant fear related to their children's possible death, loss of control over their lives, and helplessness (Wonkam et al., 2014). Pelchat (1993) found that the way in which one perceives the problem is an important predictor of adaptation. The impact of chronic illness on family functioning and how the family adapts is contingent on a range of factors including limitations to finances, spouse and sibling relationships, extended family relationships and the ability to cope (Ashton, 2004). As families transition from the initial diagnosis to regular, ongoing care, one study showed that every-day life is often more stable in families that have dealt with a child's illness for a relatively long period of time (Trzcieniecka-Green, Bargiel-Matusiewicz, Wilczynska, & Omar, 2015). Moreover, parents of children with a chronic illness eventually develop a shared view of the illness, its management, and its impact on family life (Knafl & Zoeller, 2000), which

helps with the overall transition and adaptation of families coping with a chronic illness.

CHAPTER 3: RESEARCH DESIGN AND METHOD

This chapter describes the research procedures for the study including the research design, methodology, the data collection, human subjects, and analysis procedures. This chapter also addresses the rationale for using qualitative methodology and issues of trustworthiness, credibility, and ethics.

Research Design

Parents who give birth to a child suffering from a genetic disorder, i.e., an affected child, have many concerns and must cope with many struggles. Not only can the parents face anxiety and guilt for passing on a genetic disorder to their child (Dinc & Terzioglu, 2006), but they also face other sources of stress, including a lack of understanding of risk, delay between diagnosis and available treatment, and a fear of stigma or possible discrimination in employment or with insurance carriers (Knafl, Knafl, Gallo, & Angst, 2007; Kelly, 2009). Moreover, parents who are considering having additional children need to take into account the possible disruption of parent-child and sibling-sibling relationships that may result from the diversion of time and energy towards the medical management of the current or future child's genetic disorder (Knafl, Knafl, Gallo, & Angst, 2007).

The overall purpose of this qualitative study was to understand the lived experiences of couples in the Midwestern United States who have a child suffering from a genetic disorder (affected child). The results have been divided into two studies: The purpose of Study 1 was to understand what variables contribute to a couple's decision to have additional children after already having a child diagnosed with a genetic disorder, how couples communicate with each other about having additional children and what factors contribute to their decision-making

processes; the purpose of Study 2 is was to understand the couple's experience of how their family adapted to the diagnosis in a child of an inherited genetic disorder and what factors were helpful or a hindrance to the adaptation process. All couples will participated in one joint, face-to-face, semi-structured, in-depth interview and completed a demographics questionnaire.

The interviews were designed to explore the couple's experiences of raising an affected child and how the couple communicated with each other to decide about having additional children. Interview questions were written in a way to capture how the couple's experience with the medical system as well as the family belief systems, resources, and spirituality shaped the couple's decision-making process concerning having additional children as well as the role it played in shaping the family's adaptation process. See Appendix A for interview guide.

Rationale for Qualitative Methodology

Qualitative research involves an interpretative, naturalistic approach to the world wherein it positions the researcher in the world that he/she is trying to understand (Denzin & Lincoln, 2005). It is the study of the meaning individuals or groups ascribe to a social or human problem (Creswell, 2007; Bloomberg & Volpe, 2008). Qualitative research helps us understand the complex details of problems that cannot always be accurately reflected in quantitative research. Qualitative research is well suited for research in health care because of its emphasis on people's lived experiences, especially in capturing the interaction between multiple parties (i.e., between patient and provider), which is cannot be easily reduced to a statistical number (Al-Busaidi, 2008; Pascal, Johnson, Dore, & Trainor, 2011). The popularity of qualitative research has been growing in health care studies, as evidenced by its own scholarly journal, Qualitative Healthcare Research, which has been in existence since 1991.

Thematic Analysis

Thematic analysis was used in both Study 1 and Study 2 as the main tool for analysis. Thematic analysis is a method for identifying, analyzing, and generating rich insights into the data (Braun & Clarke, 2006; Knapton, 2015) and is a useful research tool because of its flexibility to identify common threads that extend across and entire set of interviews (Braun & Clarke, 2006) and allows the researcher to analyze the themes while considering the broader context influencing the stories (Vaismoradi, Turunen, & Bondas, 2013). The flexibility of the approach has brought up concerns about the rigor of the research conducted, but some researchers argue thematic analysis is a good fit for qualitative researchers at the beginning of their careers due to the detailed sequence of analytical stages (Vaismoradi, Turunen, & Bondas, 20130). Braun and Clarke (2006) provide an detailed step-by-step guide, which is summarized in the Data Analysis section and reflects the process the researcher followed during analysis.

Role of the Researcher, Biases, Credentials

In keeping with the qualitative tradition, the researcher is involved in multiple phases of the research process. It is important for the researcher to be mindful of his/her beliefs and preconceived notions that may have an effect on the research being conducted. To this extent, the researcher bracketed, or set aside, any personal views or notions so that the meaning gleaned from the data comes directly from the participants (Groenwald, 2004). The researcher kept a detailed journal throughout the course of the research process to monitor and reflect on any epistemological assumptions and the ways in which her beliefs and experiences may impact the research (King & Horrocks, 2010). The researcher is a licensed Marriage and Family Therapist and has been practicing in a private practice setting for seven years. She is a married, Caucasian woman in her early 30s with no children who has been struggling with infertility for about eight

years. The researcher has never had any genetic testing done, nor has anyone else in her family. Therefore she cannot relate to the experience of genetic testing, for herself or for her children, nor does she have any experience as a parent. Through her battle with infertility the researcher has been a patient with several doctors and has had both positive and negative experiences. She understands the importance and benefit of having a trusting relationship with a health professional. The researcher bracketed any personal views and experiences during the analysis process and noted in her journal any feelings that came up.

Participants

Inclusion Criteria

Couples were eligible to participate if they met the following criteria: (a) had at least one affected child and considered having another child, (b) expressed an interest to participate in the study, (c) provided written consent, and (d) spoke English.

Exclusion Criteria

Couples were excluded from the study if they did not meet any of the eligibility requirements, and if they were currently involved with child protective services due to confirmed child abuse or neglect. The sample size goal of nine couples was met (Smith & Osborn, 2003; Creswell, 2007).

Procedures

Following approval from the Michigan State University (MSU) Institutional Review Board (IRB), couples were recruited through a "call for participants" message posted on a Facebook group associated with a particular disease or disorder. Three disorders are represented in the two studies: Spina Bifida, Fragile X, and Tuberous Sclerosis Complex. Participants came from one of the following three genetic disorders: Spina Bifida, Tuberous Sclerosis, and Fragile

X. Spina Bifida is the most common permanently disabling birth defect in the United States and happens when the baby is in the womb and the spinal column does not close all of the way. It is sometimes called the "snowflake disorder" because no two cases look the same, and can result in mobility problems, bladder, bowel, or gastrointestinal disorders, and learning disabilities (www.spinabifidaassociation.org). Tuberous sclerosis complex (TSC) is a genetic disorder that causes tumors to form in many different organs, primarily in the brain, eyes, heart, kidney, skin and lungs. The aspects of TSC that most strongly impact quality of life are generally associated with the brain: seizures, developmental delays, intellectual disabilities, and autism spectrum disorder (www.tsalliance.org). Fragile X syndrome (FXS) is a genetic condition that causes intellectual disability, behavioral and learning challenges, and various physical impairments. Though FXS occurs in both genders, males are more frequently affected than females, and generally with greater severity. Life expectancy is not affected in people with FXS because there are usually no life-threatening health concerns associated with the condition (www.fragilex.org).

Of the nine couples interviewed, three couples had a child with Spina Bifida, three couples had a child(ren) with Fragile X, and three couples had a child with Tuberous Sclerosis Complex. Seven of the nine couples had children after their first child was diagnosed, two couples were considering adoption at the time of the interview. The researcher used both convenience sampling and purposive sampling, as these types of sampling allow the researcher to find a more closely defined group for whom the research questions will be significant (Smith & Osborn, 2003). Couples who participated in study were given a $50 Visa gift card for their participation. See Table 1 for a more complete description of the participants.

Data Collection

Data was collected through face-to-face, semi-structured joint interviews with couples that have at least one affected child and considered having another child. In all of the interviews except for one the children were present for the entire interview. This was because of the unique childcare needs of these families. Sensitive information that would have been disturbing to the child was not discussed in his/her presence. All interviews were audio recorded and later transcribed verbatim. Throughout the interview process the researcher kept a detailed journal that reflected the researcher's thoughts, observations, and experiences.

Data Analysis

After the interviews were transcribed, the researcher analyzed the data collected from the interviews according to the phases outlined by Braun and Clarke (2006) for using Thematic Analysis: 1) familiarize yourself with your data; 2) generating initial codes; 3) searching for themes; 4) reviewing themes; 5) defining and naming themes; and 6) producing the report. First, the researcher was immersed in the data, listening to the audio files, reading and re-reading the data and jotting down initial ideas. Second, the researcher coded interesting features of the data across the entire data set. Next, she collated the codes into potential themes and gathered all the data relevant to each potential theme, and then checked to see if the codes and themes fit across the entire data set. As the analysis progressed, the researcher continued to check and refine the themes, generating a clear definition and name for each theme. Finally, specific quotes that provided a thick description of the themes were collected and separated to produce the two studies.

Issues of Trustworthiness

Guba and Lincoln (1998) suggest using the terms credibility, dependability, and transferability to address issues of trustworthiness in a qualitative research study. Credibility, which is similar to validity, is concerned with whether the participants' perceptions match up with the researcher's portrayal of the participants' perceptions. Dependability, which is comparable to reliability, focuses on ability to track the procedures used to collect and interpret the data. Transferability (or generalizability) does not focus on the representativeness of a sample is, as it would be in quantitative research, but whether the reader could imagine similar processes occurring in other settings (Bloomberg & Volpe, 2008).

The researcher used several methods to ensure credibility. All transcribed interviews will be sent back to each couple for review. By using member checks, the researcher will attempt to eliminate personal biases and confirm that only the couple's perspectives are portrayed. As themes emerge from the interviews the researcher will check in with couples through email or by phone about the findings if needed. The researcher kept a detailed journal that included reflective field notes about subjective perspectives and biases throughout the data collection and analysis process.

Qualitative researchers should provide detailed descriptions of the phenomena they study and their context (King & Horrocks, 2010). These "thick descriptions" help to ensure that the analysis is consistent with the narrative being presented and that dependability is established. The researcher worked with her advisor on coding of interviews and consultation on themes that emerged, using a constant comparative method to check the consistency of the coding and establish inter-rater reliability.

Transferability can be identified in the study through thick, rich descriptions of the participants and the context provided in the discussion section (Shenton, 2004). The couple's own words were used when describing a particular theme, and all transcribed interviews are available for review.

Table 1: Demographics Table

	Couple 1		Couple 2		Couple 3	
Name	David	Janet	Chris	Jen	Oliver	Holly
Gender	M	F	M	F	M	F
Age (yr)	41	35	38	33	35	34
Length of Marriage (yr)	10	10	12	12	10	10
Ethnicity/ Race	White	White	White	White	White	White
Highest Degree Earned	Masters	Bachelors	Some College Credit/ No Degree	Some College Credit/ No Degree	Some College Credit/ No Degree	Some College Credit/ No Degree
Current Employment	Employed For Wages	Self-Employed	Employed For Wages	Employed For Wages	Employed For Wages	Home-Maker
Combined Family Income 2015	$50,000-$74,000	$50,000-$74,000	$50,000-$74,000	$50,000-$74,000	$75,000-$99,000	$75,000-$99,000
Religion	None	None	None	Non-Denom.	Atheist	Protestant
Total Number of Children	4	4	3	3	3	3
Male Child	3	3	0	0	2	2
Female Child	1	1	3	3	1	1
Birth Order of Affected Child	Third (Male)	Third (Male)	First (Female)	First (Female)	First (Male); Third (Female)	First (Male); Third (Female)
Disorder	Spina Bifida	Spina Bifida	Spina Bifida	Spina Bifida	Fragile X; Fragile X	Fragile X; Fragile X

Table 1: (cont'd)

	Couple 4		Couple 5		Couple 6	
Name	John	Katie	Robert	Nancy	Steve	Kristen
Gender	M	F	M	F	M	F
Age (yr)	34	29	44	39	37	40
Length of Marriage (yr)	5	5	9	9	4	4
Ethnicity/ Race	White	Hispanic	White	White	White	White
Highest Degree Earned	Doctorate	Associates	Some College Credit/ No Degree	Masters	Bachelors	Masters
Current Employment	Employed For Wages	Home-Maker	Self-Employed	Employed For Wages	Employed For Wages	Self-Employed
Combined Family Income 2015	$75,000-$99,000	$75,000-$99,000	$50,000-$74,000	$50,000-$74,000	$100,000-$149,000	$100,000-$149,000
Religion	Protestant	Protestant	None	None	Non-Denom.	Non-Denom.
Total Number of Children	1	1	2	2	2	2
Male Child	0	0	1	1	0	0
Female Child	1	1	1	1	2	2
Birth Order of Affected Child	First (Female)	First (Female)	First (Male)	First (Male)	First (Female)	First (Female)
Disorder	Spina Bifida	Spina Bifida	Tuberous Sclerosis	Tuberous Sclerosis	Tuberous Sclerosis	Tuberous Sclerosis

Table 1: (cont'd)

	Couple 7		Couple 8		Couple 9	
Name	Joe	Tina	Ben	Kelly	Zach	Rachel
Gender	M	F	M	F	M	F
Age (yr)	40	37	44	41	51	48
Length of Marriage (yr)	14	14	19	19	20	20
Ethnicity/ Race	White	White	White	White	White	White
Highest Degree Earned	Masters	Doctorate	Trade/ Technical/ Vocational Training	Associates	Bachelors	Bachelors
Current Employment	Employed For Wages	Employed For Wages	Employed For Wages	Home-Maker	Employed For Wages	Home-Maker
Combined Family Income 2015	$150,000 or more	$150,000 or more	$20,000-$34,000	$20,000-$34,000	$150,000 or more	$150,000 or more
Religion	Protestant	Protestant	Lutheran	Lutheran	Roman Catholic	Roman Catholic
Total Number of Children	2	2	2	2	2	2
Male Child	1	1	1	1	0	0
Female Child	1	1	1	1	2	2
Birth Order of Affected Child	First (Male)	First (Male)	Second (Female)	Second (Female)	First (Female); Second (Female)	First (Female); Second (Female)
Disorder	Fragile X	Fragile X	Tuberous Sclerosis	Tuberous Sclerosis	Fragile X; Fragile X	Fragile X; Fragile X

CHAPTER 4: STUDY ONE

"The Fear of Having Another Biological Child Versus the Fear of Not Having Another Biological Child": Decision Making Among Couples With a Child Who Has a Childhood Genetic Disorder

ABSTRACT

There is a gap in our understanding of how couples deal with genetic information, especially when a child is diagnosed with an inherited genetic condition. Also, there is a lack of understanding of the decision-making processes regarding genetic testing. The purpose of this qualitative study was to understand the lived experiences of couples who have a child suffering from a genetic disorder (affected child). For this study, the affected child is defined as a child who has been diagnosed with a genetic disorder that "causes significant morbidity and mortality". Ten couples from the Midwestern United States were interviewed using a semi-structured, joint interview process. Using thematic analysis and the couples' own words, this study found that several factors contributed to the decision to have future children after their child received a diagnosis of an inherited childhood genetic disorder. These factors include evaluating the risks and burdens of having another affected child and collecting information regarding genetic status and available resources. Implications for family therapists are addressed.

Introduction

Genetic testing in the United States started in an experimental laboratory in the late 1950s (Feetham & Thomson, 2006). Since the completion of the Human Genome Project in 2003, genetic tests have become widely available, and at relatively low cost, for more than 1,000 genetic disorders (Feetham & Thomson, 2006). As the human genome is more completely understood and new genetic tests are developed to detect both the predisposition to, and the presence of, specific diseases, medical professionals and family therapists are becoming more aware of the effect these tests will have on family systems and on the health beliefs of the family. Both the benefits and risks of genetic testing are high (Johnson, Wilkinson, & Taylor-Brown, 1999) in that families can know risks to disorders early on in the process, but can also be affected in their day to day activities due to the anxieties raised by the genetic diagnosis. Predictive genetic testing is of huge benefit to couples in that it allows them access to numerous preemptive medical care options that they likely would not have otherwise known about without the testing. While diagnostic genetic testing can confirm diagnoses, determine their type and severity, and substantiate the appropriateness of targeted therapy (Peterson, 2005), it can also lead to decisions that lead to ethical quandaries for parents of children.

The primary risk associated with all types of genetic testing is that such testing can reveal information that may either not be actionable by the couple, or if actionable, cause considerable stress and anxiety. The dilemmas that these scenarios pose for couples can alter family relationships, intensify preexisting family difficulties, and result in both perceived and actual discrimination and stigmatization (Street & Soldan, 1998; Van Riper & Gallo, 2006). For couples who give birth to an affected child, this inability to act can take on different meanings as parents face multiple decision points about treatment and prognosis. Parents of a child who has

been referred for genetic testing or whose child has a genetic disorder report an elevated level of anxiety due to the concern about the health and future of their child, and upon receiving results indicating an abnormality, may feel worried, hopeless, and guilty (Dinc & Terzioglu, 2006).

Not only must the parents of a child with a genetic disorder consider the time, energy, and costs involved in the medical management of the genetic disorder; they also face other potentially negative effects in multiple other areas including the decreased self-esteem and self-worth of the diagnosed child, disruption of parent-affected child relationship, parent-unaffected sibling/s relationship, or sibling-sibling relationships, along with an increase in parental anxiety and guilt (Knafl, Knafl, Gallo, & Angst, 2007). Several studies have identified a number of sources of stress for families who have a child with a genetic disorder, including a lack of understanding of risk, lag time between genetic discoveries and available treatments, fear of loss of privacy, stigma, and possible discrimination in employment, insurance reimbursement, or by school systems (Knafl, Knafl, Gallo, & Angst, 2007; Kelly, 2009). Research has demonstrated that once a child has been diagnosed with a genetic disorder, there are numerous implications for parental mental health and overall family functioning including parental feelings of denial, anger, guilt, and depression (Jacobs & Deatrick, 1999), lower relationship and sexual satisfaction in the couple along with greater relationship strain (Manne et al., 2004; Quaid & Wesson, 1995), and the real and perceived losses of child and family functioning (Jacobs & Deatrick, 1999). On average, families with a child with a disability face greater levels of stress and dysfunction than do other families (Collins & Williamson, 2003). However, little is known about the role these factors play in parent decision-making about future pregnancies.

In a study completed by D'Amico et al. (1992), the authors found that among couples with previous experience of a child affected with genetic disease, a significantly higher

percentage (65.5%) avoided further pregnancies as compared to couples who opted for further reproduction (34.5%). Surprisingly, couples with a first affected deceased child were far less likely (13.6%) to refrain from reproduction. In a similar study, the majority of parents of a child affected with a genetic disorder chose to avoid future pregnancies, so as to avoid any decisions about prenatal screening or testing to identify whether subsequent children may be affected (Kelly, 2009). Although prenatal diagnosis and preimplantation diagnosis are made available to families who are at higher risk for genetic abnormalities or who are carriers of genes that cause childhood onset disorders, many couples choose not to test for a number of personal, ethical, or religious reasons. Based on feelings of guilt or disrespect for the first-born, in the case of selective termination, or if the parents are unsure of whether they could make a decision about termination, many parents choose not to test and thereby wait and see if their child is affected (Gotz & Gotz, 2006). In a study done on parents of children diagnosed with cystic fibrosis (CF), Ormond et al. (2007) found that the majority of parents were likely to pursue (or had already pursued) future pregnancies. In fact, the initial experience of diagnosis and care of a child with CF lessened any anxiety about caring for future children with CF. The current study explores how the couple views future family planning and related health care decisions.

Background

Decision Making in the Family

The family strongly influences health behaviors (Peterson, 2005; Rolland & Williams, 2005). For this reason, it is important to understand the process of decision making within the couple related to behaviors affecting health. At a familial level, decision-making is a process of reducing dissonance between and among family members (Bubolz & Sontag, 1993) and is the central control system of family organization. This reduction of dissonance leads to less stress

and conflict around critical decisions. At its most basic, decision-making involves: 1) the recognition of need for decision, 2) identifying and weighing acceptable alternatives, and 3) selecting or mediating an alternative and facilitating its action (Paolucci, Hall, & Axinn, 1977). From a different perspective, Hastie (2001) describes a decision as involving three components: 1) choice options and courses of actions; 2) beliefs about objective states, processes, and events in the world, including outcomes states and means to achieve them; and 3) desires, values or utilities that describe the consequences associated with the outcomes of each action event combination. On the contrary, decisions can also carry meaning for individuals and families in ways that may not make sense to others outside the family (Patel, Kaufman, & Arocha, 2002). In short, the mutual decision making required by families, especially around stressful issues, is complex, emotional, and often stressful.

In families, decision making can involve a seeming unlimited number of possible outcomes (Bubolz & Sontag, 1993) and can be a complex process. Not only are families faced with making many types of decisions (Bubolz & Sontag, 1993), but family decisions are also influenced by the environment in which the family functions, which can either restrict decision making or offer opportunities (Paolucci, Hall, & Axinn, 1977). In many instances, family decisions involve group decisions (Bubolz & Sontag, 1993) and group decision-making processes. Since family decisions are typically made by more than one family member, it is necessary to analyze decision making from an ecological perspective, because each family member is different, with a different psychological make-up. Moreover, each member brings their unique experiences into the decision (Paolucci, Hall, & Axinn, 1977) using their own personal style of thinking to make their way through the decision-making process, with different meanings for their experiences, values, expectations, and uses of information (Anderson, 2007).

Risk, Family Decisions, and Genetics

In addition, many family decisions are made under conditions of uncertainty and risk (Bubolz & Sontag, 1993). The concept of risk is especially highlighted in regards to predictive genetic testing, where decisions of risk are paramount because the information gleaned from such testing can drastically alter an individual's sense of self, can change personal identities, and can cause extreme anxiety and stress (Hallowell, Foster, Eeles, Ardern-Jones, & Watson, 2004). Risk embodies aspects of ambiguity and uncertainty. When related to genetic inheritance, risk can be very difficult to deal with from a personal and familial perspective (Peters, Djurdjinovic, & Baker, 1999). There are two types of risk. A first type of risk is viewed negatively. Knowledge of this type of risk constitutes a heightened awareness of potential unknown dangers or hazards that then becomes a major source of anxiety within the family. A second type of risk is viewed positively. Knowledge of this type of risk helps families predict the likelihood of events occurring in the future and allows the family to feel empowered and more in control of their actions and medical destiny (Hallowell et al., 2004). Both of these types of risk are evident in families considering genetic testing. An understanding of genetic risks is considered crucial for informed decision-making (Henneman, Marteau, & Timmermans, 2008).

Decision-making in families does not rely solely on one family member. Rather, the decision is shared on some level by all family members (Paolucci, Hall, & Axinn, 1977). In making decisions about whether, when, or how genetic information should be evaluated and shared, the interests of both the individual and those with whom they are genetically linked must be considered (Van Riper & Gallo, 2006).

It is important to elicit each family member's understanding of his or her genetic risk and any previous health beliefs prior to beginning to convey risk information to the family (Peters,

Djurdjinovic, & Baker, 1999). Genetic testing and risk knowledge can disrupt core beliefs (Rolland, 2006). Bartle-Haring and Gregory (2003) emphasize that the genetic testing decision-making process has a psychological impact on everyone who is genetically linked, and not just on the proband (i.e., person of genetic interest). Several steps can be taken in communicating genetic risk information within the family. First, it is important to have an accurate family history across generations (McCann et al., 2009). Second, genetic risk information given to one member of the family should be circulated to the rest of the family members (Bowen, Bourcier, Press, Lewis, & Burke, 2004). Third, decisions about what to do relative to future genetic testing or behaviors related to healthcare decisions should be discussed among family members (Bowen et al., 2004). Shared health beliefs help family members find meaning in their illness experience, whether current or future, and help guide decisions and actions (Rolland, 2006). Research indicates that families who discuss illness and genetics are associated with genetic test uptake, or likelihood to participate in genetic testing (McCann et al., 2009). This discussion may also serve as a social support function to build relational bonds and to facilitate coping among family members (Peterson, 2005).

Risk perception correlates with decision making about genetic testing (Lerman, Croyle, Tercyak, & Hamann, 2002), and perceived risk can be more influential than objective risk (Hiraki, Chen, Roberts, Cupples, & Green, 2009). Understanding perceived risks for complex genetically based diseases can improve risk communication, optimize coping strategies, and encourage the uptake of preventive health behaviors (Hiraki et al., 2009). Decision-making is not merely dependent on obtaining accurate risk information for weighing pros and cons or to resolve or mitigate risk (Anderson, 2007). Decision-making is a complex process that can involve emotion based- and/or rational- modes of perceiving and responding to information

(Hopwood, 2000), and using these modes in the context of personal relationships (e.g. within couples and families) and professional relationships (e.g. with doctors and genetic counselors) (Anderson, 2007).

Reproductive Decision Making Related to Genetic Testing

Decisions to have children are usually not made solely by one individual, but typically as part of a couple. When considering the potential genetic risks, couples often make these reproductive decisions, not in isolation, but with input from family members, health care workers, and others (Klitzman, Thorne, Williamson, Chung, & Marder, 2007). One study showed that 67% of the women made the decision to test prenatally with their partners and no women reported making the decision alone (Humphreys, Cappelli, Aronovitch, Allanson, & Hunter 2008). Another study confirms that decision making concerning genetic testing is primarily a joint enterprise (Kenen, Smith, Watkins, & Zuber-Pittore, 2000). Genetic testing has made it possible to clarify uncertainties before or when making reproductive decisions (Downing, 2005). The decisions that couples make in situations of genetic risk are not based solely on risk information. Instead, these decisions are multifaceted and depend on factors such as desired family size, perceived risk, social context, and personal experiences with the genetic condition (Myring et al., 2011). Beeson and Golbus (1985) and Myring et al. (2011) found couples with personal experiences of a genetic condition (i.e., those who already had an affected child) had a better understanding of the issues regarding further reproductive decisions, whereas those couples with little experience of a genetic condition were unclear in their decision making about future children.

Prenatal genetic counseling and testing emphasize couple decision-making. This counseling is not only a potential stressor, particularly when spousal conflict or disagreement

exists regarding testing, but can also act as a source of comfort and support (Humphreys et al., 2008). Factors considered in reproductive decision making include interests of self, interests of current offspring, interests of future offspring, preferences of spouse, preferences of family of origin, and obligations to broader society (Klitzman et al., 2007). Religious, ethical, and intellectual values have also been shown to influence the decision making process (Pivetti & Melotti, 2013). In one study, race/ethnicity was associated with decisions about prenatal genetic testing (Learman et al., 2003). African Americans and Latinas were more likely influenced by their faith/religion, while Caucasian women were most likely to report that their health care provider would influence their decision-making. Couples may discover that making decisions about subsequent children involves finding a balance between wanting to have another child, the future needs of existing children, and the wellbeing of the at-risk parent (Downing, 2005).

Decisions About Genetic Testing in Children

Many common genetic conditions occur in families with no previous family history or experience of the disorder (Collins & Williamson, 2003). In their study on prenatal genetic testing, Pivetti and Melotti (2013) found that people who had a good understanding of genetic testing, and could count on the support of family members and friends, showed a positive attitude toward genetic testing. In making the decision about whether to undergo prenatal genetic testing, women and their partners must consider age-related risk information, the physical risks to the fetus associated with the procedure (Verp, 1992), and their own values and attitudes concerning pregnancy termination and the possibility of raising a child with a genetic disorder (Humphreys et al., 2008; Moyer et al., 1999). The most frequent reasons which lead a couple to consider genetic testing include the worry that the baby is at an increased risk of genetic disorder, the need to make a decision about family planning, a desire for relief from uncertainty,

and the intention to take better care of one's health (Moyer et al., 1999; Pivetti & Melotti, 2013). One study found that receiving a positive (i.e., confirmatory genetic) prenatal diagnosis may be experienced by couples as a traumatic event with long term consequences (Nazare, Fonesca, Gameiro, Canavarro, & Datilio, 2011).

In dealing with the uncertainty and anxiety that can come with the decision for genetic testing, it is unclear what information is necessary for couples and families to make the decision that is best for their family and situation. Broadstock and Michie (2000) discovered that an informed decision, i.e., one based on all relevant information, may not be as straightforward as one would assume, because people differ in how much information they need, want, or receive. Moreover, couples may be looking for an effective decision that is both informed and consistent with the decision maker's values, and this is not always easy or possible. Disclosure and communication about genetic testing appears to be fairly open for common, treatable conditions such as inherited cancers. However, this may not be the case for more severe and stigmatizing disorders such as sex-linked or recessive conditions (e.g., cystic fibrosis) or Huntington's Disease (Peterson, 2005).

The dynamics operating between partners, particularly the extent to which the views of each partner are respected or taken into account by the other partner are relevant to the nature of couples' decision making process (Daniels, Lewis, & Gillett, 1995). Specifically, the more men perceived that their wives valued and accepted their opinions, and felt more appreciated by their wives, the more women felt that the decision was shared and that couple's agreement was high (Nazare et al., 2011). If genetic testing is a subject that causes tension within the relationship, then it is likely that both men and women will forge alliances with others, such as extended family or medical professionals (Daniels, Lewis, & Gillett, 1995). One study found that couples

in a position to consider genetic testing revealed the genuine care and concern they expressed for their future child(ren) and their understanding of the profound significance of their decision, regardless of their decision type (Hershberger et al., 2012).

In summary, couples have much to consider when faced with a decision about genetic testing. First, couples need to think about what family members may be involved or who they would want to include in the decision making process (Bubolz & Sontag, 1993), knowing that each person brings their own experiences, values, and way of thinking to the process (Anderson, 2007). Second, understanding that inherent in genetic testing is a certain amount of risk (Lerman, Croyle, Tercyak, & Hamann, 2002) that has a psychological impact on everyone genetically linked, not just the individual being tested (Bartle-Haring & Gregory, 2003). Third, reproductive decision making is a joint enterprise by the couple (Kenen, Smith, Watkins, & Zuber-Pittore, 2000) based on numerous factors, including preferences of spouse and self, (Klitzman et al., 2007), perceived risk, (Myring et al., 2011), and religious and ethical values (Pivetti & Melotti, 2013). Finally, it is necessary for couples to gather information and assess their personal values about the possibility of pregnancy termination or raising a child with a genetic disorder when deciding about genetic testing in children (Humphreys et al., 2008).

Purpose of the Study

The purpose of the study was to understand experiences and processes related to a couple's decision to have additional children after already having a child diagnosed with a genetic disorder. I sought to explore how couples communicate with each other about having additional children and what experiences and factors contribute to their decision-making processes.

Methods

Sample

Following approval from the Michigan State University (MSU) Institutional Review Board (IRB), couples were recruited using purposive sampling through a "call for participants" message posted on a Facebook group associated with a particular inherited childhood genetic disease or disorder. Nine couples responded and were interviewed. The mean relationship length for couples was 12.8 years. Seventeen of the 18 participants interviewed identified as White, and the other 1 identified as Hispanic or Latino. See Table 1 for a more complete description of the participants.

Participants came from one of the following three genetic disorders: Spina Bifida, Tuberous Sclerosis Complex, and Fragile X. Spina Bifida is the most common permanently disabling birth defect in the United States and the genetic condition does not allow the spinal column to close all the way while the baby is in the womb. It is sometimes called the "snowflake disorder" because no two cases look exactly the same, and can result in mobility problems, bladder, bowel, or gastrointestinal disorders, and learning disabilities (www.spinabifidaassociation.org).

Tuberous sclerosis complex (TSC) is a genetic disorder that causes tumors to form in many different organs in the body, primarily in the brain, eyes, heart, kidney, skin, and lungs. The aspects of TSC that most strongly impact quality of life are generally associated with the brain: seizures, developmental delays, intellectual disabilities, and autism spectrum disorder (www.tsalliance.org).

Fragile X syndrome (FXS) is a genetic condition that causes intellectual disability, behavioral and learning challenges, and various physical characteristics. Though FXS occurs in

both genders, males are more frequently affected than females, and generally with greater severity. Life expectancy is not affected in people with FXS because there are usually no life-threatening health concerns associated with the condition (www.fragilex.org).

Of the nine couples interviewed, three couples had a child with Spina Bifida, three couples had a child(ren) with Fragile X, and three couples had a child with Tuberous Sclerosis Complex. Seven of the nine couples had children after their first child was diagnosed, two couples were considering adoption at the time of the interview.

Data Collection

After couples provided informed consent and completed a demographics questionnaire, a face-to-face semi-structured interview was conducted with both the husband and wife present. Couples who participated in study were given a $50 gift card for their participation. All interviews were audio recorded and later transcribed verbatim by the researcher. The interviews were designed to illicit the couples' experiences regarding what thoughts, feelings, and events contributed to a couple's decision to have or not have additional children after already having a child diagnosed with a genetic disorder, how couples communicated with each other about having additional children, and what factors contributed to their decision-making processes.

Data Analysis

Audiotaped interviews were transcribed verbatim, all identifying information was removed from the transcripts and each participant was assigned a study number. Thematic content analysis was used to analyze each of the interviews (Braun & Clarke, 2006) and NVivo software was used to organize codes and themes that emerged from the data. Thematic analysis is a qualitative method for analyzing data that identifies patterns or "themes" within a data set that provides rich and detailed descriptions and is well suited for health research (Vaismoradi,

Turunen, & Bondas, 2013). Braun & Clarke (2006) provide a thorough step-by-step guide to doing thematic analysis that the author followed in this study. First, the author familiarized herself with the data through reading and re-reading the interviews several times as well as listening to the audio recordings and jotting down initial impressions. Next, she read through each interview and coded interesting features of the data, followed by grouping the codes into relevant themes. Ultimately the codes were reviewed to ensure they fit within the theme and the theme was given a name.

Results

The following two major themes and related sub-themes emerged regarding decision making for future children: 1) the evaluation of the risks and burdens for the future and 2) collecting information that informed the decision.

Evaluation of the Risks and Burdens for the Future

Risks. Many of the couples interviewed reported that the risk of having another affected child was an important part of the discussion regarding whether or not to have any more children. Rachel and Zach had both of their daughters diagnosed simultaneously with Fragile X before the age of 2 when they noticed developmental delays with their oldest daughter. Both of their daughters require a lot of care and one of them has extremely limited communication. When directly asked if the possibility of future children having Fragile X played a role in their decision-making, they said:

> Zach: Oh yes, it had. We had in 1 in 2 chance of all of our children having Fragile X. That's what the doctor said. 1 in 2, 50-50, you never know. It could be a boy, totally healthy, could be a girl totally healthy or could be a boy with Fragile X.
>
> Rachel: So yeah, Fragile X -- made the decision for us.

Zach: Yeah, that's fair to say. And without Fragile X, would we have more children?

Rachel: Probably. (Zach and Rachel, two daughters with Fragile X)

With a disorder like Fragile X, where there is a 50 percent chance of each child having the disorder, it was important for couples to address the strong possibility of having another child with the same or even worse physical or behavioral issues.

Oliver: We almost didn't have a third. I was done. I was done. You really wanted to try for a girl. Which clearly worked out. But we were really, really worr--I was really worried. I think you were really worried too [to Holly]. We were both really worried that, you know, what happens if we have a second kid with Fragile X? And what if it's harder than [affected child].

Holly: So we were talking about the possibility of having two children that might be at home with us for the rest of our lives. Maybe a child that was more aggressive than [affected child]…that was another big consideration for us. (Oliver and Holly, son with Fragile X)

Holly and Oliver described their oldest son (diagnosed with Fragile X) as having severe tactile defensiveness ("you couldn't touch him") and did not speak until he was 6 years old, this experience highlight's their concern of having another affected child, someone who might be more severely affected than their son. Of the Fragile X couples went on to have another child with Fragile X after their first child was diagnosed with it, one Fragile X couple had another child that did not have Fragile X. Of those two couples who had a second Fragile X child, one couple had a child with minor physical and behavioral issues whereas the other couple had a child that was unable to communicate well and had other physical issues.

Even though there is still a 50 percent chance of each child having the disorder, the risk

for Spina Bifida and Tuberous Sclerosis Complex is more complex because in both of those disorders there is a chance for the disorder to be caused by a spontaneous mutation, which only slightly increases the chance for the mutation to happen again. In the case of Spina Bifida, if a woman has already had a child with Spina Bifida, the chance of having another affected child is 1 in 20. A woman who has Spina Bifida has a 1-5% chance of having a Spina Bifida child and that increases to 15% if both parents have Spina Bifida. In the case of Tuberous Sclerosis Complex, it is estimated that a family with an affected child by spontaneous mutation has a 1-3% chance of having another child with Tuberous Sclerosis Complex. For John and Katie, the risk of occurrence was too much:

> John and I really hashed it all out because I was definitely--definitely heartbroken when he told me he didn't want to have another biological child. That for him the risk was too high. That he didn't want to take. And I think--I'm gonna cry a little bit. I think it was higher because it made it feel like it was my fault. And I know that's not true. (Katie, daughter with Spina Bifida)

Because the risk was too high for them, John and Katie opened up about the discussions and fights they had about growing their family and how they came to the decision to adopt:

> John: I would say that the tears come from two places. One is there's the fear that's having another biological child and two, the fear of not having another biological child. And I think that the hard part with that discussion has always been, when we discuss it and we're honest with ourselves, both those fears are readily present on both sides. There was lots of fights and lots of good solid strong communication on this. One of the things we talked about was, you know, what I don't see for my wife is an all out fight that she has to have another child. Biologically.

Katie: Would I like to? Yes. Is that reality now? Has it changed? Yes. Can we still get pregnant accidentally? Yes.

John: I have friends that had abortions because their children were not necessarily medically ideal. And when you get to those type of situations where it's all or nothing and you're willing to risk what you're willing to risk. Then you have to have a different conversation. I think if you're in the situation where we were, where it's like, it'd be nice to have another child. And we don't really need it to come out our loins to be considered a child of ours. And to love it equally. We really only wanted two anyways. So maybe adoption really is the best.

Katie: We're applying and talking to social workers about it. We're figuring out all the steps. It's something. I, I look forward to it. I look forward to meeting a new child. (John and Katie, daughter with Spina Bifida)

Burdens. One consideration for many of the couples in deciding whether or not to have more children was the amount of care and resources needed for affected child. At the time of the interviews all of the families were involved in multiple therapies and programs inside and outside of school:

> We delayed the decision to have more children partially because it was so unclear as to how [child] would be affected, how that would affect us. Are we going to be at hospitals every single week for the rest of our lives? Are we going to have to deal with major medications and insurances and -- are we going to be able to keep a job that's going to keep her insured? All these things are definitely things we've had to weigh and consider. (Kelly, daughter with Tuberous Sclerosis)

Ben and Kelly came from a lower socioeconomic status and had created a 501c3 organization so

they could help fundraise for their daughter's treatments and therapies. Ben and Kelly also lived in a smaller, rural area which added to their hardship, driving for more than an hour to go to the doctor's and out of state for a Tuberous Sclerosis specialty clinic. For David and Janet, who had a son with Spina Bifida, their decision to have more children stemmed around the amount of physical care their child needed in regards to having limited mobility and needing to use a catheter and enema every day:

> We were kind of like, can we have four kids because they kind of changed our plan and how does his disability weigh into our family and how does that affect our decision. How can I have a baby and a kid that is immobile and – so when he was a year, we were nowhere near ready for another kid at that point and so we waited awhile. (Janet, son with Spina Bifida)

Robert and Nancy, who had a son with Tuberous Sclerosis, they were concerned about the amount of attention they could give their children while having to deal with the amount of care the affected child needed:

> I think the problem is, is not--My concern isn't, isn't the genetic aspect of it. I know that you [wife] have concern of it. Once they told us that neither of us had it it wasn't really a concern with me anymore. My biggest issue has been they're exhausting. I mean he was exhausting because of his conditions….Well sometimes we feel like we're not giving them enough attention. You know? (Robert, son with Tuberous Sclerosis)

Prior to surgery, Robert and Nancy's son was having multiple seizures a day and having trouble controlling the movements on one side of his body. When he was four years old he had brain surgery to help control the seizures, but now he is communicating less than he was before and is having some behavioral issues, he can be aggressive at times, hitting his head against the wall.

Many couples also discussed what would happen with their child(ren) in the future and who would take care of them, since most of the children in this study would not be able to live on their own due to the level of care they would need on an ongoing basis:

> And it kind of made us realize that we weren't going to live forever. And my husband's parents are not going to live forever. My parents are not going to live forever. Not that my parents are super involved. And we realized that what happens when we're old? Who's going to take care of her? (Jen, daughter with Spina Bifida)

Jen and Chris' daughter was confined to a wheelchair and was able to communicate utilizing an iPad. At the time of the interview the couple had 8- month-old twin girls.

> One of my concerns was care for [affected child] after we're gone. As an only child myself, I'm at a point right now where I'm feeling a strong burden to have to care for that portion of my family and to being the only one; it's rough. It's like I have a bucket with all these holes in it where everything is going and there's nothing saving. There's no backup. So I felt like…it would have been really nice to have more kids in order to have more for [unaffected child]. I don't want him to be an only child or to feel like an only child who's got to take care of the needy other family member. (Kelly, daughter with Tuberous Sclerosis)

Some couples saw future siblings as one of the best options for caretaking and support for the affected child:

> Holly: I think one of the big, one of the big factors for me in that discussion was seeing how well [affected child] responded to [unaffected younger brother]. To this day I say the best thing we ever did for those two was having them close together, because [affected child] has picked up so much from [unaffected younger brother]. Even now [affected

child] picks up stuff from [younger sister].

Oliver: It's like, you know, that second kid usually progresses faster because he's got the first one to learn from. I mean [unaffected younger brother] kinda surpassed [affected child] now. But there was a while where they were, hitting a lot some of the same milestones together, [unaffected younger brother] would push [affected child]. That was nice, that felt good to watch.

Holly: It happened actually with [younger sister] and [affected child] with her talking. He--They have progressed almost evenly, with speech and language development. From when she started talking, he had been progressing like right along with her. Which I don't know if it's coincidence or just like when it happened or maybe a little of both. (Oliver and Holly, son with Fragile X).

Collecting Information to Inform Decision Making

All of the couples in the study said it was important for them to get as much information and resources about their particular disorder as possible so they could make the best possible decisions for their family, including whether or not they as parents should be genetically tested. For Holly and Oliver, the discussion about genetic testing started when they were dating:

I'm one of those guys that when I was younger, I always said I can't handle a special kid. It's not in my skill set. I won't be able to do it. I don't wanna try to do it. I never considered it as part of my life. So when we started talking about it, we had already dating for a little bit before it came up, maybe 6 months or so. And then I didn't really know what it [Fragile X] was. She explained it to me. I just, I can't even remember what I thought it would be like. I just generally assumed it [a child] would be--dumb. Like a kid, right? I just figured it would be like, oh he's mentally handicapped. So it was a big deal.

> We had a lot of arguments while we were dating about her getting tested. And then, I don't know. I think I gave up. (laughs) I think that's what ended up happening. I gave up. I knew you [Holly] weren't gonna get tested. I knew based on kinda what we talked about with you and what you told me, that it kinda, we had a chance either way. When we started talking about it to, I think when we got really closer to having kids it was like, well what are the chances anything could go wrong? And then we just said well okay. (Oliver, son with Fragile X)

Holly got tested when she was pregnant with her first child and when she found out she tested positive for Fragile X, it did not stop them from having biological children. For some couples decision making for future children was made slightly easier when they found out the disorder was caused by a spontaneous mutation:

> So we struggled with insurance to get us tested. They [the lab] had some type of plan that if your insurance didn't pay for it, you'd pay x amount. So I was like well let's just do it. And I said [to the lab] what can we do for us to get tested because we would like to have a child but we want to know for sure that we don't have it. So bless her heart. She sent me 2 testing kits overnight. We got a notice that neither of us had it. Both of ours came back negative. (Nancy, son with Tuberous Sclerosis)

Chris and Jen reported being divided on the importance of knowing the genetic risk in regards to deciding about future children:

> Chris: I don't think it would've changed our thought process of having children one way or another. It was just kind of getting the heads up of knowing what we could be getting into.
>
> Jen: I wanted to know beforehand. I don't know. I don't know if I would have been so

gung ho if either of us were a carrier. If somebody gave me a crystal ball and said, "Yes. You're going to have another child with medical issues." Would I have opted to have another baby? Probably not. I don't know. Once we got the- - I mean once we found out neither one of us were carriers. Green light. (Chris and Jen, daughter with Spina Bifida)

For those couples whose risk of having another affected child was not as great, there was more peace of mind in moving forward with the decision to have more children. Both of the couples in this category went on to have unaffected children.

Discussion

This study reflects the experiences of nine couples that made or are in the processing of making a stressful and emotional decision regarding having another biological child knowing there was a genetic risk to future children. Many common genetic conditions occur in families with no previous family history or experience of the disorder (Collins & Williamson, 2003). Eight of the nine couples interviewed had no knowledge of genetic conditions in their families prior to having children. Holly and Oliver, who knew about their potential genetic risk, reported having some conversations about the risk prior to having children but Holly did not get tested until she was pregnant with their first child (she found out when she was 16 she was potentially a carrier for Fragile X).

Evaluation of Risks and Burdens

One of the themes echoed throughout this study is the evaluation of the risks and burdens for the future that couples used as part of their decision-making processes for future children. The couples in the study identified several positive and negative risks. Positive risks help families predict the likelihood of events occurring in the future and allow the family to feel empowered and more in control of their actions and medical destiny (Hallowell et al., 2004).

Some of the couples decided to get genetically tested and in receiving a negative result, meaning they did not have the genetic mutation, went on to have more biological children. Negative risks constitute a heightened awareness of potential unknown dangers or hazards that then became a major source of anxiety within the family (Hallowell et al., 2004). John and Katie were "hashing out" the decision to have another biological child after their first child was diagnosed with Spina Bifida and were on opposite sides, John thought the risk was too great to take and Katie was still grieving the loss of not having another biological child.

Another factor in the decision-making process of the couples in this study was the potential burdens on the family. During the interview several couples described a typical day/week, the amount of care and resources needed for their affected child. Families were dealing with several therapy appointments a week, multiple medications, and some couples were dealing with diapers and/or catheters and enemas (in the case of Spina Bifida). Other couples were dealing with behavioral issues and children who were unable to communicate. One father, Robert, specifically commented on being worried about finding balance in giving enough attention between children, which is reflected in the literature (Downing, 2005), and other couples shared the same sentiment. Several couples expressed concern about who would take care of their children after they were gone because many of them would need on-going care and they did not want to burden other children or extended family.

Collecting Information to Inform Decision Making

A second theme echoed throughout the study was the need to gather information to make an informed decision about future children. A study by Broadstock and Michie (2000) showed that it isn't clear what information is needed for an informed decision because each family may find different information important. Some of the couples thought it was important to get genetic

testing done, either for themselves or for their child. Many of those couples felt relieved when the test came back negative and were more comfortable planning for future children.

Couples Lack of Decision Making Processes

The purpose of this study was to explore how couples communicate with each other about having additional children and what experiences and factors contribute to their decision-making processes. The researcher assumed that once a couple received a genetic diagnosis for their child, there would be a process for decision-making within a couple for having additional children. This was not the case. Five of the nine couples included in the study learned that the genetic disorder came from a spontaneous mutation and was not inherited from either parent. For these couples the spontaneous mutation greatly lessened the risk of having another child with a genetic disorder and the couples proceeded with future pregnancies.

Three of the nine couples in the study were already pregnant or had more children by the time they received the genetic diagnosis, and most of the couples were done having children at the time they received the diagnosis, which eliminated the need for any conversations about future children. Several couples in the study acknowledged that having more children after the diagnosis was not a deterrent because the couples already had experience in taking care of the disorder and knew what to expect. This supports existing literature, where in a study done on parents of children diagnosed with cystic fibrosis (CF), Ormond et al. (2007) found that the majority of parents were likely to pursue (or had already pursued) future pregnancies because the initial experience of diagnosis and care of a child with CF lessened any anxiety about caring for future children with CF.

During the course of the interviews two themes emerged that were not overtly addressed, ambiguous loss and resilience. Ambiguous loss is defined as an incomplete or uncertain loss

(Boss, 2007). There are two kinds of ambiguous loss. In the first, people are perceived by family members as being physically absent but psychologically present, like a soldier who is missing in action or adopted children and their birth parents. In the second, people are perceived by family members as physically present but psychologically absent, like in cases of substance abuse, mental illness or chronic illness (Abrams, 2001). Boss and colleagues have identified several areas of ambiguity that contribute to family distress in relationship to chronic illness: a) a lack of clarity in diagnosis, b) difficulty in predicting outcomes, c) day to day changes in functioning of the ill person that affect family relationships, d) the fact that the ill individual may give an outward appearance of health thus raising expectations for his or her behavior and functioning within the family, and e) fear that important emotional relationships will be affected by the illness (O'Brien, 2007). Nancy highlights the ambiguity in predicting future outcomes, as she describes the loss she feels for her son:

> That's not--So it's been a lot of loss I think that I've, I've learned through this journey. The loss of he's not gonna go to college. And he's not gonna get married. He's not gonna have kids. And. He's not gonna be on his own. I don't know if he'll always be at home. I don't know if he's eventually gonna be in a group home. You know. So it's that, that loss of everything. (Nancy, son with Tuberous Sclerosis)

Being able to make sense out of the loss, the ability to maintain hope and to find some way to change are all needed in order to resolve the loss, which expresses the essence of resilience. Walsh (1998) defines resilience in families as the capacity to rebound from adversity, strengthened and more resourceful. Many of the couples in this study either did not live close to family or aging parents made it difficult to rely on them for much support, therefore the couples needed to be resilient in how they managed day-to-day life. Kelly and Ben are a reflection of this

resilience, in that they created a 501c3 organization in order to help fundraise for the care their daughter with Tuberous Sclerosis needs.

Limitations

One limitation of this study is that eight of the nine couples interviewed were retrospectively discussing their decision making about having more children after the diagnosis, which may make it difficult to accurately remember every detail about their decision making process. It is also possible that their current experience with raising an affected child within their family changed the way they remembered past details. Only one couple was still in the process of finalizing their decision about future children. Additionally, with a small sample size of only nine couples, the results are only generalizable to these nine couples and not to the experiences of other couples.

Another limitation was the lack of racial and ethnic diversity with 17 of the 18 participants identifying as White. Lastly, the genetic disorders that were included in this study were inherited childhood genetic disorders and findings may not be generalizable to other genetic disorders (e.g., spontaneous mutations). Five of the nine couples interviewed had a child with a spontaneous mutation, three couples had an inherited mutation, and at the time of the interview one couple had not been tested yet. All of the couples were interviewed conjointly in their homes, and although there was an opportunity to observe the dynamics of the relationship and what caretaking looked like, individual interviews might give each person the opportunity to speak freely about their perceptions or views without needed to be sensitive to others' reactions.

Conclusion and Implications

The couples in the study reported several factors that contributed to their decisions to have future children after receiving a diagnosis of an inherited childhood genetic disorder,

including the risks and burdens of having another affected child, who would take care of the affected child when the parents were gone, caretaking issues like money, resources, and time, and having determined that the parents are not genetic carriers. Several couples expressed the need for more readily available resources during and after the diagnosis to help with decision-making for future children, such as: accurate information about the disorder, the genetic risks, and subsequent care, so that couples are not left on their own to search the Internet; links to local and national groups related to their disorder; and relationships with trusted health professionals that can help them evaluate the risks of genetic testing balanced with the family's health beliefs. More research is needed in order to help genetic counselors and therapists to be a potential resource for couples during a difficult time.

CHAPTER 5: STUDY TWO

Couples' Experiences in Family Adaptation After Their Child Receives a Diagnosis of Childhood Genetic Disorder

ABSTRACT

As the human genome is more completely understood and new genetic tests are developed to detect both the predisposition to, and the presence of, specific diseases, medical professionals and family therapists are becoming more aware of the effect these tests will have on family systems and on the health beliefs of the family. Once a diagnosis has been received, families now have the task of making sense of the diagnosis and deciding on how the family system needs to change or adapt in order to accommodate the genetic disorder.

The purpose of this qualitative study was to understand the couple's experiences of receiving the diagnosis of an inherited genetic disorder in a child, and how they as a couple, and by extension, their family, adapted to that diagnosis. Nine couples from the Midwestern United States were interviewed using a semi-structured, joint interview process. Using thematic analysis and the couples' own words, this study provides insight into couples' experiences in family adaptation after their child receives a diagnosis of an inherited genetic disorder. Three themes emerged from the interviews in regards to what the family's adaptation looked like after the diagnosis: 1) the couples' experiences of the diagnosis; 2) how families educated themselves about the disorder; and 3) how the couple dealt with the transition. Implications for family therapists are addressed.

Introduction

Genetic testing in the United States started in an experimental laboratory the late 1950s (Feetham & Thomson, 2006). Since the completion of the Human Genome Project in 2003, genetic tests have become widely available, and at relatively low cost, for more than 1,000 genetic disorders (Feetham & Thomson, 2006). As the human genome is more completely understood and new genetic tests are developed to detect both the predisposition to, and the presence of, specific diseases, medical professionals and family therapists are becoming more aware of the effect these tests will have on family systems and on the health beliefs of the family. Families can know about a genetic diagnosis sooner than ever before and they can also learn about the expected course of a disorder over time. Once a diagnosis has been received, families now have the task of making sense of the diagnosis and deciding on how the family system needs to change or adapt in order to accommodate the disorder. A genetic disorder diagnosis brings an array of challenges to parents of a child with a genetic disorder, and depending on the severity and prognosis, can lead to considerable stress on all in the family system. The purpose of this study is to understand the couple's experiences of receiving the diagnosis of a genetic disorder in a child, and how they as a couple, and by extension, their family adapt to this diagnosis.

Background

Emotional Roller Coaster for Parents

The literature points to how stressful this type of diagnosis is for parents. Studies describe a wide array of emotions including anguish, bitterness, confusion, despair, devastation, disbelief, distress, fear, grief, guilt, loneliness, numbness, sadness, shock, or upset (Strehl & Middlemiss, 2007, Nusbaum et al., 2008). Upon receiving the diagnosis of Fragile X Syndrome (FXS), most

mothers recounted being on a "huge emotional rollercoaster" consisting of being devastated on the one hand about the seriousness of the condition, and relieved on the other that their child had a diagnosis (Visootsak et al., 2012). In a study done by Lord, Wastell, & Ungerer (2005), researchers reported parents of a child diagnosed with Phenylketonuria, a manageable, inherited childhood genetic disorder, as experiencing a mild trauma reaction when first learning about the diagnosis and the related challenges. Fonseca, Nazare, and Canavarro (2014) found that for parents, adjustment to a diagnosis i and a reduction in symptoms related to anxiety and depression took about six months post diagnosis of a chronic illness.

Activities to Manage the Diagnosis

Managing the diagnosis requires many activities that invariably include frequent visits with medical professionals. The literature suggests that the role medical professionals play during the diagnosis and subsequent care of children is crucial. In several studies, it is very common for parents to report that initial difficulties in obtaining clear, balanced, and useful information about their unborn baby's condition and future functioning contributed significantly to their feelings of distress, confusion, and frustration. These findings support other studies which have concluded that parents benefit most from receiving clear, balanced, up-to-date written and verbal information, with follow-up appointments for further clarification (Helm et al., 1998; Schuth et al., 1994; Statham et al., 2000). Parents now experience contact with health-care providers as a vital connection to expertise, advice, reassurance, and healthcare instructions (Grob, 2008). In one study, parents reported that positive encounters with doctors had a significant impact on their coping, while negative encounters had an inverse effect on coping (Chaplin, Schweitzer, & Perkoulidis, 2005). In another study, participants reported both negative and positive experiences with their medical professionals; however, the negative experiences

outnumber positive experiences 2.5 to 1, and as a result, this was still a net negative experience for these parents (Goff et al., 2013).

The Long Term Burden of a Genetic Diagnosis on Families

Several studies suggest that for many genetic disorders, substantial burdens are created for families, and these burdens are not time limited, but often endure for years and decades. Some families report they live in a state of hyper vigilance, heightened anxiety, and emotional exhaustion (Kratz et al., 2009). The psychosocial burden experienced by family members of affected children may begin to wear on families over time. For example, some couples report marital disharmony, limitations on their time, financial distress, and hindrance from enjoyment of life (Panepinto et al. 2005; Tunde-Ayinmode 2007). For example, mothers of children with Sickle Cell Disease (SCD) have commonly reported daily emotional challenges of constant fear related to their children's possible death, loss of control over their lives, and helplessness (Wonkam et al., 2014). Pelchat (1993) found that the way in which one perceives the problem is an important predictor of adaptation. The impact of chronic illness on family functioning and how the family adapts is contingent on a range of factors including limitations to finances, spouse and sibling relationships, extended family relationships, and the overall ability to cope (Ashton, 2004). As families transition from the initial diagnosis to regular, ongoing care, one study showed that every day life is often more stable in families that have dealt with a child's illness for a relatively long period of time (Trzcieniecka-Green, Bargiel-Matusiewicz, Wilczynska, & Omar, 2015), and parents of children with a chronic illness eventually develop a shared view of the illness, its management, and its impact on family life (Knafl & Zoeller, 2000), which helps with the overall transition and adaptation of families coping with a chronic illness.

Family Systems Illness Model

The Family Systems Illness Model (FSI) provides a systemic framework for assessing the interaction of a disease or illness between an individual and the family system (Rolland, 1987b). The model emphasizes the interface between time relative to disease progression and family developmental phases, illness type, and components of family functioning (Rolland, 1987a). Not unlike Bronfenbrenner's Ecology Theory (Bubolz & Sontag, 1993), the FSI model highlights the interactive nature between the type of illness/disability/loss, the individual, family, and illness life cycles, and the family's ability to cope. This model also highlights the family's belief systems, culture, ethnicity, spirituality, and gender configuration, wherein each one is nested in the other (Rolland & Williams, 2006). The FSI model was originally developed to provide a framework for the psychosocial types of chronic illness and disability before there was a need to emphasize the genetic components of illnesses. Having an understanding of the family system's reactions to, behavior, and adaptation to illness over time and in relationship to different points in the family life cycle, is crucial for the practitioner to help the family to navigate current and future illness crises (Rolland, 1987a; Rolland & Williams, 2006). There are four key elements of the family illness belief system: 1) the family's sense of mastery and control over the illness; 2) the family's multigenerational evolutionary process with illness, loss, and crisis; 3) the family's ethnic, cultural, and religious beliefs; and 4) the family's assumptions about the etiology of illness (Rolland, 1987b).

Purpose of the Study

The purpose of the current study is to achieve three specific goals in interviews of a sample of nine couples. These goals were all informed by Rolland's Family Systems Illness Model (1987). First, to understand the couple's experience of how their family adapted to the

diagnosis of an inherited genetic disorder in one of their children, while taking into consideration the families' history, cultural background, and religious background. Second, how the family faced and overcame the transitions and related psychosocial challenges that came with a diagnosis of a chronic illness. Third, how the families worked to overcome the assumption about illness and educated themselves as to the etiology and life course of the illness and how this helped them in adaptation to the diagnosis as a family unit.

Methods

Sample

Following approval from the Michigan State University (MSU) Institutional Review Board (IRB), couples were recruited using purposive sampling through a "call for participants" message posted on a Facebook group associated with a particular inherited childhood genetic disease or disorder. Nine couples responded and were interviewed. See Table 1 for a more complete description of the participants.

Participants came from one of the following three genetic disorders: Spina Bifida, Tuberous Sclerosis, and Fragile X. Spina Bifida is the most common permanently disabling birth defect in the United States and happens when the baby is in the womb and the spinal column does not close all of the way. It is sometimes called the "snowflake disorder" because no two cases look the same, and can result in mobility problems, bladder, bowel, or gastrointestinal disorders, and learning disabilities (www.spinabifidaassociation.org).

Tuberous sclerosis complex (TSC) is a genetic disorder that causes tumors to form in many different organs, primarily in the brain, eyes, heart, kidney, skin and lungs. The aspects of TSC that most strongly impact quality of life are generally associated with the brain: seizures, developmental delays, intellectual disabilities, and autism spectrum disorder

(www.tsalliance.org).

Fragile X syndrome (FXS) is a genetic condition that causes intellectual disability, behavioral and learning challenges, and various physical impairments. Though FXS occurs in both genders, males are more frequently affected than females, and generally with greater severity. Life expectancy is not affected in people with FXS because there are usually no life-threatening health concerns associated with the condition (www.fragilex.org).

Of the nine couples interviewed, three couples had a child with Spina Bifida, three couples had a child(ren) with Fragile X, and three couples had a child with Tuberous Sclerosis Complex. Seven of the nine couples had children after their first child was diagnosed, two couples were considering adoption at the time of the interview.

Data Collection

After couples provided informed consent and completed a demographics questionnaire, a face-to-face semi-structured interview was conducted with both the husband and wife present. Couples who participated in study were given a $50 gift card for their participation. All interviews were audio recorded and later transcribed verbatim by the researcher. The interviews were designed to elicit the couples' experiences regarding what thoughts, feelings, and events contributed to a couple's decision to have additional children after already having a child diagnosed with a genetic disorder, how couples communicated with each other about having additional children, and what factors contributed to their decision-making process. Questions were also focused around couples experiences after receiving the diagnosis, cultural, religious, and multigenerational factors in their decisions around the diagnosis, and their approach to overcoming barriers and challenges related to the diagnosis.

In all of the interviews the children were present for the entire interview. This was

because of the unique childcare needs of these families. Sensitive information that would have been disturbing to the child was not discussed in his/her presence.

Data Analysis

Audiotaped interviews were transcribed verbatim, all identifying information was removed from the transcripts, and each participant was assigned a study number. Thematic content analysis was used to analyze each of the interviews (Braun & Clarke, 2006) and NVivo software was used to organize codes and themes that emerged from the data. Thematic analysis is a qualitative method for analyzing data that identifies patterns or "themes" within a data set that provides rich and detailed descriptions and is well suited for health research (Vaismoradi, Turunen, & Bondas, 2013). Braun and Clarke (2006) provide a thorough step-by-step guide to doing thematic analysis that the author followed in this study. First, the author familiarized herself with the data through reading and re-reading the interviews several times as well as listening to the audio recordings and jotting down initial impressions. Next, she read through each interview and coded interesting features of the data by providing labels to these features, followed by grouping these codes/labels into relevant themes. Ultimately the codes were reviewed to ensure they fit within the theme and the theme was given a name.

In order to establish trustworthiness, the author worked closely with another colleague on the coding of interviews and consulted on themes that emerge, using a constant comparative method to check the consistency of the coding and establish inter-rater reliability. Qualitative researchers should provide detailed descriptions of the phenomena they study and their context (King & Horrocks, 2010). These "thick descriptions" help to ensure that the analysis is consistent with the narrative being presented and that dependability is established. The couple's own words are used when describing a particular theme.

Results

Experiences of Diagnosis

Realizing the Permanence of the Condition: Processes Leading to Acceptance. This study sample incorporated two time phases of the Family Systems Illness Model (FSI), the crisis phase and the chronic phase. According to Rolland, one of the transitions couples face in the crisis phase in having a child with a chronic illness is the recognition of the permanence of the condition. All of the disorders represented in this study had different manifestations and different levels of required care which affected the experiences of the couples. For Oliver and Holly, talking with a medical professional helped them understand more about their son's Fragile X condition. Holly had unofficially know she was a carrier for Fragile X since she was 16 years old (her dad was a carrier for disorder so they assumed Holly was as well) but never got officially tested until after she was pregnant with their first child, who ended up with a Fragile X diagnosis. As a carrier she never expressed and signs or symptoms of the disorder, so talking with the doctor allowed Holly and Oliver to face up to the hard reality that this was a diagnosis that had a great deal of stressful ramifications:

> It was the first time we ever talked to anyone. I mean it was emotional because it was a lot of like hard news. But it was the first time we ever talked to anyone that was like, oh all these things that your kid does that are, you don't know how to deal with, here's how to deal with them. Here's what--yes, he's not weird. There's a bunch of other kids that do this. So, he had some weird tics and quirks that were like, yeah that's [son]. And she [doctor] was like no that's all Fragile X. So it was interesting to kind of get--to get that, you know, to get some good tools to help us out with that. (Oliver, son with Fragile X)

Some of the couples in the study shared how important it had been to meet with other families who were dealing with same disorder, and how this helped them to assimilate to the news of the diagnosis:

> We met with some people that had kids that were supposedly of like similar lesion levels with our son…we met with the Spina Bifida community at the zoo and met some great families and kind of started to see things kind of – like that was kind of our – like that first week was just horrendous and then that weekend, we kind of started to pull in some pieces that got us through the next couple of weeks. (Janet, son with Spina Bifida)

Katie and John also said they met with another Spina Bifida family while Katie was pregnant, which helped initially to calm their anxiety about care so soon after receiving the diagnosis, but it wasn't until John's parents started to share their research "findings" that the reality of the level of care needed for their daughter started to set in. Katie and John viewed this support as helpful, even though some of the information communicated by family members was inaccurate or misunderstood:

> John: First thing my mom told me after she Googled it was not all the doors in my house are 30 inches wide. We will fix that before she gets old enough. (Katie laughs)
>
> Katie: And I'm like (laughs)
>
> John: I don't know if she's gonna be old enough. (laughs)
>
> Katie: I think we were first like for what? She's not gonna be fat. (laughs) Oh! For wheelchairs. And then it hit me like oh wait she's gonna be in a wheelchair. (laughs) Like maybe possibly. Okay. (John and Katie, daughter with Spina Bifida)

For Rachel and Zach, as they began to get settled into managing both of their children's chronic illness, Rachel realized how much her role was changing. Acceptance of these new roles was a big component of acceptance of the diagnosis in the child:

> I think for me, I lost the role as parent because parent was a privilege. Now at that point when they were diagnosed, the doctor came first. So I had to become a doctor. I had to become a therapist. I had to learn speech therapy, occupational therapy, physical therapy. Again, how does [daughter's] brain work, how does [other daughter's] brain work. And I remember sitting down one day and said I just want to be a mom. (Rachel, two daughters with Fragile X)

The loss Rachel shared in not feeling like a mom the majority of the time due to the level of care both of her daughters required is echoed throughout many of the interviews and highlights the next transition in the crisis phase of the FSI Model, processing loss and redefining normality.

From Diagnosis to Acceptance: Processing Loss and Redefining Normality. Another transition couples face in having a child with a chronic illness is finding a new normal, what the day to day activities may look like, from daily medications and therapy appointments to regular medical care. Janet describes what is needed in order for their son simply to use the bathroom every day:

> At some point, a lot of these kids undergo surgery where there's a hole in the belly button or something on their part of the stomach where they insert a cath and then push the water in, so it's pushing everything out as oppose to right now, we literally hold the cone in his butt that we then push water up and then it sits there for 5 to 10 minutes and then we pull it out and stuff comes out over the next course of 30 minutes to an hour. So, yes, that's something that he will have to do for the remainder of this life. We do a morning

> routine with him. And then we change diapers all day for him because he doesn't have bladder controlled so he leaks all day. He does not cath. A lot of these kids have to be cathed. So, he's not trainable in terms of a potty and won't ever be to our understanding of how everything works. (Janet, son with Spina Bifida)

David confirmed that he gets up extra early with their son to give him the enema and sit with him for 30 minutes before he has to go to work while Janet gets the rest of their children ready for the day. This process was just one described activity and as is evident, it represents a high level of stress for families, activities that needed to be built into already stressful lives. Nancy and Robert described the amount of planning it takes to simply get out of the house:

> I can't just oh let's go to my friend's house and take you know a change of clothes and their tennis shoes. It's, you know, diapers, wipes. He doesn't eat everything. So I gotta make sure do I have snacks for him to eat? When you go out in public…are there people nearby that he's gonna swat at? Just, you know, just talking. Or are his noises gonna bother people? I went to a water splash pad. My friends were sitting on the bench chit chatting with each other. I'm chasing my kid around because I can't let him by himself. And nobody noticed. Nobody came near me. They didn't move up and talk to me. (Nancy, son with Tuberous Sclerosis)

Nancy particularly highlights the loss that is felt as the relationships with her friends have changed as she continues to adapt to the new normal of life with a child who has a chronic illness.

While all of the couples reported a level of ongoing medical or therapeutic treatment they needed to attend to every day, many of the couples were looking into the future as the redefined normality, and had to readjust their views of dreams and expectations for their child:

> I mean I had all these intentions of taking her to classes at the library and, you know, taking her to swim lessons and play dates with friends. There are still the things we haven't, that haven't come to yet, you know? The things you think that are going to happen as a parent. Seeing your child walk across the stage in their cap and gown. See them go on their first prom date. See them get married. I'm never going to see those things. Sucks. Never say never but. (Jen, daughter with Spina Bifida)

Jen's daughter is nonverbal and confined to a wheelchair, although she is able to communicate via a type to speak program on her iPad. She is currently in a mainstream classroom with children her age because it is still developmentally appropriate but Jen realizes her daughter may have to stay behind or switch to a special needs class as the other children move up in grades.

> That's not--So it's been a lot of loss I think that I've, I've learned through this journey. The loss of he's not gonna go to college. And he's not gonna get married. He's not gonna have kids. And. He's not gonna be on his own. I don't know if he'll always be at home. I don't know if he's eventually gonna be in a group home. You know. So it's that, that loss of everything. (Nancy, son with Tuberous Sclerosis)

> Holly: [Son] didn't like birthdays. He doesn't like it when people sing happy birthday. He didn't it even like opening presents. It was so much anxiety for him. So for his fifth birthday we took him to the store and let him pick out toys. Which is like a weird thing. Like it's not what you expect of a parent but--but we didn't sing happy birthday to him. We did what he wanted though and everyone had a great time. But that's not something you would expect, you know? Changing your expectations to fit the reality of your situation.

> Oliver: What she [Holly] was talking about with the expectations, that was a big thing. It was like every event we had for so long. She would just come home and cry and just be like sad. (Oliver and Holly, son with Fragile X)

How Families Educated Themselves

Relationships With Health Professionals. One key aspect to coping with the chronic illness and finding meaning for the disorder lies in the relationship the couples have with health professionals. All of the couples reported at least one good relationship with a health professional, whether it was a doctor, speech therapist, or occupational therapist:

> We really value Dr. Z and he's-- I mean, I, I saw him one day in a parking lot. Like what? Two years ago? I think. I was out grabbing lunch. He came out of the store next door. I said hey Dr. Z. He's like hey [wife], how are ya? How's [son]? [Son] was nowhere around. And I said oh he's okay but he's developed these new seizures. And he goes well tell me about them. This is in a parking lot. So he knows his patients. (Nancy, son with Tuberous Sclerosis)

Ben and Kelly explained the benefit of being linked in to a specialty clinic and the differences in care between the clinic and local doctors:

> They are so stinking fantastic, I don't know -- they've tried to do a clinic at [local town] which the doc who is the head of it worked under the group in [major city]. So he is familiar with it. And they are so epically failing at this clinic and I'm like okay, I'm going to give you the benefit of our being here so that you can learn. But I'm not getting -- you are not allowed to boot me out of this other office. I won't give that up because [major city] knows -- they're involved in such a deeper level of stuff with TSC, than our local --

> even the local pediatric doctors. So it's just a whole different level down there. It's just amazing. (Kelly, daughter with Tuberous Sclerosis).

Kelly and Ben live in a rural area where they have to travel an hour to visit their local doctor, let alone having to stay overnight when they travel to the specialty clinic, but the couple recognizes the importance of the level of care and peace of mind they receive from the specialty clinic.

Personal Research. Another key aspect to coping with the chronic illness and finding meaning for the disorder is found in being well-informed about symptoms, behaviors, and treatment options. For many of the couples in this study, their child's diagnosis was their first introduction to the disorder and they needed to learn as much as they could about their child's condition. While some of the health professionals were a helpful resource, for Rachel and Zach, the doctors knew as much as the couple did about the disorder:

> So the answer the doctors gave us was Mr. and Mrs. Simon, we're going to be very honest with you. We don't know. We don't know about girls. We can't give you any answers. …at night the kids were sleeping, when they finally did sleep, I read every research paper there was. I tried to figure this out and first I had to figure out how a normal brain functioned. And then I had to figure out how the girls' brain is functioned and I figured out they don't work at all the same. (Rachel, two daughters with Fragile X)

Many couples in the group also turned to the national organization or local, specialized Facebook groups for information and resources about managing the day-to-day care aspect of their disorder:

> We belong to the Spina Bifida Association, Spina Bifida Association of Michigan, so we have some people in like situations that we can bounce ideas off and stuff like that. Some

of the Facebook groups are extremely helpful. I mean there's a whole Facebook group on just bowel management. All we talk about is poop. (Janet, son with Spina Bifida)

Facing and Overcoming Transitions

Managing Relationship Imbalances. A major transition couples faced in living with the diagnosis was in dealing with relationship imbalances that occurred with every transition brought upon by the diagnosis. Once couples and families have made it through the disruptions of the crisis phase, they move into the "long haul" or chronic phase, wherein which the couple has to face and address the day to day living with the chronic illness, while at the same time carrying out the tasks of their regular lives. One important task from this phase is how couples will manage relationship imbalances. Two of the husbands, David and Steve, shared how the relationship with their wife shifted after the diagnosis, and how they no longer felt as important as they had before the diagnosis:

Janet: I mean I definitely think we give more to our kids than each other, for sure. I mean that's a change in our roles.

David: For dynamics wise, she's definitely [son's] mom first, the rest of their mom, second and so I think what I said my wife third because I think that's how it probably plays out in our family. (David and Janet, son with Spina Bifida)

David described with such sadness how his relationship with Janet has changed, as if he wished things could be different, and implied that they might need couples' therapy to work on improving their relationship. On the other hand, Steve described the relationship imbalance with Kristen in a very matter of fact manner:

You figure out a way to do it between the two of you. And you kind of get used to coming last. I mean Kristen feels the same is that, throughout the day, the girls always

> come first. Then you try and take care of your problem and then you come last. That's just the way it is. Even the dogs come before us because they're also going get fed and…So you kind of just get used to it. And then your relationship certainly goes into the back burner. I mean we have not spent a night away from the girls in 3 years. (Steve, daughter with Tuberous Sclerosis)

In contrast, and in spite of these role imbalances, most of the couples in the study reported the relationship with their spouse had grown stronger, and that they were closer and they are more supportive of each other than they were prior to the diagnosis. Several of the couples shared that one of the reasons they are more supportive of each other is because they have no only else to rely on. Two of the couples did not live close to family, while others felt a little more insulated from the rest of the world:

> I know that it's changed, but I know we -- I think we do, I hope we do rely on each other. I feel like we're more isolated from other families. We don't get a chance to -- we don't have the friendships that most families have. You see the neighbors going out and they're chumming with each other and I'm like -- if my kids are having a bad night, I can't go out and visit because I've got to sit here and monitor for behaviors or seizures or it's almost med time. (Kelly, daughter with Tuberous Sclerosis)

Several couples also shared how important communication was in maintaining relationship balance within the marriage:

> We really slow down. I think we really slow down on the--when we're having arguments because we wanna make sure we're both communicating very clearly. Especially with her care. We both, at least I think, we both take the time to research and form our own

> opinions and our own process through everything basically. John's much more on the logical side and the scientific side. (Katie, daughter with Spina Bifida)

Rachel and Zach shared how they initially had difficulty as they were moving into the chronic phase and learning how to maintain their connection. This couple has two teenage daughters living with Fragile X who have severe mental delays and one does not communicate verbally. They received the girls' diagnoses almost simultaneously and immediately jumped into crisis mode, learning how to manage the girls' disorder. Rachel said for a period of time she didn't feel as close to her husband:

> And so in a way that sent me down at the time almost a road of depression towards him and anger saying how can you give up. And he wasn't giving up. But to me, I felt like he was giving up and it's like okay, I've got to do this all on my own. I've got to figure this out. And I would get so frustrated because he would come home and I would have, maybe a half an inch of paper work saying, okay, this is all medical jargon. Teach me what this means. It's like I just worked 14 hour days. I can't do this. So I think -- maybe 3 years there that I was very angry because I felt like I was in this healing process alone. Now that's not what his thoughts were, but that's where my thoughts were. (Rachel, two daughters with Fragile X)

Zach recognized his wife's feelings at the time, especially as they were trying to make sense of the diagnosis for the family. When Rachel was feeling alone, he had this response:

> I said sweetheart, I told you I loved you. Whatever day I told you I loved you the first time, I don't remember when it was, this was way before we got married. And I told you I'd love you forever. And I didn't say I love you if our kids were all running around

> quoting scripture and winning Nobel Prizes. I said I would love you and there was period at the end of that sentence. (Zach, two daughters with Fragile X)

Figuring Out Family Identity. Another component of managing transitions and the diagnosis was figuring out a new identity as a family and adapting this identity throughout the life cycle. Couples and families also need to learn how to make sense of the diagnosis and what it meant for their family. Two of the families, both with a Tuberous Sclerosis diagnosis, spoke of how their identity is connected to the diagnosis and day-to-day care. In essence, the family organized itself and its identity around the illness and the activities required by the diagnosis. Kelly and Ben, who did not have a lot of family support to help them out and who worked hard to get the necessary treatment for their daughter, saw themselves as an official "Team", became a 501c3 organization and put on several organized fundraisers under this "Team" identity. Steve and Kristen had the opposite opinion in terms of identity:

> I think a lot of these families, who have a member who is affected with TS, they kind consumes them up, it's kind of like the identity, I have a child with TS and I'm going to go first and then they are so focused on this TS they tend to lose sight of what's going on around them. They are the people who have children with the same condition, but they remain with one little enclave and that is their sole focus, is they lean on these people to support them and then look after the child with TS. They forget that there's a big world out there, TS does not define you. (Steve, daughter with Tuberous Sclerosis)

Discussion

A genetic disorder diagnosis brings an array of challenges to parents of a child with a genetic disorder, and depending on the severity and prognosis, can lead to considerable stress on all in the family system. This study provides insight into couples' experiences in family

adaptation after their child receives a diagnosis of an inherited genetic disorder. Three themes emerged from the interviews in regards to what the family's adaptation looked like after the diagnosis: 1) the couples' experiences of the diagnosis; 2) how families educated themselves about the disorder; and 3) how the couple dealt with the transition.

Experiences of Diagnosis

The couples' experiences of the diagnosis can be broken down into two parts: Realizing the permanence of the condition, the processes leading to acceptance and processing loss and redefining normality. Many couples in this study found talking with doctors about the diagnosis and subsequent treatment was helpful in understanding the permanence of the condition and accepting how life might have to change. Some of the couples were able to meet with other families who had the same disorder and got a glimpse into what the day-to-day care could look like. Those couples said the meetings were helpful because the unknown became less scary and they were able to visualize what it might be like to care for their child's condition.

Another part of the diagnosis experience was processing loss and redefining normality. During this phase many families experienced anticipatory loss, which refers to the experience of living with possible, probable, or inevitable future loss (Rolland, 2006). Many of the couples addressed the loss of future dreams for their children, the idea that their child may never get married, have children, or even live on their own. Nancy described how sad she felt that her friendships were changing, that she wasn't able to have a "normal" friendship because her son required so much care when they went out, and most of her friends did not know how to relate to her anymore. In redefining normality, every family's "normal" was different and organized around the care needed for the affected child(ren), whether it was simply helping them to use the bathroom, as was the case for the Janet and David, or for most of the families it was education

plans at school or taking them to multiple therapy and doctor's appointments on a weekly basis.

How Families Educated Themselves

For most of the couples in this study, this was the first encounter they had with this disorder and they had no idea what they were to face in the coming years. The literature reflects that parents who reported positive encounters with doctors had a significant impact on their coping, while negative encounters had an inverse effect on coping (Chaplin, Schweitzer, & Perkoulidis, 2005). Several couples described how important the relationship with their health professionals was in not only understanding the diagnosis but also understanding what kind of ongoing care would be needed for their child. A few couples described a negative experience with health professionals, receiving inaccurate information about inheritance rates and doctors who knew little about the disorder. Chris and Jen, whose daughter has Spina Bifida, shared that the doctor who diagnosed their daughter used the out of date term "water head" to describe the disorder. Several couples also reported feeling pressured to terminate a pregnancy or consider not having any more children after receiving a diagnosis.

During this time of uncertainty, most of the couples also turned to the Internet to find out more information about their particular disorder and to start to locate resources. All of the couples were recruited through the Facebook group related to their disorder, and many of these couples felt these groups were invaluable resources for information, especially relating to the day-to-day care of their child.

Facing and Overcoming Transitions

According to the literature, a transition's time span extends from the first signs of change (diagnosis), through the period of instability (crisis phase), until a new stability has been achieved (chronic phase) (Messias et al., 1995). Two major transitions couples face in living

with the diagnosis of a genetic disorder are managing relationship imbalances and figuring out family identity. In managing relationship imbalance, some of the fathers, like David and Steve, pointed out how the relationship with their spouse had changed and they felt less of a priority. Some of the couples also described a lack of connection with their spouse, but all of the couples alluded to being okay with imbalance because the needs of their children should come first. Despite these imbalances most of the couples reported their relationship was stronger and they were more supportive of each other. One reason for this is many of the families felt isolated and had no one else to rely on for help, as highlighted by the quotes above. Several couples mentioned how important communication was with each other as a way to overcome the relationship imbalances.

Another transition experienced by couples in this study was figuring out a new family identity in light of the diagnosis. Two families, both with a Tuberous Sclerosis diagnosis, had differing points of view in terms of their family identity: for Kelly and Ben, the entire family organized themselves around the illness and activities required by the diagnosis, proudly identifying as a "team" for their daughter and the illness. The other family, Kristen and Steve were very bothered by families whose entire identity was wrapped up in the disorder and was making sure that disorder would not be what defines their daughter but only a part of who she is.

Limitations

The purpose of this study was to understand the couples' experiences in adaptation after a diagnosis of a childhood genetic disorder, and although disorders that were either inherited or spontaneous mutations were included the study was not meant for a direct comparison of the two and therefore may be a limitation to this study. There was a diverse sample in terms of reported resources (income, support) and we were not looking at whether or not more resources aided in

better adaptation. Additionally, with a small sample size of only nine couples, the results are only generalizable to these nine couples and not to the experiences of other couples. Another limitation of this study is the lack of racial and ethnic diversity with 17 of the 18 participants identifying as White.

All of the couples were interviewed conjointly in their homes, and although there was an opportunity to observe the dynamics of the relationship and what caretaking looked like, individual interviews might give each person the opportunity to speak freely about their perceptions or views without needed to be sensitive to others' reactions.

Implications

This study emphasized the significance of having a good relationship with a health professional the families trust in learning to accept the permanence of the disorder of their child and in redefining the family's normality. Since family therapists are trained to work with multiple relationships, they can help guide families who are working with an interdisciplinary health care team to help meet the families' needs at any point during the process, from considering to test to ongoing medical care.

Communication about genetics and chronic illness is an ongoing process, not a one-time conversation (Wilson, et al., 2004), and family therapists can help provide a forum for the family to discuss their feelings and their needs as more details about an illness emerge or other relatives may need to be contacted. In this study, communication was a vital component in the couples' processing of their child's diagnosis and in redefining the family's normality. Shared health beliefs help family members find meaning in their illness experience, whether current or future, and help guide decisions and actions (Rolland, 2006). Family therapists offer families an opportunity for ongoing discussion about the families' health beliefs and communication

approaches to disseminating genetic information to extended family members. These discussions may also serve as social support functions to build relational bonds and to facilitate coping among family members (Peterson, 2005).

Conclusion

A genetic disorder diagnosis brings an array of challenges to parents of a child with a genetic disorder, and depending on the severity and prognosis, can lead to considerable stress on all in the family system. Having a trusted relationship with health professionals and access to accurate and informational resources helped these families navigate through the process of adaptation after a diagnosis and through the transition of finding the normal.

CHAPTER 6: CONCLUSION

Contributions of the Current Studies to Existing Research

As the human genome is more completely understood and new genetic tests are developed to detect both the predisposition to, and the presence of, specific diseases, medical professionals and family therapists are becoming more aware of the effect these tests will have on family systems and on health beliefs of the family. Scientists have discovered the genetic link to over one thousand disorders and medical conditions and continue to work on finding new links every year. This is important for families because we are now able to understand even more about our health, sometimes before ever experiencing any symptoms of disease. Prenatal testing for genetic conditions/mutations is also available to individuals and families, which allows couples the opportunity to learn if their child has a genetic condition and what treatment options are available for such condition. The literature points to how stressful a genetic diagnosis in children is for parents (Strehl & Middlemiss, 2007, Nusbaum et al., 2008). The results of the current two studies explain the factors that contribute to the decision-making processes of families regarding genetic testing and the process of adaptation families experience after the diagnosis of a genetic disorder.

Decision Making Processes of Families Regarding Genetic Testing

Evaluation of the Risks and Burdens for the Future. This study reflects the experiences of nine couples that made or are in the processing of making a stressful and emotional decision regarding having another biological child knowing there was a genetic risk to future children. Many common genetic conditions occur in families with no previous family history or experience of the disorder (Collins & Williamson, 2003). Eight of the nine couples

interviewed had no knowledge of genetic conditions in their families prior to having children. Holly and Oliver, who knew about their potential genetic risk, reported having some conversations about the risk prior to having children but Holly did not get tested until she was pregnant with their first child (she found out when she was 16 she was potentially a carrier for Fragile X).

Risks. One of the themes echoed throughout this study is the evaluation of the risks and burdens for the future that couples used as part of their decision-making processes for future children. The couples in the study identified several positive and negative risks. Positive risks help families predict the likelihood of events occurring in the future and allow the family to feel empowered and more in control of their actions and medical destiny (Hallowell et al., 2004). Some of the couples decided to get genetically tested and in receiving a negative result, meaning they did not have the genetic mutation, went on to have more biological children. Negative risks constitute a heightened awareness of potential unknown dangers or hazards that then became a major source of anxiety within the family (Hallowell et al., 2004). John and Katie were "hashing out" the decision to have another biological child after their first child was diagnosed with Spina Bifida and were on opposite sides, John thought the risk was too great to take and Katie was still grieving the loss of not having another biological child.

Burdens. Another factor in the decision-making process of the couples in this study was the potential burdens on the family. During the interview several couples described a typical day/week, the amount of care and resources needed for their affected child. Families were dealing with several therapy appointments a week, multiple medications, and some couples were dealing with diapers and/or catheters and enemas (in the case of Spina Bifida). Other couples were dealing with behavioral issues and children who were unable to communicate. One father,

Robert, specifically commented on being worried about finding balance in giving enough attention between children, which is reflected in the literature (Downing, 2005), and other couples shared the same sentiment. Several couples expressed concern about who would take care of their children after they were gone because many of them would need on-going care and they did not want to burden other children or extended family.

Collecting Information to Inform Decision Making. A second theme echoed throughout the study was the need to gather information to make an informed decision about future children. A study by Broadstock and Michie (2000) showed that it isn't clear what information is needed for an informed decision because each family may find different information important. Some of the couples thought it was important to get genetic testing done, either for themselves or for their child. Many of those couples felt relieved when the test came back negative and were more comfortable planning for future children.

Couples Lack of Decision Making Processes. The purpose of this study was to explore how couples communicate with each other about having additional children and what experiences and factors contribute to their decision-making processes. The researcher assumed that once a couple received a genetic diagnosis for their child, there would be a process for decision-making within a couple for having additional children. This was not the case. Five of the nine couples included in the study learned that the genetic disorder came from a spontaneous mutation and was not inherited from either parent. For these couples the spontaneous mutation greatly lessened the risk of having another child with a genetic disorder and the couples proceeded with future pregnancies.

Three of the nine couples in the study were already pregnant or had more children by the time they received the genetic diagnosis, and most of the couples were done having children at

the time they received the diagnosis, which eliminated the need for any conversations about future children. Several couples in the study acknowledged that having more children after the diagnosis was not a deterrent because the couples already had experience in taking care of the disorder and knew what to expect. This supports existing literature, where in a study done on parents of children diagnosed with cystic fibrosis (CF), Ormond et al. (2007) found that the majority of parents were likely to pursue (or had already pursued) future pregnancies because the initial experience of diagnosis and care of a child with CF lessened any anxiety about caring for future children with CF.

During the course of the interviews two themes emerged that were not overtly addressed, ambiguous loss and resilience. Ambiguous loss is defined as an incomplete or uncertain loss (Boss, 2007). There are two kinds of ambiguous loss. In the first, people are perceived by family members as being physically absent but psychologically present, like a soldier who is missing in action or adopted children and their birth parents. In the second, people are perceived by family members as physically present but psychologically absent, like in cases of substance abuse, mental illness or chronic illness (Abrams, 2001). Boss and colleagues have identified several areas of ambiguity that contribute to family distress in relationship to chronic illness: a) a lack of clarity in diagnosis, b) difficulty in predicting outcomes, c) day to day changes in functioning of the ill person that affect family relationships, d) the fact that the ill individual may give an outward appearance of health thus raising expectations for his or her behavior and functioning within the family, and e) fear that important emotional relationships will be affected by the illness (O'Brien, 2007). Nancy highlights the ambiguity in predicting future outcomes, as she describes the loss she feels for her son:

> That's not--So it's been a lot of loss I think that I've, I've learned through this journey. The loss of he's not gonna go to college. And he's not gonna get married. He's not gonna have kids. And. He's not gonna be on his own. I don't know if he'll always be at home. I don't know if he's eventually gonna be in a group home. You know. So it's that, that loss of everything. (Nancy, son with Tuberous Sclerosis)

Being able to make sense out of the loss, the ability to maintain hope and to find some way to change are all needed in order to resolve the loss, which expresses the essence of resilience. Walsh (1998) defines resilience in families as the capacity to rebound from adversity, strengthened and more resourceful. Many of the couples in this study either did not live close to family or aging parents made it difficult to rely on them for much support, therefore the couples needed to be resilient in how they managed day-to-day life. Kelly and Ben are a reflection of this resilience, in that they created a 501c3 organization in order to help fundraise for the care their daughter with Tuberous Sclerosis needs.

Family Adaptation Process After a Diagnosis

Experiences of Diagnosis. The couples' experiences of the diagnosis can be broken down into two parts: Realizing the permanence of the condition, the processes leading to acceptance and processing loss and redefining normality. Many couples in this study found talking with doctors about the diagnosis and subsequent treatment was helpful in understanding the permanence of the condition and accepting how life might have to change. Some of the couples were able to meet with other families who had the same disorder and got a glimpse into what the day-to-day care could look like. Those couples said the meetings were helpful because the unknown became less scary and they were able to visualize what it might be like to care for their child's condition.

Another part of the diagnosis experience was processing loss and redefining normality. During this phase many families experienced anticipatory loss, which refers to the experience of living with possible, probable, or inevitable future loss (Rolland, 2006). Many of the couples addressed the loss of future dreams for their children, the idea that their child may never get married, have children, or even live on their own. Nancy described how sad she felt that her friendships were changing, that she wasn't able to have a "normal" friendship because her son required so much care when they went out, and most of her friends did not know how to relate to her anymore. In redefining normality, every family's "normal" was different and organized around the care needed for the affected child(ren), whether it was simply helping them to use the bathroom, as was the case for the Janet and David, or for most of the families it was education plans at school or taking them to multiple therapy and doctor's appointments on a weekly basis.

How Families Educated Themselves. For most of the couples in this study, this was the first encounter they had with this disorder and they had no idea what they were to face in the coming years. The literature reflects that parents who reported positive encounters with doctors had a significant impact on their coping, while negative encounters had an inverse effect on coping (Chaplin, Schweitzer, & Perkoulidis, 2005). Several couples described how important the relationship with their health professionals was in not only understanding the diagnosis but also understanding what kind of ongoing care would be needed for their child. A few couples described a negative experience with health professionals, receiving inaccurate information about inheritance rates and doctors who knew little about the disorder. Chris and Jen, whose daughter has Spina Bifida, shared that the doctor who diagnosed their daughter used the out of date term "water head" to describe the disorder. Several couples also reported feeling pressured to terminate a pregnancy or consider not having any more children after receiving a diagnosis.

During this time of uncertainty, most of the couples also turned to the Internet to find out more information about their particular disorder and to start to locate resources. All of the couples were recruited through the Facebook group related to their disorder, and many of these couples felt these groups were invaluable resources for information, especially relating to the day-to-day care of their child.

Facing and Overcoming Transitions. According to the literature, a transition's time span extends from the first signs of change (diagnosis), through the period of instability (crisis phase), until a new stability has been achieved (chronic phase) (Messias et al., 1995). Two major transitions couples face in living with the diagnosis of a genetic disorder are managing relationship imbalances and figuring out family identity. In managing relationship imbalance, some of the fathers, like David and Steve, pointed out how the relationship with their spouse had changed and they felt less of a priority. Some of the couples also described a lack of connection with their spouse, but all of the couples alluded to being okay with imbalance because the needs of their children should come first. Despite these imbalances most of the couples reported their relationship was stronger and they were more supportive of each other. One reason for this is many of the families felt isolated and had no one else to rely on for help, as highlighted by the quotes above. Several couples mentioned how important communication was with each other as a way to overcome the relationship imbalances.

Another transition experienced by couples in this study was figuring out a new family identity in light of the diagnosis. Two families, both with a Tuberous Sclerosis diagnosis, had differing points of view in terms of their family identity: for Kelly and Ben, the entire family organized themselves around the illness and activities required by the diagnosis, proudly identifying as a "team" for their daughter and the illness. The other family, Kristen and Steve

were very bothered by families whose entire identity was wrapped up in the disorder and was making sure that disorder would not be what defines their daughter but only a part of who she is.

Implications for Clinical Practice of Marriage and Family Therapy

The psychosocial and interpersonal aspects of genetic illness are a natural venue in which family therapists can participate as part of the healthcare team in the evolving practice of helping patients to: 1) understand their risk for genetic illness, 2) cope with this information and the results of testing other family members, 3) make decisions regarding preventive measures, treatment, and testing, and 4) improve individual coping and family relationships in the face of these new challenges (McDaniel, 2005). Ideally, family therapists work collaboratively on an interdisciplinary health care team that would include primary care physicians, specialists, genetic counselors, and possibly clergy (McDaniel, Peters, & Acheson, 2006). Parents experience contact with health-care providers as a vital connection to expertise, to advice, to reassurance, to instructions (Grob, 2008), and report that positive encounters with doctors have a significant impact on their coping (Chaplin, Schweitzer, & Perkoulidis, 2005).

Study 2 emphasized the significance of having a good relationship with a health professional the families trust in learning to accept the permanence of the disorder of their child and in redefining the family's normality. Since family therapists are trained to work with multiple relationships, they can help guide families who are working with an interdisciplinary health care team to help meet the families' needs at any point during the process, from considering to test to ongoing medical care.

Communication about genetics and chronic illness is an ongoing process, not a one-time conversation (Wilson, et al., 2004), and family therapists can help provide a forum for the family to discuss their feelings and their needs as more details about an illness emerge or other relatives

may need to be contacted. In Study 2, communication was a vital component in the couples' processing of their child's diagnosis and in redefining the family's normality. Shared health beliefs help family members find meaning in their illness experience, whether current or future, and help guide decisions and actions (Rolland, 2006). Family therapists offer families an opportunity for ongoing discussion about the families' health beliefs and communication approaches to disseminating genetic information to extended family members. These discussions may also serve as social support functions to build relational bonds and to facilitate coping among family members (Peterson, 2005).

Study 1 highlighted that couples felt some level of uncertainty in deciding whether to move forward with genetic testing, either for themselves or for their children. Some couples disagreed whether genetic testing was even necessary for their family, whereas other couples felt genetic testing was necessary in order to mitigate the risk of a having a genetic disorder in the family. Risk embodies aspects of ambiguity and uncertainty. When linked with genetic inheritance, risk can be very difficult to deal with from a personal and familial perspective (Peters, Djurdjinovic, & Baker, 1999).The concept of risk is especially highlighted relative to predictive genetic testing, where decisions of risk are paramount because the information gleaned from such testing can drastically alter an individual's sense of self, can change personal identities, and can cause extreme anxiety and stress (Hallowell et al., 2004). Family therapists, acting as a trusted health professional, can help families process any anxiety or negative feelings they may have regarding the decision-making process for genetic testing within the family.

Another theme identified in Study 2 that may be pertinent to family therapists is the anticipatory loss many couples identified when dealing with the diagnosis and subsequent care of an affected child. Anticipatory loss refers to the experience of living with possible, probable, or

inevitable future loss (Rolland, 2006). The losses described by families with members tested for an inherited genetic disorder differ from those losses described by families with a member who suffers from a chronic or terminal illness of a non-hereditary nature. When a member tests positive, it is a loss without a foreseeable end (Sobel & Cowan, 2003). The anticipatory losses described by the couples in the study were twofold. On one hand, couples described the loss of future dreams for the affected child, such as attending college, getting married, or moving out of the house. On the other hand, some couples described the unanticipated loss of friendships and friendships that changed because the caretaking needs of the affected child dictated a lot of the family's time. Family therapists are in a unique position to assist families in identifying and coping with potential losses.

Future Considerations

The existence of genetic conditions produces a need for psychosocial and family interventions throughout the lifespan (Peters, Djurdjinovic, & Baker, 1999). Although genetic testing may be a one-time event for a family, the results of the test have implications for all family members across all life cycle stages. Family therapists should consider working collaboratively as part of an interdisciplinary health care team to provide the best possible care to families. As a part of this team, family therapists can highlight the impact of predictive genetic testing and the associated psychosocial issues on families and offer a space for exploring the families' needs and feelings about the results of the test. Taswell & Sholtes (2003) emphasize that, "[b]ecause knowledge of their genetic status will affect every aspect of their life, from the time of testing and into the future, they not only should receive immediate post-test counseling but periodic long-term counseling at life cycle mileposts" (p. 121).

Family therapists who are interested in the intersection between medical issues, the medical community, and family therapy should consider furthering their education in specialized programs like Medical Family Therapy. There are several degree and post-degree certificate programs in Medical Family Therapy. For those training programs that do not have an emphasis on Medical Family Therapy, those programs should consider adding elective course that address the needs of families with genetic disorders or chronic illness.

At the outset of the study, the researcher anticipated that a couples' spiritual or religious beliefs would play a significant role in the decision-making process for genetic testing and in the adaptation process after the diagnosis. The interviews did not show this to be the case, and it is unknown if the participants in the study did not happen to be spiritual/religious or they did not think it was an important part of either process. Further research is needed to understand what role spirituality/religious beliefs play in the decision-making process for genetic testing, especially for testing done in utero, when the couple may face the decision of whether to terminate the pregnancy based on the diagnosis.

APPENDIX

Interview Guide

Thank you for agreeing to participate in this interview today. The purpose of this interview is to learn more about your experiences with genetic testing for your child(ren) and how you communicated as a couple about the results. This interview will take approximately one hour to complete. This interview will be tape recorded today and later transcribed. All identifying information will be eliminated from the transcribed interview to ensure confidentiality. Do you have any questions?

I'd like to first start with getting to know more about you and your family. Please share a little bit about how you met, who is in your family, and what a typical day looks like for you.

- Who are your children? Ages?
- Where do you work?
- What are your typical daily routines?

What were participant experiences with the medical system before, during, and after the genetic testing process for their affected child?

- Tell me about how you found out your child has _____.
    - Describe the story about how the diagnosis came about and the steps you went through to receive that diagnosis.
    - Describe your interaction with the doctor; genetic counselor

How did the diagnosis and related medical system interfaces affect you and your family in your roles as spouse/parents?

- In what ways has your relationship with your spouse changed or not changed since the diagnosis of your child?
- In what ways has your role as a parent changed or not changed since the diagnosis of your child?

How did the diagnosis of your child affect your medical decision making processes especially around having additional children?

- Have you and your spouse communicated about wanting to have another child? If yes, ask them to describe in detail the process. If no, explore why not.
- How have you and your spouse communicated about genetic testing for future children?

What types of supports and resources did you as a couple utilize as you moved through the diagnosis and related decision making processes?

- Extended family

    - Could you tell me about the reaction or response your extended family had to your child's diagnosis?
    - How have they shown or not shown support for your family? Please explain
- Community Support
    - What role has your workplace had in supporting or not supporting you and your family?
    - What role has your spirituality/religion/church community had in supporting or not supporting you and your family?
    - How have you utilized national and/or local organizations associated with your child's disorder?
- Medical system (doctors, genetic counselors, etc.)
    - Can you tell me about any resources you were or were not offered during or after testing?
    - Describe the supports you received from the medical system.
    - What resources did you have to get for yourself and where did you get them?

Can you think of anything else in regards to your experience with genetic testing, the medical system, or decision making that we should discuss?

Thank you for your time.

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