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An Exploration of the Genetic Counselor-Patient Relationship Following a Life-limiting Prenatal Diagnosis

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An Exploration of the Genetic Counselor-Patient Relationship
Following a Life-limiting Prenatal Diagnosis

by

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Bachelor of Science
Wofford College, 2013

Submitted in Partial Fulfillment of the Requirements

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Dedication

I would like to dedicate my thesis work to my loving family and friends, especially my mother, Susan Curtis, and my little sister, Elizabeth Anderson. They have been the support and encouragement I have always and only needed. Without them there to laugh with and fight with, I wouldn't be who and what I am today.

Acknowledgements

I would first of all like to graciously thank my committee members Janice Edwards, Kathryn “Katie” Berrier and Krista Redlinger-Grosse who have been my beacons of light through this process. They have offered guidance, encouragement, enthusiasm, and inspiration from the first moments of this project’s fruition (and even before). Janice Edwards rescued me from multiple failed ideas and helped me discover my “passion,” Katie was my spark who made me believe this was possible, and Krista kept me grounded and thinking from all sides, at all angles. I will be forever grateful to them for making me the type of person who is capable of producing something that can, hopefully, help others. I would also like to also thank Peggy Walkers who is probably the most dedicated and generous person I’ve ever met. She is dedicated to empowering others which in turn empowers her; it is certainly a beautifully perfected cycle! I would like to thank my classmates who stressed out with me and complained with me when we all felt overwhelmed. I always knew I was not on this island of anxiety alone. Finally I would like to thank all the other faculty members who were a constant reminder that the process of creating a thesis project is survivable and that you can be better for it.

Abstract

The proposed Reciprocal-Engagement Model of genetic counseling is comprised of five core tenets with the relationship between the genetic counselor and patient being central to the process and success of genetic counseling. This study sought to explore the relationship between the genetic counselor and patient during and after a prenatal diagnosis of a “life-limiting” diagnosis that resulted in a major loss (termination, stillbirth/miscarriage, or neonatal death). Eight genetic counselor and patient pairs were individually/separately interviewed about their mutual relationship and asked about the development and maintenance of the relationship, the contributing factors, and the long-term effects on both the genetic counselor and patient. The length of these relationships ranged from four months to fourteen years following their initiation. All clinical relationships established required extensive follow-up via multiple modes of communication with more frequent communication in the beginning and less as the relationship evolved with more equal initiation of communication over time. The content of conversation expanded to include more personal elements from both the genetic counselor and the patient perspectives. Most participant pairs fell somewhere on a spectrum of professional relationship to deeply personal relationship as one pair maintained an exclusively personal relationship. The support offered by the genetic counselor during the time of crisis was both essential and unique to the patient compared to other healthcare providers and family/friends. Strategies employed and/or characteristics of the genetic counselor and patient did contribute to the development and

maintenance of the relationship as did the life-limiting nature of the diagnosis, which was thought to overall strengthen the connection. The long-term effects on participants reveal clinical implications for genetic counseling. This exploratory study highlights the unique service of support offered by genetic counselors, as well as potential avenues for future research and training implications.

Preface

“Trust in the other to grow and in my own ability to care gives me courage to go into the unknown.... And clearly, the greater the sense of going into the unknown, the more courage is called for in caring,” (Mayeroff, 1990, pp. 34-35)

“We learn about suffering from witnessing stories that teach us about resilience, courage, and grace while confronting adversity. We learn from essays and autobiographies that stimulate reflection and deepen our understanding of the breadth of human experience. And we also learn from art and music, which provide a conduit to emotion and nurture our empathic abilities. The integration of these experiences enlightens us about the uniquely personal and subjective experience of suffering,” (Shapira 2013, p. 1609).

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List of Abbreviations

CHD.....	Congenital Heart Defect
CDH.....	Congenital Diaphragmatic Hernia
FTS	First Trimester Screen
MMS.....	Multiple Marker Screen
REM	Reciprocal-Engagement Model
VSD.....	Ventricular Septal Defect

Chapter 1. Background

1.1 Introduction

Genetic counselors often work through difficult situations with their patient, which subsequently develops into a reciprocal and lasting relationship. The personal and/or professional characteristics of the patient and the genetic counselor as well as the patient and genetic counselor perspectives of the crisis/significant life event may provide valuable insight into important aspects of this relationship. To understand how this relationship is developed and maintained, research about contributing factors must be explored. This study sought to explore the unique connection between the prenatal genetic counselor and patient who have worked through a crisis of a life-limiting prenatal diagnosis together.

Providing background on some of the factors which may contribute to this relationship is important to understanding the relationship between genetic counselor and patient. Some of these factors include the nature of the genetic counseling profession and the elements of practice employed, as well as the unique characteristics about both counselor and patient that individually and collectively contribute to the relationship's development and maintenance. This background will specifically explore the nature of helping professions, like genetic counseling. This background will also explore the Reciprocal-Engagement Model (REM) of genetic counseling and its emphasis on the patient-counselor relationship and the commitment to and development of the reciprocal relationship itself. In addition, the professional and personal characteristics of the genetic

counselor, the characteristics and needs of the patient, and psychosocial support offered throughout the crisis event will be explored.

1.2 Helping Professions

Thinking more generally of helping professions may provide some insight into the roles and goals of these providers in a crisis situation since little information is available that addresses these elements in the genetic counseling literature. A helping profession is one that nurtures or addresses the problems of a person's well-being, whether these problems are described as physical, psychological, emotional, or spiritual. Much of this helping involves the development of a relationship between the helper and those in need of help (Skovholt, Yoo, & Hall, 1999).

Making a positive difference... it is the joy of practice, which results from the close connection to other people and the opportunity to help individuals in need. Successful helping can produce a sense of satisfaction and a joy of practice. It can be great work. But to do this well, we must constantly attach and separate successfully, over and over again, with person after person, (Skovholt et al., 1999, p. 49).

Being able to form professional attachments to people in need is essential in a helping profession, as is being able to then subsequently detach when the role is complete. At the core of making these attachments, the most crucial element, is the ability to care (Skovholt, 2005). At one end of the spectrum, these attachments

may be short lived, the task may be simple and the goal may be met with both speed and efficiency. At the other end, the relationship may be longer, the task may be laced with challenges for the client, challenges that require more from the client and the professional. “The idea at this phase is to share a vision together and work toward that,” (Skovholt, 2005, p. 88).

Skovholt described the value and challenges of working in a helping profession, which can be extrapolated and applied in the genetic counseling profession. Genetic counselors desire to provide useful genetic information, offer psychosocial support, and facilitate decision making are some of the “helping” qualities involved in the profession (Hartmann, Veach, MacFarlane, & LeRoy, 2015). Exploring and describing the nature of helping professions reveals the fact that people who belong to these professions tend to value the connections and relationships they can build with patients, or those they work with, as well as the benefits and challenges of forming such relationships.

1.3 Reciprocal-Engagement Model

The Reciprocal-Engagement Model (REM) of genetic counseling is a first step in describing our roles and goals as helping professionals. Historically, genetic counseling was thought to be described by two models: the “education” model and the “counseling” model. The education/teaching model focuses on the genetic counselors’ primary role as an educator and the counseling model describes the genetic counselors’ role as primarily related to meeting the psychosocial needs of the patient (Kessler, 1997).

Neither the education nor counseling model was thought to be sufficient because both did not fully appreciate the complexities of the genetic counselors’ actual role in the

patient's care. The scope of genetic counseling practice is challenging to define simply as it stretches beyond what is considered purely medical into something deeper, something that is sometimes intangible. The National Society of Genetic Counseling defines genetic counseling as the "process of helping understand and adapt to the medical, psychological and familial implications of genetic contributions to disease," (National Society of Genetic Counselors, 2005). At a meeting of North American genetic counseling program directors, a new model of genetic counseling was developed that was believed to better encompass all the roles of a genetic counselor. This model was called the Reciprocal-Engagement Model (REM). As opposed to the former models, the REM incorporates both education and counseling, and how, in fact, the two are used together in a session and throughout the course of the relationship (Hartmann et al., 2015).

This model was recently proposed as a model for the genetic counseling profession. The REM is composed of five tenets with each tenet having a mutual influence on the other; yet, the counselor-patient relationship is central to how they interact (Veach, Bartels, & Leroy, 2007). This tenet states that the *Relationship is Integral to the Genetic Counseling Process*, which emphasizes the importance of the relationship between the counselor and patient. It reflects the belief that an alliance between counselor and patient is the most effective way to address the psychosocial, medical, and emotional concerns that the patient has when receiving genetic counseling (Veach, LeRoy, & Bartels, 2003). When genetic counseling services are being provided, a connection between counselor and patient is essential and has a large impact on the outcomes of the experience. "In other words, the quality of the genetic counselor-patient

relationship is as important to genetic counseling outcome as the genetic information provided,” (Veach et al., 2007, p. 721).

In Genetic Counseling Practice, the model is composed of five tenets that are defined and described below by Gaff & Bylund (2010):

1. *Genetic information is key.* Providing information about perceived or known genetic contributions to disease and engaging in discussion with patients about this information is a particularly unique aspect of genetic counseling.
2. *Relationship is integral to genetic counseling.* The quality of the relationship developed between genetic counselor and patient is just as important as genetic information. Genetic counseling is a relationally-based helping activity whose outcomes are only as good as the connection established between the counselor and patient.
3. *Patient autonomy must be supported.* Patients should be as self-directed as possible regarding genetic counseling decisions. The counselor is an active participant, working with the patient’s individual characteristics and family and cultural context to facilitate informed decisions. An essential aspect of this tenet is that “*the patient knows best.*”
4. *Patients are resilient.* Most patients have the strength to deal with painful situations. Genetic counselors therefore, encourage patients to draw upon their inner resources (coping strategies) and support systems

and resources to make decisions and arrive at an acceptance of their situation.

5. *Patient emotions make a difference.* Patients experience a multitude of emotions that are relevant to genetic counseling processes and outcomes, for instance, affecting their desire for information, their comprehension of information, the impact of information on their decisions, their willingness and ability to connect with the counselor, their desire for autonomy, and their perceived resilience, (p. 185).

Each tenet has a mutual influence on the other with the counselor-client relationship being the conduit for how they interact (LeRoy et al., 2010). Here, again, the relationship is emphasized. If the relationship is the tool used to connect and enhance the integration of all other tenets in the model, its importance cannot be curtailed. “This tenet reflects a belief that a patient’s genetic concerns are best addressed when the counselor and patient form a strong alliance...” (Veach et al., 2007, p. 721). They form this alliance in order to be able to fully address all the needs of the patient including those that are psychosocial, medical, and educational. “This tenet further reflects a belief that people need connection, especially in times of distress. Genetic counseling therefore is a relation-ally based helping activity whose outcomes are only as good as the connection established between the counselor and patient” (Veach et al., 2007, p. 721). This means, simply, that the relationship formed between the genetic counselor and the patient is just as critical to the outcome as the educational information itself (Veach et al., 2007, p. 721).

1.4 The Relationship

The act of relating to patients (or forming a reciprocal relationship) is a major component of helping professions, like genetic counseling (Skovholt, 2005). Described in the nursing profession by Pierson in 1999, the relationship between the patient and professional is an active relationship in which a path is formed that allows for the professional to know and participate in the experience of others. Pierson expresses that the relationship involves constant interaction: “Understanding the meaning of situations is achieved through a continuing pattern of verbal and non-verbal interactions with others” (Pierson, 1999, p. 300). When a patient and genetic counselor interact in counseling sessions, the patient does not simply receive what the genetic counselor provides, but a dynamic encounter unfolds that involves both sides gaining something through the experience, whether it be knowledge, insight, or simply greater appreciation for others’ experiences (Evans, Bergum, Bamforth, Macphail, 2004). In this case, rather “than [counseling] being a prescriptive activity, counseling is informative, supportive and enables counselors to assist people who are making life changing decisions” (Evans et al., 2004, p. 463). In these sessions the process of communication involves learning about each other so that they, together, structure the session (Evans et al., 2004).

The relationship formed between counselor and client is integral to success when working in a helping profession—a positive relationship contributes to a positive outcome for both the helped (client or patient) and the helper (therapist or counselor) (Jackson, Dimmock, Taylor, & Hagger, 2012). Research in the field of psychology and rehabilitation also reveals that a negative relationship actually contributed to slower progress outcomes, demonstrating the importance of the relationship between the two

individuals (Kivlighan, Gelso, Ain, Hummel, & Markin, 2014; Jackson et al., 2012). Sexton & Whiston (1994) even went as far to describe that the quality of the relationship has the most significant impact on a successful outcome for the patient. Because these relationships are so important, it is necessary to delve into and subsequently understand the process of its creation.

There are several concepts that can be used to enhance our understanding of how these relationships are developed and maintained. Two of these concepts are the “working/therapeutic alliance” and “relational ethics," which have attempted to explore elements of these relationships.

Research in the therapy and counseling fields have explained that part of what promotes development of the relationship between genetic counselor and patient is a “working” or “therapeutic” alliance, a concept commonly applied to counseling and psychotherapeutic professions. The working alliance is composed of three parts: goals, tasks, and bonds that are collaborated on and established between the counselor and patient (Shick Tryon, Collins Blackwell, & Felleman Hammel, 2007). “The working alliance is a positive, collaborative relationship between patient and therapist and is composed of the relational bond, agreement on the tasks of therapy (in-session activities), and agreement on goals (targets of intervention, objectives),” (Busseri & Tyler, 2004, p. 78). This alliance is influenced by many factors such as attachment, counselor experience, length of therapy, personality, etc. (Taber, Leibert, & Agaskar, 2011; Bachelor, Meunier, Laverdière, & Gamache, 2010; Shick et al., 2007). Without a positive working alliance that involves a great deal of collaboration on the part of the patient and counselor, a positive outcome will be more difficult to achieve (Taber et al., 2011).

Research has exposed some of the attributes and behaviors that contribute to a positive therapeutic alliance; the specifics will be explored in more detail in the discussion about the individuals involved in these relationships.

The second concept that can be applied to the patient and genetic counselor relationship is “Relational Ethics.” This concept is described as an action ethic with focused interest being on the relationship between people. This focus requires that interactions move beyond objective and rational interests to encompass “emotion, imagination, memory, language, the body, and even other selves” (Gadow, 1999, p. 62). Relational ethics “implies that it is within human-to-human relationships that the origin of ethical commitment to self and other is expressed,” (Gadow, 1999, p. 63). This ethical commitment likely translates into an investment, from both parties, in both the outcome of the service and the outcome of the relationship. This approach emphasizes a focus on the relationship shared between two people as well as the knowledge that is transferred. “In this way, instead of being a unilinear process, from a relational perspective, genetic counseling is reciprocal; both the counselor and the client learn, grow and change through the experience,” (Evans et al., 2004, p. 463). The reciprocal relationship encourages both genetic counselor and patient to learn from each other and promotes the development of a relationship that will impact both of them. When a client and counselor interact in the counseling sessions the focus from this perspective insists that the encounter is not only what the counselor is giving the patient and what the patient is receiving, it is a dynamic interaction in which the relationship unfolds. In these sessions the process of communication involves learning about each other so that they, together, structure the session.

In 2004, a qualitative study of six adult genetic counseling patients undergoing predictive genetic testing were asked about relevant aspects of the genetic counselor-patient relationship; the elements of relational ethics were revealed to be engagement, dialogue and presence, which work together to create an evolving and fluid relationship. Engagement is the counselor's responsiveness to the patient, as well as the patient's responsiveness to the counselor. This relationship is often grounded in "ambiguity, uncertainty, openness, trust and respect," (Evans et al., 2004, p. 465). "Dialogue [rapport] is seen as the initial step towards developing a connection between a client and counselor," (Evans et al., 2004, p. 466). This connection begins as purely information being passed back and forth between the counselor and the patient as they become acquainted with each other. During this process they listen, hear, and share in order to establish this dialogue; to accomplish knowing each other in a relationship, you must be open to listening to the other person (Evans et al., 2004; Leach, 2005). There must be an atmosphere of openness in order to accomplish "presence," or the sense of being present as a whole person who is committed to forming a relationship. To build this relationship, it requires treating each other as a whole person and not merely as a function (Evans et al., 2004, p. 468; Skovholt, 2005, p. 86). Presence is the way (an atmosphere) in which a person's individuality (not merely their role or expertise) is shared," (Evans et al., 2004, p. 468).

1.5 The Genetic Counselor

Genetic counselor specific strategies or behaviors that may promote development of a relationship include active listening, recognizing the impact of genetic information on the patient's lives, ethical behavior and maintaining objectivity and boundaries (Veach

et al., 2007). In a 1999 study of former genetic counseling patients, three behaviors or strategies were identified by patients that were performed by the counselor during the genetic counseling session, which included (1) listening adequately to patient concerns, (2) answering patient questions, and (3) offering support. Support was defined in different ways, including answering questions, taking time with them, being “understanding,” providing encouragement, and making referrals to support groups (Veach, Truesdell, LeRoy, & Bartels, 1999). Behaviors used to establish a bond with the patient may include “sitting quietly, reflect patient thoughts and feelings, summarize patient statements, rephrase, and use similar body language (Veach et al., 2007).

It is clear that therapist characteristics are an essential aspect of developing the genetic counselor-patient relationship. In the realm of cancer treatment, an editorial was written that discusses some of the attributes of an outstanding health professional, which include being compassionate, empathic, respectful, genuine, and trustworthy, being present fully, and valuing the patient. Some strategies of genetic counselors include being gentle, having good judgment, not rushing, but instead, remaining attentive to the patient and adjusting immediately to cues (Schapira, 2013).

Research of the therapeutic alliance reveals certain characteristics and strategies described through this research include dependability, the ability to instill confidence and trust, and the therapist’s ability to connect and convey an adequate level of competence to effectively help patients under distress. The therapist displaying obvious benevolence and being responsive are related directly to the development and maintenance. It is also incredibly important that the therapist feel confident in his/her ability to help patients. Communication with the patient should be clear and coherent laced with an overt

supportive stance, sensitivity toward the patient’s feelings and being empathic, (Ackerman & Hilsenroth, 2003; Castonguay, Constantin, & Grosse Holtforth, 2006). Ackerman’s meta-analysis allowed him to generate a summary table of attributes and techniques used by therapists that result in a positive alliance. This table is shown in Table 1.1.

Table 1.1 Summary of therapist’s attributes and techniques found to contribute positively to the alliance	
Personal Attributes	Technique
Flexible	Exploration
Experienced	Depth
Honest	Reflection
Respectful	Supportive
Trustworthy	Notes past therapy success
Confident	Accurate interpretation
Interested	Facilitated expression of affect
Alert	Active
Friendly	Affirming
Warm	Understanding
Open	Attends to patient’s experience

The genetic counselor has two main goals in a session and those are to meet the education and psychosocial needs of the patient (Biesecker, 2001). Professional goals that relate to the relationship that occur in the session include “(1) the genetic counselor and patient establish a strong working relationship; (2) the genetic counselor and patient engage in good communication with each other; and (3) the counselor knows how to intervene in order to build rapport and foster communication.” (Veatch et al., 2007, p. 721). Out-of-session goals aimed at the relationship are patient follow-up, which refers to “maintaining a relationship with patients beyond the counseling session, specifically, making oneself available for further contact,” (Hartman et al., 2015, p. 231).

1.6 The Patient

Patient contributions promote development and maintenance of the relationship as well. Levels of anxiety increase when a genetic diagnosis is made or an anomaly is found. Patients have to deal with not only the stress of becoming a parent, but also the new stress of a diagnosis (Fonseca, Nazaré, & Canavarro, 2012). When it comes to reactions to the diagnosis, levels of information seeking vary greatly among parents. Regulation of the level of information sought is an attempt to cope for most patients. In a longitudinal study of 42 women following a prenatal diagnosis, Lalor et al. (2008) revealed that mothers will display one of two types of information seeking following a prenatal fetal anomaly diagnosis; these types are termed ‘monitoring’ and ‘blunting.’ Monitoring is defined as having high information needs and preferring to have as much information as is available. Blunting is defined as the preference to avoid the information or the mindset of “I’ll cross that bridge when I come to it,” (Lalor et al., 2008).

When services are being provided, the needs of the patient include (1) the need for certainty, (2) the personal relationship between patient and counselor, and (3) the integration of lay and scientific explanations (Berkenstadt, Shiloh, Barkai, Katznelson & Goldman, 1999). Each need was essential to the outcome related to psychological adaptation to the genetic condition. The need for certainty is often the desire to regain control of the situation, a concept called perceived personal control (Berkenstadt et al., 1999). A positive relationship between counselor and patient was preferable to the patient because of how it made the patient feel about the situation. A study by Shiloh and Berkenstadt in 1992 of former genetic counseling patients and their families emphasized the importance of effective communication between patient and counselor. The cases in

this study appear to confirm that effective communication at least partly depends upon the patient believing that the counselor is interested in their personal circumstances (Skirton, 2001). Strong sensitivity to the patient's need to "be family" or seek a deeper connection with the counselor, or at least to "be friends" (Bailey, 1988) is a common element of the relationship.

Research related to the therapeutic alliance identified four elements of the patient experience from his/her perspective that have been found repeatedly to be associated with the therapeutic alliance. Depending on these elements patient satisfaction with the alliance can range from negative to positive. These elements are 1) patient involvement, 2) how comfortable the patient is with the therapist, 3) how much progress the patient makes throughout the course of the relationship (e.g., reaching new insights), and 4) the patient "real" relationship (how much the patient likes that therapist and how the therapist is perceived through the patient lens). (Eugster & Wampold, 1996).

1.7 Psychosocial Support

One of the major components of genetic counseling is the offering of psychosocial support (Veach et al., 2007). Psychosocial support aims at easing resumption of normalcy and to prevent negative consequences of potentially traumatic situations (Ager, Fiddian-Qasmiyeh, & Ager, 2015). Psychosocial support in the context of genetic counseling was characterized by the encouragement that they received and the attempts on the behalf of the genetic counselor to make the patient feel better. Immediate psychosocial support commonly occurs during the crisis event, such as a receiving a prenatal diagnosis. Genetic counselors felt that "listening to and validating concerns,

empowering the patient, and providing supportive counseling with regard to decision making,” were ways in which they offered psychosocial support to patients (Bernhardt, Biesecker, & Mastromarino, 2000). Patients also spoke commonly of the encouragement they receive during the genetic counseling visit (Bernhardt et al., 2000).

Long-term psychosocial support occurs after the crisis event. “Genetic counselors may initiate a process that continues beyond the genetic counseling session,” (Hartmann et al., 2015, p. 234). “Many genetic counseling patients value their genetic counseling because they have been able to have an on-going supportive relationship with their genetic counselor,” (Bernhardt et al., 2000). This relationship may persist through telephone or letter contacts rather than through follow-up appointments (Bernhardt et al., 2000).

In regard to genetic counseling practices, the literature offers sufficient examples of aspects of psychosocial support which influence patient satisfaction (Bernhardt et al., 2000; Moore et al., 2012; Skirton, 2001; & Veach et al., 1999), as well as perspectives of genetic counselors related to goals and approaches to achieve success (Fox, Weil, & Resta, 2007; Hartmann et al., 2015; & Veach et al., 2007).

While literature exists in other helping professions regarding provider-patient relationships (Skovholt et al., 1999; Taber., 2011; Bachelor., 2010; Shick et al., 2007; Busseri & Tyler 2004; & Pierson, 1999) and the importance of the genetic counseling relationship is alluded to in regards to characteristics of the genetic counselor and patient (Veach et al., 2007; Esterling, L’Abate, Murray, & Pennebaker, 1999; Veach et al., 1999; Lalor et al., 2008; Skirkton, 2001; Bailey, 1988; & Hartmann et al., 2015), as well

as psychosocial support (Bernhardt et al., 2000; Moore et al.; 2012; Skirton et al., 2001; & Veach et al., 1999), the patient-genetic counselor relationship has not been fully and exclusively explored. This study aims to describe this relationship that forms between patient and genetic counselor patients who received a potentially lethal prenatal diagnosis resulting in a major loss via pregnancy termination, miscarriage, stillbirth or neonatal death.

Chapter 2. Manuscript

The Genetic Counselor-Patient Relationship Following a Life-limiting Prenatal Diagnosis: An Exploration of the Reciprocal-Engagement Model¹

¹ Anderson, S., Edwards, J., Berrier, K. L., & Redlinger-Grosse, K. To be submitted to *Journal of Genetic Counseling* 2015

2.1 Abstract

The proposed Reciprocal-Engagement Model of genetic counseling is comprised of five core tenets with the relationship between the genetic counselor and patient being central to the process and success of genetic counseling. This study sought to explore the relationship between the genetic counselor and patient during and after a prenatal diagnosis of a “life-limiting” diagnosis that resulted in a major loss (termination, stillbirth/miscarriage, or neonatal death). Eight genetic counselor and patient pairs were individually/separately interviewed about their mutual relationship and asked about the development and maintenance of the relationship, the contributing factors, and the long-term effects on both the genetic counselor and patient. The length of these relationships ranged from four months to fourteen years following their initiation. All clinical relationships established required extensive follow-up via multiple modes of communication with more frequent communication in the beginning and less as the relationship evolved with more equal initiation of communication over time. The content of conversation expanded to include more personal elements from both the genetic counselor and the patient perspectives. Most participant pairs fell somewhere on a spectrum of professional relationship to deeply personal relationship as one pair maintained an exclusively personal relationship. The support offered by the genetic counselor during the time of crisis was both essential and unique to the patient compared to other healthcare providers and family/friends. Strategies employed and/or characteristics of the genetic counselor and patient did contribute to the development and maintenance of the relationship as did the life-limiting nature of the diagnosis, which was thought to overall strengthen the connection. The long-term effects on participants reveal

clinical implications for genetic counseling. This exploratory study highlights the unique service of support offered by genetic counselors, as well as potential avenues for future research and training implications.

2.2 Introduction

The Reciprocal-Engagement Model (REM) has been put forth as a model of practice for genetic counseling (Veach et al., 2007). This model has been empirically investigated in only one study (Hartmann et al., 2015) and thus additional validation is needed. The REM is composed of five tenets that can be used to describe the genetic counseling session, including goals, behaviors, and elements considered to be important to the success of the counseling session (Hartmann et al., 2015). The current study explored the value of the second tenet: Relationship is Integral to the Genetic Counseling Process. This tenet is considered to be the conduit through which all other tenets interact and is therefore essential to the genetic counseling process and its success (Veach et al., 2003). The relationship between genetic counselor and patient depends on both parties as each contributes to the development and maintenance of the relationship. Despite its importance, few studies in genetic counseling have explored the nature of this relationship. The purpose of this study was to describe the relationship developed and maintained between genetic counselors and prenatal patients whose relationship was centered on a crisis event, which in this study was defined as a potentially lethal/life-limiting prenatal diagnosis (e.g., chromosome aneuploidies, major fetal anomalies, life-limiting single-gene disorders, etc.) resulting in a major loss via pregnancy termination, miscarriage, stillbirth or neonatal death. The specific aims of this study were: 1) Understand and describe aspects of the genetic counselor-patient relationship in the

context of a potentially lethal or life-limiting prenatal diagnosis, 2) Identify characteristics and actions of the genetic counselor that influenced the relationship, and 3) Identify characteristics and actions of the patient that influenced the relationship.

2.3 Materials & Methods

2.3.1 Participants & Recruitment. Eligible genetic counselor participants included formerly or currently practicing prenatal genetic counselors who had maintained a relationship with a former prenatal patient beyond the prenatal diagnosis that resulted in a major loss via pregnancy termination, miscarriage, stillbirth or neonatal death, and whose respective patient was willing to participate. Genetic counselors were invited to participate through a study advertisement posted on the National Society of Genetic Counselors (NSGC) email mailing list and the NSGC Prenatal Special Interest Group (SIG) discussion forum. Eligible patients were identified by their respective genetic counselor and invited to complete a web-based survey disclosing contact information to the principal investigator. Only English-speaking participants were included in this study as interpretation from English to other languages was not provided due to limited resources

2.2.2 Study Method. Ethical approval for this study was sought and obtained from the University of South Carolina Institutional Review Board. An amendment to the proposal was made to expand eligibility from relationships established after September 2011 to relationships established any time in the past to gain additional participants.

Semi-structured interviews were conducted with eight genetic counselor-patient pairs (eight genetic counselors and at least eight former prenatal patients-female) in the prenatal setting who developed a relationship in the past and had maintained this

relationship beyond the crisis event (point of diagnosis). Interested participants were asked to complete a short web-based questionnaire to establish eligibility. The study questionnaires were created, collected and managed using Qualtrics—a secure, web-based application. Eligible genetic counselor participants were contacted by telephone or email directly by the principal investigator and invited to share with their patient a separate specific web-based questionnaire via email, which collected personal patient health information including prenatal diagnosis and personal contact information. The principal investigator contacted patient participants and scheduled telephone interviews. Following the completion of the patient interview, the principal investigator arranged a scheduled 60 minute telephone interview with the genetic counselor participant.

Genetic counselor and former patient participants were asked similar or parallel questions from an interview guide that was piloted with a genetic counselor and patient pair. Minor changes to the interview guide were made following this pilot interview to include necessary follow-up questions. The interviews were conducted over the telephone during a scheduled time based upon the participant's preference and availability. Participants could choose not to answer a question or to end their participation in the study at any time without penalty. Aside from participants' voices and demographic information, no other personal identifying information was collected. The interviews were audiotaped and transcribed verbatim. Recordings were scheduled to be destroyed upon transcription (within six months of the study interview).

2.2.3 Data Analysis. A qualitative methodology was used to explore the relationship between genetic counselor and patient during and after the prenatal diagnosis. Thematic analysis methods were used for systematic coding and identification of emergent themes

(Hsieh & Shannon 2005; Bradley, Curry, & Devers, 2007). Transcriptions were reviewed by the co-investigators to establish a code list. Data was independently coded by the principal investigator (SA), with regular inter-coder consistency checks and discussion of discrepancies with the co-investigator (KB).

2.2.4 Study Content

2.2.4.1 Web-based questionnaires. The web-based questionnaire specific to the genetic counselors asked four questions to determine eligibility. To be considered eligible, genetic counselor participants had to answer 1) “Have you maintained a connection with a patient following the provision of prenatal genetic counseling for a potentially lethal diagnosis that resulted in a major loss via pregnancy termination, miscarriage, stillbirth or neonatal death?

Yes/ No,” 2) “When was this relationship formed (mm/yyyy),” 3) “Would you be willing to contact this patient and invite their participation in this study for a 60 minute interview? Yes/ No,” and “Are you also willing to complete a separate telephone interview lasting up to 60 minutes? Yes/ No.” Skip logic was embedded in the questionnaire in which a “no” or failure to answer any of the above questions would immediately dismiss them from the questionnaire. If all of the above questions were answered with a “yes,” the genetic counselor participant would be prompted to disclose his/her contact information.

The web-based questionnaire specific to the patient participant asked three questions to determine eligibility. To be considered eligible, patient participants were asked 1) “What was the prenatal diagnosis in your pregnancy?_____,” 2) “What was the pregnancy outcome?” with the options “Pregnancy Termination, Spontaneous

Abortion/Miscarriage, Stillbirth, Infant Death, Other _____,” and 3) would he/she be willing to participate in a 60 minute interview, Yes or no. If the patient participant selected “yes,” then he/she would be prompted to disclose his/her contact information.

2.2.4.2 Interview Guides. The patient and genetic counselor interview guides consisted of seven identical general questions addressing aspects of the development, maintenance, and factors contributing to the relationship as well as demographic information gathered following the conclusion of the interview. A checklist of topics needing to be discussed was generated to create follow-up questions specific to participant type (genetic counselor or patient) for each general question. The specific questions asked were 1) how did you meet (patient/genetic counselor): how was your relationship developed, how has it been maintained, 2) what about the circumstances of the prenatal diagnosis encouraged the relationship, 3) what are the specific characteristics about the patient that encouraged the relationship, 4) what are the specific characteristics about the genetic counselor that encouraged the relationship, 5) what are specific actions from the genetic counselor that strengthened the relationship, 6) what made the genetic counselor unique compared to other providers, family, and friends, and 7) how has the relationship influenced the genetic counselor/patient long-term? Consistent prompts were used for each question.

2.4 Results

2.4.1 Participants. A total of 35 genetic counselors started the web-based questionnaire (Tables 2.1 and 2.2). Eleven of those questionnaires were not completed. A total of nine genetic counselors agreed to contact their former patient to participate in this study. Due to cessation of contact from one genetic counselor participant, eight total pairs were interviewed. All genetic counselor and patient participants were female. Demographics of the genetic counselor participants are shown in Table 2.1.

Genetic Counselor	1	2	3	4	5	6	7	8
Age	33	38	27	25	50	64	33	52
Race/Ethnicity	Caucasian	Caucasian	Caucasian	Caucasian/Middle Eastern	Caucasian	Caucasian	Caucasian	Caucasian
Marital Status	Married	Married	Married	Single	Married	Married	Married	Married
#of living children	0	2	0	0	2	2	1	2
Years of Practice	3.5	13.5	2.5	1.5	25	7	5	28
Years in Prenatal	3.5	10	2.5	1.5	20	5	5	28
Location	Tennessee	Oregon	Florida	Alabama	Maine	South Carolina	South Carolina	South Carolina
Length of Relationship ¹	1y 7m (05/2013)	1y 8m (06/2013)	2y 1 m (11/2012)	4m (08/2014)	2y 1m (11/2012)	4y 5m (07/2010)	4y (2011)	14y (2001)

The ages of the genetic counselor participants ranged from 25 to 64 years old, with a median age of 33, and clinical experience ranged from 1.5 to 28 years, with the median years of practice being approximately 6 years. All of the genetic counseling participants were of Caucasian ancestry, one of whom also had Middle Eastern ethnicity. Most of the genetic counselors were married. Of note, one genetic counselor disclosed during the telephone interview that she experienced a major loss comparable to the losses explored in our patient participants. Geographically, genetic counselors practiced in six states of the United States. The length of the relationship maintained between counselor and patient ranged from four months to fourteen years.

Table 2.2. Patient participants								
Patient	1	2	3	4	5	6	7	8
Age	38	39	28	27	29	37	38	35
Race/Ethnicity	Caucasian	Caucasian	Caucasian	Caucasian	Thai	Caucasian	Caucasian	Caucasian
Marital status	Married	Married	Married	Married	Engaged	Married	Married	Married
Total # of pregnancies (disclosed)	4	3	2	1	3	3	2	14
# of (living) Children	2	2	1	0	1	2	1	4
Highest level of Education	Masters	Some college	Bachelors	Associates	Bachelors	Bachelors	Associates	Bachelors
Indication for referral	Abnormal FTS	Abnormal anatomy scan	Abnormal anatomy scan	Abnormal anatomy scan	Abnormal FTS	Abnormal MMS	Abnormal FTS	After loss
Prenatal Diagnosis Pregnancy outcome	Trisomy 21 plus cystic hygroma & CHD	Trisomy 18	Prune Belly syndrome plus VSD	CDH+	Meckel Gruber Syndrome	Smith-Lemli-Opitz	Trisomy 18	Surfactant protein B deficiency
Pregnancy Outcome	Termination	Stillbirth	Neonatal Death	Termination	Termination	Neonatal Death	Stillbirth	Neonatal Death

Participant demographics are shown in Table 2.2. Seven of eight of the patients were of Caucasian ancestry and one was of Thai ancestry. All patient participants were in a long-term romantic relationship. Most had children. All patients had some level of higher education beyond a high school degree. The patients were referred to genetic counseling following abnormal first trimester screens (FTS), multiple marker screen (MMS) screens, abnormal anatomy scans, or after a major loss defined as a termination, miscarriage, stillbirth or neonatal death. The pregnancy outcomes included medical terminations, stillbirths, and neonatal deaths. Of the eight, five were natural losses. The specific prenatal diagnoses were Trisomy plus a large cystic hygroma and congenital heart defect (CHD), two trisomy 18 diagnoses, prune belly syndrome plus a ventricular septal defect, congenital diaphragmatic hernia plus other anomalies (CDH+), Meckel Gruber syndrome, Smith-Lemli-Opitz, and surfactant protein B deficiency.

2.4.2 Communication. In exploring the relationship, several communication strategies emerged related to characterizing the relationship and how it changed over time. The first question, “how did you meet,” prompted the participant to elaborate on the series of events. They were encouraged to describe the communication (mode, frequency, content, and who initiated), the evolution of the support provided by the genetic counselor, and the evolution of the relationship from those first clinical encounters to present day.

The mode of communication established during the start of the relationship included in person, phone, email, and cell phone (as needed). This expanded to also include Facebook and letters.

“On Facebook she'll comment on my pictures of new baby boy, or something that I post on Facebook. Then it makes me think of her, and I think, oh I should call [genetic counselor] and see how she is.”-Patient.

Even if the baby was unaffected, she would call and make sure I was doing okay, she would send me a card, she always sent me cards for losses and for healthy babies. So she was always very good about keeping in contact and checking up on me. -Patient.

At the start of the relationship, the frequency of communication was daily, weekly, monthly and unknown.

“So the emails gradually became shorter and shorter because we were literally writing each other books probably the first two weeks...”-Patient.

At the present, or more recently, the frequency of communication included weekly, monthly, and a few times a year.

We don't call each other all the time, but I do email her every now and then, like over the holidays or when she pops in my mind, or I work in the medical field too and so when something would just hit me, I would email. -Patient.

“I think a couple times a year we would think of each other and send each other a letter or give each other or call.”-Patient.

Other frequencies included annually, varies and during the subsequent pregnancy for clinical management.

“I talked to her after they came back and just maintained a relationship with her. When she got pregnant again, she called me... Like right away.”-Genetic Counselor.

“When she was pregnant again, I was the second person that she told, her husband was first and I was second and then she told her mother and sister.”- Genetic Counselor.

“Every time that I got pregnant, she would be the second person that I called, yeah it would be my husband and then her”- Patient.

Much of the content in the beginning of the relationship revolved around the medical and educational aspects of the situation as well as socially “checking-in”, which was mostly superficial. Fewer pairs indicated having more in-depth, emotional or faith-based conversations, and minimally included the personal disclosure by the genetic counselor.

I told her that I've lost a baby, a number of years ago, for very different reasons, but I know about that on a very personal level, and I think she sort of looked at me like a survivor. I was able to help her by saying that you will be able to go on and have a life, and live a good life. I had to say, I have two kids, I had to get brave and be willing to try again after we lost our first child. -Genetic Counselor.

Since the establishment of the clinical relationship, the content of conversation evolved to include more content about the personal life of the genetic counselor.

So we've kept in touch, I sent her pictures of my kids and when I think of her I give her a call and then we just talk about each other's lives, how her family is, how my family is, how work is going. She also had one of her students interview me in my home, so I would frequently ask about her.

Once the pregnancies were out of the way, it was just more of a social interaction. -Patient.

The initiation of communication also evolved from predominantly patient-initiated to both genetic counselor and patient initiated contact.

2.4.3 Support offered during the development of the relationship. The support provided by the genetic counselor during and after the clinical relationship evolved with some features being present throughout the course of the relationship and others being present only during the crisis. Three main categories of support emerged: 1) the role of education or information-giving, 2) the professional aspects exhibited by the genetic counselor, and 3) the psychosocial elements addressed throughout.

The importance of education and information was critical for all the patients and genetic counselors. Patients and counselors said that education and information was useful for preparing the patient and facilitated decision-making.

I coached her about some of the really difficult questions that she would be asked after the baby was born. What do you want done with the baby's body after she passes, you can have a memorial service, you can have a burial, it's your choice, and here is some information. I gave her lots of information. For some patients this is relevant so please make the most of it or call me with specific questions and I will try to find out, if it is not relevant to you for some reason, then you can just put it to the side and don't think about it. One of her main concerns was how to tell her daughter, her step-son too but he was older, but her daughter was pretty young, five or six; how do you tell your daughter that her baby sister is not

going to come home from the hospital? We talked about that and I got some resources that may have been helpful. I don't know what she used and what she didn't but I made them available for her. -Genetic Counselor.

Patients mentioned specifically that the genetic counselor made the complex information understandable and most patients said the genetic counselor was available for any follow-up questions throughout the crisis experience.

The fact that she gave me her email address because I could usually just shoot her an email of an article saying hey, I read this one and it said this, what are your thoughts, have you seen this or have you not seen this. That was really helpful. -Patient.

Whenever I needed to reach her, she made herself available and we just didn't feel like we were left hanging in the wind," and, "She would give me answers that she knew, but she was available for the questions that we had as we went through the process and she was present at numerous appointments from that point going forward as well. -Patient.

I always let her know that I was there if she ever needed anything at all, it's just showing that the door is always open. Even if they had the baby for months and they just need someone to talk to, I always want my patients to know that just because our clinical contact ended doesn't mean I won't listen to them like if they don't have anyone to speak to or they just felt like they were more comfortable. I know others and genetic counseling people are very strict about patient contact, but in the world of fetal

anomalies where you see these families multiple times it makes it hard to, not necessarily get attached, but just keep it so cut and dry where we're only going to talk about the baby's anomaly. I wanted them to know that I understand that they're going through a very hard time in their life; there are not a lot of people that could just grasp what they're going through. - Genetic Counselor.

There were many professional elements displayed by the genetic counselor. Several patient participants said the genetic counselor acted as an advocate or liaison for the patient's medical needs, some commented on how the genetic counselor was non-directive throughout the decision-making process, many discussed the provision of resources for the family. Some patients emphasized that the genetic counselor made the patient feel like a priority and that she was both factual/complete but also sensitive in her delivery and discussion of information.

I think just in the beginning and what I ended up appreciating, and I don't know if I knew at the time what I needed but I ended up appreciating was the factual information [was provided] without it being delivered in a cold manner. I really appreciated feeling like we weren't just another patient. I think during these life altering situations with this life-altering diagnosis that we were making some very heavy decisions based on the information we were getting and I didn't feel like we were being treated as if "okay where is the next person in line?" I really appreciate that. On the other hand it wasn't sugar coated. They didn't withhold information from us, they totally explained everything. I think that's what I really appreciated

because in a short amount of time, I mean 20-30 minutes, someone can give you genetics 101 and tell you what you need to know and make sure that you understand that this could mean this or this could mean that. And made sure you're armed with the next step. That's a lot to do in a short amount of time. -Patient.

One patient commented that she felt like the genetic counselor was committed to taking this journey with the patient and her family, which provided her with a great sense of support.

"She was our only connection to that diagnosis and somebody who's going to be moving forward with us to help us understand what was going on in the beginning." – Patient.

A lot of times when it comes to pregnancy, people have questions, but I don't want them to come to this appointment thinking that they're just going to talk to me and that's it. I know that this is a partnership in trying to get through the rest of the pregnancy and it's not just an information session. -Genetic Counselor.

Both patient and genetic counseling participants mentioned many psychosocial support pieces to their relationship as it was first developing during the crisis. Most pairs mentioned that the genetic counselor was always willing to listen, and several talked about how the genetic counselor made a point to check-in with the patient.

"...over time just providing an active ear to listen to what they were going through."-Genetic Counselor.

The genetic counselor showed support during the time of crisis in ways that included making the patient feel comfortable or confident, showing sympathy, offering hope, validating feelings, normalizing feelings, displaying advanced empathy, contextualizing information, mirroring the hopes and positivity of the family, offering emotional support to family members, or reflecting patient emotions.

I did give her, in addition to, like advanced empathy, and just my more advanced genetic counseling skills in the sense that helping her to recognize, helping her do some metacognition as she was coping, and also addressing like, there was a lot of reflecting back the feelings that she is expressing to me. A lot of it was about the feeling she was having about the ambivalence of the decision and to acknowledge the feelings that yes, if this baby had Down syndrome but didn't have a large pocket of fluid attached to it, you probably would have different feelings, because she really struggled with the fact that Down syndrome itself does not bother her it was just this combination of Down syndrome and this abnormality. She thought like are we keeping this baby suffering.... she also, I use a lot of advanced empathy and normalizing for her because she really just wanted a healthy baby and she's 39 or whatever. It is okay to feel like this isn't going to work; it is okay to feel like I want to move on from it. -

Genetic Counselor.

Also, the genetic counselor allowed the family to talk about the loss without redirecting the conversation.

We are a family with a huge emotional thing that we are going through and to allow something like when she was meeting with us for nonmedical talk to share our hopes and the emotional side of what we were going through. Granted she is not a therapist, but there has to be space for that in those moments that are so heavy. In those moments that are so heavy there has to be room for people to exhale a little bit, to not be so tightly wound and all they can ask about is the omphalocele and run out the door, but to allow some space for the reality of what's actually happening to a family that is looking at the possible life of their child. That is a heavy big thing and to create space for what that means is so compassionate. I don't think everyone has a bedside manner that would do that but it is very important for the family. – Patient.

The genetic counselor encouraged the patient to self-advocate, alleviated feelings of guilt, provided physical presence after the loss, shared personal experiences of loss, and respected the family's connection to the pregnancy.

Also, they are emotionally attached to this pregnancy as if it is a baby girl, they lost a baby girl. They knew that the chromosome showed that it was XY, but they requested that the entire staff refer to the baby as a girl; she would say I know that it's XY but in our hearts that was the baby girl. So we respect that, and say 'girl.'-Genetic counselor.

The genetic counselor provided support that the patient could feel good about. *So to me, [genetic counselor] was the first step of our entire experience for having the groundwork for having a positive outcome, whether our*

child survived or not. Our family felt respected and so supported, for our daughter with her particular anomalies and her particular situation and what was right for us. That was kind of the biggest deal that she was a part of, because it really could have gone both ways, dramatically, it could've been either. - Patient.

2.4.4 Support versus Others: Other Healthcare Providers. Patients and genetic counselors were also asked about how the support offered by the genetic counselor differed from other providers or health professionals (Table 2.3). There were both professional and psychosocial differences mentioned. One of the most frequent comments made was that the genetic counselor was accessible and available for follow-up.

It's really simple, but we were more than just a listening ear, or like a counselor but I think the thing that I see unique about our position, you know, I have just doesn't go to the wayside with the changing landscape of our job, we spent a lot of time with patients and to be honest, and to be honest no one else is going to spend that much time with them, and they need that much time. So to me that's a unique situation. - Genetic Counselor.

Patients and counselors also mentioned that genetic counselors made the information understandable, acted as a liaison or advocate, offered resources, and helped guide the patient through the process.

“I think those families appreciate you walking through everything with them as opposed to you just giving them information and then walking away you're really making

an effort you're always there, which is important in such a serious time.”-Genetic Counselor.

They were unbiased and non-directive, committed to helping the patient, and had a unique skillset.

“I think we offer a unique skillset and we have enough medical knowledge peppered with the right amount of desire to feel the need to support people as an educator that I think is rare for physicians.”-Genetic Counselor.

The genetic counselor was also willing to have an active role between pregnancies, and listened to the patient.

Because I had to the time to sit with her and talk with her and listen to her, and to try to understand what the impact was of losing a child, trying to understand what her thinking was when she at first we decided she would not do any prenatal testing and then eventually change her mind, what a thought process was that made her change her mind to want to do prenatal testing. Then to decide that she would terminate a pregnancy, and then after that termination say I would never do it again, but then yet do it again with another pregnancy. It's just listening, I was able to listen to her, give her the chances to tell me where she's coming from, and again not to be judgmental but I know other people have said to her why do you keep on having children. I think I had a conversation with one of her doctors down where she was and they asked, 'Why does she keep getting pregnant? Why does she keep chancing it?' And I would say, '...because she wants a big family, she's always wanted a lot of children. You know,

like can't you understand it, it's that simple. That desire, that passion, it's that simple and that strong. '-Genetic Counselor.

The genetic counselor was able to build the relationship quickly during such a sensitive time and treated the patient like an informed autonomous person by showing respect.

There were several people on the team we had to convince that we understood that [the baby could die]. Because they think at first, maybe you only get 30 minutes in the meeting with the specialist and they start hearing you asking for these options and those options, it might be their inclination to go, hmmm does this family even know what they are dealing with, do they understand that only 5 to 10% survive the first year. Then we would say yes we do understand, but we do not want you coming out and doing your job on-the-fly if she survived. My only thing was that, because there are so many people in the medical profession who are refusing care for trisomy babies, we want to know if you're going to do this so we have time to find someone else if you won't. We wanted to make sure that we had team who were ready for what could happen for our daughter, what she might need, and are we going to be on the same page on going through with these treatments. We never had to convince counselor that we knew what we were getting into. We were treated by her as informed and intelligent parents asking for something that was valid. That was a very big deal; that was important. Unfortunately we did have to convince others on the team. -Patient.

The genetic counselor also helped interpret and contextualize the information and results.

Every time we got a result whether it was from the ultrasound or a blood test, she helped us interpret it and she gave us our options if you or just told us, this is what this test means, this is the next test you can do, this is the next decision you can make...where we never saw anyone else a second time in her office. - Patient.

The psychosocial support offered by the genetic counselors that was unique compared to other providers included the fact that the genetic counselor addressed the psychosocial aspects of the situation as well as the medical.

I don't know of any other medical professional that I've run into that sort of serves that advocacy, counselor, medical provider role because most of the other medical providers have that one hat that they wear and then you're on your own for the other items but she can give you advice about the medical side of things, she offered former patients for us to talk to who had terminated pregnancies and then those who kept pregnancies. She was willing to get in touch with other people that we could talk to, which was really interesting. It was more of a holistic approach to a medical diagnosis if you will, which I really appreciated because that kind of diagnosis is a multifaceted diagnosis. -Patient.

The genetic counselor empathized, acted as a source of comfort at other doctor's appointments, and was supportive of the patient's hopes.

[Genetic counselor] I would say though was supportive emotionally from the very, very beginning because she understood what we wanted, and once we said again what we wanted, there were some other people on the team that we really had to convince that we knew what we were looking at and that we were not just in denial that our baby might die, that moment had passed. There were several people on the team we have to convince that we understood that. -Patient.

One patient felt a lack of support from other providers.

To be routinely let down by so many different areas of the medical community because your child has a trisomy diagnosis, that is definitely impacting how many people feel like they had a negative experience. I certainly don't want you to think that, I have not tapped into genetic conditions as a whole. That is really specific to the trisomy community. They feel like once they get the diagnosis they are hung out to dry with very few options for people who want to actually support them. -Patient.

The genetic counselors delivered the information compassionately, and one patient mentioned she appreciated the female “comradery” between her and her genetic counselor.

2.4.5 Support versus Others: Family and Friends. Patients received support from family, friends, faith-based origins, and support groups, but to understand what made the support offered by the genetic counselor different, both patients and genetic counselors were specifically asked how the support offered by the genetic counselor was unique (Table 2.3). The support that was discussed could be divided into medical and

psychosocial. Medical support was support that could only be gained due to the medical training of the counselor. One major difference between the support of family or friends and genetic counselors was that the genetic counselor was medically educated.

Well I guess it was just that it was someone that I had not been connected to prior to being pregnant. There were a bunch of people who were supportive of us when I was pregnant like friends and family, it was just that she knew exactly what was going on with the pregnancy, she knew all the scientific side of it, but with everybody else they didn't really know what the condition was or anything, it was more of a professional opinion that I could get as opposed to just a shoulder to cry on with friends and family. -Patient.

The genetic counselor knew the details of what was going on, could explain potential outcomes for the patient, and could fully appreciate the gravity of the situation when family and friends just wanted to stay positive.

I mean obviously they have a lot of support in people being able to listen to their story and help them go through the emotions, but I think it helped them having someone who can listen but also understand exactly what we were dealing with in a realistic way. I know there was a point where, from my perspective, they knew they were up against very bad risks or a bad prognosis when he was born, and I think a lot of those situations people want to stay positive but I know there was a point where they just didn't want to see him suffer anymore, and they had been through so many other... They had a better understanding. I think it helped having someone

listen but also understand the medical side of it and why it was just so serious. -Genetic Counselor.

The genetic counselors were also able to anticipate patient needs due to professional experience and training.

I'd been around that enough to know that I could anticipate that it would be hard for her in terms of coming back to our office; you know the PTSD thing that people get when they have to come into our office after another hard pregnancy. - Genetic Counselor.

A psychosocial difference between family and friends and genetic counselors was that the genetic counselor seemed more comfortable talking about the affected pregnancy and potential loss.

The sad thing to me is that you can literally google what to do in that situation and get that information in an instant. But they still say, 'I didn't know what to say, I didn't know what to do.' That is what is really sad. -

Patient.

The genetic counselor was non-judgmental, she focused exclusively on the patient, was neutral, objective, and she did not have the same invested “needs” as family and friends did.

I think it differs because, um, it comes from a different place; I'm a neutral person who is not asking anything of the patient other than to take care of herself and her needs. Where family members might be coming from a place where, I mean of course her mother was upset, I mean this was her daughter she was seeing go through a very hard time, and this was her

granddaughter she would only get to know for a very short time so the grandmother had needs of her own, the husband had needs of his own. He, like other husbands wanted to fix it, but there was no way to fix it and so different family members and even friends have their own needs and need to be helpful and she didn't need help or she needed a certain type of help, or they felt the need to say something when she would rather have silence and I came from a neutral place where I am not living with her or around her so the needs that I talked about were always her needs, they were not her husband's needs, these are not your mothers needs, I want you to focus on your needs so you can be healthy on the other side of this.-

Genetic Counselor.

Table 2.3 Genetic counselor support offered versus others: friends and family and other healthcare providers	
Versus Other healthcare providers	Versus Family and Friends
<i>Professional</i> Offered follow-up/accessible Made information understandable Liaison/advocate Offered resources Helped guide through process Unbiased/non-directive Committed to journey Willing to have active role between pregnancies Able to build relationships quickly to deal with sensitive information Helped interpret/ contextualize information <i>Psychosocial</i> Met emotional needs Listened Empathized Source of comfort at other appointments Supportive of patient's hopes Compassionate/warm delivery of information Female "comradery"	<i>Medical</i> Explained potential outcomes Medically Educated Knew all the details Appreciated gravity of situation Able to anticipate patient's needs <i>Psychosocial</i> Comfortable talking about loss Non-judgmental Family/friends have needs Focuses exclusively on patient Neutral/objective Strong Limited discloser

2.4.6 Support throughout the maintenance of the relationship. There are several strategies that both counselors and patients mentioned when asked about how the genetic counselor continues to provide support to the patient beyond the initial clinical relationship. The most important factor mentioned by essentially all of the participants was that the genetic counselor remained indefinitely accessible to the patient.

“Even once our job is done we are still there for them because it's not an easy road that they're going to have to go down.”- Genetic Counselor.

The genetic counselor helped facilitate decision making in subsequent pregnancies, made the patient feel supported in her decisions, was perceptive to patient needs, and continued to listen. The genetic counselor acted as a professional opinion that offered guidance,

insight, and advice to the patient; she was comfortable with the patient talking about the past loss and did not attempt to redirect the conversation.

“She was incredibly compassionate about it, she allowed us to talk about, she never tried to redirect or say let's talk about this new baby, which would have been devastating for her to do that. She would just listen.” – Patient.

The genetic counselor offered encouragement and offered recognition of the patient's effort and strength, she provided education and information, stayed updated and connected to the patient's life, tried to ease patient anxiety and fear, offered friendship, and was present for the patient, both physically (i.e., for ultrasounds in the subsequent healthy pregnancy) and emotionally, as needed. The genetic counselor offered continuity of care and was willing to coordinate care even when, in some cases, she was not clinically involved in the patient's medical care at the time.

She wanted me to... even though I was not the genetic counselor involved, she wanted me to be the one to call her with the results. So the two times that she went down to the hospital, she wanted me to call her with the results, and of course the genetic counselor down there was more than willing to let me give her that bad news. I never even saw her in those pregnancies, because she didn't even live here she lived in another city at the time, so I never even saw her but yet I was still involved. -Genetic Counselor.

One genetic counselor even collected monetary resources for the patient's family.

2.4.7 The Relationship. The genetic counselors and patients were asked to describe their relationship and its evolution from those initial clinical experiences to the present.

The descriptions of the relationships included it presently being more of a kinship, consultant/consultee relationship, a relationship that is mutually supportive, or a form of friendship. All of the relationships had moved past one that is purely clinical. Most of the relationships explored fit somewhere between clinical and personal/social. One genetic counselor described her relationship with her patient as a friendship but different from those formed outside of the clinical setting. The most recently formed relationship (approximately four months) was still in the process of evolving.

I think that I might send her a Facebook message, maybe in a month and say I hope you're doing okay, if you ever have any questions, let me know. One thing I'm worried about is that i don't want her to feel weird that she friended me, and has my cell phone number... I don't want her to feel bad. She's said to me before, " I probably take up so much of your time, can you charge my insurance every time you talk to me please," which would in some ways be lovely hahaha. So I think in probably a month I will Facebook message her, not from my work phone, but through Facebook and say I hope you're doing okay. I think that will let her know, a) I'm not concerned about how much time I've spent with her, b) this relationship can go where ever you want to it to go. If she says, I never want to see you again, you've ruined my life, that's cool, that's understandable. -Genetic Counselor.

Almost all pairs still have some clinical aspects woven into the relationship. Of all the relationships explored in this study, only one has moved exclusively past the clinical realm into a more traditional "friendship," (Patient/Genetic Counselor Pair 6).

“She would support me as a friend now rather than a genetic counselor...It’s like a normal friendship now versus me being a patient of hers. I don’t even consider that anymore”-Patient.

Every time we talk, that’s [connection is] there, even if we don’t talk for six months. I mean I have several best friends and most of them I’ve known for 30 years, but [patient] I have known for not even four years and I feel like we have such a good fit together that every time we are together, we lose total track of time and just share, and share and share our experiences and I think it’s because we kind of come from the same place. We were both raised by our families to have a very strong faith and family is first in everything and every way and we want to be living according to that and acting out our faith rather than reacting to the seemingly unimportant things that happen in our lives.- Genetic Counselor.

The feelings that motivated the desire to maintain a relationship included the patient’s desire to reciprocate the support and kindness offered by the genetic counselor, the re-establishment of a clinical relationship during the subsequent pregnancy, and the genetic counselor being physically and/or emotionally present for the patient after the loss. Some of the participants said they could not think of a particular moment in time when they realized they surpassed the clinical relationship, and mentioned that there was just a gradual increase in concern for the patient’s psychosocial well-being as opposed to just the medical elements. Others could comment on the exact event. Many pairs said that there was a moment when the realization arose—each of those had to do with the genetic counselor being physically or emotionally present (or attempting to be present) during a

deeply emotional time for the patient and her family (i.e., attending the memorial service, visiting the patient in the hospital after delivery, etc.).

When it was time for them to exit [the memorial service] I didn't have a chance to talk to her because they ushered them out first, but when I did go out they were sitting in the limo and waiting for everyone to get into their cars. She saw me and she waved to me and I waved to her and I could read her lips that said I love you [genetic counselor] ... thank you. - Genetic Counselor.

Most of the pairs commented specifically that they also had a connection due to similarities between them and had an affinity for the other. Some participants felt that they also had a connection through faith. One pair explained that it was just nice to have a stable and consistent care provider.

The relationships ranged from relatively superficial and comprised of mostly clinical conversation with personal aspects subtly woven in. Most of these pairs felt a need to maintain some professional boundaries. Only a few pairs admitted that they no longer felt any need for professional boundaries due to the fact that they would not be clinically involved in this patient's clinical care again in the future as a result of geography, no future pregnancies, or that their relationship is now exclusively social and the boundary has switched to those a counselor maintains with a traditional friendship. All of the pairs established a trust between each other with the genetic counselor being able to anticipate the patient's needs. Participants knew details about each other's personal lives and some of the patients act as a resource or source of support in some way for the genetic counselor.

Some limitations on the relationship moving completely into the personal realm included geography and physical distance or a sense of hesitancy due to uncertainty on the part of the genetic counselor to cross professional boundaries. Several genetic counselors explained that they sought peer advice on maintaining relationships with patients. Some inquiries were met with approval while others were decidedly discouraging.

2.4.8 Patient. After exploring what the relationship looked like in terms of communication, support, and its evolution, the contributing factors that motivated its development were explored. Starting with the patient, we asked both the patient and the genetic counselor to describe the patient's personality, behaviors, and what her needs were during that time that contributed to the development and maintenance of the relationship. All of the pairs described the patient as being an information seeker.

She definitely was a binding force for our relationship because saw me as not just a support person but more of just a way for her to learn, and then through that she became sort of an expert on what it's like to go through something like that. She had a diagnosis and some expertise and she wanted to share that with other people. -Genetic Counselor.

The patient wanted to know and understand the details of what was going on. The patient was also described as someone who was talkative and outgoing, nice or kind or loving, receptive to what the genetic counselor was trying to offer, resilient, faith-driven and accepting of God's plan, and family-oriented.

Here is another ultrasound where she is thriving and she is doing well and she is still surviving. And that becomes a story where those things become

the traits for their child, look our child is strong, she is surviving, and that feels really good too, especially if you don't get those moments after birth.

– Patient.

The most frequent characteristics mentioned by all participants were that the patient was always very appreciative.

I think it's important for people who made that difference for us to know how much of a difference it made. I think it's important to step out of what is the norm and show what a difference they can make and how many people hunger for that. -Patient.

The patient was determined and strong, easy to talk to, emotionally open or, less frequently, emotionally reserved.

“I think part of it is that she’s just very easy to get to a point of emotional connection in conversation with. She is quite verbal with her feelings.”- Genetic Counselor.

So I got to meet with her... I knew that I needed to open up a little bit more and share with her because it had to do with my daughter. I think that’s what made me open up with her, because it wasn’t about me... it wasn’t about me. We wanted to do everything we could for our daughter and that is when I became open to [genetic counselors] and opened up about things that either concerned me, things that she probably wouldn’t have asked but I felt the need to share because it was on my heart at the time.-Patient.

Some of the participants commented on how resolute the patient was in her decisions or plans for the pregnancy, how complimentary she was toward the genetic

counselor, and was self-directed or proactive. One patient had a past pregnancy history that precipitated anxiety for the patient prior to this affected pregnancy which contributed to the patient's feelings. Three counselors mentioned specifically that the patient was pleasant in spite of the situation which contributed to the development of a positive relationship. Most patients were well-adjusted at present following the loss.

The needs of the patient included needing to feel supported.

I mean she is intelligent enough, and medically savvy enough to research trisomy 18 and make her own medical decisions. She needed somebody to tell her okay, if you're going to make the medical decisions, you go ahead...and not judge her for that. -Genetic Counselor.

Patients needed to be given as much information as possible for preparedness and understanding, to be given hope, to have someone to guide them through the process, or confirmation that it is okay to take control of your care. Two patients felt the deep desire to reciprocate on some level the help that the counselor had provided. One pair described the need to have a female provider, which the counselor helped.

2.4.9 Genetic Counselor. Patients and genetic counselors were asked to describe the genetic counselor's personality and behaviors or strategies that contributed to the relationship or that the patient found appealing. The genetic counselor was described as understanding, someone who valued long-term patient relationships, patient, genuine and sincere, positive and reassuring, and compassionate.

"We knew that as an office we wanted to send something but I wanted to show that in some way that we were truly sorry for her loss and it was a really hard day."- Genetic Counselor.

The genetic counselor was described as approachable or relatable; as tenacious and persistent.

Strategies or professional aspects of the genetic counselor included self-reflective, wanting to be physically present after loss, and being emotionally invested in the relationship.

...I completely understand and my heart broke for her because I just didn't want her, it was hard for me to see her go through more struggle and anxiety about what has already been such a hard decision to get you in the first place. – Genetic Counselor.

“There were a couple of times when like she [GC] went to our funeral and she was crying”- Patient.

Other descriptors for the genetic counselor included being comforting, warm, and personable.

“She was still someone I did not know anything about. My first major interaction with counselor was her holding my hand for the amniocentesis, which was huge to me because she was being very personable and very nonclinical.”-Patient.

They were also described as adaptable and flexible, and patient-centered or patient-focused. One genetic counselor extrapolated on her role during the crisis which may be simply providing the genetic information, or which may expand to the necessity of an ongoing relationship.

I feel like that's what a genetic counselor does, we build relationships with people, we support them and finding them what they need. I don't know what people need right away but if I build a relationship with them then

you can figure that out. Those are the most fulfilling patient relationships or patient interactions. Sometimes people just need a little information and statistics about what are the chances, other times they need an ongoing relationship. -Genetic Counselor.

2.4.10 Crisis. The crisis following a prenatal diagnosis or the realization that there may not be a healthy baby to take home is a severely intense time in a person's life. Patients and counselors were asked how they felt the actual situation contributed to the development of the relationship. All pairs admitted that they had a deeper connection with each other because of their shared experience. All the participants felt that it was challenging to tease apart the most important parts. Some participants felt that the process of helping someone through the experience is what creates the bond or connection. One counselor clarified that it was not the nature of the diagnosis itself that contributed to the diagnosis but was the amount of support the patient needs that determines the possibility for a connection. One of the counselors said that the possibility to develop a relationship has a great deal to do with the patient and how much they feel that they need the counselor.

Pairs declared that because the diagnosis was life-limiting or complex that it required more extensive follow-up both medically and psychosocially, which would bring the pair into contact more frequently, thus providing opportunity to develop a deeper connection.

...since it was so serious we knew not only that it more sensitive but they have more needs, they needed more support. I wouldn't say more handholding but I think those families appreciate you walking through

everything with them as opposed to you just giving them information and then walking away, you're really making an effort, you're always there which is important in such a serious time. - Genetic Counselor.

Participants stated that the diagnosis not only required more follow-up but that the patients might have more psychosocial needs. These patients are dealing with the potential or imminent death of their child.

"I think if it wasn't a diagnosis quite, even something like Down syndrome, it doesn't quite go to that level as far as planning for death"- Genetic Counselor.

One patient explained that the circumstances of the situation forced her to open up to the counselor when she would not have otherwise wanted to. Another expanded her response to say that such a diagnosis requires you to learn, process, and make decisions based on a lot of information in a short period of time so they needed the counselor to help facilitate that.

"It's just like the scariest possible information you could get. So when we were given that information, it was almost like you have been blessed with a genetic counselor that can come in and see you right now."-Patient.

Some patients felt as though she was unable to navigate this new and heartbreaking territory and needed the help of the counselor to make it through.

I've never been through this before, I didn't have any idea. I think with her support, that helped me through because you go through something without having any experience before, you're kind of lost. I didn't know if I was going left or right or if it was okay to feel the way that I was feeling. – Patient.

Seven of the eight patients had another pregnancy following the loss in which they immediately tried to reconnect or reunite with the counselor. They explained that the history made them fearful and anxious about another pregnancy.

Patients and counselors were asked if they thought they might have had a connection with the patient regardless of a prenatal diagnosis and most said that there would have been a connection regardless of the diagnosis due to similarities or complimentary personalities.

We would have had a positive relationship with her no matter what the diagnosis, but it would not have been as intense. To go through a child dying, and for someone to be a part of that story, is probably the most intense of a relationship that you can have with someone. When you are given the threat of your child dying for several weeks or months, that is an opportunity for a strong connection. -Patient.

I've had patients where the diagnosis was awful right, though they don't, for whatever reason, they don't lean on me as much, they don't require as much psychosocial support, I don't have those long-lasting relationships with those patients. I think they may be a repeat customer not necessarily a bond. I feel for those people but I don't get as connected to them. So I wonder if it's more rooted in the support, the give and take that happens between the patient and provider like whole relationship. That's what continues on. - Genetic Counselor.

Few said it was unlikely without the shared experience.

I don't know... that's what sort of made it hard for me [to become completely engaged in a personal relationship] because why do I not want to do that? Why am I holding back from saying yes I would be happy to come and do that? I don't think that we have a lot in common so I don't think that we would be friends outside having this shared experience...so I'm not really sure. - Genetic Counselor.

Some of the patient needs during the crisis included the need for certainty, whether it was confirmation that they are making the right choices for the pregnancy...

And one thing that she wanted, and this is not atypical, what she wanted was for me to say that "you should have that abortion. I am your healthcare provider and your baby is very abnormal, it will not have a normal life and so for your baby you should have an abortion." I can't say that, that is a medical decision on her part... But she could have wanted one and was searching in her mind and she wanted for me to say that yes you're right this is the right thing to do, which I can't say.-Genetic Counselor.

...or that the information they were given is both complete and accurate, “I wanted more data. I wanted more information on outcomes”-Patient.

Although many of the patient participants appreciated the depth of relationship established through the experience, none of the patient participants admitted the need to establish a personal relationship.

“No, I wouldn't say I had a deeper need [to connect with her past a clinical relationship]”-Patient.

Patients also greatly appreciated that the information provided was made understandable.

...in a short amount of time, I mean 20-30 minutes, someone can give you genetics 101 and tell you what you need to know and make sure that you understand that this could mean this or this could mean that. -Patient.

“The way the she spoke to you, she didn't speak to you like a patient, she spoke to you like a person, no medical jargon, just being real, I appreciate that.” -Patient.

2.4.11 Long-term Effects. The effects of the relationship and influence on the patients and counselors long-term both personally and/or professionally were explored.

The genetic counselors were asked what some of the long-term effects are of having this lasting relationship with the patient or having shared in this experience with the patient. Two genetic counselors said that the patient had become a source of support for the genetic counselor in either her personal or professional life. Several counselors realized that patients have the power to impact and change providers' approaches to patient care.

“So patient's case really brought a lot of attention to some major people and helped us get together to create a more clear understanding of what we can offer to people for patient, especially in this terrible situation.”-Genetic Counselor.

“I knew that she was gonna be one of those patients that I was gonna remember, someone who was going to shape me as a genetic counselor, help me help other people that come into my life later.” –Genetic Counselor.

When you have a patient that is going through a pregnancy and has a goal and you just know in your heart that goal isn't going to happen, that they

are not going to meet that goal but you're rooting for them anyway. And when they don't reach their goal, they look at you and say that they wouldn't change a thing and that this has been a wonderful experience, then you know you really succeeded, you didn't let them down. Just because they didn't meet their goal, it doesn't mean things went wrong for them. - Genetic Counselor.

One genetic counselor said that the experience increased her professional self-awareness.

My job isn't to help them make their decisions; my job is to give them information so they can make informed decisions. Like I said, I don't think I would have done or had the strength to do what she did but I'm glad that she did because I think it reminded me that my job is to advocate for peoples informed decisions, not to tell them what decision to make.-

Genetic Counselor.

Several other genetic counselors stated that they realized the impact that a genetic counselor can have in a patient's life and how satisfying it can be.

I learned from [patient] on another level of what a difference I can make, I think that I listen more closely. I ask specific questions about support and who they have in their lives. I think that is how we can make a difference. - Genetic Counselor.

Some of the other effects mentioned were that they had the opportunity to learn from her mistakes, not to judge a book by its cover, and the strength that faith can have in

a patient's life. Genetic counselors also felt that they were better able to appreciate the patient's perspective and the experience of loss.

Just going to the funeral had the biggest impact because if you work with fetal anomalies you know it's really hard for these families but seeing it in person was definitely something unlike anything I've ever experienced before. It gave me deeper appreciation for what these families go through.

– Genetic Counselor.

The counselors were impressed by the patient's resilience and one said that the experience validated her current practice.

One counselor stated that the experience changed her personal perspective on life and reminded her to be more positive.

"I think about her a lot when people in my life, like family or friends, make decisions that I don't agree with and I just have to remind myself again that they're not my decisions to make."-Genetic Counselor.

Counselors also learned that providers should respect patient views and caution them about proposing a diagnosis with little evidence because the patients may latch onto that. Some realized that they should take more steps to discover who the woman really is, and many said that this experience challenged perceptions of being friends with patients, albeit remaining cautious, but not closed to the potential.

[This experience] challenged my perceptions of being able to be friends with your patients, which first of all I was very against. I think the reason I was initially against it was because the relationships that I have with

patients are usually very short, pregnancy is a transient state. So I think it's changed my ideas of how appropriate it is.-Genetic Counselor.

It's sort of made me aware of that fact that it's sometimes very easy to potentially sort of want to be friends with people, or that it can go there and that there is potential for that, whether it's okay for that to happen or not...it's definitely made me more aware of that. Not that that's bad but sometimes it's inappropriate to, when you're discussing such close personal things but it's like, where is it going? I need to be careful about that I think. - Genetic Counselor.

These counselors proved the value of maintaining contact with a patient past the clinical experience. One counselor commented that she was able to see firsthand how a story can end happily.

The patients were asked what some of the long-term effects are of having maintained this relationship with their genetic counselor or having had this experience. One patient said that her standards for patient care were raised due to the excellent care she received from the genetic counselor and her team.

"I think [GC] set the bar high for me knowing now that there are medical professionals who can exhibit concern at the same time that they're doing their job. I think I've come to expect that now."- Patient.

One patient learned how to better advocate for herself and others through and following the experience. One patient realized that the patient is allowed to take control of her care. Two patients felt a comfort knowing that they will always have the support of their counselor and another felt that the genetic counselor was going to be a part of their life in

some was indefinitely. Several patients gained a deeper appreciation and understanding of the value of life and family.

Her [baby girl] story is what it was, you don't get anything else after that, that's something that I don't think everyone realizes, if the baby does not survive, those moments that the family has when that baby is alive in the womb...that becomes that child's life, those moments, those are the memories. You remember the ultrasound when she did this with her fingers or do you remember when counselor said she could survive that if blah blah blah. Those are the moments of your child's life forever, that's it, that's all you have of her. - Patient.

Two patients gained a confidence in growing their family after the loss; one realized the value of carrying a child who would not survive. One patient said that she realized that there are those people out there who still care; another patient realized that there can be a happy ending after such sadness and heartbreak. One patient described the value of the overall experience in the context of her life and the lives of others.

Yes in that moment all you really want is for your baby to survive, but guess what, when your baby does not survive, there are going to be things that you wanted or needed that you did not realize at that moment, that you could have asked for or that you needed. Those are the things that people can do that are different. Those are the things that people can give you without giving it a guarantee that your child is going to live. That is not what makes the difference, what makes the difference is the respect and the dignity that your child deserves...for people who have a genetic

diagnosis I think there is sometimes so much that is not getting done that we look back on it and say, God I just wish that we had that be part of our experience. There are some of us who have that, and it is amazing that there are people who know what to give.-Patient.

2.5 Discussion

2.5.1 Communication. There is little to no documented literature that extensively explores the modes of communication utilized by genetic counselors following the establishment of the genetic counselor-patient relationship. Much of what is known discusses only the modes used during the clinical establishment of the relationship which are well established in the healthcare profession (telephone, email, in person appointments). These typical modes were expanded to include more personal, social venues (Facebook, cellphones). Social media is being used by some medical practices to recruit patients (i.e., plastic surgery) (Wheeler, 2011) and other practices utilize mobile technologies to assist physicians with medical decision making, clinical tasks, and other computing functions (Putzer, 2012). This study effectively chronicles that information and provides some insight into what modes are utilized. The content of conversation also expanded to include more personal elements of both the patients' and the genetic counselors' lives. Medically related topics have not been completely evicted from the conversations for most of the pairs, in fact, the counselor acts as somewhat of a resource for the patient even to the present time. Communication amongst the pairs overall decreased as time progressed, unless there was another clinical interaction (i.e., subsequent pregnancies). The initiation of who would contact whom changed over time, the majority of patients would contact the genetic counselor first, but that has shifted to

be slightly more balanced with both the patient and the genetic counselor contacting each other.

2.5.2 Support. The support offered by the genetic counselor in this study aligns with Bernhardt et al. (2000) study of psychosocial support offered in genetic counseling. Participants thought that the genetic counselors listened and validated concerns, empowered and encouraged the patient, and facilitated decision making. This study complemented Bernhardt et al. study as well as expanded on some of the elements mentioned. The participants in this study expressed in more detail some of the education, professional, and psychosocial areas of support they were offered. The genetic counselor acted as an advocate for the patient, treated the patient like a priority, offered resources and committed herself to the journey with the patient. The genetic counselors also checked-in with the patient, mirrored the patient's hopes, respected the patient's connection to the pregnancy, contextualized the information, and made the patient feel comfortable and confident moving forward.

Bernhardt et al. also mentioned that some relationships that are established between the pair may involve on-going support, often in the form of telephone or letters rather than appointments. This study also expanded on the long-term or on-going relationship Bernhardt mentioned. The exploration of this relationship revealed that genetic counselors continued to be present for the patient, to listen, and serve as a resource, attempt to ease anxiety and fear, offer friendship, and were willing to coordinate care even when not clinically involved in patient care. The genetic counselor allowed the patient to determine the content of conversation without redirecting, which

also serves as a means of support to the patient (i.e., the patient was free to talk about loss).

Compared to other health care providers, the genetic counselor was always available and helped bridge the gap between the medical information and the patient's life. The depth of psychosocial support was also markedly different between other providers and genetic counselors with the genetic counselor spending a great deal of time and energy addressing those needs. The genetic counselor listened to and advocated for the patient, made information understandable, was committed to the journey with the patient, helped the patient contextualize and integrate the information provided in order to make informed decisions the patient could feel good about.

The support offered was unique compared to family and friends as well as other providers. Compared to family and friends, genetic counselors had medical knowledge, were able to provide information, address specific psychosocial needs that family members and friends were not always capable of. Another notable theme was that family and friends of the patients' have needs too when the patient is experiencing crisis. Being patient-centered is known to be valuable in the medical field (Victoor, Delnoij, Friele, & Rademakers, 2012) and may be equally important when dealing with family members and friends.

2.5.3 Relationship. The relationship between the genetic counselor and patient evolved from one that was clinical to include more personal elements amongst all of the pairs. The relationships ranged in length from four months to about fourteen years. Some relationships developed slowly due to frequent interaction, for example, limited to subsequent pregnancies. Other relationships transitioned after a single event, such as

attending the memorial services for the affected pregnancy or sharing other deeply personal moments for the patient and/or her family. These relationships ranged from relatively superficial to deeply personal. Most of the counselors felt a need to maintain some professional boundaries but a few felt as though those boundaries had expired after their clinical relationship with the patient ended.

2.5.4 Contributions. The patient, genetic counselor and the situation itself all contributed to the development of the relationship. Lalor et al. (2008) explained that there are two main types of patients when facing a prenatal diagnosis, “monitoring” and “blunting.” Monitoring patients have high information needs while blunters are the opposite and would prefer to avoid the information. The patients in this study were all information seekers. Berkenstadt et al. (1999) explained, the needs of the patients in a genetic counseling session were indeed, 1) the need for certainty, whether it be for the confirmation that they are making the right choices for their pregnancy or that the information they were given is both complete and accurate. The second need Berkenstadt et al. described was the need for a personal relationship. This was not something necessarily expressed by the participants in this study. Although many of the patient participants appreciated the depth of relationship established through the experience, none of the patient participants admitted the need to establish a personal relationship. The third need Berkenstadt et al. included was the need for the integration of lay and scientific explanations.

Eugster and Wampold (1996) explored some of the patient contributions to the therapeutic alliance in a therapy setting that fit well for the patient participant population in this study. The patient contributions include 1) patient involvement, 2) how

comfortable the patient feels with the therapist, 3) how much progress the patient makes through the course of the relationship, and 4) how much the patient likes the therapist and how he/she is perceived through the patient lens. This is consistent with the findings of this study. The patient participants were all involved in the process of genetic counseling; they were deeply invested in establishing a positive relationship with their genetic counselor. All of the patients expressed feeling comfortable with their genetic counselor and admitted the impact the genetic counselor had on their experience...

I could not have done it without her... we could not have done it without her. We could not have felt good about where we stand, we still don't feel good, we are still angry, we are still hurt, but we can only move on because she held our hand.-Patient.

All of the patient participants liked and appreciated their genetic counselor and saw them in a positive light.

Hartmann et al. (2015) and Veach et al. (2007) described the goals of a genetic counseling session and specific strategies necessary to achieve some of these goals from the perspective of the genetic counselor. The genetic counselor's ability to build a relationship and foster good communication with the patient during and after the session was critical to the session's perceived success. The genetic counselors in this study did exactly that with their patients. They established a relationship, fostered good communication, and were expressly available to the patient. Schapira (2013) asked psychological oncology clinicians what attributes and strategies are used by a "good quality health professional" which was also supported in the current study. Genuine, engaged, compassionate, nonjudgmental, etc. were considered highly valuable attributes

to both Schapira and the participants in this study. The 2003 meta-analysis by Ackerman & Hilsenroth's of attributes and strategies/techniques of therapists which contributed to a positive working alliance also proved to be relevant to the genetic counseling participants. Flexible, warm, friendly, open, interested/invested, etc., were all deemed very important for both the therapist population he studied as well as the population explored. The techniques employed by both the therapists in his study and the genetic counselors in the current study prove to be complimentary. The genetic counselor participants, like the therapists, were supportive, understanding, active participants in the relationship, attended to the patient's experience, etc. The strategies and attributes/characteristics of the genetic counselors in this study contributed to the development and maintenance of the relationship.

The diagnosis itself did not play the largest role in the development and maintenance of the relationship. The supportive needs of the patient motivated the relationship and the crisis allowed for the opportunity to develop a strong relationship. The patient and genetic counselor personalities and behaviors solidified and strengthened the relationship during the crisis and beyond. Pierson (1999) stated that constant interaction in the field of nursing contributed to the development of the relationship between the professional and the patient. Our participants agreed with that sentiment. The crisis required a great deal of follow-up, both medical and psychosocial (Fonseca et al., 2012), which makes for the perfect setting when two relatively similar personalities come together by circumstance.

2.6 Conclusion

2.6.1 The Reciprocal-Engagement Model (REM)

The REM emphasizes the role of the relationship in the success of the genetic counseling session. Veach (2007) described in detail the importance of a positive relationship in the REM by theorizing the relationship developed between counselor and patient is just as important to the success of the session as the information itself. In our study we were able to see how successful a relationship and therefore genetic counseling experience can be. The fact that the patient wanted to maintain a relationship with the genetic counselor after what can only be considered a heartbreaking ordeal for the patient and the patient's family speaks to the importance of developing a positive relationship with the patient. As a result of the development of such a relationship, the genetic counselor-patient pairs were able to navigate the experience and grow together in complimentary ways.

This exploration supports the concept put forth in Tenet 2) "relationship is integral to genetic counseling." This study also supported all of the other tenets Veach et al. (2007) and Gaff & Bylund (2010) describes. Tenet 1) "genetic information is key," Tenet 3) "patient autonomy must be supported," Tenet 4) "patients are resilient" and Tenet 5) "patient emotions make a difference," (Veach et al., 2007). The genetic information was important to the patient participants; in fact, all of them expressed that the genetic counselor providing the information was one of the greatest sources of support during the time of crisis. The genetic counselors support of patient autonomy was also something mentioned often. The participants described the genetic counselor as being non-directive/unbiased, supportive of the patient's hopes, and empowering and

encouraging of the patient's desire to direct their own care. Many of the genetic counselors mentioned how impressed and inspired they were with the patient's resilience throughout the process. These impressions were translated, for some genetic counselors, into a new or re-established personal philosophy or outlook on life. All of the genetic counselors knew that the patient's emotions made a difference. Although not directly stated, the genetic counselors empathized with the patient, went at the patient's pace in terms of informational and psychosocial needs, and offered as much support as possible or necessary. This provides further validation to how the REM represents the genetic counseling process.

The relationship itself was the conduit for all the other tenets of the REM to be met. The participants viewed the relationship positively, at least to some degree, and were therefore able to fully satisfy the other tenets while strengthening the relationship simultaneously.

2.6.2 Implications for Genetic Counseling Practice

There are many implications of this study including clinical, professional training, and research implications. Clinically, we have captured some of the complex and intangible elements of a lasting relationship that develops between a patient and genetic counselor during a crisis situation. This provides both direction and value for genetic counselors as they need to understand characteristics that contribute to this relationship, communication strategies and support needs of the patients. The characteristics of the patient, genetic counselor, and crisis all play an important role in the relationship, such as the genetic counselor being understanding, sincere, compassionate, relatable, and open to developing long-term relationships, and the patient being an information seeker,

appreciative, emotionally communicative, and proactive/self-motivated. The crisis situation (regardless of the specific diagnosis) offers a unique opportunity for the patient and genetic counselor to unite on a shared journey. These potential relationships are not restricted to a specific subset of patients; it instead seems to revolve around the supportive needs of the patient. Communication during the establishment of the relationship most strongly suggests that the genetic counselor make her/himself available using multiple modes (in person, phone, email, etc.), as frequently as the patient requires (daily, weekly, monthly, etc.), and to cover all the content that the patient needs addressed (medical/educational, psychosocial/emotional, etc.). The supportive needs of the patient plays an important piece because this study revealed many of the ways that a genetic counselor's support differed from other healthcare providers as well as family and friends. Genetic counselors have provided medical input, made themselves available to the patient, and addressed the patient's psychosocial needs.

Other clinical implications can be gained by revisiting the long-term effects (or lessons learned) on genetic counselors. For instance, patients have the power to affect healthcare by challenging the status quo for patient care. The patient can also have an impact on the genetic counselor's personal perspectives on life and medicine. Genetic counselors also came to realize that they can form deeply engaged relationships with a patient and that a patient relationship can alter the genetic counselor's approach to other patients.

Implications for patient outcomes can also be gained by exploring their long-term effects. Patients mentioned that their standards of patient care have increased due to this

relationship and that an experience like one they have been through with the genetic counselor can have some positive outcomes in the patient's life.

Though limited in scope, this study may validate the professional education, both medically and psychosocially, genetic counselors receive as genetic counselor participants offered something unique and invaluable to the patient participants in this study. The fact that the genetic counseling participants sought peer advice and were met with differing opinions overall displays the potential need for more guidance. This study may suggest the need for more training and supervision of genetic counselors as they manage relationships with patients and learn to appreciate situations in which professional boundaries can be more flexible

This is a preliminary study exploring a relatively specific patient population. Future research could include a larger population of the same patient group (life-limiting prenatal diagnosis) or even be applied to other prenatal populations (non-life-limiting conditions, etc.). Research in other specialty areas such as pediatrics, adult and cancer genetics could also offer an interesting perspective on the patient-counselor relationship and the unique role of the Genetic Counselor. How might the relationship established in these specialties resemble those explored in this study and in what ways might they be different?

2.6.3 Limitations

There are several limitations to this study, one being the relatively homogeneous population (all women, most Caucasian, all educated). A qualitative investigation of eight relationships in a homogeneous population may not necessarily yield generalizable information. Observations about communication, support, relationships, etc., gathered in

this study may not represent the full depth of these elements in other patient-counselor relationships, especially in other specialty areas.

Another interesting limitation is among those who completed the genetic counselor specific online questionnaire to determine eligibility; seven were not willing to contact their former patient to invite them to participate. This could speak to the possibility that perhaps the counselors did not think the patient would participate for some reason (i.e., reopening patient wound), or perhaps the genetic counselor feels uncertain about opening themselves up or feels uncertain about the deep and more personal relationship that can develop from the experience.

In conclusion, this study demonstrates that the relationship established between the genetic counselor and patient participants proved to add valuable insight into an essentially unexplored area in genetic counseling. The participants in this study explained that the crisis provided the opportunity for the genetic counselor and patient to establish a deeply personal relationship. The relationship developed and maintained was built on and strengthened by the unique and essential support offered by the genetic counselor during and after the life-limiting prenatal diagnosis. The personal and/or professional characteristics of the genetic counselor and patient, as well as patient needs were important contributing factors. The experience and relationship had lasting effects on the participants which have affected their personal and/or professional lives. The relationships between these participants exposed some of the intangible and invaluable elements of genetic counselors and the role and impact these professionals can have.

Chapter 3. Conclusion

3.1 The Reciprocal-Engagement Model (REM)

The REM emphasizes the role of the relationship in the success of the genetic counseling session. Veach (2007) described in detail the importance of a positive relationship in the REM by theorizing the relationship developed between counselor and patient is just as important to the success of the session as the information itself. In our study we were able to see how successful a relationship and therefore genetic counseling experience can be. The fact that the patient wanted to maintain a relationship with the genetic counselor after what can only be considered a heartbreaking ordeal for the patient and the patient's family speaks to the importance of developing a positive relationship with the patient. As a result of the development of such a relationship, the genetic counselor-patient pairs were able to navigate the experience and grow together in complimentary ways.

This exploration supports the concept put forth in Tenet 2) "relationship is integral to genetic counseling." This study also supported all of the other tenets Veach describes. Tenet 1) "genetic information is key," Tenet 3) "patient autonomy must be supported," Tenet 4) "patients are resilient" and Tenet 5) "patient emotions make a difference," (Veach, 2007). The genetic information was important to the patient participants; in fact, all of them expressed that the genetic counselor providing the information was one of the greatest sources of support during the time of crisis. The genetic counselors support of patient autonomy was also something mentioned often. The

participants described the genetic counselor as being non-directive/unbiased, supportive of the patient's hopes, and empowering and encouraging of the patient's desire to direct their own care. Many of the genetic counselors mentioned how impressed and inspired they were with the patient's resilience throughout the process. These impressions were translated, for some genetic counselors, into a new or re-established personal philosophy or outlook on life. All of the genetic counselors knew that the patient's emotions made a difference. Although not directly stated, the genetic counselors empathized with the patient, went at the patient's pace in terms of informational and psychosocial needs, and offered as much support as possible or necessary. This provides further validation to how the REM represents the genetic counseling process.

The relationship itself was the conduit for all the other tenets of the REM to be met. The participants viewed the relationship positively, at least to some degree, and were therefore able to fully satisfy the other tenets while strengthening the relationship simultaneously.

3.2 Implications for Genetic Counseling Profession

There are many implications of this study including clinical, professional training, and research implications. Clinically, we have captured some of the complex and intangible elements of a lasting relationship that develops between a patient and genetic counselor during a crisis situation. This provides both direction and value for genetic counselors as they need to understand characteristics that contribute to this relationship, communication strategies and support needs of the patients. The characteristics of the patient, genetic counselor, and crisis all play an important role in the relationship, such as the genetic counselor being understanding, sincere, compassionate, relatable, and open to

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Appendix A – Web-based Questionnaires

University of South Carolina Master's Thesis Project: Genetic Counselor Web-Based Survey

Q1 Occasionally prenatal genetic counselors work through a difficult situation with their patient that develops into a reciprocal and lasting relationship. Thank you for your interest in this study which seeks to explore that unique connection between the prenatal genetic counselor and patient who have worked through a crisis together.

Q2 Have you maintained a connection with a patient following the provision of prenatal genetic counseling for a potentially lethal diagnosis that resulted in a major loss via pregnancy termination, miscarriage, stillbirth or neonatal death?

Yes/ No

Q3 When was this relationship formed (mm/yyyy).

Q4 Would you be willing to contact this patient and invite their participation in this study for a 60 minute interview?

Yes/ No

Q5 Are you also willing to complete a separate telephone interview lasting up to 60 minutes?

Yes/ No

Q6 Please provide your preferred contact information to get involved in this study. Your participation is voluntary. Research studies include only people who choose to take part. By providing the information below you are indicating your willingness to participate in this study. Disclaimer: Information provided below will be used for contact purposes ONLY. No identifying information will be used for this study.

Name

Email

Office Phone

Cell Phone

Home Phone

Best Time to Contact (00:00 AM/PM)

Q7 You will be contacted with more information within the next couple weeks regarding participation in the study.

University of South Carolina Master's Thesis Project: Patient Web-Based Survey
Patient Participation

Q1 Occasionally genetic counselors work through a difficult situation with their patient which develops into a reciprocal and lasting relationship. Thank you for your interest in this study which seeks to explore that unique connection between the prenatal genetic counselor and patient who have worked through a crisis together.

Q2 What was the prenatal diagnosis in your pregnancy?

Q3 What was the pregnancy outcome?

- Pregnancy Termination
- Spontaneous Abortion/Miscarriage
- Stillbirth
- Infant Death
- Other _____

Q4 This study involves an interview that may last up to 60 minutes, would you be willing to participate in this study?

- Yes
- No

Q5 Please provide your preferred contact information to get involved in this study. Your participation is voluntary. Research studies include only people who choose to take part. By providing the information below you are indicating your willingness to participate in this study. *Disclaimer: Information provided below will be used for contact purposes ONLY. No identifying information will be used for this study.*

Name

Email

Office Phone

Cell Phone

Home Phone

Appendix B – Interview Guides

Interview Questions: **Patient**

1. *Tell me how you met _____ (genetic counselor)*
 - How was your relationship developed? Communication (mode, frequency, content, initiation)
 - How was it maintained? Communication (mode, frequency, content, initiation)
 - How has it changed over time?
2. *What about the circumstances of the prenatal diagnosis encouraged the relationship.*
 - What crisis factors/actions influence the relationship?
3. *What are some personal characteristics about yourself that encouraged the relationship?*
 - What patient factors/actions establish & maintain the relationship?
4. *What were some specific characteristics about ____ (GC) that encouraged relationship?*
 - What personal characteristics establish & maintain the relationship?
5. *What about actions—is there something specific that ____ (GC) did to strengthen or encourage the relationship?*
 - What personal actions establish & maintain the relationship?
 - What professional actions establish & maintain the relationship?
 - Emotional support
 - Facilitate decision-making
 - Educator
6. *What makes the GC unique compared to other providers (during and after the crisis)?*
 - What personal and professional factors/actions make them unique?
 - Was additional or alternate support offered? If so, describe.
7. *How has this relationship influenced you long-term?*
 - Do you desire similar relationships with other providers?

Demographics:

Patient

Gender: _____

1. Age: _____
2. Ethnicity: _____
3. Marital Status: _____
4. Number of Children: _____
5. Education: _____
6. Prenatal Diagnosis: _____
7. Pregnancy Outcome: _____

Interview Questions: Genetic Counselor

1. Tell me how you met _____ (patient)
 - How was your relationship developed? Communication (mode, frequency, content, initiation)
 - How was it maintained? Communication (mode, frequency, content, initiation)
 - How has it changed over time?
2. What about the circumstances of the prenatal diagnosis encouraged the relationship.
 - a. What crisis factors/actions influence the relationship?
3. What are some personal characteristics about _____ (patient) encouraged the relationship?
 - What patient factors/actions establish & maintain the relationship?
4. What were some specific characteristics about you that encouraged relationship?
 - a. What personal characteristics establish & maintain the relationship?
5. What about actions—is there something specific that you did to strengthen or encourage the relationship?
 - What personal actions establish & maintain the relationship?
 - What professional actions establish & maintain the relationship?
 - Emotional support
 - Facilitate decision-making
 - Educator
6. What makes a GC unique compared to other providers (during and after the crisis)?
 - What personal and professional factors/actions make them unique?
 - Was additional or alternate support offered? If so, describe.
7. How has this relationship influenced you long-term?
 - Do you desire similar relationships with other patients?

Demographics:

Genetic Counselor

1. Gender: _____
2. Age: _____
3. Ethnicity: _____
4. Marital Status: _____
5. Number of Children: _____
6. Years of practice: _____
7. Years of practice in prenatal genetic counseling: _____

8. Location (state) where patient-counselor relationship began:

9. When patient-counselor relationship began: _____

Appendix C- Data

Communication (then versus now)

When relationship was being developed (then)	Percent	<i>n</i>	As relationship has been maintained (now)	Percent	<i>n</i>
<i>Mode</i>			<i>Mode</i>		
In person	100	8	In-person (clinical only)	37.5	3
Email	62.5	5	In-person (clinical & social)	50	4
Phone	87.5	7	In-person (social only)	12.5	1
Cellphone (as needed)	37.5	3	Email	62.5	5
<i>Frequency</i>			<i>Frequency</i>		
Daily	25	2	Phone	100	8
Weekly	37.5	3	Cellphone	12.5	1
Monthly	50	4	Cards/letters	12.5	1
Unknown	12.5	1	<i>Frequency</i>		
<i>Content</i>			<i>Content</i>		
Clinically-related	100	8	During subsequent pregnancy	75	6
Social/check-in (superficial)	62.5	5	Weekly	12.5	1
Social/check-in (in-depth/emotional)	37.5	3	Monthly	25	2
Faith	12.5	1	Few times a year	25	2
GC personal life			Varies	25	2
<i>Initiation (excluding GC protocol-results, testing)</i>			<i>Content</i>		
GC	25	2	Clinically-related	87.5	7
Patient	50	4	Social/check-in (superficial/minimal emotional)	62.5	5
Both	25	2	Social/check-in (depth/emotional)	62.5	5
			Faith	37.5	3
			GC personal life	75	6
			<i>Initiation</i>		
			GC	12.5	1
			Patient	67.5	5
			Both	75	6