Assessment of Genetic Provider and Parent Communication Patterns in Pediatric Genetic Counseling Sessions

THESIS

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By

Nicole Louise Sophia Lahner

Graduate Program in Genetic Counseling

The Ohio State University

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Master's Examination Committee:

Dawn C. Allain, Advisor. MS, LGC

Matthew Pastore, MS, LGC

Susan Havercamp, PhD
Abstract

Background: Communication and counseling are critical to helping parents cope with a genetic diagnosis in their child, and there are various models suggesting the ideal manner in which to communicate information about a diagnosis. Previous studies using parent surveys and simulated genetic counseling sessions demonstrate healthcare providers emphasize medical and genetic information over quality of life issues at the time of a diagnosis and attend less often to parents’ psychosocial concerns.

Methods: Researchers audio recorded pediatric genetic counseling sessions during which a family was receiving an initial diagnosis in a child. Researchers also surveyed parents about their counseling experience.

Results: Analysis of twenty sessions indicates providers’ focus is on educating parents/guardians about technical medical and genetic aspects of the condition. Providers discussed quality of life issues less often in our study and usually did so in response to parents/guardians’ concerns or in the context of referrals to other families. Additionally, providers and parents were cooperating partners in the exchange of biomedical information about the pediatric patient. Providers also attended to parents/guardians’
emotions via empathic statements and by proactively addressing genetic guilt. Finally, the majority of parents reported helpful aspects about their genetic counseling session and found their experience to be positive.

Limitations: Limitations of our study include parents/guardians with a serious diagnosis in their child not being invited to participate. All subjects were Caucasian, so our findings are not generalizable to other patient populations. In addition, parents may have received brief telephone counseling about the diagnosis prior to their initial visit, and we did not account for this aspect of communication in our analysis. Furthermore, the parent/guardian survey asked about helpful aspects before unhelpful aspects, which may have biased participants’ responses. Future research includes determining if a similar communication strategy is used by groups of genetic providers in other pediatric genetics clinics. Other studies might also focus on patients from non-Caucasian ethnic backgrounds and/or clinics in other states or countries. It would also be useful to re-contact the parents/guardians and ask if their perceptions of their genetic counseling session have changed from their initial survey responses. Finally, video-recording genetic counseling sessions would allow for further characterization of providers’ and parents’ psychosocial behaviors.

Conclusions: Genetic providers’ communication pattern emphasizes education and information giving and involves some engagement of parents/guardians’ emotions. Providers expressed empathy but directly assessed parents’ concerns less often.
Providers also appear to not view quality of life issues as important to discuss at the time of an initial diagnosis. Parents were given resources, most often information about their child’s disorder and the genetic test results. Providers and parents were also cooperating partners in the exchange of biomedical information about the child. Finally, the majority of parents/guardians perceived this pattern of communication in their genetic counseling sessions to be helpful.
Dedication

To all the children who have received a genetic diagnosis and their parents.
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Vita

June 2010 ...................................................... Eden Prairie High School

2014 ......................................................... B.A. Biology, Northwestern University

2016 ........................................................... M.S. Genetic Counseling, The Ohio State University

Fields of Study

Major Field: Genetic Counseling
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Chapter 1: Background

Models of Communication in Genetic Counseling

Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process consists of interpretation, education, and counseling (Resta et al., 2006).

Two models have been suggested as the primary means of communication during genetic counseling sessions: the teaching model and the counseling model (Kessler, 1997). The teaching model is a form of health education with the goal of creating educated counselees. This model assumes that humans act and make decisions in a more or less rational manner and fully informed clients should be capable of making their own decisions. Thus, the objectives of the genetic counselor are to provide impartial and balanced information and to correct misinformation. The relationship between counselor and counselee is based on authority rather than mutuality. The teaching model approaches genetic counseling as a “one size fits all” process where the same information (more or less) is given to each client. This is an advantage when there is a high volume of patients, but the unequal relationship between counselor and counselee may increase dependency on the professional’s authority and suppress autonomy (Kessler, 1997). The counseling model seeks to create a client who is psychologically more autonomous and
functional. The genetic counselor assumes that counselees need more than just information. The relationship between counselor and counselee aims for mutuality, and genetic counselors seek to engage and understand the client. Education is not an end in itself but a means of helping clients deal with the personal meaning of genetic information and make decisions. While individuals receiving genetic counseling have been found to assimilate information they receive surrounding risk and diagnosis, the teaching model can lead to excessive information giving, leading counselors to provide more information than what people can understand. The teaching model also does not account properly for how to assimilate clients’ personal meaning of information into a genetic counseling session.

The counseling model can help address some of these concerns as it seeks to help clients understand and work through difficult problems (Kessler, 1997). However, given the increasing demands for counselors to see many patients in a short time period, it is not realistic to rely only on this model. Many counselors are unable to put in the emotional effort the counseling model requires in order to use it for every patient since the counselor must individualize the session to fit the client’s needs and actively participate in eliciting clients’ concerns. This compels genetic counselors to combine both models in the short-term interactions of genetic counseling. Veach et al., advocate for a combination of the teaching and counseling models called the Reciprocal Engagement Model that combines both provision of genetic information and active engagement of clients’ feelings during the education process (Veach, Bartels, & LeRoy, 2007).
Use of Communication Models in Genetic Counseling Sessions

One method of researching the practice of genetic counseling is via process studies, which aim to determine what happens in genetic counseling practice and why it works. Process studies can assess behavior, including how providers and patients communicate with one another during genetic counseling sessions (Biesecker & Peters, 2001).

A 2008 article reviewed studies that assessed the process and content of genetic counseling communication. This review identified two major themes: genetic counseling is provider driven and information giving is central to the genetic counseling process (Meiser, Irle, Lobb, & Stewart-Barlow, 2008). A large proportion of communication in genetic counseling was also found to be biomedical rather than psychosocial, indicating that the teaching model is the most prevalent communication model. This is not a recent phenomenon given that a 1982 study assessing pre-amniocentesis genetic counseling sessions found that genetic counselors conveyed more factual/information content than affective/expressive content (Kessler & Jacopini, 1982). Examples of factual content are the medical significance of the diagnosis, prognosis, treatment, recurrence risk, reproductive alternatives, inheritance, tests, and population risk (Lippman-Hand & Fraser, 1979; Michie, French, Allanson, Bobrow, & Marteau, 1997). A recent process study explored genetic counseling communication by video recording 177 simulated cancer and prenatal genetic counseling sessions (Roter, Ellington, Erby, Larson, & Dudley, 2006). After analyzing 152 of the sessions, four patterns of counseling practice
emerged: clinical teaching, psycho-educational teaching, supportive counseling, and psychosocial counseling. Among these four patterns, counselors again showed a preference for the teaching models over counseling models (57% of cancer and 58% of prenatal sessions). Roter and colleagues also developed an overall communication profile of the video recorded genetic counseling sessions. Based on their analysis, counselor dialogue was found to primarily encompass clinical information (47%) while psychosocial issues made up 9% of the dialogue. In addition, the researchers found that although counselors engaged in question asking behaviors, the majority (68%) of questions asked were close-ended. Ultimately, it was surmised that the counselors’ communication behaviors led to less active engagement of clients, which is consistent with the teaching model of genetic counseling (Roter et al., 2006). While most of the studies to date analyzing genetic counseling communication have been in cancer and prenatal specialties, a study published in 2010 explored genetic counseling communication at the time of a new genetic diagnosis in a child (Babul-Hirji, Hewson, & Frescura, 2010). This study used a qualitative discourse analysis from 10 audio recorded sessions between the parents of children with a genetic disorder and genetic counselors. As in other specialties, the authors found a tendency for genetic counselors’ use of the teaching model (Babul-Hirji et al., 2010). Overall, it is clear that research on genetic counseling communication demonstrates a preference for the teaching model in both simulated and real-time genetic counseling sessions.
Communication of “Bad News”: Guidelines and Parents’ Preferences

Breaking bad news is a common occurrence in the medical field and something that healthcare providers are often called upon to do. Various models and guidelines exist to help facilitate this process, and there is an assortment of literature on parents’ preferences for how the process should ideally be conducted. A broad definition of bad news is information that adversely alters individuals’ expectations about their current situation and their future (Minichello, Ling, & Ucci, 2007). Since breaking bad news is a difficult skill but one that healthcare providers are often called upon to use, several different models and guidelines on how to approach this type of dialogue are described in the literature (Minichello et al., 2007). Two of these models, the nondisclosure and full disclosure models, are on opposite ends of the spectrum. The former model assumes that it is appropriate for the doctor to decide what news is best for the patient while the latter model assumes the patient has a right to full information. Another model, the individualized disclosure model, is a combination of the full and nondisclosure models and treats the delivery of bad news as a negotiation between doctors and patients as to how much information is desired and subsequently conveyed. This model suggests a tailored approach to breaking bad news. However, determining what information patients want to know can be difficult (Girgis & Sanson-Fisher, 1995). These models are largely based on personal opinions and clinical experience rather than on evidence from systematic study. To that end, medical professionals collaborated to develop guidelines on how to break bad news based on a critical review of the existing literature. These guidelines include ensuring privacy and adequate time, assessing patient understanding,
providing information simply and honestly, encouraging patients to express feelings, discussing treatment options, and providing information about support services (Girgis & Sanson-Fisher, 1995).

Several studies have found similar themes regarding what parents would like to experience during a conversation about their child’s diagnosis (Boyd, 2001; Krahn, Hallum, & Kime, 1993; Myers, 1983; Sloper & Turner, 1993). Parents’ primary preferences include being given detailed and accurate information in language that is easy to understand. Parents also prefer providers to demonstrate warmth, empathy, and compassion when delivering a diagnosis. In addition, parents desire opportunities to ask questions (Boyd, 2001; Krahn et al., 1993; Myers, 1983; Sloper & Turner, 1993) and are more satisfied with their diagnostic experience when they are given these opportunities (Sloper & Turner, 1993). Thus, providers should periodically stop and inquire if parents understand what they are being told and if they have any questions (Myers, 1983). Parents desire information to be given in a balanced manner with providers discussing both positive and negative aspects of the pertinent condition (Krahn et al., 1993; Myers, 1983). The information should also be tailored to each individual family. This can be accomplished by assessing parents’ concerns and understanding throughout the session (Sloper & Turner, 1993). Providers should also outline management and follow-up appointments (Myers, 1983). Resources such as written or printed information on their child’s diagnosis and referral to support groups should be provided at the time of the appointment (Boyd, 2001; Krahn et al., 1993), and providers should also send a follow-
up letter (Myers, 1983). Ideally, parents should also be allowed to display their emotions and not be forced to comprehend more technical information until they are ready. Thus, adequate time is required for the delivery and discussion of a diagnosis (Myers, 1983). Awareness of parents’ preferences and implementation of practices on the part of providers to meet these preferences leads to parents being less anxious and more informed (Boyd, 2001) as well as more satisfied with their diagnostic experience (Sloper & Turner, 1993).

In the context of genetic counseling, specific guidelines for providing a diagnosis of Down syndrome have been developed (Skotko, Capone, & Kishnani, 2009). A postnatal diagnosis of Down syndrome should be shared as soon as the diagnosis is suspected, preferably to both parents at the same time. The news should be given in a private space away from other healthcare providers, patients, and visitors. The individual delivering the news should be knowledgeable about Down syndrome and comfortable answering questions, ideally the child’s pediatrician or mother’s obstetrician. As with a prenatal diagnosis, the primary questions to answer include a discussion about what Down syndrome is and its cause, as well as what the diagnosis means practically for a family. The information provided should be balanced and not vague, inaccurate or outdated. It should also not be overly pessimistic or use offensive terms such as “mongolism.” These guidelines caution that the conversation should be started with positive words because the initial words healthcare providers use frame the atmosphere for the rest of the discussion. The individual delivering the news should also limit the information to the most
immediate or common medical concerns. Thus, limited information should be provided about possible medical conditions later in life like Alzheimer disease. Parents should also be provided with up to date resources including national and local support groups. Finally, the information should be conveyed using nondirective language (Skotko et al., 2009).

A review article on models and recommendations for giving bad news (Harrison & Walling, 2010) references a 1984 study by Cunningham and colleagues that generated a model for delivering a diagnosis of Down syndrome (Cunningham, Morgan, & McGucken, 1984). This model also recommends having a private, appropriate location in which to deliver the news and that the clinician most responsible for management decisions provides the information. In addition, it advises that both parents be informed at the same time and as soon as possible after the diagnosis is confirmed. Providers should consider having the infant/child present and should ensure adequate, uninterrupted time for questions and discussion as well as a private location for family reflection and discussion after the encounter. Follow-up calls and/or interviews should be arranged within days and resources such as written information on the condition and providers’ contact information should be provided. This model has since been revised for general use in other new diagnoses in children (Boyd, 2001; Harrison & Walling, 2010). Finally, a recent communication model was developed based on a study of parental experiences of receiving a genetic diagnosis for their child in a medical genetics clinic. This model combines different factors relating to both parents and providers to suggest a mechanism
by which parents’ diagnostic experiences can be positive or negative. Parental factors are their readiness for and emotional reaction to information. Provider factors are hope and perspective, verbal dominance, attendance to emotions, follow-up plans, and language. Together, these factors determine whether or not parents are empowered to articulate their needs and subsequently whether providers can respond appropriately to parents’ needs (Ashtiani et al., 2014).

How “Breaking Bad News” Occurs in Practice

The literature on parent/patient satisfaction with how healthcare providers break bad news reveals a level of dissatisfaction that is striking considering the many models and advice available on how to go about this process.

Much of the literature on the topic of parent satisfaction is based on studies where parents are receiving a diagnosis of or learning their child is at risk for Down syndrome (Cunningham et al., 1984; Roberts, Stough, & Parrish, 2002; Skotko, 2005). These studies surveyed or interviewed parents about their diagnostic experiences and found the majority of parents had negative experiences. In fact, the majority of women surveyed felt frightened and anxious (Skotko, 2005). Common criticisms from parents included receiving unbalanced descriptions of Down syndrome with an emphasis on negative aspects and a lack of sympathy and concern by the providers who were delivering the diagnosis (Cunningham et al., 1984; Roberts et al., 2002; Skotko, 2005). Parents also reported that the information provided lacked discussion about quality of life issues for
people with Down syndrome (Roberts et al., 2002). A similar tendency was found in simulated sessions where nearly 100% of genetic counselors mentioned the physical and cognitive aspects of Down syndrome while only 63% of genetic counselors discussed quality of life issues (Farrelly et al., 2012). More satisfying diagnostic experiences were associated with parents receiving up to date resources such as parent support groups and being told more positive aspects of having a child with Down syndrome (Skotko, 2005).

Dissatisfaction with communication about a diagnosis of a disability is not limited to Down syndrome. Prior studies interviewing or surveying parents of children with other kinds of disorders or disabilities have identified several prominent themes (Ashtiani, Makela, Carrion, & Austin, 2014; Firth, 1983; Hare, Laurence, Payne, & Rawnsley, 1966; Mulligan, Macculloch, Good, & Nicholas, 2012; Strauss, Sharp, Lorch, & Kachalia, 1995; Tluczek et al., 2006; Waxler, Cherniske, Dieter, Herd, & Pober, 2013; Woolley, Stein, Forrest, & Baum, 1989). One theme is parents’ desire for healthcare providers to be more empathic, show concern, and show appreciation for their worries and anxieties (Ashtiani et al., 2014; Graungaard & Skov, 2006; Hare et al., 1966). Parents want emotional support (Tluczek et al., 2006) and are frustrated when providers do not give them time to talk and show their feelings (Strauss et al., 1995). Parents would like to be given hope that raising a child with a disability is not a completely negative task (Waxler et al., 2013). In addition, more negative diagnostic experiences are associated with parents’ being passive receivers of information rather than active
participants (Ashtiani et al., 2014). Rather, parents want to be seen as co-operating partners with their child’s doctors (Graungaard & Skov, 2006).

Another theme related to parent dissatisfaction is the language used by providers. Parents frequently recall that providers’ language is too difficult to understand (Ashtiani et al., 2014; Woolley et al., 1989) and the information is delivered too quickly leaving little to no time for clarification or for parents to ask questions (Woolley et al., 1989). Additionally, parents report receiving less information than they would have liked (Firth, 1983; Strauss et al., 1995). Parents are also dissatisfied when the information they receive is merely a “problem list,” instead of a discussion about the implications of a genetic diagnosis for the entire family (Waxler et al., 2013). Parents also prefer to be told what their child will be able to accomplish rather than receiving information that focuses on their child’s limitations (Mulligan et al., 2012).

Lack of resource provision such as written materials about the disorder and contact information for other families is another complaint of parents (Mulligan et al., 2012; Waxler et al., 2013). In one study, 16 out of 100 (16%) parents recalled being referred to other families (Strauss et al., 1995). Finally, parents want a concrete follow up/management plan for their child. They desire information about “next steps,” and not being connected with appropriate healthcare providers for ongoing management also leads to frustration and dissatisfaction (Firth, 1983; Waxler et al., 2013).
The Importance of “Breaking Bad News” In a Satisfying Manner

How “bad news” is delivered influences parents’ ideas on what it means to have a child with disabilities as well as parents’ abilities to cope. An initial diagnosis of a neurological disability is a key point at which parents’ perceptions of their child’s disability are defined (McLaughlin, 2005). McLaughlin describes how disability can either be viewed as a tragedy to be avoided at all costs or as limitation of opportunities to participate on an equal level with others. While delivering a diagnosis, providers should frame the child’s condition in a comprehensive manner that acknowledges the child’s abilities and possible futures. In other words, medical professionals should guard against viewing children with disabilities purely through a medical lens (McLaughlin, 2005) since how a diagnosis is delivered can influence parents’ ability to cope (Myers, 1983). Parents’ coping responses upon receiving a diagnosis are in response to a perceived threat. Motoric adaptive responses constitute actions to reduce the threat such as joining a parent support group. Other defense mechanisms include intellectualization, denial and blame. Parents also experience a variety of emotions, with the kind and intensity varying over time, including anger, anxiety, grief, sadness, shame, and guilt. Medical professionals can assist with coping by helping parents not see their child as a defect and helping parents in adapting to their new way of life (Myers, 1983). Providers can further assist with coping by inspiring trust and communicating effectively when delivering a diagnosis (Strauss et al., 1995). Providers can also heed the research done on parents’ preferences for delivering a diagnosis. Tluczek and colleagues developed a theoretical model of parents’ experience of genetic counseling, which included the consequences of matching or mismatching with
parents’ preferences for counseling about their child’s diagnosis. Matching parents’ preferences reduces uncertainty and emotional distress while mismatching with parents’ preferences increases emotional distress and confirms parents’ assumptions about abnormal results, such as their child has a “death sentence” (Tluczek et al., 2006). Affective expressions, particularly negative ones such as anxiety and guilt, influence parents’ perceptions of threat and therefore their coping mechanisms (Myers, 1983). Thus, delivering a diagnosis according to parents’ preferences can help parents avoid unnecessary emotional distress and assist with their coping. A healthcare professional’s counseling also has the potential to positively shape parents’ future parenting behaviors and perceptions of their child’s vulnerability to illness (Tluczek et al., 2006).

Since genetic counselors are medical professionals often at the forefront of providing diagnoses, they have a prominent role in framing disability and helping parents cope with their child’s diagnosis. However, discrepancies between how genetic counselors frame disabilities yet advocate for disability rights are thought to contribute to a tenuous relationship between genetic counselors and the disability community (Madeo, Biesecker, Brasington, Erby, & Peters, 2011). The information genetic counselors provide is likely accurate from a medical standpoint, but the question remains whether the information presented includes a balanced description of positive and negative aspects of conditions (Madeo et al., 2011). Since how a disability is described has long lasting effects beyond the initial counseling session, it is important to conduct further
research into the communication process between genetic providers and parents/guardians at the time of a genetic diagnosis.

Aims

The present study seeks to supplement the literature on communication, “breaking bad news” and the diagnostic experience. This study will qualitatively evaluate the communication process between genetic providers and parents at the time of an initial diagnosis of a genetic disorder in the pediatric setting. This study also aims to analyze parents’ perceptions of the genetic counseling session by surveying parents/guardians after receiving genetic counseling about what they perceived as helpful and not helpful during their counseling session. The goal of this study is to determine the communication strategy used by genetic providers in real-time pediatric genetic counseling sessions and how this strategy compares to communication patterns found in previous research using parent/guardian interviews and simulated patient sessions. In addition, as the majority of research on this topic within the pediatric specialty has been on Down syndrome, the present study aims to explore the communication process across other diagnoses. By assessing parent/guardian perceptions of their genetic counseling experience immediately after their genetic counseling session, this study aims to reduce the bias inherent in studies where perceptions are assessed months or years later. Finally, audio recording pediatric sessions in real-time will assist in understanding how communication patterns compare across genetic counseling specialties.
Chapter 2: Study Design and Methods

Study Design

This prospective study recruited parents of children receiving a new genetic diagnosis. The individuals eligible for study were identified by genetic counselors at Nationwide Children’s Hospital in Columbus, Ohio. Prior to their genetic counseling appointment, potential study subjects were approached and offered an invitation to participate in the study. Participation was voluntary and written informed consent was obtained from the parents/guardians (Appendix A). Children over nine years of age and who had the mental capacity to assent to the study signed an assent form (Appendix B). In addition, genetic professionals who were providing the counseling were also consented to the study (Appendix C). Once consent/assent was obtained, the genetic counseling session was audio recorded.

Following completion of the genetic counseling session, each parent/legal guardian present during the appointment was asked to complete an anonymous survey. The questionnaire, modeled after a published survey used by Waxler and colleagues (Waxler et al., 2013) consisted of demographic questions followed by two free-response questions (Appendix D). The first question asked whether the parents/guardians recalled anything specific that was told to them during the genetics consultation that they found particularly
helpful, supportive, or caring and if so, to list what was said and how they felt about it. The second question asked whether they recalled anything specific that was told to them during the genetics consultation that they felt was not helpful, inappropriate, or insensitive. Again, they were asked to describe what was said and how they felt about it. All study families then received a $10.00 Target gift card as an incentive for their participation in the study. Each audio recording was numbered in the order in which it was recorded (number 1 for the first session recorded, number 2 for the second session recorded, etc.) After recording a session, the audio recording was uploaded from the device onto a password-protected computer for transcription. All audio recorded sessions were transcribed for final data analysis. All personal health information was removed from the final transcripts. The parent/guardian surveys were coded in the same manner as the audio recordings. The Nationwide Children’s Hospital Institutional Review Board approved this study.

Participants

Parents or legal guardians of a child receiving a new genetic diagnosis were invited to join this study. To be eligible for inclusion, the genetic counseling sessions had to involve the communication of an initial diagnosis of a genetic disorder where the diagnosis was obtained either from a clinical assessment or due to identification of a pathogenic mutation(s) that explained the patient’s phenotype. Disclosure of the diagnosis had to be provided by a medical geneticist or a licensed genetic counselor. Sessions involving a diagnosis of Down syndrome or a variant of uncertain significance,
where the parents or legal guardians did not speak English, and where counseling was not provided by a geneticist or licensed genetic counselor were ineligible for inclusion.

Data Analysis

An undergraduate student employed as a research assistant at The Ohio State University transcribed the audio recordings using an online tool from https://transcribe.wreally.com/. Transcripts were then analyzed using NVivo Qualitative Data Analysis software. Nvivo calculates coverage or the percentage of characters coded under a specific theme across all transcripts. Nvivo also determines the number of references, which refers to the number of times a particular theme or subcategory is coded across all transcripts. Researchers, NL and DA, independently coded each session according to an initial codebook. NL and DA met after coding each transcript to compare codes and revise the codebook until reaching greater than 90% agreement and kappa values greater than 0.70. This occurred after coding the first 5 sessions. A final version of the codebook was then agreed upon, and NL coded the remaining sessions. Transcripts were coded using content analysis, specifically a deductive approach where questions derived from other studies and models are the basis for the codebook. During analysis, researchers created different subcategories within the bounds of the questions being asked, following the principles of inductive content analysis. The final codebook consisted of six overarching themes: medical aspects, genetic aspects, resource provision, quality of life, communication, and psychosocial. The research questions guiding our analysis were as follows: what information do genetic providers communicate, do genetic providers
prompt for questions, do parents/guardians ask questions, what psychosocial behaviors do genetic providers exhibit, and what are parents/guardians’ perceptions of the helpfulness of their genetic counseling session.
Chapter 3: Results

Demographics

Researchers recorded a total of 21 sessions. One session was excluded from analysis because it was a provider giving a new diagnosis of Down syndrome. Twenty sessions were transcribed and used for analysis. Across these twenty transcripts, 32 parents/guardians of a child receiving a new genetic diagnosis participated in the study. Two families were invited to participate in the study but declined. Twenty of the parents were females (62.5%) and 12 were male (37.5%). The average age of the child receiving the diagnosis was 5.81 years (range of 3 weeks to 18 years old), and all children were Caucasian. None of the subjects were able to provide assent due to age or their intellectual level. Median household income was $50,740 based on household income information collected from 13 of the 20 sessions.

There were a total of 8 genetic providers audio recorded across the 20 sessions. Of these providers, there were 3 were male medical geneticists, 1 female medical geneticist, 3 female genetic counselors, and 1 male genetic counselor. The average years of practice experience among the medical geneticists was 5-10 years and was 7.25 years for the genetic counselors. The genetic counseling sessions ranged in duration from 10 minutes 1 second to 57 minutes 50 seconds with an average session duration of 28 minutes 32 seconds.
seconds. The subjects’ diagnoses varied, and the categories are listed in Table 1. A molecular diagnosis refers to genetic disorders caused by mutation(s) in single genes. A chromosomal diagnosis refers to partial chromosome deletions and duplications and aneuploidies. A clinical diagnosis is made based on the subject meeting diagnostic criteria with no genetic testing performed.

Overview of the Sessions
The typical structure of a session consisted of brief contracting, which included an assessment of what the parents/guardians remembered about their previous visits/conversations with the genetic providers. The provider then reviewed the genetic test results, which typically included a discussion covering basic concepts such as cells, chromosomes, and genes. The conversation about the genetic test result concluded with a discussion of the specific mutation/genetic cause of the diagnosis. Following the result explanation, the diagnosis including medical issues associated with the disorder and genetic inheritance was conveyed. Follow-up/management was presented and then parents/guardians were given opportunities to ask questions. Resources, such as handouts on the child’s diagnosis and copies of the genetic testing report, were also provided. The genetic providers spoke more than the parents/guardians in 19 out of 20 sessions. The average amount of time the providers spoke was 21.05 minutes (range of 6.63 minutes to 40.46 minutes) versus an average amount of time of 6.60 minutes (range of 0.38 minutes to 19.45 minutes) for parents/guardians.
Major Themes

Four of the six overarching, major themes were present in all of the transcripts (n=20; 100%): genetic aspects, medical aspects, communication, and resource provision. The psychosocial theme was present in nearly all of the transcripts (n=19; 95%), and the quality of life theme was present in the fewest transcripts (n=9; 45%). Table 1 delineates the six themes and the number of transcripts they occurred in.

Each major theme contains subcategories, which are shown in Table 2. The medical aspects theme is comprised of 14 subcategories representing characteristics of a disorder from a medical standpoint. Subcategories include medical issues, treatment, and follow up/management. The genetics aspect theme consists of 9 subcategories referring to genetic characteristics of a disorder. Prominent subcategories include genetic tests and basic concepts. The theme of resource provision has 6 subcategories, which describe the different types of resources given to parents. These resources include handouts on the disorder, copies of the genetic test reports, advocacy group information, referral to other parents, a genetic counseling letter, and invitation to call the genetic provider. The quality of life theme is divided into two subcategories to delineate between quality of life topics brought up by providers and topics brought up by parents/guardians. These topics include lifespan and schooling. The communication theme consists of providers’ and parents/guardians’ question asking behaviors, as well as what kind of language is used to describe medical and genetic concepts. Finally, the psychosocial theme contains 4 subcategories of providers’ behaviors such as empathy and addressing parents/guardians’
concerns. It also contains the subcategory of parent/guardian psychosocial, which was a prominent theme in our data but is not discussed further in our analysis as it did not directly pertain to our research questions.

Medical Aspects

Genetic providers discussed medical information in all transcripts (n=20; 100%). The most common subcategories, appearing in more than 50% of transcripts, related to medical aspects are displayed in Table 3. Medical information was most often presented as medical issues associated with the diagnosis; there were 100 references across 19 transcripts (95%). Information about medical issues was typically coupled with a discussion of follow-up/management (Excerpt 1).

Excerpt 1

Provider: Well about 1/3 to 1/4 of people who have [disorder] have some sort of bleeding difference about themselves...it can be a lot of different things but generally what I recommend is if [patient’s] gonna have some sort of elective surgery to let us know and we’ll do more of a bleeding workup or send you over to hematology.

Subcategories discussed in less than half of transcripts include treatment (n=9; 45%) in the form of therapy, medications, surgery, and/or diet, severity (n=7; 35%), physical characteristics (n=5; 25%) in the form of facial features and/or body habitus, prognosis (n=3; 15%), behavior (n=3; 15%), and other tests performed (n=5; 25%).
Genetic Aspects

Genetic providers discussed genetic information in all transcripts (n=20; 100%). The genetic aspects theme had the highest coverage (37.79%). Table 4 illustrates the most common subcategories related to genetic aspects. The subcategory of genetic tests appeared in every transcript (n=20; 100%) and frequently occurred in conjunction with review of the genetic testing report. The number of references for genetic tests varied depending on the type of testing that was done. For example, patients who underwent whole exome sequencing had over five times as many references for “genetic tests” than patients who had a karyotype. When reviewing the genetic test results, providers frequently used similes to explain basic genetic concepts. Commonly used similes included comparing the cell nucleus to an egg yolk and comparing genes to books in a “genetic library.” Providers also defined genetic terms such as “cis meaning that they are on the same chromosome” and “incomplete penetrance [means] not everyone who has a mutation in this gene will have symptoms.” In addition, providers defined medical terms such as “that bigger blood vessel called the aorta on the top of the heart” and referring to the thyroid as “the master gland that regulates your metabolism.” Other genetic testing subcategories discussed in every transcript include inheritance pattern and specific mutation/genetic cause (n=20; 100%). Subcategories related to genetic aspects that were discussed in less than half of the transcripts include family history (n=6; 30%) and history of disorder (n=3; 15%).
Resource Provision

The theme of resource provision occurred across all transcripts (n=20; 100%). The most common subcategories related to resource provision are displayed in Table 5. No specific type of resource provision occurred across all transcripts. Resource provision subcategories occurring in less than half of the transcripts include discussion of an advocacy group (n=8; 40%) and connecting parents/guardians to other parents who have a child with the same disorder (n=3; 15%).

Quality of Life

Genetic providers discussed quality of life issues in the fewest number of transcripts (n=9; 45%). The most common subcategory of quality of life was schooling (n=4; 20%). Excerpts 2 and 3 in Table 6 illustrate quality of life discussions about schooling.

Discussions surrounding lifespan (n=3; 15%) occurred in three transcripts. Of these three transcripts, the discussion occurred 2 times (66.6%) specifically in response to a parent/guardian concern (Excerpts 4 and 5). In one transcript (11.1%) where quality of life was discussed, it occurred in the context of connecting the parents/guardians to other parents of a child with the same disorder (Excerpt 6).

Communication

The theme of communication occurred in all transcripts (n=20; 100%). The subcategories included in this theme were provider questions and parent/guardian questions and are further described in Table 8 below. Genetic providers’ questions were grouped into three
categories: provider prompting for questions (n=17; 85%; Excerpt 7), provider clarification questions (n=17; 85%; Excerpt 8), and provider assessing understanding (n=14; 70%; Excerpts 8 and 9). Provider clarification questions occurred most often (159 references). Parents/guardians’ questions were divided into female parent/guardian questions (n=17; 85%) and male parent/guardian questions (n=8; 40%). Examples of female and male parent/guardian questions are illustrated in Excerpts 11 and 12. Female parent/guardian questions were more common than male parent/guardian questions (184 verses 45 references). The majority of parent/guardian questions revolved around follow up/management. Two examples describing this are Excerpts 13 and 14 below. Other topics parents/guardians asked about include clarification regarding genetic test results, medical issues, resources, prognosis, and risk to family members.

Psychosocial

Psychosocial behaviors by genetic providers were identified in nearly all transcripts (n=19; 95%). The most common behavior was empathy (n=17; 85%), an example of which is illustrated in Excerpt 15 in Table 9 below. The least common psychosocial behavior was genetic providers directly assessing parent/guardians’ concerns (n=2; 10%). Excerpt 16 is an example of when this did occur. Genetic providers also addressed genetic guilt (n=12; 60%). An example of this is described in Excerpt 17.
Parent/Guardian Survey Responses

The parent/guardian survey consisted of demographic questions followed by two open-ended questions. The first open-ended question asked what the parents/guardians found helpful during their genetics consultation. Twenty-eight parents/guardians (87.5%) provided responses to the first open-ended question. Table 9 contains excerpts of these responses along with the parent/guardian’s gender.

The second open-ended question asked what the parents/guardians found unhelpful during their genetics consultation. Three parents/guardians (9.3%) responded to the second open-ended question. Two responses were from a male and female parent/guardian in the same session. The male parent/guardian mentioned being misled by a medical provider about the genetic testing results. The female parent/guardian from that same session said it was unhelpful when the genetic provider referred to her daughter as being “the lucky one that started the gene.” The third response to question two occurred in a different session and was made by a female parent/guardian relating to not receiving a “definitive answer.” Finally, four parents/guardians (12.5%) circled “no” for both questions, indicating no response.
<table>
<thead>
<tr>
<th>Subjects</th>
<th>n=20</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Gender</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Female</td>
<td>14</td>
<td>70</td>
</tr>
<tr>
<td>Male</td>
<td>6</td>
<td>30</td>
</tr>
<tr>
<td><strong>Race</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Caucasian/white</td>
<td>20</td>
<td>100</td>
</tr>
<tr>
<td><strong>Diagnosis category</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Molecular</td>
<td>13</td>
<td>65</td>
</tr>
<tr>
<td>Chromosomal</td>
<td>6</td>
<td>30</td>
</tr>
<tr>
<td>Clinical</td>
<td>1</td>
<td>5</td>
</tr>
<tr>
<td><strong>Age (y)</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>0-5</td>
<td>13</td>
<td>65</td>
</tr>
<tr>
<td>6-10</td>
<td>2</td>
<td>10</td>
</tr>
<tr>
<td>11-15</td>
<td>4</td>
<td>20</td>
</tr>
<tr>
<td>16-20</td>
<td>1</td>
<td>5</td>
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</table>

Table 1. Study Subjects' Demographics
Figure 1: Major themes verses number of transcripts
<table>
<thead>
<tr>
<th>Medical Aspects</th>
<th>Genetic Aspects</th>
<th>Quality of Life</th>
</tr>
</thead>
<tbody>
<tr>
<td>Physical Characteristics</td>
<td>Family history</td>
<td>Parent/Guardian</td>
</tr>
<tr>
<td>Body habitus</td>
<td>Specific mutation, genetic cause</td>
<td>Lifespan</td>
</tr>
<tr>
<td>Facial features</td>
<td>Inheritance pattern</td>
<td>School</td>
</tr>
<tr>
<td>Treatment</td>
<td>Recurrence risk</td>
<td>Day to day living</td>
</tr>
<tr>
<td>Therapy</td>
<td>Risk to family members</td>
<td>Friendships/relationships</td>
</tr>
<tr>
<td>Medications</td>
<td>Basic concepts</td>
<td>Effects on family</td>
</tr>
<tr>
<td>Diet</td>
<td>Incidence</td>
<td>Provider</td>
</tr>
<tr>
<td>Follow up/Management With genetics</td>
<td>History of disorder</td>
<td>Lifespan</td>
</tr>
<tr>
<td>Other specialties</td>
<td>Genetic tests</td>
<td>School</td>
</tr>
<tr>
<td>Severity</td>
<td></td>
<td>Day to day living</td>
</tr>
<tr>
<td>Development</td>
<td></td>
<td>Effects on family</td>
</tr>
<tr>
<td>Behavior</td>
<td></td>
<td>Independence</td>
</tr>
<tr>
<td>Other tests performed</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Variation</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Medical issues</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Normal test/scan results</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Resource Provision</th>
<th>Communication</th>
<th>Psychosocial</th>
</tr>
</thead>
<tbody>
<tr>
<td>Handout</td>
<td>Visual aids</td>
<td>Provider empathy</td>
</tr>
<tr>
<td>Referral to other parents</td>
<td>Language</td>
<td>Provider hope</td>
</tr>
<tr>
<td>Copy of test reports</td>
<td>Defining medical terms</td>
<td>Genetic guilt</td>
</tr>
<tr>
<td>Genetic counseling letter</td>
<td>Similes, metaphors</td>
<td>Provider assessing concerns</td>
</tr>
<tr>
<td>Advocacy group</td>
<td>Provider question asking</td>
<td>Parent/guardian psychosocial</td>
</tr>
<tr>
<td>Call provider</td>
<td>Assessing understanding</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Clarification questions</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Prompting for questions</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Parent/guardian question asking</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Male</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Female</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Topic</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Insurance</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Follow up/management</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Recurrence risk</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Risk to other family members</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Resources</td>
<td></td>
</tr>
</tbody>
</table>

Table 2. Codebook of Major Themes and Subcategories
<table>
<thead>
<tr>
<th>Subcategory</th>
<th>Number of transcripts</th>
<th>Example</th>
</tr>
</thead>
<tbody>
<tr>
<td>Medical issues</td>
<td>19</td>
<td>&quot;So when you have enough damage to some of the different parts of the brain cells it will predispose you to seizures and so that's kind of a feature of this.&quot;</td>
</tr>
</tbody>
</table>
| Follow up/management        | 16                    | with genetics: "I for sure should see you back here once in a while to see how things are progressing okay? So how about we see you in one year okay?"
other specialties: "I think hearing, especially with some speech concerns and things, is definitely something we should check out…so I can put in that referral to audiology…" |
| Variation                   | 15                    | "And just like any genetic syndrome, just having, you know, this little duplication doesn’t guarantee that he’s going to have all of these. Um we know patients who have this duplication have had these so we know he’s at higher risk to develop some of these features but it doesn’t guarantee that um he will have all of these. Some patients with duplications have lots of symptoms like I said and some have fewer." |
| Development                 | 12                    | "It's associated with global developmental delay even in early childhood, that fits, which stays about the same...static...um slow motor and cognitive gains until and into adolescence and adulthood." |
| Normal test/scan results    | 11                    | "And then finally you guys have already had a kidney ultrasound. Sometimes there are problems with how the kidneys are formed or how the tubes connect the kidneys to the bladder. She had that done and everything there seems to be ok." |

Table 3. Most common subcategories of medical aspects theme
<table>
<thead>
<tr>
<th>Subcategory</th>
<th>Number of transcripts</th>
<th>Example</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genetic tests</td>
<td>20</td>
<td>&quot;And when we take a blood sample to look at the individual chromosomes, we take the blood cells out of the sample, actually open up the cells, and look at them under a microscope. So that was the first test that they did.&quot;</td>
</tr>
</tbody>
</table>
| Inheritance pattern         | 20                    | "Just like everyone else he has two copies of [chromosome]. He just has the one that has the extra piece of genetic information on it. Um every time you have a child you pass one or the other, but not both. It's a total flip of a coin which one you pass. So essentially he has a 50 percent chance each time he has a child which one he passes on."

"Just like everyone else he has two copies of [chromosome]. He just has the one that has the extra piece of genetic information on it. Um every time you have a child you pass one or the other, but not both. It's a total flip of a coin which one you pass. So essentially he has a 50 percent chance each time he has a child which one he passes on."

| Specific mutation, genetic cause | 20 | "Then here is the exact result. So it’s saying that she has 46 chromosomes, 2 X chromosomes, saying she’s female, and then del is for deletion or missing. And then just as [doctor’s name] was describing, the exact area on [chromosome] that told us yes this is why she has [disorder]."

| Risk to family members       | 19 | "Um could it have bearing for your daughter? Well it's not something that we would rush in and test her. If she's not showing any symptoms whatsoever then we would probably just delay it until maybe she was an adult and say well if she wanted to know... she's like I knew my brother had this so I wonder if I could pass it on and maybe I just never showed symptoms."

| Basic concepts               | 19 | "...our body is made up of millions and millions of cells. And each one of our cells, you know, is shaped like an egg and inside the yolk of the egg, the nucleus, is our genetic information, our DNA. And we package our DNA nice and tightly um wound up in the form of these strands called chromosomes."                                                                                                                                 |
| Recurrence risk              | 14 | "... would another baby have a problem like this? It would be extremely unlikely. Your risk to have that is the same as anyone else in the general population. Just because he has this difference doesn't mean that your risk is higher to have another baby with a similar problem."

| Incidence                    | 10 | "...incidence of about 1 in 900 to 1 in 1,000 live births so it's, you know for genetics that's relatively common um compared to most of the things that we see that are really rare."

Table 4. Most common subcategories of genetic aspects theme
### Table 5. Most common subcategories of resource provision theme

<table>
<thead>
<tr>
<th>Subcategory</th>
<th>Number of transcripts</th>
<th>Example</th>
</tr>
</thead>
<tbody>
<tr>
<td>Handout</td>
<td>14</td>
<td>&quot;I printed off a little bit of information about this syndrome.&quot;</td>
</tr>
<tr>
<td>Copy of test reports</td>
<td>14</td>
<td>&quot;Of course you guys are gonna get a copy of it for your records as well&quot;</td>
</tr>
<tr>
<td>Call provider</td>
<td>12</td>
<td>&quot;If anything looks different than anything we talked about, just give us a call and we’ll go over it with you and clarify.&quot;</td>
</tr>
<tr>
<td>Genetic counseling letter</td>
<td>12</td>
<td>&quot;We'll send a letter that kinda summarizes all of this so that you have it, if your kids ever need it for, if someone asks questions about it, it's available to them as well.&quot;</td>
</tr>
<tr>
<td>Excerpt Number</td>
<td>Subcategory</td>
<td>Transcript Excerpt</td>
</tr>
<tr>
<td>----------------</td>
<td>------------------------------------------</td>
<td>---------------------------------------------------------------------------------------------------------------------------------------------------</td>
</tr>
<tr>
<td>2</td>
<td>Schooling</td>
<td>Provider: “Umm one more thing to go over on people with [disorder] and this is the part that’s usually the toughest. Children who have [disorder] will have delays. They’ll have delays in not only what you’re seeing with her rolling and sitting but delays in how they’re able to think and learn. So as time goes by she will learn and acquire things but she’s gonna do it a little bit slower than other kids and she’s likely gonna need help all the way along through her schooling.”</td>
</tr>
<tr>
<td>3</td>
<td>Schooling</td>
<td>Provider: “Where you might be more likely to notice concerns might be getting closer to school age. Whether there's um difficulties with attention you know kinda ADHD stuff or just um needing a little bit of extra help, a little bit of therapy that sort of thing. It's not to say that that definitely will happen […] But most kids that I've seen are able to um...attend a mainstream school as opposed to a special needs school. I can't say that 100% you know that a special needs school wouldn't be completely out of the realm of possibility but most when you do formal IQ testing are gonna be still within that sort of normal range that we…that we accept.”</td>
</tr>
<tr>
<td>4</td>
<td>Lifespan in response to parent/guardian concern</td>
<td>Parent/Guardian: “Yeah, I was just happy that it wasn’t something…that’s gonna… that’s fatal or…she was telling me about a bunch of these—my doctor was telling me about somebody, um, one of her clients that had a syndrome that um, that children normally don’t live to be very old and all that and I was happy that it just wasn’t anything like that.” Provider: “No, it’s nothing like that. So I’m not sure who’s my oldest one at the moment, but, um, people with [disorder] live pretty normal lives in terms of length of life. They will need some help but certainly in terms of length, they’re pretty good, so this is not one that’s more serious where a child will die as an infant or as a you know as a young child.”</td>
</tr>
<tr>
<td>5</td>
<td>Lifespan in response to parent/guardian concern</td>
<td>Parent/Guardian 1: “Do we um...is there any life expect...you know I mean do you know?” Provider: “That’s a good question um I don't know if we have good information on that I mean I think that just with the um I mean we have the progression of the involuntary movements but I think that's the main thing that's actually changing otherwise she's pretty static and so um let's get the doctor's take on that.”</td>
</tr>
<tr>
<td>6</td>
<td>Context of referral to other parents</td>
<td>Provider: “I do have a number of people who have this. If you’re interested, I can ask some of these people if they would be interested in talking to you about it. I can tell you all the medical stuff and the genetic stuff but I don’t have a child with [disorder] so I can’t tell you how things day to day sometimes so…that’s where when you meet other families with children with the same thing, they can provide you with lots of good information…what’s it like every day? What do you go through? How do you manage it?...that can be very helpful.”</td>
</tr>
</tbody>
</table>

Table 6. Quality of Life Excerpts
<table>
<thead>
<tr>
<th>Excerpt Number</th>
<th>Subcategory</th>
<th>Transcript Excerpt</th>
</tr>
</thead>
<tbody>
<tr>
<td>7</td>
<td>Provider prompting for questions</td>
<td>“Do you have any other questions or concerns we didn't address?”</td>
</tr>
<tr>
<td>8</td>
<td>Provider clarification question</td>
<td>“So right now she’s getting therapies monthly?”</td>
</tr>
<tr>
<td>9</td>
<td>Provider assessing understanding of testing process</td>
<td>“Can you guys tell me kind of what you remember from what we’ve done and the process of things so far?”</td>
</tr>
<tr>
<td>10</td>
<td>Provider assessing understanding of basic concepts</td>
<td>“Did you guys get to have biology class in school, you know talking about genes and chromosomes and things like that?”</td>
</tr>
<tr>
<td>11</td>
<td>Female parent/guardian question</td>
<td>“...if she would be the first person to get it, would I be able... like if I have another baby...will that baby have a chance of getting it?”</td>
</tr>
<tr>
<td>12</td>
<td>Male parent/guardian question</td>
<td>“But was the syndrome a contributing factor to her having that stroke and thus causing that damage to her corpus callosum?”</td>
</tr>
<tr>
<td>13</td>
<td>Parent/guardian question about follow up/management</td>
<td>“We will look for the signs and symptoms whatever but other than that we will only see you once a year?”</td>
</tr>
<tr>
<td>14</td>
<td>Parent/guardian question about follow up/management</td>
<td>“And he’ll be needing to get [cardiac MRIs] at what frequency?”</td>
</tr>
</tbody>
</table>

Table 7. Communication Excerpts
<table>
<thead>
<tr>
<th>Excerpt Number</th>
<th>Subcategory</th>
<th>Transcript Excerpt</th>
</tr>
</thead>
<tbody>
<tr>
<td>15</td>
<td>Provider empathy</td>
<td>“There are many people who get very flummoxed when I present stuff like this and people like certainty and dealing with uncertainty is really hard.”</td>
</tr>
<tr>
<td>16</td>
<td>Provider assessing parent/guardian concerns</td>
<td>“But this is kind of the age where people tend to start tailing off [in height], so, I guess the question would be whether that’s something you’re worried about.”</td>
</tr>
<tr>
<td>17</td>
<td>Genetic guilt</td>
<td>“It’s one of those things that’s just purely an accident…that happened…in the formation of her. Ok? It’s not anything that you could’ve done or anything that you did not do during pregnancy that would’ve caused this it’s just pure chance. Ok?”</td>
</tr>
</tbody>
</table>

Table 8. Psychosocial Excerpts
<table>
<thead>
<tr>
<th>Excerpt Number</th>
<th>Parent/Guardian Gender</th>
<th>Response</th>
</tr>
</thead>
<tbody>
<tr>
<td>18</td>
<td>Female</td>
<td>The explanation of what my daughter has was easy to understand; list of symptoms was also helpful.</td>
</tr>
<tr>
<td>19</td>
<td>Male</td>
<td>Everything was described very well with great detail. The list of websites to research will be very helpful.</td>
</tr>
<tr>
<td>20</td>
<td>Female</td>
<td>The counselor was very thorough and explained how the specific mutation could affect our daughter. [Counselor] was very supportive and answered all of our questions.</td>
</tr>
<tr>
<td>21</td>
<td>Male</td>
<td>Clearer picture of what to look for.</td>
</tr>
<tr>
<td>22</td>
<td>Female</td>
<td>I appreciate the details that I was told, and I felt more knowledgeable about my son’s case.</td>
</tr>
<tr>
<td>23</td>
<td>Female</td>
<td>Everything!! They break down everything for me to understand. Always very helpful and answers all my questions!</td>
</tr>
<tr>
<td>24</td>
<td>Female</td>
<td>I felt the doctor was thoughtful and caring about everything.</td>
</tr>
<tr>
<td>25</td>
<td>Female</td>
<td>The few questions we had they explained very well! Done very great—such a great, helpful team!</td>
</tr>
<tr>
<td>26</td>
<td>Female</td>
<td>Information to take is always helpful! Very easy going and understanding.</td>
</tr>
</tbody>
</table>

Table 9. Excerpts of Responses for First Open-ended Question from Parent/Guardian Surveys
Chapter 4: Discussion

Few studies have explored communication across a variety of genetic diagnoses or surveyed parents immediately following their genetics consultation. It is important to examine communication patterns and parents’ perceptions in order to understand the process of genetic counseling and provide the best overall patient care. The primary aim of this study is to analyze the communication patterns between genetic providers and parents/guardians at the time of a genetic diagnosis. This study also aims to analyze parents/guardians’ perceptions of their genetic counseling session.

These data reveal the provider-parent/guardian communication process at the time of a genetic diagnosis is one-sided with an emphasis on highly technical, detailed information. On average, providers talk more than the study participants during the sessions, supporting the idea that genetic counseling is provider driven (Meiser et al., 2008). Providers act as educators and emphasize genetic and medical information about the diagnosis. Thus, it is likely that these particular aspects are deemed to be the most important to discuss with parents in order to help them comprehend their child’s diagnosis. It is also probable that providers are most comfortable communication this information since they are specifically trained in medicine and genetics.
The pattern of provider-driven communication with an emphasis on information giving is consistent with the teaching model of genetic counseling. This pattern is apparent in our study across all the providers and diagnoses. Within the major themes of genetic and medical aspects, providers discuss the subcategories of genetic testing, inheritance, and the specific mutation or genetic cause of the diagnosis in 100% of sessions and medical issues related to the diagnosis in 95% of sessions. While providers give priority to educating parents/guardians on medical and genetic information, they attempt to convey this information in a simple manner. Their strategy involves informing parents/guardians by breaking down complex information. For example, providers frequently used similes to explain concepts related to genetic and medical subcategories such as genes and the function of the thyroid. These concepts are likely not encountered by the parents or guardians on a day-to-day basis, therefore making them difficult to comprehend. Providers’ use of similes appears to be an attempt to aid in parents’ comprehension of complex terms and to enable parents/guardians to better understand their child’s diagnosis. This in turn may lead to parent satisfaction with their diagnostic experience. Since complicated language is problematic and leads to parent dissatisfaction (Ashtiani et al., 2014), the strategy of using similes and definitions suggests providers are aware of the difficulty of understanding medical and genetic concepts. This emphasis on information giving is not unique to our study (Mesier et al., 2008), and the very definition of genetic counseling involves helping individuals adapt to medical and familial implications of disease. Therefore, it is not surprising that the majority of the factual
information provided in the sessions is focused on medical and genetic aspects of the diagnoses.

The genetic providers discussed quality of life issues less often (45% of sessions) than medical and genetic information (100% of sessions). This is consistent with communication patterns found in discussions about Down syndrome (Farrelly et al., 2012; Roberts et al., 2002). The present study excluded a diagnosis of Down syndrome so as to determine whether a similar pattern of emphasizing medical and genetic aspects over quality of life issues occurs in the context of other diagnoses. The data show that this is indeed the case. Discussions about quality of life issues occurred less often across a variety of diagnoses in our study, including partial chromosome deletions and duplications and syndromic disorders such as Beckwith-Wiedemann syndrome and Noonan syndrome. The data show genetic providers communicate quality of life issues less often regardless of the particular diagnosis.

There are several possible reasons for this apparent lack of discussion about quality of life issues. It is possible that providers’ training emphasizing skills required to convey medical and genetic information rather than information that goes beyond these aspects and addresses the patient’s day to day life. Conveying information to parents about quality of life issues requires skills more consistent with the counseling model such as eliciting parents’ concerns and individualizing information giving. Therefore, if providers are not adequately equipped with these skills or are taught to communicate via the
teaching model, then quality of life issues would be less likely to be discussed. Indeed, Kessler writes that genetic counselors who are working professionals may find themselves relying heavily on a teaching approach (Kessler, 1997). Providers’ use of a certain methodology to deliver “bad news” is supported by the fact that the pattern of communication was uniform across our study. If providers were simply choosing to omit quality of life topics, we would expect a more varied communication pattern. Overall, providers’ training may contribute to skewed descriptions of genetic disorders by not emphasizing skills necessary to broach quality of life issues.

It is also conceivable that quality of life issues are discussed less often because providers do not feel comfortable discussing these topics with parents/guardians. This hypothesis is supported by the context in which quality of life issues were discussed in our study. A couple discussions occurred in response to parents/guardians expressing a particular concern such as about a shortened lifespan (excerpt 4) or asking a question about life expectancy (excerpt 5). The provider was more than willing to discuss these topics once they were broached, but the fact that they were brought up by the parents/guardians themselves suggests that providers may have a low level of comfort with discussing quality of life. While the providers readily discussed medical and genetic aspects related to the diagnosis without much prompting by the parents, this did not hold true for quality of life issues.
Providers also may not consider themselves to be the experts on quality of life topics. This is supported by excerpt 6 where one provider defers discussion of quality of life issues to other parents who have a child with the same disorder. The provider asserts that other parents are better able to give details about day-to-day living. This suggests while providers know information about quality of life issues is helpful to parents, they may not feel qualified to provide this information. Instead, they view other parents or guardians as the “experts.” In some sense, this is true: parents/guardians who have a child with a genetic disorder are very knowledgeable about day-to-day issues, but whether they are the best source of quality of life information for parents receiving a new diagnosis is not clear. Given that parents desire information about quality of life issues, including how the diagnosis will affect the family as a whole (Waxler et al., 2013), it is essential for providers to assess from whom parents want this information. Parents may view genetic providers as the experts and expect to learn about quality of life issues during their genetics consultation. On the other hand, parents/guardians may view genetic providers as their source of information solely on medical and genetic aspects. Providers should convey the information that parents expect to the best of their ability; they should not shy away from topics they are less comfortable with but discuss them as thoroughly as possible. Discussing quality of life may make providers uncomfortable as it often involves departing from an educator role and engaging more with parents’ concerns, but building rapport and a relationship with parents should make providers more comfortable broaching quality of life issues. Talking about quality of life may also include referring parents to other families. Providers considering this approach should keep in mind that
not all parents/guardians have the time or resources to connect with other parents. While families can be a good source of knowledge about quality of life, this may not be a feasible avenue for all parents.

Another reason why providers discuss quality of life issues less often is the context of the parents/guardians’ visit. Our study only analyzed the initial discussion about the diagnosis, but not all topics may be broached in the initial discussion of a diagnosis. Indeed, Skotko recommends to only discuss the most immediate and common medical concerns at the time of a diagnosis of Down syndrome (2005). Pediatric genetics care is ongoing, so it is possible that discussions during follow-up appointments include further information about medical and genetic aspects as well as quality of life topics. This is in contrast to the prenatal genetic setting where decision making is time sensitive and providers have a limited number of appointments with patients during which to present information. The pediatric genetics setting is less time sensitive, so providers have more opportunities to discuss issues as they arise, including quality of life. Since our study only includes analysis of initial visits, it is important to consider the availability and likelihood of follow-up discussions between genetic providers and parents/guardians as a means of further communication about quality of life issues. Even though the number of opportunities to discuss quality of life varies depending on the specialty, providers must first and foremost be comfortable and knowledgeable about quality of life topics so that when the opportunities to discuss these arise, parents are provided with accurate and relevant information. Increasing providers’ exposure to individuals with disabilities
during their training is a sure way to accomplish this as it will provide a better understanding of day to day issues families face as well as how genetic disorders impact families as a whole. Providers who share their personal experiences working with individuals with disabilities will also be better able to connect with parents and convey empathy, leading to overall parent satisfaction. Training programs also should consider increasing opportunities for students to participate in advocacy groups and using standardized patient encounters where the focus is on discussing quality of life issues related to a specific diagnosis. Increased exposure to individuals with disabilities, more practice discussing quality of life topics, and assessing parents’ expectations for what information they want during the genetic counseling session are all methods of working towards conveying a more balanced description of a genetic diagnosis.

Genetic providers also communicated information about resources; they gave resources in 100% of the sessions. The most common resource provided were handouts on the child’s disorder and copy of test reports (70% of sessions), while the least common resource offered was a connection to other parents or families (15% of sessions). It is possible that referrals to other families occurred with such low frequency due to the very nature of the diagnoses. For instance, some chromosomal abnormalities have specific breakpoints and varying phenotypes, making connection to other families with a similarly affected child less feasible and/or not as helpful. Similarly, the diagnoses identified via whole exome sequencing in our study are rare diseases, which also would make it difficult to connect with other families. Additionally, providers may not know other
families who are willing to talk to others, so they did not mention it as an option. Finally, providers may also assume that providing the parents with information about advocacy groups will allow for connection to other affected families, so the provider themselves did not make a direct referral to another family. Yet, if this was the case, it is important to note that the discussion of advocacy groups within the counseling sessions also occurred at a low frequency (40% of sessions). Therefore, parents/guardians’ awareness of opportunities to be connected with other parents was not necessarily increased in this manner. It is possible that opportunities to connect with other families may have been described in a summary letter, which we did not have access to for our study. In the end, 85% of the sessions the provider did not make parents or guardians aware of the option to connect with other families, either by a direct referral or advocacy group. If, as discussed earlier, providers see other parents as experts on quality of life issues, then their lack of referrals to these “experts” is puzzling. It is also important to consider that quality of life issues may not be as critical to discuss during an initial visit. Instead, these discussions and referrals may occur during follow-up visits, which we did not record. Overall, it is critical to address the lack of this specific referral, and providers should attempt to facilitate this connection since parents and guardians desire connections to other families with the same diagnosis (Mulligan et al., 2012; Waxler et al., 2013).

Occurring in 60% of the sessions, another common resource given was encouragement to call the genetic provider if the parents had any questions. Establishing this open line of communication likely gives parents/guardians the opportunity to ask questions in the
future, which can aid in coping and adjustment by inspiring trust in the provider (Strauss et al., 1995). This is also important because parents and guardians may or may not have had any background about the results ahead of time, so they may not have had time to prepare questions for the providers during their initial visit. Therefore, providing parents/guardians with contact information gives them a means of asking questions as they arise. Overall, providers in our study gave varying types of resources to all study participants, which likely aid in providing information, helping parents cope, and establishing an ongoing relationship with the genetic provider.

In assessing question-asking behaviors of both providers and parents/guardians, we found the majority of questions asked by providers were clarification questions to aid in the providers’ understanding of the child’s medical situation. Excerpt 8 is an example of a clarification question where the provider asks how much therapy the child is receiving. Providers’ use of close-ended questions like in this example served as a method of engaging parents/guardians and allowing for the exchange of biomedical information. For example, the parent/guardian’s response to the provider’s question about therapies was that the child was receiving weekly therapies. This response gave the provider information used to analyze the current management plan and respond that this was “a pretty good amount of therapy.” Understanding the child’s current management allowed the provider to communicate the most pertinent management information. If the child were not receiving any therapy at all, the provider would likely have spent more time discussing why therapy was needed and its benefits. Instead, in this session, the provider
moved on to discuss behavioral issues associated with the child’s diagnosis. This method of engagement fits with the Reciprocal Engagement Model of information exchange where asking close ended questions is recommended to achieve the goals of informing patients and allowing the counselor to know what information to impart (Veach et al., 2007). In addition, this exchange of information between providers and parents allows parents and guardians to be co-operating partners rather than simply passive receivers of information. Parents and guardians were seen as experts on their child, indicated by the providers asking them questions about their child’s medical care. This dynamic is desired by parents and helps avoid a negative genetic counseling experience (Ashtiani et al., 2014; Graungaard & Skov, 2006).

Parents and guardians were given opportunities to ask questions in 85% of the sessions (Excerpt 7). Interestingly, the majority of study participants’ questions arose when they were not specifically prompted by the provider but instead after a pause in the conversation or after the provider had talked about a particular topic. One could hypothesize that this is due to parents needing time to process information. Thus, providers should not assume parents or guardians do not have any questions if they do not ask when prompted. Models for delivering bad news include providing adequate time for parents/guardians to ask questions (Harrison & Walling, 2010). Providers in our study accomplished this by behaviors such as prompting for questions and leaving time for parents/guardians to process information and formulate questions. Finally, providers assessed parents/guardians understanding in 70% of sessions. This included their
understanding of what kind of testing their child had been through (Excerpt 9) as well as their understanding of basic science concepts (Excerpt 10). These questions likely allow providers to tailor their information giving to the parents/guardians’ level of understanding and assess what information is most pertinent to discuss. This assessment of understanding is also part of models for breaking bad news (Girgis & Sanson-Fisher, 1995). Overall, providers used question-asking behaviors to actively engage parents/guardians in providing information, answering parents/guardians’ questions, and personalizing the information-giving process.

Analysis of providers’ psychosocial behaviors indicates engagement with clients’ feelings through behaviors such as showing empathy and offering hope. Results indicate that the most common psychosocial behavior expressed by providers was empathy (85% of sessions), which is illustrated in Excerpt 14 where the provider empathizes with the difficulties of dealing with uncertainty. Raising the issue of genetic guilt was the next most common psychosocial behavior exhibited by study providers (60% of sessions). In these instances, providers were attempting to assure the parents that there was nothing they did to cause the genetic condition in their child (Excerpt 16). Interestingly, this behavior often occurred while genetic providers were providing genetic information rather than in response to a parent expressing feelings of guilt, suggesting providers may be taught in training to proactively address this issue rather than waiting for it to be brought up by parents. Providers may also be using previous experiences with clients where feelings of guilt at the time of diagnosis were raised to inform how they
subsequently deliver a diagnosis. The least common psychosocial behavior exhibited was the provider directly assessing parent/guardian concerns. Occurring in only 10% of the sessions, it was typically used when attempting to discern whether parents or guardians were particularly worried about an aspect of their child’s diagnosis (Excerpt 15). Empathy is a characteristic desired by parents/guardians who are receiving bad news (Boyd, 2001). However, as providers’ exploration of overall parental concerns was limited, it is conceivable that their actual concerns at the time of the diagnosis may not have been addressed. Further engagement of parents and guardians’ feelings may have occurred if providers directly assessed their concerns more often instead of simply asking if they had questions. Again, given the ongoing nature of pediatric genetics care, providers could conceivably engage more with parents’ feelings during future visits as they face medical issues and deal with the task of caring for a child with a disability.

Overall, our data shows that providers engage on a limited psychosocial level with parents/guardians, and the manner in which they do so involves less emotional and facilitative talk. This type of communication pattern is more consistent with the teaching model of genetic counseling and with healthcare providers’ psychosocial behaviors in other medical specialties (Krippeit, Belzer, Martens-Le Bouar, Mall, & Barth, 2014).

Our survey of parents and guardians after their genetic counseling session aimed to assess their perceptions of the genetic counseling session at the time of diagnosis (Appendix D). To our knowledge, no other studies assessing communication in the pediatric setting have surveyed parents immediately after their genetic counseling sessions. The survey
responses indicate that 87.5% of the parents and guardians found some aspect of their genetic counseling session to be helpful (see Table 10 for example responses). Study participants reported that the genetic providers’ explanations were detailed (Excerpts 19 and 22), thorough (Excerpt 20), easy to understand (Excerpts 18 and 23), and gave a clear picture of the diagnosis (Excerpt 21). This suggests that an emphasis on more technical information, as seen in our study, is appreciated by parents. This information can be perceived as helpful if the information is explained well, perhaps via similes/metaphors such as the ones used by providers in our study. Therefore, the providers’ communication strategy may actually contribute to the helpfulness of genetic counseling. Study subjects also reported receiving resources as helpful (Excerpts 18, 19, 26), which further indicates that providing resources is conducive to satisfaction (Skotko, 2005). In addition, parents and guardians described the providers as thoughtful, caring, and supportive (Excerpts 20, 24). This suggests that providers’ psychosocial behaviors, while more consistent with the teaching model, were noticed and appreciated by study parents and may have contributed to their positive perceptions. Finally, parents and guardians noted that their questions were answered (Excerpts 20, 23, 25), indicating that providers’ addressing questions and concerns is important to parents/guardians and creates a more positive perception. Given our observation that providers did not often directly assess parents’ concerns, one may expect the counseling sessions to not be as helpful. However, parents’ responses indicate that this was not necessarily the case; the questions that parents and guardians asked appear to have been addressed by providers in a satisfying manner.
Only 3 parents/guardians (9.3%) found some aspect of their genetic counseling session to be unhelpful. A female study participant wrote the following: “no definitive answer but also very interesting and hope it is helpful for others in the future.” This parent/guardian had a child with a whole exome sequencing result that found a mutation suggestive of a diagnosis but required testing of other family members to aid in the result interpretation. One could postulate that having the genetic testing process drawn out further explains why she found the result disclosure counseling session to be unhelpful. Perhaps if the provider had explored her feelings this parent/guardian would have been able to express her frustration during the session. This may have led to a more thorough discussion about the need to test other family members and made the genetic counseling session more helpful. A male and female parent/guardian reported unhelpful aspects of the same genetic counseling session. Based on the male parents/guardian’s survey response and the session transcript, a different healthcare provider had previously discussed the symptoms associated with his child’s disorder, and they were in contrast to what the genetic provider discussed with him during the session. The male parent found this miscommunication unhelpful. He did not directly express his frustrations during the session, making it difficult for the genetic provider to acknowledge his confusion. However, if the provider had spent more time assessing parents’ concerns, then this frustration would perhaps have come to the surface and able to be acknowledged via an empathic approach. The female parent/guardian in the same counseling session made a comment on her survey that her child being referred to as “the lucky one” was unhelpful. The provider made this statement while explaining the concept of de novo inheritance.
The parent, however, did not find this description helpful in thinking about her daughter’s diagnosis. Perhaps she felt it was insensitive to insinuate that someone with a genetic disorder is “lucky.” When discussing basic concepts, providers should carefully think through how their words may be interpreted by parents/guardians. Overall, the providers’ communication strategy in our study is conducive to positive parent and guardian perceptions. The finding that the majority of study participants viewed their genetic counseling session as helpful is not supported by previous research where many parents/guardians were dissatisfied with the way they received their child’s diagnosis (Ashtiani et al., 2014; Waxler et al., 2013). It is also important to note that the 3 parents/guardians who found an aspect of their counseling session unhelpful also reported aspects that were helpful. Thus, the unhelpful aspects did not seem to entirely negate the helpfulness of their genetic counseling session. It is also possible, however, that parents/guardians came up with answers for both questions simply because they appeared on the survey. However, if that were the case, we would expect a more equal number of responses to each question. Instead, all parents who responded to the survey described a helpful aspect of their genetic counseling session.
Study Limitations

There are several limitations to this study. First, parents/guardians had often received their child’s genetic test result by telephone prior to their in-person genetic counseling session. As this conversation was not recorded, we cannot determine what type of information was conveyed in this discussion or what questions parents asked at that time. However, it is likely that the family received some “preliminary” counseling and had the opportunity to ask questions. After this initial telephone discussion, parents/guardians may have investigated their child’s diagnosis on the Internet. Therefore, parents/guardians may have been more educated about their child’s diagnosis and less anxious by the time they had their in-person result disclosure genetic counseling session. This could affect their perceptions of the helpfulness of their genetic counseling session and may also explain why parents/guardians tended to not ask questions when they were specifically prompted to do so. Parents/guardians’ emotions may also not have been as negative, thereby giving them a more positive perception of their child’s diagnosis and their genetic counseling experience. Another limitation of the study is that parents/guardians may have thought that the providers would be reading their survey responses (although they were told this was not the case). Therefore, they may have provided mostly positive comments when they in fact found more aspects of their sessions to be unhelpful. In addition, the parent/guardian survey asked about helpful aspects before unhelpful aspects, which may have biased responses. Another limitation is whether audio recording affected providers’ counseling since they knew their sessions were being analyzed. While providers reported forgetting that their session was even
being audio recorded, it is difficult to determine whether they subconsciously changed their communication pattern. This study also involved subjects who were all of Caucasian ethnicity, which makes it difficult to determine whether the providers’ communication strategy would be used for patients of other ethnicities. In addition, a limitation of our study is that the sessions were not video recorded, so we could not assess other factors such as facial expressions, body language, and other nonverbal cues in determining psychosocial behaviors. Furthermore, many parents/guardians of patients with serious quality of life concerns were not invited to participate in the study due to the seriousness and context of the conversation. Thus, our study sample consists of discussions where quality of life issues were less likely to be discussed to begin with. Finally, this study is not generalizable to other populations as only patients at this particular hospital in Columbus, OH were enrolled in the study.
Conclusion

In our study, genetic providers used a communication strategy similar to the teaching model of genetic counseling in real time pediatric genetic counseling sessions. The communication strategy involves the provider acting as educator (consistent with findings by Babul-Hirji et al.) and emphasizing medical and genetic information over quality of life issues. This communication process was found in all transcripts, suggesting this pattern is not unique to a particular diagnosis or provider. Providers defined terms and used similes to make complex topics easier for parents/guardians to understand. We recommend incorporating this into clinical practice as easy to understand explanations were reported by multiple parents to be helpful. In addition, providers give parents/guardians resources to aid in understanding the diagnosis and to establish an ongoing relationship with the parents/guardians. Furthermore, providers’ question-asking behaviors allowed parents/guardians to disclose biomedical information and providers to present pertinent medical and genetic information, which is consistent with the reciprocal engagement model (Veach et al., 2007). Providers also gave parents/guardians opportunities to ask questions. Parents asked questions in every session but not necessarily when specifically prompted by providers. This further supports the importance of providing parents time to process information and ample opportunities to ask questions so they do not leave the session with unresolved worries (Sloper & Turner, 1993). Therefore, providers should create opportunities for question asking and be aware that just because parents do not respond when prompted does not mean they do not have questions. Finally, providers engaged with parents/guardians’ emotions mainly through
empathic statements and proactively addressing feelings of genetic guilt. Directly addressing parents/guardians concerns was limited. While the communication strategy demonstrated in this study is more biomedical than psychosocially focused, parents/guardians’ perceptions of the helpfulness of their genetic counseling session were overwhelmingly positive. In addition, the majority of aspects described by Ashtiani et al. in their diagnostic experience model were present in the providers in our study. These aspects include the use of easy to understand language, outlining follow-up plans, and attending to parent emotions. This suggests the most important factors for a positive parent/guardian experience are present in the providers’ communication strategy and focusing on highly technical, detailed information and less on parents/guardians’ emotions and quality of life issues need not necessarily lead to dissatisfaction. Methods for making discussions more balanced include increasing providers’ exposure to individuals with disabilities during training and opportunities to participate in advocacy groups. Training programs should also consider using standardized patient encounters to make their students more comfortable discussing quality of life issues and using the counseling model. During genetic counseling sessions, providers can assess parents’ expectations as to what information they want to receive in order to have a better gauge of what information to impart.

Future research is needed to determine if a similar communication strategy is used by groups of genetic providers in other pediatric genetics clinics. Other studies might focus on patients from non-Caucasian ethnic backgrounds and/or clinics in other states or
countries. Future research may also include re-contacting the parents/guardians who participated in our study to ask them if they would have preferred more discussion of quality of life topics during their genetic counseling session. It would also be useful to ask the parents/guardians if their perceptions of their genetic counseling session have changed from their initial survey responses. This would help assess whether and how the passage of time affects parents/guardians’ perceptions of genetic counseling. Finally, video-recording genetic counseling sessions would allow for further characterization of providers’ and parents/guardians’ psychosocial behaviors.
References


Appendix A: Parent/Guardian Consent Form

CONSENT TO PARTICIPATE IN A CLINICAL RESEARCH STUDY

STUDY TITLE: An Assessment of Genetic Providers and Parent Communication Patterns in Genetic Counseling Sessions

PRINCIPAL INVESTIGATOR: Dawn C. Allain, MS, LGC

CONTACT TELEPHONE NUMBER: 614-293-9713

CONTACT EMAIL: Dawn.Allain@osumc.edu

SUBJECT’S NAME: ______________________________

DATE OF BIRTH: _________________

NOTE: The words “you” and “your” are used in this consent form. These words refer to the study volunteer whether a child or an adult.
1) INTRODUCTION

We invite you to be in this research study because you are the guardian of a child who is being seen for genetic counseling at Nationwide Children’s Hospital OR you are an adult present at this genetic counseling session.

Participation is voluntary. Using this form as a guide, we will explain the study to you. If you have any questions about the study, please ask. Once you understand this study, we will ask you to decide whether you would like to participate or not. By signing this form, you agree to be in this study. If you do not want to be involved with this study, all regular and standard medical care will still be available to you here or at another institution. You also have the right to leave this study at any time, even if you agree to join now.

If this study involves a child between 9 and 18 years of age, the child will receive an explanation of the study in a separate form, called an Assent form. If they agree to be in the study, they will be asked to sign this form.

You will be given a signed and dated copy of the consent and the assent form.

2) WHY ARE WE DOING THIS RESEARCH STUDY?

This is a study to find out how genetic counselors and parents interact during pediatric genetic counseling sessions. We are interested in how genetic counselors explain information and what kinds of questions parents ask during the session. We are also interested in what is helpful and unhelpful for parents during genetic counseling sessions.
WHERE WILL THE STUDY BE DONE AND HOW MANY SUBJECTS WILL TAKE PART?

The first part of this study will be done at Nationwide Children’s Hospital and we hope to record up to 50 genetic counseling sessions.

The data will be analyzed by research staff at The Ohio State University Division of Human Genetics.

WHAT WILL HAPPEN DURING THE STUDY AND HOW LONG WILL IT LAST?

Your genetic counseling session will be audio recorded using an audio recording device. The recordings will then be transcribed and thematically analyzed by the researchers. You will also be asked to complete a 3 question survey after the genetic counseling session.

The audio recording portion of this study will last as long as your genetic counseling session lasts. The survey will take 5-10 minutes to complete.

WHAT ARE THE RISKS OF BEING IN THIS STUDY?

We believe that there is very little chance that bad things will happen as a result of being in this study.

It is possible that you could feel uncomfortable having the genetic counseling sessions recorded. You may also be uncomfortable answering questions on the survey about your genetic counseling experience. If you do find any of the questions upsetting or do not want
to answer a question, you do not have to, and the study coordinator will be available to discuss this with you further.

Since the initial audio recordings will contain patient information, there is a small risk you could be identified based on the audio recordings. However, the audio recording devices will be stored in a secure, locked location until the recordings are transcribed. When the sessions are transcribed, all patient information will be removed, so you will not be able to be identified in the final transcript. After the sessions are transcribed, the audio recordings will be erased from the devices.

There may be other risks of being in this research study that are not known at this time.

**ARE THERE BENEFITS TO TAKING PART IN THIS STUDY?**

Although there may be no benefit to you from being in this study, we hope to learn something about the genetic counseling process that could help genetic counselors and their patients.

**WHAT ARE THE COSTS AND REIMBURSEMENTS?**

All costs related to the research parts of this study (audiorecording) will be covered by the research team. However, the parts of the study that would be done for routine clinical care (your genetic counseling session) will be billed to you and to your insurance company or third party payer. You may have to pay any costs that the insurance company or third party payer does not pay. The study team will discuss these costs with you.
After having your genetic counseling session recorded and completing the survey, your family will be provided with a $10.00 Target gift card. This is our way of thanking you for taking the time to participate in our study.

8) **WHAT HAPPENS IF BEING IN THIS STUDY CAUSES INJURIES?**

We believe that there is very little chance that injuries will happen as a result of being in this study.

**WHAT HAPPENS IF I DO NOT FINISH THIS STUDY?**

It is your choice to be in this study. You may decide to stop being in this study at any time. If you decide to stop being in this study, simply tell your genetic counselor, and he/she will stop recording the session. Your audio recording will then be erased, and none of your information will be used in the study. If you stop being in the study, there will not be a penalty or loss of benefits to which you are otherwise entitled.

**OTHER IMPORTANT INFORMATION**

If you are an employee of Nationwide Children’s Hospital or the Research Institute at Nationwide Children’s Hospital, your job or performance appraisal will not be affected in any way if you decline to participate or withdraw your consent to participate in this study. The final study results will not be shared with you individually.

Nationwide Children’s Hospital is a teaching hospital and we are committed to doing research. Doing research will enable us to learn and provide the best care for our patients.
and families. You may be asked to participate in other research studies in the future. You have the right to decide to participate or decline to participate in any future studies. We will not share your contact information with researchers outside Nationwide Children’s Hospital.

11) **HOW WILL MY STUDY INFORMATION BE KEPT PRIVATE?**

Information collected for this study may include information that can identify you. This is called “protected health information” or PHI. By agreeing to be in this study, you are giving permission to Dawn Allain and the study staff to collect, use, and disclose your PHI for this research study and for future research purposes (including purposes that are currently unknown) unless otherwise allowed by applicable laws. Information collected is the property of Nationwide Children’s Hospital or one of its affiliated entities or the Sponsor.

The reason why this PHI is collected, and what information will be used is listed below. The PHI will only be shared with the groups listed, but if you have a bad outcome or adverse event from being in this study, the Principal Investigator and staff or other health care providers may need to look at your entire medical records. In the event of any publication regarding this or any future studies, your identity will not be revealed.
The PHI collected or created under this research study will be used or disclosed as needed until the end of the study. The records of this study will be kept for an indefinite period of time and your authorization to use or disclose your PHI will not expire.

**PHI that may be used or disclosed:** Names and voice prints.

**People or Companies authorized to use, disclose, and receive PHI collected or created by this research study:**

- PI and study staff
- The Nationwide Children’s Hospital Institutional Review Board (the committee that reviews all human subject research)
- Nationwide Children’s Hospital internal auditors
- Other research sites: The Ohio State University Division of Human Genetics

Because of the need to give information to these people, absolute confidentiality cannot be guaranteed. Information given to these people may be further disclosed by them and no longer be protected by federal privacy rules.

**Reason(s) why the use or disclosure is being made:**

PHI will be collected because we are audio recording your genetic counseling session. Therefore, if any identifiable information (including the information mentioned above) is mentioned during the genetic counseling session, it will be included on the audio recording device. These devices will be kept in a secure location at Nationwide Children’s Hospital.
When the sessions are transcribed, the PHI will not be included. Thus, the final data that the research team will analyze will not contain any PHI. The recordings will also be erased from the devices once the session has been transcribed.

You may decide not to authorize the use and disclosure of your PHI. However, if it is needed for this study, you will not be able to be in this study. If you agree to be in this study and later decide to withdraw your participation, you may withdraw your authorization to use your PHI. This request must be made in writing to the Principal Investigator at:

Dawn Allain, MS, LGC
Division of Human Genetics
The Ohio State University
2001 Polaris Parkway, Suite 1000
Columbus, OH 43240

If you withdraw your authorization, no new PHI may be collected and the PHI already collected may not be used unless it has already been used or is needed to complete the study analysis and reports.

There is a risk that someone could get access to the information (data) we have collected about you. If those data suggested something serious about your health, it could be
misused. For example, it could be used to make it harder for you to get or keep a job or insurance. The Genetic Information Nondiscrimination Act of 2008 (GINA) says that group and individual health insurers may not use your genetic information to determine whether you are eligible for insurance, how much you have to pay, nor can they request or require that you take a genetic test. We cannot guarantee that this will fully protect you. Your privacy and the confidentiality of your data are very important to us. We will make every effort to protect them.

As stated above, your PHI may be used or disclosed for future research purposes, and as part of such future research purposes, your PHI may even be disclosed to people or entities that are not listed above, such as other researchers not involved with this study, government agencies, research foundations, or pharmaceutical or device companies sponsoring future research. This future research may be related to your medical problem, but it may be related to other diseases or conditions as well. Any future research projects, however, will be reviewed and approved by an Institutional Review Board, which protects the rights, welfare, and safety of human research subjects.

I agree to allow my PHI to be stored and used for future research as described above:

(initial your choice)

______ YES ______ NO
12) WHOM SHOULD I CALL IF I HAVE QUESTIONS OR PROBLEMS?

If you have questions about anything while on this study or you have been injured by the research, you may contact the Principal Investigator, Dawn Allain, MS, LGC at 614-293-9713 Monday – Friday, between 8:00 AM-5:00 PM or by email at Dawn.Allain@osumc.edu

If you have questions, concerns, or complaints about the research; if you have questions about your rights as a research volunteer; if you cannot reach the Principal Investigator; or if you want to call someone else - please call (614) 722-2708, Nationwide Children's Hospital Institutional Review Board, (IRB, the committee that reviews all research involving human subjects at Nationwide Children’s Hospital).

Subject’s Name ________________________________

Date of Birth________________________

SUBJECT or SUBJECT’S PARENT OR PERSON AUTHORIZED TO CONSENT ON BEHALF OF THE CHILD (SUBJECT TO THE SUBJECT’S GENERAL MEDICAL CARE)

I have read this consent form and I have had an opportunity to ask questions about this research study. These questions have been answered to my satisfaction. If I have more
questions about participating in this study or a research-related injury, I may contact the Principal Investigator. By signing this consent form, I certify that all health information I have given is true and correct to the best of my knowledge.

I have been given a copy of the Nationwide Children's Hospital Notice of Privacy Practices. If allowed by law, I understand that my right to any information that is created or collected by Nationwide Children's Hospital for this study can be temporarily suspended if necessary for the purposes of this research project. I also understand that my right to access to this information from this study will be reinstated upon completion of this research unless I have been told by the Principal Investigator that I will not receive study results.

I agree to participate in this study or I give permission for my child to participate in this study. I will be given a copy of this consent form with all the signatures for my own records.

CONSENT SIGNATURES

____________________________
SUBJECT or SUBJECT’S LEGAL REPRESENTATIVE

____________________________
DATE & TIME AM/PM
Permission of the second parent not obtained because (select all that apply):

_____ Not required by the IRB (risk level 1 or 2).

_____ Other parent is deceased.

_____ Other parent is unknown.

_____ Other parent is not reasonably available.

_____ Only one parent has legal responsibility for the care and custody of subject.

________________________________________

PERSON OBTAINING CONSENT

________________________________________

DATE & TIME AM/PM

I certify that I have explained the research, its purposes, and the procedures to the subject or the subject’s legal representatives before requesting their signatures.
Appendix B: Subject Assent Form (ages 9-18 years)

ASSENT TO PARTICIPATE IN RESEARCH

(FOR SUBJECTS 9 YEARS UP TO 18 YEARS OF AGE)

Study Title: An Assessment of Genetic Providers and Parent Communication Patterns in Genetic Counseling Sessions

Study Investigator: Dawn C. Allain, MS, LGC

Subject’s Name: ____________________________

Date of Birth: _____________

You are being asked to be in a research study. Studies are done to find better ways to treat people or to understand things better.

This form will tell you about the study to help you decide whether or not you want to volunteer to participate.

You should ask any questions you have before making up your mind. You can think about it and discuss it with your family or friends before you decide.

It is okay to say “No” if you don’t want to be in the study. If you say “Yes” you can change your mind and stop being in the study at any time without getting in trouble.
If you decide you want to be in the study, an adult (usually a parent) will also need to give permission for you to be in the study.

1. What is this study about?

This study is about what happens during genetic counseling sessions and what people think of genetic counseling sessions.

2. What will I need to do (what will be done to me) if I am in this study?

Your genetic counseling session will be recorded using a machine that records people’s voices.

3. How long will I be in the study?

You will be in the study for as long as your genetic counseling session lasts today.

4. Can I stop being in the study?

You may stop being in the study at any time—just tell the genetic counselor in the room.

5. What bad things might happen to me if I am in the study? You might feel uncomfortable being recorded. Some information about your health might also be seen by people who are not part of the study, but there is a very small chance of that happening.
6. What good things might happen to me if I am in the study? You will not benefit from being in this study but we might learn something about the genetic counseling process that could help others.

7. Will I be given anything for being in this study?
You will not be paid to be in this study.

8. Who can I talk to about the study?
For questions about the study you may contact Dawn by telephone at 614-293-9713 or by email at Dawn.Allain@osumc.edu

To discuss other study-related questions with someone who is not part of the research team, you may contact the Institutional Review Board Office (the group that reviews all human subject research) at 614-722-2708.
Signing the assent form

I have read (or someone has read to me) this form. I have had a chance to ask questions before making up my mind. I want to be in this research study.

______________________________  __________________________  AM/PM
Signature or printed name of subject

Date and time

Investigator/Research Staff

I have explained the research to the participant before requesting the signature above.

There are no blanks in this document. A copy of this form has been given to the participant or his/her representative.

_________________________________
Printed name of person obtaining assent

_________________________________
Signature of person obtaining assent

______________________________  AM/PM
Date and time

This form must be accompanied by an IRB approved consent form signed by a parent/guardian.
CONSENT TO PARTICIPATE IN A CLINICAL RESEARCH STUDY

STUDY TITLE: An Assessment of Genetic Providers and Parent Communication Patterns in Genetic Counseling Sessions

PRINCIPAL INVESTIGATOR: Dawn C. Allain, MS, LGC

CONTACT TELEPHONE NUMBER: 614-293-9713

GENETIC PROVIDER’s NAME: ____________________

DATE OF BIRTH: _________________

NOTE: The words “you” and “your” are used in this consent form. These words refer to the study volunteer whether a child or an adult.

1) INTRODUCTION

We invite you to be in this research study because you are a genetic provider involved in a counseling session at Nationwide Children’s Hospital.

Participation is voluntary. Using this form as a guide, we will explain the study to you. If you have any questions about the study, please ask. Once you understand this study, we will ask you to decide whether you would like to participate or not. By signing this form, you agree to be in this study. If you do not want to be involved with this study, all regular
and standard medical care will still be available to you here or at another institution. You also have the right to leave this study at any time, even if you agree to join now.

You will be given a signed and dated copy of the consent form.

2) **WHY ARE WE DOING THIS RESEARCH STUDY?**

This is a study to find out how genetic providers and parents interact during pediatric genetic counseling sessions. We are interested in how genetic providers explain information and what kinds of questions parents ask during the session. We are also interested in what is helpful and unhelpful for parents during pediatric genetic counseling sessions.

**WHERE WILL THE STUDY BE DONE AND HOW MANY SUBJECTS WILL TAKE PART?**

The first part of this study will be done at Nationwide Children’s Hospital and we hope to record up to 50 genetic counseling sessions.

The data will be analyzed by research staff at The Ohio State University Division of Human Genetics.

**WHAT WILL HAPPEN DURING THE STUDY AND HOW LONG WILL IT LAST?**

The genetic counseling session that you are involved in will be audio recorded.
The audio recording portion of this study will last as long as the genetic counseling session lasts.

**WHAT ARE THE RISKS OF BEING IN THIS STUDY?**

We believe that there is very little chance that bad things will happen as a result of being in this study.

It is possible that you could feel uncomfortable having the genetic counseling sessions recorded. If you don’t want to have the session recorded, you don’t have to, and the study coordinator will be available to discuss this with you further.

Since the initial audio recordings will contain your name and voice, there is a small risk you could be identified based on the audio recordings. However, the audio recording devices will be stored in a secure, locked location when they are not in use. When the sessions are transcribed, all identifying names will be removed, so you will not be able to be identified in the final transcript. After the sessions are transcribed, the audio recordings will be erased from the devices.

There may be other risks of being in this research study that are not known at this time.

**ARE THERE BENEFITS TO TAKING PART IN THIS STUDY?**

Although there may be no benefit to you from being in this study, we hope to learn something that could help others. This includes genetic providers and their patients.

**WHAT ARE THE COSTS AND REIMBURSEMENTS?**
There are no costs or reimbursements for genetic providers involved in this study.

8) **WHAT HAPPENS IF BEING IN THIS STUDY CAUSES INJURIES?**

We believe that there is very little chance that injuries will happen as a result of being in this study.

**WHAT HAPPENS IF I DO NOT FINISH THIS STUDY?**

It is your choice to be in this study. You may decide to stop being in this study at any time. If you decide to stop being in this study, simply tell the genetic counselor, and he/she will stop recording the session. The audio recording will then be erased, and none of your information will be used in the study. If you stop being in the study, there will not be a penalty or loss of benefits to which you are otherwise entitled.

**OTHER IMPORTANT INFORMATION**

If you are an employee of Nationwide Children’s Hospital or the Research Institute at Nationwide Children’s Hospital, your job or performance appraisal will not be affected in any way if you decline to participate or withdraw your consent to participate in this study. The final study results will not be shared with you individually.
Nationwide Children’s Hospital is a teaching hospital and we are committed to doing research. Doing research will enable us to learn and provide the best care for our patients and families. You may be asked to participate in other research studies in the future. You have the right to decide to participate or decline to participate in any future studies. We will not share your contact information with researchers outside Nationwide Children’s Hospital.

11) **HOW WILL MY STUDY INFORMATION BE KEPT PRIVATE?**

Information collected for this study may include information that can identify you. This is called “protected health information” or PHI. By agreeing to be in this study, you are giving permission to Dawn Allain, MS, LGC and the study staff to collect, use, and disclose your PHI for this research study and for future research purposes (including purposes that are currently unknown) unless otherwise allowed by applicable laws. Information collected is the property of Nationwide Children’s Hospital or one of its affiliated entities or the Sponsor.

The reason why this PHI is collected, and what information will be used is listed below. The PHI will only be shared with the groups listed, but if you have a bad outcome or adverse event from being in this study, the Principal Investigator and staff or other health care providers may need to look at your entire medical records. In the event of any publication regarding this or any future studies, your identity will not be revealed.
The PHI collected or created under this research study will be used or disclosed as needed until the end of the study. The records of this study will be kept for an indefinite period of time and your authorization to use or disclose your PHI will not expire.

**PHI that may be used or disclosed:**

Names and voice prints.

**People or Companies authorized to use, disclose, and receive PHI collected or created by this research study:**

PI and study staff

The Nationwide Children’s Hospital Institutional Review Board (the committee that reviews all human subject research)

Nationwide Children’s Hospital internal auditors

Other research sites: The Ohio State University Division of Human Genetics

Because of the need to give information to these people, absolute confidentiality cannot be guaranteed. Information given to these people may be further disclosed by them and no longer be protected by federal privacy rules.

**Reason(s) why the use or disclosure is being made:**

PHI will be collected because we are audio recording the genetic counseling session you are involved in. Therefore, if any identifiable information (including the information mentioned above) is mentioned during the genetic counseling session, it will be included
on the audio recording device. These devices will be kept in a secure location at Nationwide Children’s Hospital.

When the sessions are transcribed, the PHI will **not** be included. Thus, the final data that the research team will analyze will not contain any PHI. The recordings will also be erased from the devices once the session has been transcribed.

You may decide not to authorize the use and disclosure of your PHI. However, if it is needed for this study, you will not be able to be in this study. If you agree to be in this study and later decide to withdraw your participation, you may withdraw your authorization to use your PHI. This request must be made in writing to the Principal Investigator at:

Dawn Allain
Division of Human Genetics
The Ohio State University
2001 Polaris Parkway, Suite 1000
Columbus, OH 43240

If you withdraw your authorization, no new PHI may be collected and the PHI already collected may not be used unless it has already been used or is needed to complete the study analysis and reports.
There is a risk that someone could get access to the information (data) we have collected about you. If those data suggested something serious about your health, it could be misused. For example, it could be used to make it harder for you to get or keep a job or insurance. The Genetic Information Nondiscrimination Act of 2008 (GINA) says that group and individual health insurers may not use your genetic information to determine whether you are eligible for insurance, how much you have to pay, nor can they request or require that you take a genetic test. We cannot guarantee that this will fully protect you. Your privacy and the confidentiality of your data are very important to us. We will make every effort to protect them.

I agree to allow my PHI to be stored and used for the research as described above: (initial your choice)

_____ YES  _____ NO

12) WHOM SHOULD I CALL IF I HAVE QUESTIONS OR PROBLEMS?

If you have questions about anything while on this study or you have been injured by the research, you may contact the Principal Investigator at 614-293-9713 Monday – Friday, between 8:00 AM-5:00 PM.
If you have questions, concerns, or complaints about the research; if you have questions about your rights as a research volunteer; if you cannot reach the Principal Investigator; or if you want to call someone else - please call (614) 722-2708, Nationwide Children's Hospital Institutional Review Board, (IRB, the committee that reviews all research involving human subjects at Nationwide Children’s Hospital).

Subject’s Name __________________________________________

Date of Birth________________________

I have read this consent form and I have had an opportunity to ask questions about this research study. These questions have been answered to my satisfaction. If I have more questions about participating in this study or a research-related injury, I may contact the Principal Investigator. By signing this consent form, I certify that all health information I have given is true and correct to the best of my knowledge.

I have been given a copy of the Nationwide Children's Hospital Notice of Privacy Practices. If allowed by law, I understand that my right to any information that is created or collected by Nationwide Children's Hospital for this study can be temporarily suspended if necessary for the purposes of this research project. I also understand that my right to access to this information from this study will be reinstated upon completion of this
research unless I have been told by the Principal Investigator that I will not receive study results.

I agree to participate in this study or I give permission for my child to participate in this study. I will be given a copy of this consent form with all the signatures for my own records.

CONSENT SIGNATURES

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DATE & TIME  AM/PM

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DATE & TIME AM/PM

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SUBJECT or SUBJECT’S LEGAL REPRESENTATIVE

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DATE & TIME AM/PM

____________________

PERSON OBTAINING CONSENT

____________________

DATE & TIME AM/PM

I certify that I have explained the research, its purposes, and the procedures to the subject or the subject’s legal representatives before requesting their signatures.
Appendix D: Parent/Guardian Survey

Parent/Guardian Survey
Session: _______

Please fill out this survey regarding your genetic counseling experience as accurately and thoroughly as possible. It is estimated to take 5-10 minutes to complete this survey.

1) What is your child’s gender? (please circle)  Male  Female

2) What is your child’s age?  ______ years  ______ months

3) What is your child’s ethnic background? (please circle)
   Caucasian  Asian
   Black or African American  Native Hawaiian or Other Pacific Islander
   American Indian or Alaskan Native  Other (please specify)

4) What is your current household income? (please circle)
   Less than $25,000  $75,000-$99,999
   $25,000-$34,599  $100,000-$149,999
   $35,000-$49,999  $150,000 or more
   $50,000-$74,499  Do not wish to answer

5) What is your gender? (please circle)  Male  Female

6) Do you recall anything specific that was told to you during your genetics consultation at the time you received your child’s diagnosis that you found particularly helpful, supportive, or caring?

   a. Yes → Please describe what was said to you and how you felt about what was said in the box below.
7) Do you recall anything specific that was told to you during your genetics consultation at the time you received your child’s diagnosis that you found not helpful, inappropriate, or insensitive?

a. Yes → Please describe what was said to you and how you felt about what was said in the box below.

b. No

Please return the completed survey to your genetic counselor.

Thank you for taking the time to participate in this study!

Version Date: 06/25/2015