Patients with Intellectual Disabilities: The Genetic Counseling Perspective

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Beth Rosen Sheidley, MS, LGC, Advisor

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Jacquelyn E. Magner

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ABSTRACT

Patients with Intellectual Disabilities: The Genetic Counseling Perspective

A thesis presented to the Genetic Counseling Program

Graduate School of Arts and Sciences
Brandeis University
Waltham, MA

Authors: Jacquelyn Magner, Sharyn Lincoln, Jonathan Picker, Marisa Likhite, Beth Rosen-Sheidley

Genetic counselors are trained to adapt their counseling skills to meet the needs of their patients. However, counselors often lack specific training regarding how to counsel patients with intellectual disabilities (ID), and literature regarding the optimal methods for counseling these patients is sparse. The purpose of this study was to explore genetic counselors’ experiences when counseling patients with ID. We interviewed ten counselors who practice in the U.S. and represent prenatal, pediatric, cancer, adult, and research specialties. We used a qualitative, semi-structured interview guide to gather information about the nature of a genetic counseling session in which the patient had ID, the challenges and feelings that counselors experience, whether counselors are comfortable counseling patients with ID, the ways in which counselors are able to assess their patients’ level of understanding, and suggestions they have for others. The interviews were transcribed and coded using ATLAS.ti software. Overall, the counselors we interviewed reported being comfortable counseling patients with ID. However, they experienced a wide range of emotions including reward and frustration. They also
identified several challenges, including concern that their word choice would lead
patients to feel as though something was “wrong with them” and anxiety about
communicating abnormal test results. Participants identified strategies for assessing their
patients’ level of understanding, which included asking patients to repeat back
information and recognizing when their patients were asking inappropriate questions.
Finally, participants provided suggestions for working with patients with ID, including
simplifying information and talking to patients alone. Our findings suggest that
counselors might benefit from small group training to enhance their ability to work with
this patient population. Future research should focus on examining the perspectives of
counselors in different specialty areas and the experiences of patients with ID who have
pursued genetic counseling.
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INTRODUCTION

As many as three out of every 100 individuals in the United States have intellectual disability (ID) and over 580,000 children between the age ages of 6 and 21 have some level of ID and need special education (National Dissemination Center for Children with Disabilities). Although now referred to as ID, individuals with below average intelligence were once deemed as having “mental retardation” (IQ <70). However, the transition from using ID instead of “mental retardation” has not been accomplished because both federal and state governments still use mental retardation for early care and education, civil and criminal justice, employment, income support, health care, and housing (Schalock et. al. 2007). It is important to understand that both of these terms are still used when referring to individuals with below average intelligence, but ID is becoming more widely accepted.

The definition of ID has long been debated, but most would agree that there is not a clear-cut answer because it is difficult to determine whether someone has a mild form of ID or cognitive abilities that are simply on the lower end of the “normal” spectrum. Although there is debate regarding the definition, ID is currently defined as “cognitive functioning that is significantly below average and begins in the developmental stage” (Accardo et. al. 1998). The diagnosis of ID requires that an individual have significant delay in at least two areas of adaptive functioning, which includes communication, self-direction, health and safety, functional academics, leisure, and work (The American Association of Intellectual and Developmental Disabilities (AAIDD)).
The AAIDD definition is used to define the severity of ID in an individual by delineating how a person functions in two different categories: intellectual functioning and adaptive behavior. Intellectual functioning refers to general mental capacity, such as learning, reasoning, and problem solving and is measured by an IQ test. Any score below 70 indicates that an individual has limited intelligence and falls into the category of having ID.

Adaptive behavior is comprised of three skill types: conceptual, social, and practical. Conceptual skills include language, literacy, money, time, numbers, and self-direction; social skills include interpersonal skills, social responsibility, self-esteem, social problem solving, and the ability to follow rules; practical skills include daily living activities, personal care, occupational skills, healthcare, transportation, scheduling, safety, and the use of money and a telephone. Like intellectual functioning, adaptive behavior can also be measured by standardized tests. However, a child cannot be tested for ID until he is at least five years of age. Once it is determined that an individual has ID, a plan can be created to maximize the strengths and potentials of that individual.

Although standardized tests are able to identify people who have ID, it is not always apparent when speaking to someone that he or she has impaired cognitive ability. According to Finucane (1998), many individuals with ID will try to downplay their disability in an attempt to pass for “normal” in society. These individuals oftentimes do not perceive themselves as having ID, but rather as having a learning disability, if they choose to admit anything at all. They may try to conceal their disability as much as possible and even lie about attending a special school or having special help because they do not want others to know that they are different.
In recent years, there has been an increased recognition that people who have ID are capable of participating in many activities and making decisions on their own behalf. There has been considerable research focused on counseling individuals with ID regarding mental health issues (Greenspan 2006). However, there is a dearth of research regarding genetic counseling for patients with ID.

Many of the strategies that genetic counselors use in a traditional counseling session, such as focusing on information giving and discussing risks, are not effective when working with individuals who have ID (Finucane, 1998). However, Finucane (1998) proposed some key strategies that may be useful to genetic counselors. One strategy is to provide a discussion of important psychosocial issues rather than following the more traditional genetic counseling model. Another strategy is to use visual aids when discussing risks. Re-evaluating the goals of the session can also be useful. Information giving is often the focus of a genetic counseling session, but shifting that focus to the patient’s thoughts and feelings has proven to be a more successful approach when counseling an individual with ID.

As the literature available to genetic counselors on how to counsel patients with ID is extremely limited, genetic counselors currently must rely on resources that do not necessarily pertain directly to genetic counseling. The purpose of this study was to explore genetic counselors’ experiences when counseling patients with ID, in an effort to gain insight into current practice. The long-term goal was to share the challenges that counselors encounter, as well as strategies for working with patients with ID in order to provide guidance for counselors and improved service for patients in this population.
METHODS

Sampling Methods

We conducted qualitative telephone interviews with genetic counselors to examine their experiences counseling patients with ID. Eligible participants were male or female genetic counselors working in any specialty who have counseled at least one patient with ID, regardless of years of experience. Participants were also required to speak fluent English. We offered counselors a $25 gift certificate to Amazon.com as a gesture of appreciation for their time and participation. We recruited genetic counselors by emailing a recruitment notice (Appendix A) to the National Society of Genetic Counselors (NSGC) listserv. Twenty-two counselors responded to the recruitment notice, twelve of whom were initially selected at random to participate.

Design of Interview Questions

We designed a semi-structured interview guide with open- and closed-ended interview questions (shown below and Appendix B) designed to capture the scope of study participants’ views and experiences counseling patients with ID. Questions were designed to ascertain a specific counseling session, counselors’ feelings, challenges they encountered, methods used to assess patient understanding of the information being presented, whether they are comfortable counseling patients with ID, and suggestions they have for other counselors preparing for a session with a patient who has ID.
Interview Guide

1) So, tell me about a counseling situation that sticks in your mind that involved a patient with ID.
   
Prompt: Why was your patient referred?
Prompt: What, if anything, did you know about the patient’s ID in advance?
Prompt: Who came to the session with your patient?
Prompt: Can you describe what the challenges of the session were?
Prompt: How were you able to assess the patient’s competency/understanding of the information?

2) How did you feel about the session at the time?

3) How do you feel about the session now, looking back with hindsight?
   
Prompt: what would/could you have done differently?

4) How comfortable were you counseling a patient with ID?
   
Prompt: (Only for those who answer “yes” to question 5 in the demographic section) How do you think having a relative with ID influenced your experience/the patient’s experience of the counseling session?

5) Was there anything in particular that you learned from the session?

6) Do you have any suggestions for other counselors who are preparing for a session with a patient who has ID?

Data Collection

Before proceeding with interviews, the PI obtained informed consent from participants (Appendix C), as well as background information (Appendix B) that included name, years of experience, area of specialty, contact information, and whether the counselor has a family member with ID. We interviewed study participants for approximately 15 to 30 minutes each via telephone and each interview was audiotaped. A semi-structured approach allowed the interviewee to respond freely and without interruption. We kept our responses and the order of questions flexible, which allowed the interviewee and interviewer to explore ideas as they arose. We generated questions that attempted to capture thoughts through a conversation, although the overall interview content remained consistent such that we did not ask interviewees questions that were beyond the scope of the study. In order to protect participant confidentiality, we kept all records containing identifying information, such as names, email addresses, telephone
numbers, and home or work addresses strictly confidential during the study. All study related documents and materials (consent forms) were kept in a locked, filing cabinet drawer in the Genetic Counseling Program’s secure space and were only accessible to the Principal Investigator (PI) and Beth Rosen-Sheidley (thesis advisor and departmental professor). Any databases containing identifiers were password protected using a password known only to the PI. Upon completion of this study, all data including audiotapes, transcripts, and databases were destroyed. The Brandeis Genetic Counseling Program will keep informed consent documents for five years in a locked filing cabinet, only accessible to Program faculty.

Data Analysis

Upon completion of each interview, the PI sent the audiotape to the transcriptionist using Pando software and the transcriptionist converted them into written transcripts. Due to unforeseen technical problems with Pando, 7 of the initial 12 interview audiotape files were unable to be used for data analysis. The PI therefore contacted nine additional counselors (who had previously responded to the study recruitment notice, but who were not selected in the initial group of twelve counselors) and invited them to participate in the study. Two of the nine counselors were not able to participate, and thus seven additional interviews were conducted, making a total of ten interviews that were ultimately used for analysis in this study.

We imported transcripts as rich text files into the qualitative analysis software, ATLAS.ti (version 5.0), for analysis. The PI determined codes to identify and group sections of text that represented a similar feeling, experience, method of assessing understanding, comfort level, or suggestion that were viewed as significant. After
completing transcript analyses, the PI selected five themes that were indicative of study participants’ most common and significant experiences. The themes selected were:

1. Counselors experience a wide range of emotions when counseling patients with ID
2. Counselors are comfortable counseling patients with ID
3. Counselors encounter many challenges when counseling patients with ID
4. Counselors use a variety of methods to assess patient understanding
5. Counselors employ a variety of strategies when counseling patients with ID
RESULTS

Demographics

As noted previously, we ultimately utilized data collected from 10 genetic counselors as study participants, and who are hereafter referred to as counselors. All of the counselors were female and practice in the United States. At the time of the interviews, two counselors saw only prenatal patients, two counselors saw both prenatal and pediatric patients, three counselors saw only pediatric patients, one counselor saw both adult and pediatric patients, one counselor saw cancer patients, and one counselor worked in research. Table 1 summarizes the counselors’ current type of work setting, years of clinical experience, and whether they reported having a family member with ID.

<table>
<thead>
<tr>
<th>Participant</th>
<th>Work Setting</th>
<th>Years of Experience</th>
<th>Family Member with ID</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Prenatal/ Pediatrics</td>
<td>1</td>
<td>No</td>
</tr>
<tr>
<td>2</td>
<td>Prenatal</td>
<td>4</td>
<td>No</td>
</tr>
<tr>
<td>3</td>
<td>Cancer</td>
<td>2</td>
<td>Yes</td>
</tr>
<tr>
<td>4</td>
<td>Pediatrics</td>
<td>11</td>
<td>Yes</td>
</tr>
<tr>
<td>5</td>
<td>Pediatrics</td>
<td>7</td>
<td>Yes</td>
</tr>
<tr>
<td>6</td>
<td>Pediatrics</td>
<td>25</td>
<td>No</td>
</tr>
<tr>
<td>7</td>
<td>Prenatal</td>
<td>4</td>
<td>No</td>
</tr>
<tr>
<td>8</td>
<td>Adult/Pediatrics</td>
<td>1</td>
<td>No</td>
</tr>
<tr>
<td>9</td>
<td>Prenatal/Pediatrics</td>
<td>17</td>
<td>Yes</td>
</tr>
<tr>
<td>10</td>
<td>Research</td>
<td>7</td>
<td>No</td>
</tr>
</tbody>
</table>

Table 1. Study participants’ current work setting, years of experience, and those who have a family member with ID.
Case Summaries

Each of the counselors was asked to describe a counseling situation that involved a patient with ID. The cases counselors chose to describe represent a broad range of diagnoses and indications. The following vignettes are brief descriptions of the cases that the counselors chose to share with the PI.

Counselor 1: A 17-year-old girl who was living in a group home was referred because she has a 22q11 deletion and there was concern that she was pregnant. The patient experienced difficulty in social situations, anxiety, and had some psychiatric issues. The counselor discussed what it means to have a 22q11 deletion and that it was the cause for some of her symptoms. The counselor also attempted to discuss the recurrence risk for her future children.

Counselor 2: A pregnant woman and her husband, both of whom have ID and previously had a son together, were referred to discuss advanced maternal age and recurrence risk for the baby to have ID.

Counselor 3: A woman in her thirties or forties with ID secondary to a stroke, who lives in a group home, was referred for BRCA1 and BRCA2 testing. Upon completion of the session, the woman seemed to understand the information and the benefits/limitations of testing, but decided to explore insurance coverage before proceeding with testing. The patient returned a month later to have her blood drawn for BRCA1 and BRCA2 testing and could not recall any of the information that was discussed in the first session.

Counselor 4: A pregnant woman and her partner were referred to discuss recurrence risks for ID and Sotos syndrome because the mother had mild ID and the father was diagnosed with Sotos syndrome. When the baby was born, he was also
diagnosed with Sotos syndrome.

Counselor 5: A boy with fragile X, whose mother was also affected, was referred to discuss the diagnosis. The session focused on the information regarding both the mother and her son’s diagnosis because it became aware to the counselor that the mother was not well educated and did not understand a lot of the information about fragile X. The counselor had to figure out what the key things were that she wanted the mother to know and had to explain them in the most simple of terms.

Counselor 6: A 16 or 17-year-old girl with Turner syndrome and mild to moderate ID was referred to discuss the diagnosis. The counselor discussed information about both medical and reproductive issues.

Counselor 7: A patient who was pregnant and had myotonic dystrophy was referred to discuss the diagnosis and risk to the pregnancy. The patient’s mother and the father of the baby’s mother accompanied her to the visit and overpowered the session with interruptions and questions. The patient’s mother and the father of the baby’s mother kept trying to answer all of the questions.

Counselor 8: A man is his twenties was referred for autism and a question of Marfan syndrome. The patient had ID, but his mother was referring to it as autism. The counselor was trying to figure out what he needed, while at the same time being sensitive about his disabilities and acting professionally, while the patient persisted in asking her out on a date throughout the session.

Counselor 9: A young woman in her mid twenties who was pregnant was referred because she had ID and there was concern for the risk to the pregnancy. The father of the baby also had ID.
This counselor also described another case in which a woman with mild ID was pregnant with a baby known to be affected with spina bifida and the patient and her husband came in to discuss their options regarding the pregnancy. The patient was not comfortable choosing termination. However, the husband, who did not have ID, did not feel the same way and began to verbally assault the wife during the session.

Counselor 10: A woman with a 22q11 deletion was referred to discuss her diagnosis.

**Theme 1: Counselors experience a wide range of emotions when counseling patients with ID**

Counselors were asked how it feels to counsel someone with ID. Eight counselors answered this question and eight different emotions were reported, which are listed in Table 2.

<table>
<thead>
<tr>
<th>Feeling</th>
<th>Number of Counselors</th>
</tr>
</thead>
<tbody>
<tr>
<td>Uncomfortable</td>
<td>3</td>
</tr>
<tr>
<td>Rewarding</td>
<td>3</td>
</tr>
<tr>
<td>Frustrating</td>
<td>3</td>
</tr>
<tr>
<td>Difficult</td>
<td>2</td>
</tr>
<tr>
<td>Uncertainty</td>
<td>1</td>
</tr>
<tr>
<td>Scary</td>
<td>1</td>
</tr>
<tr>
<td>Maternal</td>
<td>1</td>
</tr>
<tr>
<td>Fun</td>
<td>1</td>
</tr>
</tbody>
</table>

Table 2. Feelings counselors experience when counseling patients with ID

Several counselors expressed more than one feeling. Three counselors reported feeling uncomfortable when counseling their patients and one of those counselors also mentioned that it was rewarding. Two different counselors commented that it was a rewarding experience. One counselor said,
“I think to me most of the time it’s actually rewarding, because… I like feeling like I have… taught someone something or if I’ve helped them understand something. And when you feel like you do make that connection with someone, and that you’ve been able to successfully explain advanced maternal age or a quad screen to them, that can feel really good, because I think a lot of the times no one else has been able to take the time to really sit down with them and explain things.”
~ Participant 7

Another counselor said,

“What I learned in general from the session was that working with people with ID can actually be very enjoyable. I found myself laughing during the session, and not at her, but with her.”
~ Participant 10

Three counselors reported feeling frustrated. One counselor who was speaking with a woman who has ID at a second visit about BRCA1 and BRCA2 testing said,

“[It] was frustrating for me, because the way that I was approaching it is that when she left she was really following along…some people they just look so confused during the whole session that you can tell that they are having a really hard time with it. But she really seemed like she was following along and understanding this, and so it was really kind of frustrating for me because when she came back in and just really did not remember anything, first of all, I felt like I needed to do the entire session over again. Make sure that she really was told again, reminded of why she was here and what we were talking about.” ~ Participant 3

Three counselors found it difficult to counsel a patient with ID. One counselor said they experienced feelings of uncertainty, one said it was scary, one said they had a maternal feeling, and one counselor said it was fun to counsel someone with ID. The counselor who reported having a maternal feeling said,

“What it feels like for me is sort of maternal, because you’re talking to someone who in certain ways is childlike and in certain ways is adult-like. And they sometimes have adult-like problems and decisions, like if they do find themselves pregnant, or they’re not using birth control and they don’t really understand. So they can have adult-like problems, but you’re talking to them often at a level that you might talk to a child. So I actually find myself feeling sort of maternal towards them in that I care about them.
and I want the best for them, and I do understand their limitations.” ~ Participant 6

All of the feelings that counselors reported may be grouped largely into two categories: positive and negative feelings. The positive feelings include rewarding, maternal, and fun, while the negative feelings include uncomfortable, frustrating, difficult, uncertainty, and scary. We used these two groups of feelings to determine whether there was a difference in years of experience between the positive and negative feelings and whether participants who responded with a negative feeling had less years of experience than those who responded with a positive feeling. In total, there were five counselors who had less than five years of experience. Three of those five counselors responded with negative feelings and the other two responded with both positive and negative feelings. Of the two counselors who had between five and ten years experience, both responded with positive and negative feelings. Of the three counselors who had more than ten years experience, one responded with a positive feeling and the other two participants were not specifically asked the question regarding how they feel when counseling patients with ID (see limitations). To summarize, less experience may correlate with more negative feelings.

Theme 2. Counselors are comfortable counseling patients with ID

Counselors were also asked if they feel comfortable counseling patients with ID. Of the nine counselors who were asked this question (see limitations), eight of the nine said that they were comfortable counseling this patient population. Only one counselor reported that she did not feel comfortable counseling patients with ID. The counselor who reported not feeling comfortable said,

“Honestly, probably not, because I felt like I didn't have as many tools in my genetic counseling toolkit to help them. I felt really stuck at times in the sense of what was the appropriate thing to do” ~ Participant 2
One counselor who reported being comfortable said,

“I think I’m comfortable for the most part, but I think anytime you get too comfortable you miss things. So I do like to prepare a lot...for my cases in general. I like to have background on people. I don’t like to go in cold, because I feel like they’ve been through so much and they’ve told their story so many times that it’s kind of rude not to. So on that level, I make myself as comfortable as I can be, but knowing that anything can change at any moment, so just not getting complacent with what I’m doing.”
~ Participant 8

One of the questions that was asked of counselors prior to the interview was whether they had a family member with ID. The purpose of this question was to determine if the counselors who had a family member with ID were more comfortable counseling this population than counselors who did not have a family member with ID.

Four counselors reported having a family member with ID. Counselor 3 has a cousin with ID that is about eight years younger and the counselor grew up with her. Counselor 4 has two cousins with ID: one has mild ID and the other has moderate to severe ID. However, she never spent a lot of time with either cousin because they live out of the country. Counselor 5 has a cousin in her fifties with severe ID whom she spent a lot of time with growing up. Counselor 9 did not describe her family member with ID, but mentioned that she grew up with her relative.

After analysis, we determined that having a family member with ID did not affect whether the genetic counselors in this study were comfortable counseling individuals with ID. Some of the counselors felt that it was difficult to determine if their family experience had any effect on them being comfortable because they did not have the experience of growing up without that family member. Others were not close with their family member and thus did not think that it affected their comfort level with counseling these patients.
Theme 3: Counselors encounter many challenges when counseling patients with ID

Participants were asked what challenges they face when counseling a patient with ID. A total of nine counselors answered this open-ended question (see limitations). Table 3 lists the challenges and the number of counselors who identified each specific challenge.

<table>
<thead>
<tr>
<th>Challenge</th>
<th>Number of Counselors</th>
</tr>
</thead>
<tbody>
<tr>
<td>Choosing words that would not make the patient feel like they were different</td>
<td>4</td>
</tr>
<tr>
<td>Concern about how to counsel for an abnormal result</td>
<td>3</td>
</tr>
<tr>
<td>Decision Making</td>
<td>2</td>
</tr>
<tr>
<td>Explaining Information</td>
<td>2</td>
</tr>
<tr>
<td>Audience</td>
<td>1</td>
</tr>
<tr>
<td>Family History</td>
<td>1</td>
</tr>
<tr>
<td>Overpowering</td>
<td>1</td>
</tr>
<tr>
<td>Satisfaction</td>
<td>1</td>
</tr>
</tbody>
</table>

Table 3. Challenges counselors face when counseling a patient with ID

The most common challenge that counselors faced was finding the right words to use so that the patient would not feel as if something was wrong with her. The responses of three counselors are provided below:

“...I didn't want to say it in a way that made her feel like something was wrong with her. I wanted to say it in a way that she knew she had this little gene change that was responsible for some of the symptoms that she was having, or behaviors, or whatever you want to call it, and that she had a 50% chance to pass this on to children when she had them, and if they did have these changes it was something that we could help her get services...and those sort of things.” ~Participant 1

“When you're dealing with adults with mental retardation...[the challenge] is how to talk about their disability with them, especially since not all the time, but sometimes, they don't perceive that they have a disability. So how do you talk about a person's disability with them when they don't perceive a disability?” ~Participant 4

“You want to be sensitive about it and you don’t want them to feel any sort of shame or feel bad about anything.” ~Participant 7
Three counselors reported the biggest challenge was concern about how to counsel a pregnant patient if her baby was found to have a problem or how to explain the significance of a diagnosis in a patient. Counselor 2 said,

“I think genetic counselors are really trained quite well to deal with [patients who have ID], but when you’re in a setting where the patient has to make independent decisions about something that will impact their life, whether it’s a prenatal or a cancer session, I think that’s an area where a lot of genetic counselors probably feel lost and could use some guidance about how to proceed in those situations.”

And then later commented,

“My major problem with counseling them was that I was really worried that if the baby had a problem, I didn’t know how we would be able to explain that to them, and explaining to them the different things that each test would test for was almost impossible, because they probably had the intelligence of about a second to a third grader.”

Two counselors reported that it was challenging to determine whether the patient was able to make their own decisions. One counselor said,

“Having experience being around people with developmental disabilities like in the Special Olympics or in past positions doesn’t really prepare you [for] that type of situation where these people have disabilities but are making very important adult decisions, and it can be unclear whether they are capable of making them.” ~Participant 2

Two counselors also reported that it was a challenge trying to find the right words to explain genetic information or a particular concept to the patient. One counselor describes the dialog between her and her patients when trying to describe Down syndrome:

“I didn't know what to do once...[I was] showing them the picture with the extra chromosome, and I'm like, "See how this one has an extra one? And they're like, no, I don't know what you're talking about. And I'm like, Okay, and then went back and said, see all the other ones, you can see one, two, and this one there's an extra? And they were like, No.” ~ Participant 2
Counselor 4 reported having difficulty identifying who the proper audience was to address during the session. She said,

“I think one of the other challenges...is when you're counseling several people together, like a family...and one person has MR and the others don't, is knowing who you're addressing. Who is your audience. Because if you keep it just to the person with MR, the other people in the room might not be getting everything that they want. But if you're talking to them, it might be overwhelming or confusing to the person with MR.”

Challenges that other counselors reported included the patient not being able to provide a detailed family history, talking to the patient when family members become overpowering, and not feeling satisfied that the patient understood the information.

Counselor 7 said,

“She had her mother with her and also the father of the baby’s mother, and so all three of them came back to the session. And it was a challenging session, because she obviously...had mental retardation, and she was able to speak with me, but her mother really took over...both the mothers did. The patient’s mother and then the father of the baby’s mother kept trying to answer all of the questions. And they weren’t even concerned about myotonic dystrophy. They had a lot of other issues that they wanted to talk about...I tried to figure out if the patient’s mother had guardianship over her, or...who was making medical decisions, because I really felt it was important for the patient’s needs to be heard, but she kept getting overshadowed by the other two people there, so it was difficult.”

**Theme 4: Counselors use a variety of methods to assess patient understanding**

We asked participants to describe how they were able to assess whether their patients were able to understand the information that was provided. Seven counselors were able to comment on strategies they used to assess the patient’s level of understanding of complex genetic information. There were a total of five ways in which counselors reported being able to assess their patient’s understanding. These methods included asking the patient to repeat back information, determining if the patient was
asking inappropriate questions, if the patient was able to make connections between the information and the risks or potential implications, paying attention to body language, and if the patient was able to answer questions about the information (Table 4).

<table>
<thead>
<tr>
<th>Method Used to Assess Level of Understanding</th>
<th>Number of Counselors</th>
</tr>
</thead>
<tbody>
<tr>
<td>Repeat Information</td>
<td>6</td>
</tr>
<tr>
<td>Asking Inappropriate Questions</td>
<td>3</td>
</tr>
<tr>
<td>Inability to Make Connections</td>
<td>1</td>
</tr>
<tr>
<td>Body Language</td>
<td>1</td>
</tr>
<tr>
<td>Ability to Answer Questions</td>
<td>1</td>
</tr>
</tbody>
</table>

Table 4. Number of counselors who reported using each method to assess patient level of understanding.

Four counselors reported using more than one method during a session. For example, participants 4 and 9 both reported recognizing that their patients were asking inappropriate questions and asked the patient to repeat back the information. Repeating back the information was the most common method used to assess a patient’s level of understanding, which six of the counselors implemented during the session they described. Three counselors’ responses are provided below:

“I’ll say what do you understand about this? What do you think is going on?” ~ Participant 8

“When I’m particularly concerned about someone’s understanding, often times I jokingly quiz...I’d joke about it, but it was really necessary just to see if certain very minimal things she could recall.” ~ Participant 5

“Okay, so we talked about this, how would you put it into words...or if you’re going to go home and tell this to your husband or tell this to your mom or brother or sister, how would you tell them about this? ~Participant 3

Three counselors mentioned that their patient asked inappropriate questions, which indicated to the counselor that the patient did not fully understand the information. For example, counselor 4 who was meeting with a couple with ID who just had a baby with
Sotos syndrome said,

“Okay, so what do you understand about Sotos syndrome?...Right after I had said it they would repeat it, but then they would ask questions that didn’t really make sense.”

Counselor 8 said,

“A lot of the time...based on the questions they ask you...you have a pretty good idea of where they’re coming from. If their questions totally don’t make any sense from what you just told them, then you know that you need to go back to the drawing board. If they seem to be appropriate questions for the conversation...that’s a pretty good gauge for me.”

Counselor 2 reported that she was able to assess her patient’s level of understanding by recognizing that her patient was unable to make appropriate connections between the information and the potential implications of having a child with ID that was similar to the patient’s. She said,

“In terms of their ability to deal with emotions, in talking to them about if the baby had a problem, what would you do in that situation, it was similar to having that discussion with a child. Most adults are able to think about those things a little bit more. Like connect the dots a little bit more...some people might say “Oh, yeah I know someone who has a baby with disabilities which is really difficult. That might be really difficult for me.” But it seemed like they weren’t able to make this advanced connection between thoughts. They said, no, no, we want to keep the baby no matter what, but I don’t know that they were even thinking about themselves having a disability, or maybe other people they had met with a disability. They didn’t say things like “our son is normal, so could this baby be normal too? They didn’t ask advanced questions. They were very simple.”

Counselor 1 reported that she used her patient’s body language to assess whether or not her patient understood the information that was discussed.

“I walked in and told her about her chromosomes and she wouldn’t really look at me.”

We also asked counselors whether they thought that by the end of the session their patients were able to understand the information that was discussed. Five counselors
reported that their patient was not able to understand the information, while the other five felt that their patients did understand the information. One counselor was talking with a woman who has fragile X syndrome about her own and her son’s recent diagnosis of fragile X. She reported her patient was able to understand the information:

“I fortunately had the opportunity to see her again afterwards. She just showed up at our clinic with some more questions. So I know…that she did get it enough to have additional questions after the fact.” ~Participant 5

One counselor who was counseling a couple, in which both partners had ID, about advanced maternal age and attempting to explain that individuals with Down syndrome have an extra copy of chromosome 21 said,

“They didn’t understand the difference between a karyotype when you have the right number of chromosomes and the one that shows three copies of 21 where you just have a box around the extra chromosome…eventually the husband got it and saw the extra chromosome, and I think that she just said she understood when I don’t think she did. For them to understand that [the chromosome] is in your body and that’s what would cause mental retardation in some cases…I really don’t think that they made that connection. So had that situation been a situation where there was an abnormal screen or markers on ultrasound, I don’t know how we would have discussed that information with them, because just the basic information was really difficult to discuss.” ~Participant 2

Theme 5. Counselors employ a variety of strategies when counseling patients with ID

Counselors were asked at the end of the interview if they had any suggestions for other counselors who are preparing for a session with a patient who has ID. All ten counselors had at least one suggestion, shown in Table 5.

Seven counselors suggested simplifying the information. One counselor met with a pregnant couple regarding advanced maternal age and the risks to the pregnancy of having ID because both the husband and wife had ID. She said,
“I don't think that she actually understands that Down syndrome means someone has an extra chromosome. However, so, I asked, we had talked about things in the broader sense in terms of babies being born with problems that would give them difficulty to learn or health problems, would you want to know about that when the baby was still inside of you, or when the baby came out? And if you found out when the baby was inside of you, if the baby had disabilities or had health problems, would you want to keep the baby or not? We had a discussion like that, and they were very, very adamant that they would never consider ending the pregnancy, and they would never consider giving the baby to someone else to raise the baby, and that they wanted to keep the baby. Based upon that, I felt comfortable that doing an amnio would be probably okay for what they were telling me, but I felt like the discussion of these are chromosomes and this is an amnio, “we talked about the basics of an amnio involved taking fluid out from around the baby and things like that, but I felt like some of the basic information in terms of describing the tests was too advanced for them. I don't think they would have been able to understand, for example, that a quad screen was looking at proteins made by the pregnancy in her blood. All I had really said about that was that you had some blood testing already that showed a lower possibility of the baby having some common issues we could see in older women, but that doesn't tell us for sure. Very, very basic information about everything. And the information that I would normally share with a patient, some of that was missing, because when I tried to discuss they seemed lost.”
~ Participant 2

Five counselors suggested using visual aids to help explain the information, three suggested trying to talk to the patient one-on-one, three suggested asking closed-ended questions, three suggested using analogies to help explain the information, and three suggested not focusing on the information. One counselor said,

“As a student you are learning, so you're trying to learn all of this information and then give it to the patient, but really what the patient wants is maybe like one tenth of what you've prepared, sometimes, or something completely different.” ~ Participant 1

Two counselors each suggested having someone the patient has a relationship with come to the session, asking the patient how she feels about the information and if she has any concerns, asking the patient what she wants to get out of the session, speaking with someone beforehand about the patient’s level of ID and other important information,
providing the patient with written materials to take home or sending them a letter summarizing what was discussed during the session, and taking one’s time during a session. One counselor said,

“Asking the follow-up questions is important. How are you feeling about this? Does this seem like something you’re really concerned about? Because sometimes...you can tell how well people are following along by if they are having the...emotional reactions to the information that you would expect them to have. And so I think that sometimes just checking to see if someone is reacting the way that you might expect is a good indicator too.” ~ Participant 3

<table>
<thead>
<tr>
<th>Suggestion</th>
<th>Number of Counselors</th>
</tr>
</thead>
<tbody>
<tr>
<td>Simplify information</td>
<td>7</td>
</tr>
<tr>
<td>Use visual aids</td>
<td>5</td>
</tr>
<tr>
<td>Get to know the patient</td>
<td>3</td>
</tr>
<tr>
<td>Talk to the patient alone</td>
<td>3</td>
</tr>
<tr>
<td>Ask close-ended questions</td>
<td>3</td>
</tr>
<tr>
<td>Use analogies</td>
<td>3</td>
</tr>
<tr>
<td>Don’t focus on the information</td>
<td>3</td>
</tr>
<tr>
<td>Adjust the goals of the session</td>
<td>2</td>
</tr>
<tr>
<td>Suggest someone to come to the session who know the patient well</td>
<td>2</td>
</tr>
<tr>
<td>Ask the patient how they are feeling</td>
<td>2</td>
</tr>
<tr>
<td>Ask the patient what they want to get out of the session</td>
<td>2</td>
</tr>
<tr>
<td>Speak with someone who knows the patient prior to the session</td>
<td>2</td>
</tr>
<tr>
<td>Provide the patient with written information</td>
<td>2</td>
</tr>
<tr>
<td>Don’t rush/take your time</td>
<td>2</td>
</tr>
<tr>
<td>Be careful with analogies</td>
<td>1</td>
</tr>
<tr>
<td>Surround yourself with individuals who have ID</td>
<td>1</td>
</tr>
<tr>
<td>Read Brenda Finucane's book</td>
<td>1</td>
</tr>
<tr>
<td>Listen</td>
<td>1</td>
</tr>
<tr>
<td>Determine who has power of attorney</td>
<td>1</td>
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</table>

Table 5. Strategies counselors suggest for others preparing for a session with a patient with ID.
One counselor suggested being careful with the use of analogies, another suggested surrounding oneself with individuals who have ID as a way to learn how to best communicate with them, another suggested making sure to listen to the patient, and one suggested trying to figure out beforehand who has power of attorney to make medical decisions for the patient. The counselor who recommended being careful with analogies said,

“I think some genetic counselors are trained to use a lot of analogies, and I think that when you are working with somebody with ID you can lose people. So I think...there are some, and I say that because the program that I teach in has a geneticist who really loves them, and so right away the students start going towards the recipes and the books, or the bricks in the house, or the train and the guy...And it's like whoa, I'm having trouble keeping up and I'm fairly with you. And so I just think that the use of analogies should be used sparingly and with caution, is what I would say. Speaking, that I don't usually speak metaphorically very much with people with ID. I stay present and I stay more concrete.” ~Participant 4

The counselor who suggested using analogies said,

“It’s also...important to have some parallels when you talk with people with ID...even to use the word gene. I often say something like it’s sort of like a factory and it makes something that’s important for your body. So once I say factory, I might even say to them have you ever been to a factory? Like a candy factory. And that candy factory’s job is to make something. And genes are inside your body and their job is to make something that’s important for your body. So to use parallels in everyday life to help them understand something that you feel is important for them to understand, even though it may be a pretty complex topic.”

~ Participant 6
DISCUSSION

The majority of the research that has been published concerning how genetic counselors should counsel patients with ID has been the work of Brenda Finucane. Many of the results found in the current study are consistent with the research that she has published, including several of the recommendations made by counselors and the idea that many individuals with ID try to downplay their disabilities. However, there are a few novel concepts from this study that help to supplement her research. These include the emotions experienced by counselors when they counsel patients with ID, the notion that counselors are comfortable counseling these patients, the challenges they encounter, the techniques they use to assess their patient’s level of understanding, and a handful of suggestions that have not been previously reported.

Theme 1. Counselors experience a wide range of emotions when counseling patients with ID.

The data from this study that supplements the published research includes the emotions experienced by counselors when counseling a patient with ID. The wide range of emotions that counselors experience when they counsel a patient with ID indicates the difficulty in counseling this patient population. It is interesting that the majority of the feelings that counselors reported were negative in connotation, yet almost all of the counselors reported feeling comfortable counseling their patients. Three counselors said they felt uncomfortable during their counseling session, but later indicated they felt comfortable counseling this patient population. These results indicate conflicting
viewpoints among the same counselors and may be explained by several reasons. The cases that each of the three counselors described may have had certain characteristics that made them particularly uncomfortable, but the counselors in general feel comfortable counseling patients with ID. Another explanation is that the cases they described may have been from earlier in their careers when they were less comfortable in general in their role as a counselor. This data also suggest that even though there may be several challenges to this type of session, counselors are comfortable enough with their skills to adapt their sessions to all types of individuals.

When we compared feelings to years of experience, we found that all five of the counselors with fewer than five years of experience felt one of the feelings that were grouped into the negative category (uncomfortable, frustrating, difficult, uncertainty, scary) with regard to counseling patients with ID. The only participant who reported only positive emotions had more than ten years of experience. It appears there may be a trend suggesting that the more years of experience a counselor has, the more positive she feels about counseling patients with ID. One possible explanation for this is that a counselor with more years of experience has likely had more opportunities to counsel individuals with ID. However, there are limited data regarding this question from the other two counselors with more than ten years of experience. Although limited data are available for counselors with more experience, it appears that counselors with less experience are less likely to report positive feelings about counseling patients with ID.

One might assume that counselors who thought their patient understood the information might report more positive feelings, but there was no correlation between counselors who reported positive feelings and those who thought their patient understood
the information. These results suggest that the particular patient and session likely have an influence on how the counselor feels. In addition, the question of how the counselor felt when counseling a patient with ID was asked subsequent to describing a particular counseling session. The counseling sessions that the participants described were presumably the ones that stood out the most in all of their years of experience, and thus were likely cases that were more difficult, challenging, or more emotionally charged for the counselor. Based on this, their feelings may have been targeted to the particular session described and not to all counseling sessions in which the patient has ID.

All of the feelings reported emphasize the point that as counselors, we want the best for our patients and when we encounter a difficult or challenging session, we experience a range of emotions that indicate concern for the patient. As counselors, part of our profession is to help our patients understand the information, and we are trained to use a variety of techniques to accomplish this goal. However, when the techniques are exhausted and it is unclear if the patient really understands the information presented, it is natural to feel concerned or uneasy.

Theme 2. Counselors are comfortable counseling patients with ID

One hypothesis of this study was that counselors who had a family member with ID would be more comfortable counseling this population. However, this was not found to be the case among our study participants. Some counselors had a closer relationship to their family member than others, but regardless of how the close the relationship, there was no correlation between the counselors who had a family member with ID and those who did not. This may be because some counselors have had the opportunity to interact with individuals who have ID during their graduate school training, which allows them to
feel comfortable when they begin counseling patients with ID. Another possible explanation is that counselors are trained to be able to adapt their counseling styles and techniques in order to meet the needs of their patients, and thus they are prepared to counsel any type of patient. Had the results demonstrated that most of counselors were not comfortable counseling patients with ID, there may have been implications for further graduate training in this area. However, the results from this study suggest that even if counselors do not have experience with or have not grown up with someone who has ID, they are still able to feel comfortable counseling patients with ID.

**Theme 3. Counselors encounter many challenges when counseling patients with ID**

Regardless of whether a patient has ID, genetic counselors will always face challenges with certain patients as compared to others. Some of the challenges the counselors in this study reported might be similar to those counselors face in everyday practice. For example, one counselor reported that it was challenging to obtain a family history from her patient, but this challenge may also be faced when someone has difficulty discussing their family history because it may raise an intense emotional reaction or because they were adopted and don’t have any information on their biological family. However, this study suggests that there are several challenges that counselors face when counseling individuals with ID that may be unique to this situation, with the most common challenge being the attempt to explain information in such a way that does not make the patient feel as though something is wrong with her. Counselors are trained to be sensitive to their word choice, but this study suggests that this may be more of a challenge when counseling a patient with ID. An explanation for why this may be the biggest challenge is because many individuals with ID may try to downplay their
disabilities. This phenomenon has been reported previously in the literature (Finucane 1998) and several of the counselors who participated in the current study reported that their patients attempted to downplay their disabilities by referring to themselves as having a learning disability or not acknowledging that they have ID at all. It would likely be difficult to find the right words to describe a disability when the patients do not describe themselves as having one. Descriptions of a disability may cause patients to feel uncomfortable or sense that they are different, especially if they do not perceive themselves as having a disability. It is essential for patients to feel comfortable when talking to a counselor because if they are led to feel differently, then it may be difficult to build rapport and ultimately help the patient understand the information being discussed. One way to overcome this challenge, which several counselors suggested, is by using the words that the patients themselves use to describe their disabilities.

*Theme 4. Counselors use a variety of strategies to assess patient understanding*

There were five different methods that counselors used to try to assess whether their patients were able to understand the information. One of the methods, repeating back information, was the only one in which the counselor actively assessed level of understanding. The other four methods were passive, or nonverbal, and are observations that the counselor made throughout the course of a session. These passive methods are commonly used in genetic counseling sessions and are usually good indicators of whether the patient understands the information. More than half of the counselors reported that they asked their patient to repeat back the information. This method is clearly one that many counselors utilize when counseling patients with ID, and one in which students are taught to add to their handbook of genetic counseling tools. However, it is unclear from
this study whether counselors employ this method more often during sessions in which the patient has ID or whether this technique is used during the majority of counseling sessions. It is likely that asking the patient to repeat back information would be used more often when it is unclear to the counselor whether the patient understands the information being presented because it is a more direct way of determining the patient’s understanding. Therefore, this technique would likely be used more often when counseling patients with ID because they would be expected to have a more difficult time understanding the information, as evidenced by this study.

When using the mentioned techniques, only half of the counselors thought their patients were able to understand the information. If patients are not able to understand the information, then it would be difficult for them to make informed decisions. Although counselors are trained to be non-directive and present their patients with all of their options so that the patients can make their own decisions, it may be necessary to follow a more directive counseling style for individuals with ID. It may also be helpful to request a follow up visit with the patient and an individual who has developed a relationship with the patient or someone who oftentimes helps make decisions for the patient. There may also be other methods for assessing understanding that counselors who were not interviewed in this study find useful and, if identified, may be of aid to counselors.

Theme 5. Counselors employ a variety of strategies when counseling patients with ID.

Many of the recommendations that the counselors in this study proposed are similar to the suggestions reported by Finucane (1998). She suggests providing a discussion of important psychosocial issues rather than following the more traditional genetic counseling model because many of the strategies that are used in a traditional genetic
counseling setting are not compatible when working with patients who have ID. Information giving is often the focus of a genetic counseling session, but shifting that focus to how the patient feels and what she thinks has proven to be a more successful approach. Both of these strategies are ones that were suggested by counselors in this study. However, our study participants also provided additional strategies that may be helpful to counselors, including speaking with the patient alone, speaking to an individual who is familiar with the patient before the session, and to not rush the session.

The most frequent suggestion made by counselors was to simplify the information presented to the patient. It would be expected that individuals with ID would have a more difficult time understanding particular concepts and information, as it is difficult for many people in the general population to understand genetic concepts and information. It is clear from this study that there is a need to dramatically simplify the way in which counselors explain genetic information to ensure that their patients understand the major concepts. As a counselor, it can be frustrating to feel that you have not given a patient all the information. However, in cases where the patient has ID, giving them too much information may leave them feeling confused and overwhelmed. It may be difficult, albeit necessary, to accept that less information can have a greater effect on understanding for patients with ID.

The array of suggestions that counselors provided indicates that there is not one “right” way to counsel patients with ID. Each interview suggests that certain recommendations may be more useful during some sessions as opposed to others, depending on the patient. Conflicting suggestions with regard to the use of analogies support this point. One counselor found that using analogies is beneficial to the patient’s
understanding, while another found that it may not always be beneficial and may actually hinder the patient’s understanding. The counselor who suggested being careful with analogies and two who suggested using analogies had over ten years experience. This supports the point that there is no “right” way to counsel patients with ID and that the strategies used during a counseling session should differ from patient to patient.

*Utilization of genetic counseling services by individuals with ID*

More than ten years have passed since the publications of Finucane, yet much of her research still holds true as evidenced by some of the results in this study, including the recommendations made by counselors and the observation that many individuals with ID try to downplay their disabilities. These ideas may still hold true because there has not been much research on this topic since Finucane’s publications. It is possible that research has not been pursued on this topic because counselors are comfortable with the resources that are available and do not feel there is a need for more. Counselors in this study may have read Finucane’s book as a way to prepare for a session in which the patient has ID, used her strategies, and found they were useful and thus recommended them in this study. As genetic counseling becomes more available, particularly to individuals who have ID, we will be able to better understand the needs of these patients. Perhaps if this study is repeated in another ten years, counselors with have more suggestions, different emotions, and different ways of assessing patient understanding.

It is routine to encounter patients with ID in a pediatric setting, but it is becoming more common to see them in a prenatal or cancer setting. This may be because in the past, women with ID were oftentimes sterilized (Cepko, 1993) and thus were not at risk of becoming pregnant. In addition, cancer genetics has started to evolve over the past
decade. *BRCA1* (Miki et. al., 1994) and *BRCA2* (Wooster et. al. 1994) were not discovered until 1994, and therefore testing for these genes has only been in effect for several years. It is now becoming more recognized that men and women with ID are able to make informed adult decisions and thus we are seeing them more frequently in prenatal and cancer genetic counseling settings. The frequency of referrals of patients with ID may also be growing in these two settings because the medical profession recognizes that there are genetic factors responsible for ID. Therefore, we have ways to test for those genetic factors and can provide more specific recurrence risks if the genetic factor is identified. Although the published literature has proven to be useful to many counselors, there may be strategies that are more useful in a prenatal setting versus a cancer setting versus a pediatric setting, and research in this area may be warranted.

*Study Limitations*

The limitations of this study include sample size, having limited information from several interviews, and inconsistency of questions asked to each participant. We only interviewed ten genetic counselors and thus, there may be other views, opinions, and experiences from other genetic counselors that were not represented in our study population. The information from several initial interviews was rendered unusable for data analysis. The original information obtained included interviews from a handful of counselors that had more years of experience when compared with the counselors included in the current data set. Because the participants in the first set of interviews overall had more experience than the counselors in the second set, they seemed to be able to generalize more from their experiences. However, there was still substantial data to be used for this study.
Another study limitation was the manner in which particular questions were asked. For example, some counselors were asked how they feel when they counsel patients with ID, while others were asked the question, “How do you feel when you counsel people with ID? Is it frustrating, is it rewarding, etc?” The latter is more leading and may have influenced the participants’ responses.

One other important limitation to this study is that not every counselor was asked every question on the interview guide. The interview guide was meant to serve as a tool to help direct the interview with each participant. The interview was designed to flow as a conversation and not as a transcript of questions and answers. Therefore, there were a couple of questions that were not asked of every counselor, which explains why some questions do not have data from all ten participants.

A final study limitation is the validity of the responses obtained from counselors. It is difficult to determine how honest counselors were in their responses. It might be difficult for someone to admit that they feel uncomfortable or they feel like they do not have the necessary tools to counsel a patient with ID.
CONCLUSION

Interviews with genetic counselors regarding current practices for counseling individuals with ID indicate diverse perspectives and practices. There is still a great deal to be learned about how to best provide genetic counseling to individuals with ID. However, previous literature and the results of this study provide counselors with resources to help them prepare for cases in which the patient has ID. The most notable results from this study were those regarding the ways in which counselors are able to assess their patients’ level of understanding, the challenges counselors face, their level of comfort, and suggestions they have for other counselors.

The results of this study are useful for both new and experienced genetic counselors who are preparing to counsel a patient with ID. New counselors may find this information especially helpful because counselors are often not trained how to counsel patients with ID and may feel more at ease if they have some additional resources during their case preparation. Experienced counselors may find this useful because it provides the perspectives of other counselors and may help validate their own practices or provide additional techniques for them to use in their counseling sessions. Almost all of the counselors that were interviewed expressed interest in hearing the results of this study. This suggests that many counselors are interested in hearing others’ thoughts and they would like to improve their own counseling skills for this patient population. They may also not be aware of Finucane’s research or they would like to validate their own techniques and feelings.
Future studies could survey a larger group of genetic counselors that represent various specialties and years of experience. It would be interesting to see whether responses would vary depending upon either of these factors. Also useful could be a focus group in which ideas could be shared amongst genetic counselors. Finally, it would perhaps be most informative to speak with the patients who have ID who underwent genetic counseling and understand what their experiences were like. This may help genetic counselors better understand the goals, expectations, feelings, and challenges that this patient population experiences.
REFERENCES


APPENDIX A: RECRUITMENT NOTICE

Are you a genetic counselor who has experience counseling a patient with intellectual disabilities?

I am a graduate student in the Genetic Counseling Program at Brandeis University. I am conducting a study to explore experiences of genetic counselors when counseling patients with intellectual disabilities.

Participation is open to all genetic counselors who have counseled a minimum of one patient with intellectual disabilities. Participation in this study is voluntary, and participants will be asked to take part in an audiotaped phone interview that will last approximately 30 minutes. All identifying information of participants will be kept confidential and will be destroyed after completion of the study.

I plan to interview a maximum of 12 genetic counselors, and each will receive a $25 gift card to Amazon.com upon completion of the interview.

If you are interested in participating in this study, please contact me by email at Jem328@brandeis.edu.

I appreciate your willingness to participate in this study and look forward to hearing from you.

Sincerely,

Jacquelyn Magner
Genetic Counseling Graduate Student
Brandeis University
Waltham, MA
APPENDIX B: INTERVIEW GUIDE

Background Information Questions
1) Name
2) Contact Information
3) Specialty
4) Years of experience
5) Do you have a family member with intellectual disabilities?

Interview Questions
7) So, tell me about a counseling situation that sticks in your mind that involved a patient with intellectual disabilities.
   Prompt: Why was your patient referred?
   Prompt: What, if anything, did you know about the patient’s intellectual disabilities in advance?
   Prompt: Who came to the session with your patient?
   Prompt: Can you describe what the challenges of the session were?
   Prompt: How were you able to assess the patient’s competency/understanding of the information?
8) How did you feel about the session at the time?
9) How do you feel about the session now, looking back with hindsight?
   Prompt: what would/could you have done differently?
10) How comfortable were you counseling a patient with intellectual disabilities?
    Prompt: (Only for those who answer “yes” to question 5 in the demographic section) How do you think having a relative with intellectual disabilities influenced your experience/the patient’s experience of the counseling session?
11) Was there anything in particular that you learned from the session?
12) Do you have any suggestions for other counselors who are preparing for a session with a patient who has intellectual disabilities?
Informed Consent to Participate in Research

Patients with Intellectual disabilities:
The Genetic Counseling Perspective

Principal Investigator: Jacquelyn Magner
Faculty Sponsor: Beth Rosen-Sheidley, MS, LGC

INTRODUCTION
Jacquelyn Magner is a graduate student in the Genetic Counseling Program at Brandeis University conducting a research study to learn more about genetic counselors’ experiences with patients who have intellectual disabilities. Beth Rosen-Sheidley is a Professor of the Practice at Brandeis University and Co-Director of Research and Professional Development for the Brandeis University Genetic Counseling Graduate Program.

You are being invited to participate in this study because you are a genetic counselor who has experience counseling a patient (or patients) with intellectual disabilities.

Your participation in this study is completely voluntary. You should not feel any pressure to participate. You can decide to stop taking part in this research study at any time for any reason.

Please read all of the following information carefully and ask any questions that you have about this research study. Do not sign this consent form unless you understand the information in it and have had your questions answered to your satisfaction.

If you decide to take part in this research study, you will be asked to sign this form. You will be given a copy of the signed form. You should keep your copy for your records. It has information, including important names and telephone numbers, to which you may wish to refer in the future.

PURPOSE OF STUDY
The purpose of this study is to explore genetic counselors’ experiences with counseling patients who have intellectual disabilities. We hope that the counseling experiences shared by the genetic counselors participating in this study will be a useful and guiding tool for other genetic counselors.

PROCEDURES TO BE FOLLOWED
You will be asked to participate in a 30 minute audiotaped phone interview. During this interview, you will be asked questions regarding your experience(s) counseling a patient or patients with intellectual disabilities.

RISKS
Participation in this study presents no more than minimal risk. However, it is possible that by taking part in this interview, you may experience thoughts or feelings that are upsetting to you. Should that occur, Beth Rosen-Sheidley is available to talk with you.

BENEFITS
There will be no direct benefit to you for your participation in this study. We hope that in the future, information obtained from this study will help us gain a better understanding of the experiences that genetic counselors have had when counseling individuals with intellectual disabilities.

ALTERNATIVES
An alternative is to not participate in this research study.

PRIVACY AND CONFIDENTIALITY
All records containing identifying information, such as names, email addresses, telephone numbers, and home or work addresses will be kept strictly confidential during the study. All study related documents and materials (including consent forms, interview transcripts and audiotapes) will be kept in a secure location accessible only to the Principal Investigator, and any databases containing identifiers will be password protected using a password known only to the Principal Investigator. Transcripts, interview notes, and audiotapes will be labeled with a coded ID number, which will be assigned to you upon enrollment into the study. If you are quoted or referred to in any written or oral reports of the study, you will be given an alternate name. You will never be referred to by your real name or any other identifying information in any written or oral reports based on the interview.

PAYMENT
You will receive a $25 gift certificate to Amazon.com for participation in this research study as a gesture of appreciation for your time and expertise.

COST
There will be no cost to you to participate in the study, other than the time it takes to conduct this interview.
WHOM TO CONTACT
If you encounter any problems related to study participation or have questions about the study, you may contact the Principal Investigator, Jacquelyn Magner, at jem328@brandeis.edu or (203) 952-4227.

You may also contact the Brandeis University Faculty Sponsor for this project, Beth Rosen-Sheidley, at sheidley@brandeis.edu (or by phone at 781-736-2336).

If you have questions about your rights as a research study subject you may contact the Brandeis Committee for Protection of Human Subjects by email at irb@brandeis.edu, or by phone at 781-736-8136.

PARTICIPANT’S STATEMENT

I have read this consent form and have discussed with Jacquelyn Magner the procedures described above. I have been given the opportunity to ask questions, which have been answered to my satisfaction. I understand that any questions that I might have will be answered verbally or, if I prefer, with a written statement.

I understand that my participation is voluntary. I understand that I may refuse to participate in this study. I also understand that if, for any reason, I wish to discontinue participation in this study at any time, I will be free to do so.

If I have any questions concerning my rights as a research subject in this study, I may contact the Brandeis Committee for Protection of Human Subjects by email at irb@brandeis.edu, or by phone at 781-736-8136.

I have been fully informed of the above-described study with its risks and benefits, and I hereby consent to the procedures set forth above.

I understand that as a participant in this study my identity and data relating to this research study will be kept confidential.

____________________________________________
Date Participant’s Signature

____________________________________________
Date Principal Investigator’s Signature
APPENDIX D: INTERVIEW TRANSCRIPTS

Interview 1

JM: You had mentioned to me that you'd counseled a 17-year-old girl with a 22Q deletion. So, if you could tell me about that -- when she was referred, any details about that. Anything you remember.

Interviewee: When I saw the patient, I was still a student, because I had less than 1 year. So I saw her, it would have been the second semester of my second year, and I was doing the patients all by myself then, with of course a counselor sitting in there. And so when I went in there, I of course, being a student, had prepared everything on 22Q, basically had a list of all the symptoms of what they could have, and was really prepared to talk about any of that, and then when I walked in there, the patient wouldn't really look at us, and she was uncomfortable being there in the first place.

JM: Could you tell that by her body language?

Interviewee: Yeah. She was looking down at the floor. Just what I remember -- I walk in, she was sitting against the wall. And so we tried to sit close where we could show her the chromosome changes and those sort of things, but not too close. And the doctors sat in as well, and they were also concerned at the time if she was pregnant or not and was hiding it, because she was living in a group home. Didn't have contact with her family, was really having a hard time with the social situations and they were worried about her being pregnant, not taking care of herself, those sort of things. So, at the same time we were trying to educate her on this 22Q deletion, there was all these other things going on. So figuring out if she was pregnant, figuring out where she was going to live, what she was going to do with her life, and she was really just overwhelmed. Now, looking back on it, she probably could have cared less to be there at all that day. And I'm walking in there and bound and determined to tell her all about her chromosome change and so on and so forth when really I guess we should, we ended up giving her the bare minimum that we could, with, which was what she needed at the time. So, I walked in and told her about her chromosomes and she wouldn't really look at me. And it was uncomfortable for me, too, because I was a student and I was being graded at the same time. So after I kind of, I just told her that this change could be responsible for some of the symptoms that you're having, and she was having anxiety, psychiatric issues, those sort of things. And one of the things that counselor I was with told me at the time that I was really trying to be careful of was to not call them, you know, "This is the reason you are the way you are," basically. I was trying to stay away from that, so I was trying to say "symptoms," or "some of the challenges that you're having," and that was the hardest part for me was finding a good way to say "We think this is why you are having the difficulties that you are right now." So overall, she really didn't, I don't think she ever got it. So if I could
have done something differently, I probably would have just asked what she thought right away. Like, "What do you think?" And I think we did that a little bit, but I'm just trying to remember [?] [5:41]. Because she just kept looking down. I just remember that a lot. And she, when I showed her the picture of the chromosomes and things, I don't think she asked a single question. And -- I'm trying to think -- I just remember that the doctor that I work with, Dr. [Bueller?] [6:03] just piping in and he just straight up asked her, "Are you pregnant? Are you blah-blah-blah," because otherwise she just wasn't responding to us at all.

JM: So it was when you asked pointed questions that she responded more?

Interviewee: Yeah. She was very withdrawn. I mean, she was having such psychiatric issues that she was just very withdrawn. She'd had a horrible, horrible home life, like had been abused. So there was a lot going on. And I don't know if that was, if her family had the same gene change or not, because at that point we didn't know that [?] [6:45]. We just knew that she had these significant psychiatric issues. I can't remember if she had a heart defect or not.

JM: Did anyone come to the session with her?

Interviewee: Yes. The group home person, who knew nothing about her.

JM: So they probably weren't very helpful, then

Interviewee: Exactly.

JM: How long had she been in the group home?

Interviewee: She had lived there a year.

JM: So maybe not enough time to develop a relationship with that person?

Interviewee: Yeah. Well, and I think they switched all the time. I think different people work each week in the group home. And I don't think -- so you know, the way they do it here is they'll have a Mom. They live in the same house together, but I don't think that’s who brought her to the appointment. I think it was someone who weekly comes and works.

JM: Who was available that day?

Interviewee: Who knew nothing about her, yeah. So that made it worse. Sorry, my brain's sort of coming to light as to all this...

JM: So there were any other challenges in the session?

Interviewee: The biggest challenge for me at the time was just finding the right way to
say it and try to explain it to a way that I would feel comfortable that I had at least told her what was going on and what the recurrence would be. The scariest thing for me at the time was I didn't want to say it in a way that made her feel like something was wrong with her. I wanted to say it in a way that she knew she had this little gene change that was responsible for some of the symptoms that she was having, or behaviors, or whatever you want to call it, and that she had a 50% chance to pass this on to children when she had them, and if they did have these changes it was something that we could help her get services to help with the kids and those sort of things. And that was the most uncomfortable part for me, because I just, I didn't know how to say it at the time. I don't even know now, if I'd walk in and I'd be a lot better or not.

JM: Have you counseled anyone since then who's had an intellectual disabilities?

Interviewee: Yes. Well, I guess, do you consider Asperger's and those sorts of things?


Interviewee: Because we see those patients, but they also have their legal guardian, so I guess they don't really consider that. Like, we have, we had a patient the other day. We don't know what her exact gene change is, but she has self-injurious behaviors. She's about 24. Clearly has severe psychiatric concerns. Lives in a group home as well. And she came in with her Mom, and they wanted to find if there was a gene change, and she's telling us all these things that she does to herself and we were just talking to her, I guess, because she's not even her own legal guardian. But this other girl was different, because she was in a way responsible for herself.

JM: Did you find out if she was pregnant?

Interviewee: They, I think they did do a pregnancy test later, and I do not remember if she was pregnant or not. But she was gaining an immense amount of weight, and so that's why they were concerned about that.

JM: Do you have any suggestions for other counselors who are preparing for a situation like this? If you went back, is there anything you would have done differently? Do you have any suggestions for other counselors?

Interviewee: Like, newer counselors, I probably would have just watered down the information that I was ready to talk about it. I would have showed her the chromosomes, I would have shown her the little change, and I just would have given her the basic information that I could at the time. And it's not that I wanted to treat her like she was not as smart as other people or things like that, but it's just the amount of being overwhelmed was achieved faster in her than I think in others.

JM: Did you show her the chromosomes and the gene change during the session?

Interviewee: I did. I showed her the exact picture. I showed her a picture of cells and
compared them to cities. Did the whole analogy, things like that, so I was sure she got it, and I think she did, but it just seemed too overwhelming. It was too much to throw at her at one time. And they were trying to figure out these other things about her. It just seems like you kind of need a one on one setting, then have your doctor go in later. That’s probably what I would do. Separate it out. And have somebody there, ask somebody to come with them who knows what's going on, but that’s not always possible.

**JM:** Did you know her situation before you saw her as a patient?

**Interviewee:** Yeah. We did. And in a lot of those group homes or things that refer us patients, you can't really get around that. The doctor will get what I consider kind of gung ho on figuring out a genetic cause for their syndromes and send them in, and it'll be the patient who's had severe mental retardation or whatever for years, and this case worker who knows nothing about them.

**JM:** I've seen a few cases like that in my pediatric rotation.

**Interviewee:** And the parents sometimes are uninvolved. Like in that girl's case, her, I think she had been. I'm trying to think if she actually had lived with her folks and they put her out into that system. I think that might have been her. Which makes it even more difficult. And I think there was abuse there, too. So, that stuff all complicates that.

**JM:** Do you think her age played into it?

**Interviewee:** Yeah. I mean, she was clearly in between adulthood and teenage years. So that in it can be a challenge, you know, at that age. So yeah, I do.

**JM:** Anything else you want to share about that situation or other situations?

**Interviewee:** I'm trying to think of something that's actually helpful that I can tell you. I guess the one thing I've learned is just to, as a student you are learning, so you're trying to learn all of this information and then give it to the patient, but really what the patient wants is maybe like one tenth of what you've prepared, sometimes, or something completely different. So, I guess maybe just trying to listen to the patient and figure out what they want the most before you throw a bunch of information at them.

**JM:** I find you're so focused on all the information and you feel you need to get it all out, but sometimes it's not right for that situation.

**Interviewee:** Yeah. Exactly. Like you forget to look at the patient when they're looking at you very confused when you're spouting off percentages. Yeah. That would probably be the biggest thing. Because I think that patient, I should have just walked in with about one tenth of the information that I gave her.

**JM:** As a student you need to do what your supervisor wants, too.
**Interviewee:** Exactly.

**JM:** Do you think it would have been different if you didn't have your supervisor with you?

**Interviewee:** If I did it now, it would be different. I don't know. She stepped in and tried to, after I counseled the patient, my supervisor basically stepped in and did the entire thing over again. And I just think that I didn't do anything wrong. It was just that we clearly knew that the patient wasn't getting what we were saying. She was just not getting it. I mean, I think she understood that she had something wrong with her chromosomes, and it was responsible for why she was in a group home, but I don't think she got any more than that.

**JM:** Did your supervisor explain things differently?

**Interviewee:** No. She basically repeated what I said. So, I didn't feel bad or anything. It was just an awkward session the whole time.

**JM:** Were you expecting it to go differently?

**Interviewee:** I was scared, because I didn't, I had never seen a patient who I actually had to counsel about a gene change who actually has a syndrome and nobody else was there. That was different. You see little kids and their folks are there. And talking to a little kid, they know what's going on, too, and [you make sure?] [16:23] that you talk to them, but just when the, it was an adult responsible for herself, it was different. Because nobody else was there to hear the information. We didn't even know how to, who we should send, who she wanted us to send letters to and things like that, besides just who had referred her. If there was other people in the family. She couldn't give us a family history. Those sort of things.

**JM:** Did she give you any information?

**Interviewee:** She tried. She was kind of like a yes-no person. That’s what I meant with the direct questions. Dr. [Bueller?] would say, "Is blah-blah-blah this going on?” She'd be like, "Yes," "No," "I don't know," "I haven't talked to them, I don't know.” That was kind of throughout the whole session.

**JM:** That’s pretty much all of my questions unless you have anything else to add, or have any questions for me.

**Interviewee:** I just think that they can tell you all the things that they want to tell you to do, but when you walk in there and the patient absolutely just doesn't get it, I don't know how you're supposed to treat them the same way that you treat everybody else. I mean, how are you supposed to say, "Oh, they absolutely understand that"? There's no way that they understand the same thing that a person with an IQ of 160 understands. There's just no way. So I think if you at least let them know they have something and that they have,
the thing that they might pass on, I mean, I don't know how you, what you can do to do a better job. I'll be interested to see your study and see what other people have suggested, because I just have no idea.

**JM:** When you counsel someone who doesn't have an intellectual disabilities, they ask questions or nod their head.

**Interviewee:** So other people you've talked to, have they said the same thing, the patient didn't respond?

**JM:** I talked to someone this morning who is a cancer counselor. She counseled someone who lived in a group home but she was her own advocate. And the counselor had spoken with the Mom, and the Mom had told her she made all her own decisions. She thought the patient really seemed to understand what she was talking about. She used visual aids. I think it really depends on the patient. And she was a little older, too. So it depends.
Interview 2

JM: Could you tell me about the session and how it went? The challenges, how you felt about it.

Interviewee: I'm a prenatal counselor, and so the patient and her husband had come in to discuss both maternal age and the fact that both of them have developmental disabilities. So, I was just kind of surprised about how difficult it was to counsel them about the basic information. Like, for example, showing them, talking about what a chromosome problem was. They didn't understand the difference between a karyotype when you have the right number of chromosomes and the one that shows 21 where you just have a box around the extra chromosome. It took them a while to get that that picture had an extra chromosome. My major problem with counseling them was that I was really worried that if the baby had a problem, I didn't know how we would be able to explain that to them, and explaining to them the different things that each test would test for was almost impossible, because they probably had the intelligence of about a second to a third grader. So ultimately everything ended up, I think they had already had a [?] screen and their ultrasound was normal, and they had said that they didn't want an amniocentesis, but it was really challenging, because it ended up being a very basic discussion in the end where I said "If your baby had a problem, would you want to know about it in the pregnancy by doing my testing?" And they were like, "No, whatever happens happens." They knew that they had disabilities, they called it. So I was very, very basic and said "The baby might have similar disabilities as the two of you, but we don't really know why you have them." I think I'd offered them genetics, and they made an appointment, but they didn't keep two different appointments with the geneticist to do an evaluation on them. So by some miracle they already had one child, which was surprising to me that they were able to care for a child on their own, but maybe their ability to care for a baby was more advanced than their ability to have a discussion about their testing options.

JM: Do you know if they had any support?

Interviewee: They think their parents help them a little bit, because that was who was with their son when they were at their appointment. But they weren't there with them at their appointment to assist them with making decisions, and they didn't have any sort of social worker or anything like that coming along with them to the appointment, whereas sometimes situations like that there might be a social worker.

JM: Did you know before the session that they had intellectual disabilities?

Interviewee: That was one of the reasons for the referral, but I guess I didn't understand the extent of how difficult it was going to be to counsel them, because sometimes when it says that on the referral it could be the patient is mildly delayed, but you are still able to have a normal discussion with them. But this one really stood out in my mind, because I didn't know what to do once, like you were showing them the picture with the extra chromosome, and I'm like, "See how this one has an extra one?" And they're like, "No, I
don't know what you're talking about.” And I'm like, "Okay," and then went back and said, "See all the other ones, you can see one, two, and this one there's an extra?" And they were like, "No.” And then I was like "Do you have a problem...Are you able to see well without glasses?” And they said "Oh, no, yeah," and then eventually the husband got it and saw the extra chromosome, and I think that she just said that she understood when I don't think she did. For them to understand that that is in your body and that’s what would cause mental retardation in some cases, I don't, I really don't think that they made that connection. So had that situation been a situation where there was an [?] [6:18] or markers on ultrasound, I don't know how we would have discussed that information with them, because just basic information was really difficult to discuss.

**JM:** Did you feel comfortable counseling them?

**Interviewee:** Honestly, probably not, because I felt like I didn't have as many tools in my genetic counseling toolkit to help them. I felt really stuck at times in the sense of what was the appropriate thing to do. I did end up offering them to meet with a social worker in the hospital, which they thought was a good idea. To make sure that everything was okay in that respect, and I felt like that was why I passed some of that on to her, the social worker, because I didn't feel like I had the capability of knowing with the was the appropriate thing to do next. So but the patient actually ended up declining the meeting with the social worker, so that was a little difficult.

**JM:** If you had to do the session again, is there anything that you would have done differently, or wish you had done, or anything you would change?

**Interviewee:** Looking back, I probably would have explored more about, I know a both of them didn't, I had asked some questions about "Do you work during the day?" and they said that they don't work because they are both on disability. I might have just probed about more questions in terms of their social situation and how they were taking care of their son and how they'd take care of this baby. But maybe that's just my own thing that I felt a little worried about their ability to care for a baby throughout the entire discussion probably made it difficult for me to counsel them.

**JM:** Do you have any suggestions for a counselor preparing for a session with a person with an intellectual disabilities?

**Interviewee:** I think being around people with intellectual disabilities makes you better at knowing how you might communicate with them. I had experience at Brandeis being with children with intellectual disabilities in my training, and then before I was working at this position I saw adult genetics, so there were probably a handful of adults with developmental disabilities we saw to evaluate, figure out the reason for why they had disabilities. But that was a different situation in those cases, because they often came with their parents or their caretaker to the appointment, so this was a unique situation I think in a prenatal setting where the individuals are almost functioning like children, but they're having their own children. And even having experience being around people with developmental disabilities like in the Special Olympics or in past positions doesn't really
prepare you have that type of situation where these people have disabilities but are making very important adult decisions, and it can be unclear whether they are capable of making them.

JM: During the session when you were explaining the genetic concepts to them, you said that the Dad at the end was able to understand it.

Interviewee: Well, he could understand the very basic picture that people with Down's syndrome had an extra chromosome. To see that, he could see that in the picture with [?] versus a normal karyotype that there was actually an extra chromosome, but his wife didn't really get that.

JM: How did you assess that he understood?

Interviewee: I kind of got away, I don't think that she actually understands that Down's syndrome means someone has an extra chromosome. However, so, I asked, we had talked about things in the broader sense in terms of babies being born with problems that would give them difficulty to learn or health problems, would you want to know about that when the baby was still inside of you, or when the baby came out? And if you found out when the baby was inside of you, if the baby had disabilities or had health problems, would you want to keep the baby or not? We had a discussion like that, and they were very, very adamant that they would never consider ending the pregnancy, and they would never consider giving the baby to someone else to raise the baby, and that they wanted to keep the baby. Based upon that, I felt comfortable that doing an amnio would be probably okay for what they were telling me, but I felt like the discussion of "This is chromosomes and this is an amnio," we talked about the basics of an amnio involved taking fluid out from around the baby and things like that, but I felt like some of the basic information in terms of describing the tests was too advanced for them. I don't think they would have been able to understand, for example, that a quad screen was looking at proteins made by the pregnancy in her blood. All I had really said about that was that "You had some blood testing already that showed a lower possibility of the baby having some common issues we could see in older women, but that doesn't tell us for sure." Very, very basic information about everything. And the information that I would normally share with a patient, some of that was missing, because when I tried to discuss they seemed lost.

JM: How were they lost?

Interviewee: They were just staring blankly at me, and I'd back up and try again to say this similar information in a different way, or simplify it further, but even in its simplest form, I felt like they would ask questions, or they would just say "I don't understand." But you can commonly see, because we are talking about very confusing information that a lot of people have never heard before in their lives, but it was to a greater extent than you would see typical people confused, you know, discussing all the science information. And also, in terms of their ability to deal with emotions, in talking to them about "If the baby had a problem, what would you do in that situation?" it was similar to having that discussion with a child. Most adults are able to think about those things a little bit more.
Like connect the dots a little bit more. They wouldn't say, like some people might say 
"Oh, yeah, I know someone who has a baby with disabilities which is really difficult. 
That might be really difficult for me.” But it seemed like they weren't able to make those 
advanced connections between thoughts. They said "No, no, we want to keep the baby no 
matter what," but I don't know that they were even thinking about themselves having a 
disability, or maybe other people they had met with a disability. They didn't say things 
like “Our son's normal, so could this baby be normal too?.” They didn't ask advanced 
questions. They were very simple.

**JM:** Do you know if their other child had any disabilities?

**Interviewee:** According to them, he's normal, but I didn't meet him, and I don't [?] [14:40] review everyone's medical records, and they declined. So, I question how normal 
he is. I had asked if someone was coming into the house to do early intervention, and that 
sort of thing, and they said no. But sometimes they'll say no and those things are actually 
happening and they forgot to say, so I didn't really know for sure. [?] recessive or 
sporadic and not something that could be passed on, so.

**JM:** One thing I found was that a lot of people with intellectual disabilities will 
downplay their disability, and even if they have mental retardation, they will classify it 
more as a learning disability.

**Interviewee:** Yeah. That's what they had said. I almost thought when they were going to 
come in that it was going to be something like [?] [15:33] or ADHD or something like 
that. And they would never use the term, I would never, obviously, use the term with 
them like "You have mental retardation.” But they just said "We have disabilities with 
learning.” But it was clear that it was more extensive than what typically we would think 
of as a learning disability. But I don't know if they were downplaying it because, like if 
that was a conscious decision or not on their part. I feel like maybe that's what other 
people have told them that they have -- learning disabilities -- maybe to be sensitive to 
them. Because I feel like they were the type of people that if someone said, "You have 
mental retardation” that they probably would have said that to me. I don't think that they 
were downplaying it necessarily.

**JM:** I don't have any other specific questions. Is there anything you'd like to add?

**Interviewee:** I think there's just huge differences in terms of the clinical situation where 
you might encounter someone with mental retardation or intellectual disabilities. And if 
it's in a pediatric setting, I think genetic counselors are really trained quite well to deal 
with that, but when you're in a setting where the patient has to make independent 
decisions about something that will impact their life, whether it's a prenatal or a cancer 
session, I think that's an area where a lot of genetic counselors probably feel lost and 
could use some guidance about how to proceed in those situations. They happen pretty 
rarely. I can think of maybe three to four times this has happened over five years of being 
a genetic counselor, but when they do happen, you feel stuck, and it can be very stressful, 
so [?] interested in your study, because maybe you'll come up with some advice for how
genetic counselors can deal with this type of situation.
Interview 3

Interviewee: Hello.

JM: Hi, how are you?

Interviewee: Good, thanks.

JM: So, I just have a couple of questions for you. First of all, thank you so much for participating in my study. I really appreciate it.

Interviewee: You're welcome. I think it will be very interesting to see what your results are.

JM: Yeah, a lot of the counselors have been very interested in the results, and when I'm going to have them and what I'm going to do with them, so I will certainly let you know when I finish my project. So, my first question is that I know you had emailed me and said that you had counseled someone for BRCA testing that had intellectual disabilities. So, I was wondering if you could tell me about that situation and what the challenges were in the session, anything you learned from the session, that kind of thing.

Interviewee: Sure. The background was this was a patient with a [contraplasia?] who had lots of other medical complications. She had had a stroke at one point in time, and that’s what caused her intellectual disabilities. It wasn't anything that she had been born with. This was totally secondary to the stroke. And when she came in to talk to me, it was because she had also had bilateral breast cancer, and she was relatively young. I think she was in her 30's to 40's when this had happened. So a pretty young person with a very complicated history. And when she came in, she came in alone. She needed, she lives in a group home, but she did not have anyone from the group home accompanying her to the visit at all. So, someone drove her, dropped her off, and left. So, when we were talking -- I'm assuming that you've done some breast cancer counseling sessions [?] [2:04] In your rotation?

JM: I'm doing my cancer rotation next, so I haven't actually done any yet.

Interviewee: Well, there's a pretty set amount of information that we go over in these sessions. And of course, it can be tailored to the patient, but in this case, I was really just trying to use as many of the techniques as I could to try to make sure that she was really getting it and it was really sinking in. And all indications were that it was sinking in. Like, I asked her, you know, she was really close to this one particular family member. And so, I was asking her, "When you go home, are you going to tell your aunt about this?” And she said that she was, and I asked her what she was planning on saying to her, and she was repeating back the information that I would really want her to be understanding. So, everything was telling me that even with some of the troubles that she was having and some of problems that she was having that it was still sinking in. But then that day she decided not to do the testing, because the way that our clinic was set up is if
someone wanted to do BRCA testing they would not need to come back for a blood draw. We could just do it that day. But she decided that she didn't want to have it done that way. She wanted me to look into insurance coverage a little bit for her, so that was fine. I agreed to do that. So she leaves. We scheduled another appointment for her to come back. When she came back in, she had no idea what was going on. Totally did not remember anything we had talked about. Had no idea why she was being referred in. Nothing.

JM: How long after the first session was it that she came back?

Interviewee: It was about a month after the first session. And I always send people home with information, too. So that was really frustrating for me, because the way that I was approaching it is that when she left she was really following along. It didn't, you know, some people they just look so confused during the whole session that you can tell that they are having a really hard time with it. But she really seemed like she was following along and understanding this, and so it was really kind of frustrating for me because when she came back in and just really did not remember anything, first of all, I felt like I needed to do the entire session over again. Make sure that she really was told again, reminded of why she was here and what we were talking about. But also, it made me kind of worried about, when her results come, is she not going to remember, in the end, when the results come? Because the way that our clinic did results sessions was that unless a patient requested to come in in person it was all done over the phone. So, she ended up having the testing drawn, and of course, at her second visit she was there by herself again. When her results came in, I tried to call her to have her come in. She would never return my phone calls. The care providers that were at the group home that she lived in, they were wanting me to give the results directly to them, but of course I had nothing signed by her that said that I could do that, give that information to them, so I didn't feel comfortable doing that. So when it came down to it, I just had to send -- and they were negative results, but -- I ended up just having to send a copy of the results to the doctor and CC her on the letter. So I think that that was, you know, I think that it's tough. And to me that told me that it's really important for someone to have another set of eyes and ears, just someone that they trust, that they are close to who is going to be able to be another person at the session. Because having it just be you and a person with some disabilities, some delays, that's not the best situation to be in.

JM: What was her level of functioning?

Interviewee: That I could never get a clear grasp on. I tried to ask her physician, because of course she was undergoing cancer treatment at the time, and I worked with the oncologist that she had been seeing, and no one could really, no one could really explain to me whether they qualified her as mentally retarded after this stroke, or whether it was just memory problems. No one could really classify that.

JM: But there was a clear...

Interviewee: Yes. It was, I mean, it was just apparent talking to her. And her, just the level of speech that she was using and her, and just the fact that she was really repetitious
with some of the things that she was saying. Like, not in a way that the average person would be. I mean, it was pretty clear meeting her that she had some degree of difficulty.

**JM:** Did you know before the first session that she had an intellectual disabilities?

**Interviewee:** I was told that day. The day that she was coming in.

**JM:** How did you find that out?

**Interviewee:** One of the nurses just said that it was a patient that, because the oncologist that she had been seeing was in the same clinic that I was visiting, it was one my outreach days, and one of [?] [7:21] staff and the nurses just said "We're not sure what's going on with her. She had a stroke. It seems like she has some cognitive delays. We're not really sure exactly what her limitations are. And she always comes in by herself." So, it was more of just a heads up before she walked in the room.

**JM:** If you could do something differently during the session, is there anything you would have changed?

**Interviewee:** I think that I would have rescheduled it, and asked her to come in with someone that she trusted just to have another set of ears at the appointment. Just someone that she felt like might be able to help her make decisions, and, yeah, I think I would have rescheduled it for her. Because I can't think of any, I can't think of any other techniques that I could have used that would have helped her. Because really, everything that she was saying to me, it really seemed like she was getting it. Just the way that she was rephrasing what I was saying, the way that she was approaching how she was going to tell her other family members. Everything just really seemed like it was sinking in.

**JM:** What would you say was the biggest challenge with her?

**Interviewee:** The biggest challenge was the fact that it really seemed like she was understanding, and yet at her next visit it was really apparent that she was not really understanding, and so some people I think, you know, it's kind of like people that have trouble reading or are illiterate. They come up with really creative ways to hide that from people. And I think that in this woman's case, I don't think that she was purposefully trying to cover up that she had cognitive disabilities, but I think that she had come up with ways to handle that, and I think she was really good at short-term memory, and short-term making sure that, just making it seem like she was really understanding things. But I don't think that was the case.

**JM:** Would you say that you were comfortable counseling her?

**Interviewee:** I was comfortable counseling her at the time. As far as did I feel uncomfortable working with someone with an intellectual disabilities, no, I felt fine with that, and comfortable with that. I just felt like I wished that she would have gotten more out of the session.
JM: You had also mentioned to me in the background information that you have a family member with an intellectual disabilities.

Interviewee: Yes.

JM: Can you tell me about that?

Interviewee: Sure. My cousin has [?] [9:58]. So she's a younger cousin, so I pretty much have watched her grow up, because she's about eight years younger than I am, or so.

JM: Would you say that having that experience with your cousin has made you more comfortable interacting, counseling people with intellectual disabilities?

Interviewee: You know, I think that that's really tough to answer. I wish I could give you a yes or no, but I think that unless I had experienced it both ways, I'm not sure if I could answer that. I mean, I would, my instinct would be to say no, because I think our training as genetic counselors gives us some experience. I mean, especially coming from the same school, we have the Family Pals program there. So, I feel like just through my graduate education I have had experience working with people with intellectual disabilities. But I'm sure that also having that in my, having a history of intellectual disabilities in my family probably didn't hurt either.

JM: Do you have any suggestions for other counselors who are preparing for a session with someone with an intellectual disabilities?

Interviewee: I think that my, I think that my advice would be to kind of, don't, try not to feel like you're in a hurry. Because a lot of our sessions, we're in these time constraints, and it's just such a rush, and I think that for these situations really being able to take your time, use all the techniques that you were trained in, making people restate what you're saying. Just really try to make sure that that patient is actually understanding and not just nodding their head.

JM: Do you have any specific ways? I know you had your patient repeat back. Is there any other ways that you have used in your experience to assess a patient's understanding?

Interviewee: Usually...I'm trying to think...I mean, usually it is just having them either repeat back, or "How would you explain this to your family member?" Or "How..." Just asking the follow-up questions. "How are you feeling about this?""Does this seem like something you're really concerned about?" Because sometimes if, sometimes you can tell how well people are following along by if they are having the kind of, the emotional reactions to the information that you would expect them to have. And so I think that sometimes just checking to see if someone is reacting the way that you might expect is a good indicator, too. But really for people with intellectual disabilities, I think that just making sure that they are getting the facts straight is good. Because at least for this woman, she did not have any problems with expressing how she was feeling about the
information. She, it was very clear what was scary to her, what she felt comfortable with, what she felt a little bit relieved about after we talked. That was not an issue. She was very open with that. So for this particular patient, it was all about her understanding of the facts, of the actual risk of what the test is doing, what it's going to tell her, what it's not going to tell her. I just wanted to focus with her on getting the facts across. But of course there are going to be other people. Like, for pediatric counselors that might work with kids who have autism spectrum disorders. Then it might be important to focus more on the emotional reaction, if that's something that you can pick up on or not.

**JM:** Do you have any questions for me at all?

**Interviewee:** I don't think that I really...Well, actually, I do have a question that has nothing, it's very easy. How did you decide how many, what sample size you were going to use?

**JM:** I'm doing twelve interviews. And originally we had set it at eight, but when I was going through doing my IRB paperwork and everything, the woman from the IRB came in and she was like, "You know, if you are doing interviews, you want to put a maximum number." So I put a max of twelve, and then I ended up getting way more responses than I ever expected, so I ended up doing the full twelve, so.

**Interviewee:** Well, that's stressful at the time, but it'll be great when it's time to analyze it all. Well, that's exciting.

**JM:** I don't have any other specific questions for you unless you have anything else to add.

**Interviewee:** Nope, I don't have anything else. I hope that that was helpful.

**JM:** Yes, it certainly was. And thank you again for participating. And have a great holiday.

**Interviewee:** Thanks, Jacquelyn, you too. Tell Beth hi for me.

**JM:** I will, absolutely.
Interview 4

JM: Hi, this is Jacquelyn.

Interviewee: Hi! Right on time.

JM: Perfect. Thank you so much for participating. I really appreciate it. On such short notice as well.

Interviewee: No worries.

JM: So I just have a couple of questions to ask you about your experiences counseling someone with an intellectual disabilities. I was wondering if you had a particular situation that sticks out in your mind that was difficult or you learned something from or you faced some challenges during it that you could tell me about.

Interviewee: Well, gosh. I don't know if I -- I didn't really think of one that had particular challenges, but I was just trying to, you know, when you said in your email you wanted a specific, you know, you were going to talk about a case. So I was trying to think about that. And I do have, I do a lot of developmental disabilities, so I see it quite a bit. So I just kind of picked one out of my head that I felt like there was a fair of involvement with, and I, you know, there's always challenges. So we'll go with that. Do you want me to give you some background on it?

JM: Sure.

Interviewee: Just to give you some context, I do consulting for the San Diego Regional Center, and the Regional Centers in California are the state-funded agencies that coordinate services for the developmentally disabled in the state. And so from birth through adulthood. So I provide genetic consultation to individuals and families where there's a developmental disability and they have questions about the genetic part of it, I guess. And so this case I'm thinking of is not a really unusual situation and that is one where both, there were two individuals who were a couple. They were both Regional Center consumers, is what we call them. And they were having a baby together. So I was brought in to consult to address the issues of whether there was anything special that they needed to know about for the pregnancy or for the baby, given that both of the parents have intellectual disabilities. So, and one of the things that I do here, too, is I also coordinate a dysmorphology clinic. We have a limited dysmorphology clinic [?] have a dysmorphologist who comes in and sees like three patients a month, and so I coordinate that clinic for her. So both parents had mild MR, etiology unknown. And so I got the referral. I got the charts that we had on both of them. And read through there. And in reading the -- oh, and the Grandma, I should also say that the Mom's Mom was the one who was probably driving the train relative to wanted to know if there was anything that they should be concerned about, because she was seeing herself as the person who is going to be primarily responsible for this baby in the long-term, so she wanted to know what she should know in terms of that. And she's also the one that's just really involved
with everything. And so -- do you want me to just tell you what happened? I'm not too sure what you want.

**JM:** Yes, you can just tell me what happened.

**Interviewee:** In reviewing the charts, there was no etiology for either one of them, the Mom's family history was totally negative. Nobody else in the family with any kind of disabilities. And Mom was otherwise healthy but had mild MR and not much going on there. But the Dad's history was a little bit more significant. He was adopted out as a young child because his mother of origin had probably schizophrenia and was unable to care for him, and he was pulled from the home by Child Protective Services because he was severely neglected. And then he was put into foster care, and then he was raised by another family, and has grown up in that family. So he really didn't have any information about his family of origin, other than his mother had a long history of mental and intellectual disabilities as well, because she was also a consumer of the Regional Center in the Los Angeles area. So I knew she had some kind of intellectual disabilities, but there was really limited records around that. And I'm trying to remember if I met them first, or if I just went ahead and brought them in...I think I just brought them in. I went ahead and had them scheduled to see Dr. Jones, our dysmorphologist. And so they were both seen, and the Mom, there was no clear etiology. She's not dysmorphic. But the Dad, when he came in, at the beginning of just looking at him and talking to him, it became pretty clear that he probably had 22Q deletion syndrome. And we tested him, and indeed he did have that. And so then I went back and met with him and her and, let me see, both sets of their parents, and the service coordinators, like their social workers. And...

**JM:** So this was together with all of those people?

**Interviewee:** Yeah, big family meeting. So both of them -- she's pregnant -- so both of them, they're both, well, the Dad's Dad, the Mom's Mom, the service coordinators, who were the agency's social workers, and also her or one of [?] 6:30 I can't remember. One of their ILS independent living skills workers. And we all sat down to talk about his 22Q deletion and the risk to the baby and what needed to be done for the baby and testing and pregnancy and all that good stuff. And that I think went pretty well. There were a lot of people in the room. Both of the parents were very committed and very interested in doing everything that they could for this baby. Very loving towards the pregnancy, very loving towards each other. And they accepted the diagnosis of him pretty readily. It seemed to, like it made sense for him, I mean for him it made sense for some of the things that he had trouble with. He had thyroid disease, and he had had seizures, which may have been related to a calcium problem when he was little, but he hadn't had seizures in years. He's actually a pretty healthy guy. He's pretty, actually a pretty healthy guy. Doing pretty well, but mildly disabled. And they, you know, we went through the genetics of it, and they seemed to do pretty well with it, and they seemed to be not too worried about the baby, but understood the 50% risk. The grandparents, the grandma was probably the one, the maternal grandma was probably the one who asked the most questions and did an Internet search and came with a long list of questions about, "What about calcium?" "What about the thymus? What about this? What about that? What about heart, what about..." And all
of that. We coordinated to make sure the baby got a fetal echo, and the baby did, and it was normal. And then they went off and had their baby a few months later, and the baby did have [?] syndrome. And then I went again and I met with the bigwigs in the NICU for some trouble with feeding, and had a very low calcium, and so was in the hospital getting stabilized on calcium supplementation. And why else would the baby...I guess it was mostly feeding and calcium. And so when they had a family meeting at the hospital with the neonatologist and the social worker and the discharge [?] , then I went to that meeting as well to follow up and review the information with them and see how things were going, making sure that all the appropriate follow-up was done, that the people in the hospital knew the history as well. And that seemed to go okay and they went home and the last I heard I think they were planning to move out together, I mean to move in together with the baby, but I don't think that's actually happened yet. I think they're, she's still living, the baby's living with Mom and Grandma at the Grandma's house. The Dad is involved and he comes around a lot, but I don't think they've gotten it together to get a place on their own. So that's in the works.

**JM:** At that initial session with them when you had explained the diagnosis of 22Q to the husband and his wife, how were you able to sense that they understood that and the rest of the pregnancy?

**Interviewee:** I think I used a couple of different ways to explain it. I had visual, and I had a copy of his chromosomes, and I had a copy of his FISH test and I had pictures of a deletion and I also explained it. And you know, I try to ask people questions to see what they're grasping. And I'll say "What do you understand about this? What do you think is going on?" And kind of see what they, how they respond to see what they're getting. And in that case they really seemed to be getting it really pretty well. I've had cases where that's not true. But in their case they seemed, I mean, it's fairly straightforward. It was...I think my goal was for them to understand that it's a chromosome problem. That it's something that's part of yourselves that you can't control and there's a little piece that's missing. And he probably got it from somebody before him, it's nothing that he did. And that there's a, because we pass down our material to our kids probably, there's a chance that the kids could get it as well. And they seemed to do okay with that and not have any issues with that. But I guess I try to just feel them out and say things in a couple of different ways, and I gave them written information to take home with them and, you know, so that they can have that as well. They both read, and they both like to read, and like to get information that way, too. And then I sent home letters with them. So after our meeting I did a letter that went to each one of them and to Grandma that kind of reviewed everything we talked about, and then I sent another letter that was for the doctors that was kind of a "Dear Doctor" letter that was, you know, the family letter was written in very easy to grasp language. And then a "Dear Doctor" letter that said "This father has a 22Q deletion. When the baby's born, this is what you need to do." And then they took that with them to the hospital to hand to the pediatrician in the hospital. And all of that went well. It all got done.

**JM:** If you could go back to redo the session, is there anything you would have done
differently or changed?

**Interviewee:** In that case no. I think it was, everything went pretty successfully.

**JM:** It sounds like it.

**Interviewee:** I would have to say I could give you another example of a very similar case that didn't go so well. Funny that I should choose the one that went well! I had another couple, very similar situation. Both Mom and Dad are consumers. And also had them come into see Dr. Jones. Dad got diagnosed with Soto's syndrome. Big, big guy. Actually, he had been diagnosed with Soto's syndrome as a child, but nobody seemed to remember that. But it was in the records. So it was there, but he didn't seem aware of it. And also Mom had kind of, Mom in that case had mild MR of unknown etiology, but very significant family history. I mean, every, literally everybody in the family history as far back as you could go was a consumer of the regional center with kind of mild MR of unknown etiology. So a lot of history on that side of the family that was not a specific diagnosis. Whereas Dad had no significant but had this diagnosis of Soto's syndrome. And that's one where very similar scenario. I did not get to meet with them again. When Doctor Jones met with them, she clinically diagnosed Soto's syndrome. She had already actually, she was the one who had seen him as a child and had diagnosed it the first time. So we discussed it with them at that point. And for them, especially with the Dad, I don't know how well they were really understanding it. But the sense you got, it was hard to know how much they didn't understand and how much they just didn't care. In that the Dad, you know, they were both just very excited about having a baby, and very focused on the kind of more childlike version of having a baby. Like is it going to be a boy or a girl, and is it going to play sports, and what are they going to name him, and what color are they going to decorate the room, and not so interested about whether there might be health complications or whether the child's going to have an intellectual disabilities. So, that was just not on their radar. On the radar were the fun details. Do you know what I mean?

**JM:** Not so much how to care for the baby, things like that?

**Interviewee:** Exactly. But we did tell them, we explained it in a couple of different ways. I sent them a letter in language that was straightforward, explaining that there's a chance that the baby could have the same thing as Dad had. Dad didn't really perceive that he had much of a problem. He's had mild mental retardation. He had special ed. He's had a lot of, I don't think he works, though he's had job coaches he's tried, but he doesn't, he's never done very well in job situations and I think hasn't been very successful in keeping jobs. But he doesn't perceive himself, I think, as disabled. So to say that your child might have the same thing as you was not meaningful really to him. And you know, he's been healthy. It was really just the size and disability, it was size and cognitive issues. He was huge. So one thing I think that they did understand a little bit more is this might be a really big baby, and this baby might have a really big head that's not going to come out the traditional way, and you might get a C section. And indeed that's what happened. It was an enormous baby. I think this baby was like 10 pounds and had this
huge head and did have to have a C-section and did have Soto's syndrome in the baby. And they also, I went and met with them in the hospital with the social worker -- I wonder if I still have this -- hold on a second. Let me get something, because this is really interesting. I'll go grab the chart.

**JM:** My battery on the recorder is about to die, so I'm going to change the batteries. I'm going to stay on the line with you, I'm just going to change the batteries.

**Interviewee:** Okay, that's fine, because I'm going to go grab the chart. [pause] Are you there? Okay. You know what, I don't have the thing I was hoping to have, but that's okay, we can go on anyway. So, I did meet with this family in the NICU also. The baby, you know, the baby actually did not, the baby had problems at delivery that we never really understood why the baby had, but was having some problems breathing initially, was really a sick little kiddo for being a full-term kid. So had gotten transferred to the NICU from the hospital they had the delivery at. And I mean ultimately did okay, but was on a vent for like a week, and was a pretty sick little kid. In any case. So I went, when they were getting closer to discharge, the social worker asked me to come meet with this family and talk about this diagnosis, because her sense was that they didn't really get it. And indeed, they didn't really get it. And despite my best efforts, I'm not really sure that they ever really got it. And so we talked about, and I don't have the thing I was hoping for. I think it's in the chart over at Children's, not over here. But we talked about, I think my goal with them was just more focused on what they needed to know to be able to take care of the baby. And that yes, the baby has this thing called Soto's syndrome, and the most important way that that affected him was his development would probably be a little bit delayed compared to other children, and that he was bigger than other children. But that otherwise we didn't expect that that would really affect his health in a lot of other important ways.

**JM:** Did they seem to understand that?

**Interviewee:** They could repeat it when I would question them and say "Okay, so what do you understand about Soto's syndrome? Tell me what your understanding is." Right after I had said it, they would repeat it, but then they would ask questions that didn't really make sense again. So, it's not clear. Like, they would ask questions about, I'm trying to remember what it was. She, I think she, like little things again. Was he going to have glasses, and was he going to be able to play sports. And hold on, what were some of the other things? Well, the reason I said I was pretty clear it didn't work is then the baby didn't get discharged for like another week, and, mostly because I think the hospital was holding onto them to try to do as much teaching with this family, because they were really concerned about whether this family was going to be able to care for him in the home. And when they were getting discharged, the social worker had them make a list of all the questions that they had. And I got a copy of that list, and it was like 20 questions, and when you read the questions you went, "Oh, man, they really didn't get it." That's the list that I thought it would have been really interesting to be talking about but it's not in front of me. [?] [3:57] their chart. But it was a lot of the same kind of little things. Some of it was about, not very much of it was actually about the Soto's syndrome, it was more
about what he was going to do, and about, and a lot of was just normal childcare. Like "When can I feed the baby a hot dog?" and "Is the baby going to be able to do this?" and "Are they going to be able to talk?" And "What can I do, can I take the baby on a walk with me?" A lot of very basic child care kinds of things, and then a lot of things that had to do with predicting when the baby was going to do things, or if the baby was going to do things, and when we had really spent quite, a good amount of time saying that we expected the baby to be able to do a lot of the, most of the things that other babies do, relative to walking and talking, but that he might do them later than other babies, but that we didn't know exactly when it was going to be. We said that a lot of times in a lot of ways, written and verbal. And it was clear that at the end they still didn't quite get that.

JM: What did that feel like for you? Knowing that they sort of didn't get it, but you'd done all of these things to help them try to understand.

Interviewee: What does it feel like for me? I think that probably primarily it makes me feel concerned about this baby. I don't, I didn't feel like I failed. I don't know if that's the question that you're getting at. But I think because, I think a lot of people worked with this family. Not just me. But I know that they weren't hearing it from just me, that they were hearing it from the social workers, and they were hearing it from their service coordinators, and that it wasn't a failure of effort. It just, I think that there were just limitations that we couldn't get beyond. Yeah. And unfortunately, well, unfortunately or fortunately the supports were limited in this family, too, and that I think the baby was going to go home where they were both living with her family, and her family all have mild mental retardation too, and so they weren't really in a much better position to help, whereas the first family I described the Grandma was very with it and on top of everything and I didn't have a concern about that baby falling through the cracks. Whereas this baby, you just have a lot, I just had a lot more concern about how was this baby going to do and are they going to be able to do this? And so I think I felt bad for the baby but I also felt bad for them, because I felt like I could see where they might not end up being able to do it, like potentially they could, there could be neglect or they could lose this baby at some point. And that made me sad for them. Because I think that they were really excited about this baby. But I just don't know if they had what they needed to be able to parent this child successfully.

JM: Is there anything during this session that you would have changed or done differently if you could do it again?

Interviewee: Oh, gosh. I think, you know, I don't...I'm trying to remember now, did I do a second letter after that, after I met with them after the baby was born? I don't remember if I did, and maybe, you know, now that I'm thinking about it, I could have, or should have gone back and written them another letter after the baby was born saying "This is what we talked about." Though I don't know that that would have made a difference, but at least I could have, it would have been something that they could look at. Though I think that the social, I think a lot of that went, the social worker was going to do some of that. Maybe that's why I didn't do it. But maybe that would have been helpful. Part of the hard part, too, with this family is they live about 120 miles away, so they are not right
here, and they are east of San Diego basically in the middle of nowhere, so there's not a lot of services out there. It's harder to keep up and keep track and get services in place down there. And that makes it even more worrisome. So, you know, I don't know. I don't know if giving them more written information would have been more helpful. I don't know if meeting, talking to them again would be more helpful. With both of these families I did copy the letter and give information to their social workers from the agency so that the social worker would be able to follow up with this information and would know the information that they had been given. I don't know.

**JM:** Would you say that you are comfortable counseling people with intellectual disabilities?

**Interviewee:** I think so.

**JM:** One of the reasons that I ask that is that in the background information I asked about if you had any family members with an intellectual disabilities, and I was looking to compare if there was any difference in counselors who have close relatives or don't have close relatives, and how they feel about counseling this population.

**Interviewee:** Well, you got my stuff I faxed, right?

**JM:** Yeah, I did.

**Interviewee:** So I have two cousins, but the honest truth is I've never spent a lot of time with either one of these cousins. They live in France, and so I grew up, my summers I spent in France, and so my summers I spent around them when I was little. But then, after, then we only started going every other year and then every three years and so it's not like people I grew up with frequently. So I know them, I spent a little bit of time with them growing up but not...And one of them is very mild, and one of them is probably more on the moderate to severe end. And so, it wasn't somebody that I was very close with or had spent a lot of time with. So I don't know that that really makes a, I don't know if it should count or not. You know, to be totally honest with you, I never wanted to work with people with intellectual disabilities. And if you had asked me 15 years ago -- I was a social worker before I was a genetic counselor -- if I wanted, what I would have thought of working at the Regional Center with families of people with intellectual disabilities, I would have run screaming in the other direction. Just because it was not an area I felt like I had a lot of familiarity with, it was not an area that I felt super comfortable with, and honestly I thought I didn't have the patience for it. I'm not, I don't feel like I'm a particularly patient person in general. Well, I mean, I'm not horrible, but in general. And so I always thought I can't work with somebody with intellectual disabilities. It'd make me crazy. And what I've learned is that it actually doesn't. That I'm actually much more comfortable with it than I thought. Because I got thrown into this not by choice but by necessity, in terms of I needed a job and this was the job they needed filled. But I think that when I know somebody has an intellectual disabilities, I go at it with that expectation, and then I'm fine with it. If I'm dealing with somebody in the general world, and somebody's just not keeping up with what you want, you know, you go to bank or
whatever, then I don't have patience for that, but when I'm meeting somebody in a professional context and I know they have an intellectual disabilities I'm absolutely comfortable with it because I think my role and my expectations are different there. So I have learned that actually it's much better than I thought it was...it's better than I thought it was going to be, and I'm more comfortable with it than I thought I would have been. Does that make sense?

JM: Yeah.

Interviewee: Yeah. So I think I do okay with it. I think that I'm pretty comfortable. I think that, you know and I teach also. I teach genetic counseling students at UC Irvine and so sometimes this comes up, too. We talk about it, because I teach advanced counseling skills. And I think that one of the things for me is being able to adjust your counseling style and your expectations for what your objectives are for the counseling. So what my expectations are for, you know, if I was working with a couple that were both really well educated, high functioning people, what I expect them to understand about their child with a disability, and what I expect to educate them about, my list of things they're going to talk about, is different than when I'm talking to a couple where both of them have mild MR. And I know I'm not necessarily going to go into the same detail about, I'm not going to go into the molecular origins of it. And I don't need them to understand [?] [13:50] or whatever it is, but I need them to understand some more concrete concepts that they need to be able to take care of their child. What are the things their child needs? What should their expectations be? What are the special appointments or things that they need to do to help their child? Is it going to happen again if they're having more kids? So I think that that's for me the key of being able to work with people with intellectual disabilities is having realistic and appropriate objectives.

JM: So is that a suggestion that you would give to other counselors who are facing their first session with an intellectual disabilities?

Interviewee: Yes. That's what I would say.

JM: I don't have any other particular...

Interviewee: You know what else I would say? This is my little bugaboo is be careful with the use of analogies. Because I think some genetic counselors are trained to use a lot of analogies, and I think that when you are working with somebody with intellectual disabilities you can lose people. So I think, you know, there are some, and I say that because the program that I teach in has a geneticist who really loves them, and so right away the students start going towards the recipes and the books, or the bricks in the house, or the train and the guy, I don't know what. And it's like whoa, I'm having trouble keeping up I'm [?] [15:33] with you. And so I just think that the use of analogies should be used sparingly and with caution, is what I would say. Speaking, that I don't usually speak metaphorically very much with people with intellectual disabilities. I stay present and I stay more concrete.
JM: Do you have any questions for me?

Interviewee: So this is your Masters thesis process?

JM: Yeah.

Interviewee: Very good. Are you going to provide the finished product to people you interview?

JM: A lot of counselors that I've interviewed so far have been very interested in my final product, so I'm definitely leaning towards that.

Interviewee: I would love to...I mean, I would love to...because I admit that I do this in I think a bit of a vacuum in that I haven't had a lot of conversations about it with other people, and I'd be interested to hear what other people say because maybe I'm off base. It's nice to know how I compare to other people.

JM: Even when I was doing my literature search, there wasn't much information on this topic in particular. That's where my thesis project came from. So yeah, I will certainly let you know when I have a finished product.

Interviewee: Yeah. I would be interested to see it. Can I ask what your findings are so far?

JM: I've only done a couple of interviews so far, but I've gotten a lot of feedback and a lot of what you were saying as well in terms of being more concrete and setting different goals as opposed to more specific goals as you would counseling somebody who doesn't have MR. I've taken a break because of the holiday, so I need to go back and look at more of my interviews and see what other counselors have said. But yeah, that's definitely one of the big things that I've found so far. And trying to assess people's level of understanding is a big challenge that a lot of counselors have said that has been difficult in some of their situations.

Interviewee: I think one of the other challenges, and I don't know if other people have talked about this, is when you're counseling several people together, like a family or whatever, and one person has mental retardation and the others don't, is knowing who you're addressing. Who is your audience. Because if you keep it just to the person with MR, the other people in the room might not be getting everything that they want. But if you're talking to them, it might be overwhelming or confusing to the person with MR. And also when you're dealing with adults with MR, which is what I do a lot, is how to talk about their disability with them, especially since not all the time, but sometimes, they don't perceive that they have a disability. So how do you talk about a person's disability with them when they don't perceive a disability?

JM: A lot of the counselors have also said that, too. A lot of people downplay, they'll say "I have a learning disability" not "I have an intellectual disabilities" kind of thing.
Interviewee: Right. Or we talk about things like having trouble in school and needing special classes in school, and talking about it in that context. As opposed to saying, like I've said with you "people with MR, mild MR," and I wouldn't say that in front of that person, probably. I might talk about, it depends on them. I'll try to get a feel for what their sense of what their needs are. What do they understand about themselves? And then I'll try to go with whatever they say. And if it's right on, if it's, if they say "I've got a disability and I was in special ed and it's hard for me to learn things," then that makes it easy. But if they say "Nothing's wrong with me," then you feel a little bit more stuck. It's a little bit harder to have a conversation around that when, "Okay, so we're here in the genetics clinic because nothing is wrong with you." It's harder to know how to address that. But you try to pick at the things that, you know, "Did you have special classes in school?" or "How did you do in school?" And sometimes you get the truth, and sometimes you get, like this guy and his father with Soto's syndrome, if you hear him talk he was like he did great in school and everything was great and he's super smart. And his only problem is he's too smart. Then it makes it really difficult to know how to address them.

JM: Well, thank you so much for giving me all of your insight.

Interviewee: You're very welcome.

JM: And if you have anything else that pops into your mind after we hang up the phone, you can send me an email, if that works.

Interviewee: Okay. And yeah, I'd be very interested to see your finished product when it gets done.

JM: Yeah, definitely.

Interviewee: Okay.

JM: All right, enjoy the rest of your afternoon.
Interview 5

Interviewee: Hi, how are you?

JM: I'm good, how are you?

Interviewee: I'm doing all right.

JM: Is now a good time for the interview?

Interviewee: Yes.

JM: I just have a couple of questions to ask you. First, I want to thank you for participating in my study on such short notice as well.

Interviewee: No problem.

JM: I was wondering if you had any specific experience that comes to mind that involved a patient with an intellectual disabilities. If you could just tell me a little bit about that situation, how you counseled them, maybe some of the challenges that were faced in the session. Things like that.

Interviewee: Yeah, there's one that comes to mind immediately. [?] So, really it was her son that was referred. He had Fragile X, but she also does. It was interesting because I knew ahead of time from reading the notes that she was affected, but it wasn't clear exactly what level of understanding she had. And it was clear as we talked more and more that there was a lot she wasn't aware of regarding the diagnosis and everything like that. She was someone that, I basically just had to figure out what were the key things that I wanted her to know in the most simple of terms. Because I was kind of counseling her and her son, obviously. So I think it's just interesting along the spectrum of all patients that we see of really just catering to that particular person. And for her, the difficulty with Fragile X, if you know, it's very complex, the counseling, because different generations can be affected very differently, so it's extremely complex, but for her it really just needed to be broken down to the bare necessities and really as simple as possible. And it was probably more of a struggle than it should have been. I think that was more on my part of I wanted to share as much as I could, because we don't usually see people back in follow-up, but then I had to realize pretty quickly therein that some of it is just going to be [in the note?] [3:43], I just need to get the bare minimum across to her so that she can remember it, and just gets, offer resources for follow-up for additional information. Which isn't the way I typically like to do things, but it was necessary in that case.

JM: How were you able to assess what she was able to understand, from what you were telling her?

Interviewee: For some, when I'm particularly concerned about someone's
understanding, often times I, you kind of jokingly quiz. So I think for her -- what I always start off with is just asking for what has been shared before. So what information they already had. And usually from that I can gauge the context and the level of understanding or never having been informed or what sort of misunderstandings there had been. So, it was really clear just from starting off that she had some difficulties in understanding. And then I know for her that I did kind of quiz her on some things. You know, and I'd joke about it, but it was really necessary just to see if certain very minimal things she could recall.

**JM:** Did you feel comfortable counseling her, or anybody with an intellectual disabilities in general?

**Interviewee:** Yeah. I actually find it fun and challenging. And certainly necessary. I think that all too often people are dismissed when they're harder to talk with, and so I find that even that much more necessary. And it is challenging to me, and so it makes it more interesting, I think. And I think that it makes, it reminds me of, what I always tell students is know what you have to convey versus what it would be nice to convey. And so it's just a reminder of yeah, there's the need to know, and then there's the nice to know. And really just focusing on the need to know at a certain point.

**JM:** If you were able to do that session again is there anything you would change or have done differently?

**Interviewee:** I think with her, she, I didn't have as much of a conversation with her as I would have liked. I mean, a lot of it was conversation, because it always is. But I think there's more we could have talked about in an informal way. I think for -- unfortunately, after many years experience I still, I think I felt a little constrained by time. Which I never should be. So, I think that with more time and also just having a more conversational time, I think it may have resonated better with her. I fortunately had the opportunity to see her again afterwards. She just showed up at our clinic with some more questions. So I know, I had that follow-up, thankfully, to know that she did get it enough to have additional questions after the fact. But without that, before that had happened, I think there was just this unsettling of I'm not really sure of what she got from it and what should didn't. And there was a lot of focus on her son. Because I think she was displacing things herself. I think she sees it in her son but not in herself so much. And either she misunderstood what someone had told her before, or it wasn't explained very clearly to her before. So, I was trying to break down a lot of misinformation and I'm not sure that, I'm sure that I could have done a better kind of rebuilding it. I think it was adequate. I just always feel like it could be better.

**JM:** Do you have any suggestions for other counselors who are preparing for a session with a patient with an intellectual disabilities? Maybe things to do before the session, or things keep in mind during the session of how to counsel them?

**Interviewee:** I think my number one would be set aside time. Make sure that there's something to -- I mean, depending on literacy level, make sure there's something for
someone who can read that they can take home with them that's very, very simple words. I kind of thing for any of us it takes repetition. And I think having a conversation is better than lecturing, because I know for many of us even if we don't have an intellectual disabilities, a lecture we start zoning out.

**JM:** I'm definitely guilty of that.

**Interviewee:** So I think just emphasizing the conversation and really just, something we should do in any appointment, but certainly in these, is "What are the key points that I really, really want to focus on," and then "What are the things that it would be great to do, but I don't necessarily have to." And knowing how to say the same thing in five or six different ways. And also being comfortable quizzing. I think that's something that many of us are a little uneasy because we feel silly doing it, but sometimes, and particularly in these cases, it's just a necessary thing to see, really to test ourselves, not so much the patient. Make sure that we conveyed that need to know information in a way that it resonated so that if we didn't, we can try again.

**JM:** During the session was there anyone else present, or was it just the Mom and the son?

**Interviewee:** The Mom, her son, the maternal great grandmother, and someone who works, she's kind of the support person for them, but she's not their primary support person. So that was what was a little funky was later on I met the primary support person who really understood, knows the family the best. And I think that would have, it would have been nice to have someone who was really connected with the family, but whatever. And I don't remember [?] [11:23].

**JM:** During this session, or any session when you counsel someone with an intellectual disabilities, I know you said that it's a challenge, but is there any other feeling that you get when you counsel this population? Maybe frustration, or is it rewarding for you?

**Interviewee:** I think it's rewarding. Particularly so. I think that people who have more, either more just natural education or ability, they, we're more apt to have received comprehensive information either by seeking it out ourselves or by our providers or teachers sharing it with us. And I think it can be more challenging and so when people have limited resources and limited time, I think they're more likely to dismiss someone and not really share with them as much as they could. And so I feel, and this is a population that, the population that I work with now is not necessarily always intellectually with a disability, but socioeconomically at a lower status. So, I see a lot of similarities. They don't necessarily, or most often don't have the opportunity to ask the questions or have things conveyed the way that we might. And so there's even that much more benefit. And I think it's rewarding just to know that I took time out that someone else might not have.

**JM:** I don't have any other particular questions for you. Do you have anything else to add?
Interviewee: I guess one thing that I find a struggle is we have a lot of resources for support groups and things like that. And theoretically, they're supposed to be appropriate for patients, but many of them are still very advanced. So that would be my plug is just to have more resources for people. It's ironic that the Fragile X information on their web site is really very complex, whereas the patient themselves is not. So, I would just make a plug for having written information or even pictures, something that we can provide these families to take home that isn't completely overwhelming and is language appropriate for them.

JM: Do you have any questions for me at all?

Interviewee: I noticed you had the question on the questionnaire of if there was someone in the family with an intellectual disabilities. Do you find that that seems to be prevalent in the people you have spoken with?

JM: Some of the people that I have spoken with -- the reason that I asked that question originally was to see if I could make any comparison between counselors who do have a relative or close family friend with an intellectual disabilities and those who don't to see if there's any difference in either how they counsel or how comfortable they are counseling people with intellectual disabilities. And I have found that in some of the people who do have a close friend or relative with intellectual disabilities, a few of them have said that they are more comfortable counseling this population because of their experiences with that. Which I guess is what I expected to hear, because for me, anyway, I haven't had that much interaction with people with intellectual disabilities, so I'm not sure how comfortable I would be counseling someone. Does that answer your question?

Interviewee: Yeah. I thought it was a very interesting question to ask and it's something I'm sure that you've noticed with our field. There's a lot of reasons that we enter it, and many people that I come across have some experience some way with someone with special needs of some sort or a genetic condition or whatever that might be. So, I was interested to see what you had gotten back. I thought it was insightful to ask that question.

JM: My thesis committee actually came up with that question, which I thought was very interesting.

Interviewee: Yeah. I think it is because I think it's really hard to take our experiences and separate "How does that influence me?" But I can imagine the comfort level being a little bit higher when...But I actually, what's sort of interesting, I noticed this when I was at Christmas this year. My cousin has very severe mental retardation. She's like in her 50's now. And I've grown up with her my entire life, but at Christmas this year, after being in the field for almost eight years now, this Christmas I was more comfortable approaching her and talking with her to the extent that I can than I ever had been in the past. So I think actually work has made me more comfortable with my family. I mean, she's part of the reason that I'm in this field in the first place, so it all comes full circle.
But I found that interesting, because I'm much more accustomed now to working with people who either intellectually or economically are of limited means. So I think my perspective and relatability is different now than it was even when I first started working. And I'm actually less comfortable with the very highly educated people than I was before. Because I'm not accustomed to it. I keep laughing -- I saw a patient who was like 13 year old, totally typically developing boy, and having a conversation with him, which was such a foreign concept to me, because it's been so long since I've seen a typically developing person. So anyway, I thought that that was interesting.

**JM:** Thank you so much for taking your time. And if you have anything to add when we hang up the phone you can send me an email.
Interview 6

**JM:** Hi, this is Jacquelyn. My first question is if you had a specific situation involving a patient with an intellectual disabilities that you could tell me about. The background, challenges you faced, things you learned from it, etc.

**Interviewee:** You want me to start with that?

**JM:** Yeah.

**Interviewee:** Okay. I mean, I do this a lot. But I’d like to think of something that I think would be – I counsel women who have a full mutation in the fragile X gene, often, so, but you want a specific case, right?

**JM:** Yeah. If you don’t have one, that’s okay.

**Interviewee:** I have many. I’m trying to think of one where we could maybe talk about it in detail. Okay. I counseled a woman who had Turner’s syndrome and happened to also have intellectual disabilities a couple of years ago. So, it was quite complicated in that much of the information about Turner’s syndrome is quite medical as well as reproductively oriented. And, I had to also assess what her knowledge is of basic reproduction, and she was someone who was quite, I don’t know that she was promiscuous, but she was socially very active, if I remember correctly. So, this was a situation where she was referred to us in the genetics division. She was short, and hadn’t cycled, and she was, I want to say, 16 or 17. She’d been in special ed, and had I would say moderate, mild to moderate MR.

**JM:** Did anyone come to the session with her?

**Interviewee:** You know, someone came to session with her, but I’m trying to visualize it. I think they actually waited outside. They might have – she had a social worker. She might have come to session with her, she might have been there for part of it, but my memory of the situation was really between she and I, so if the other person was there, they were pretty invisible.

**JM:** Did you know she had an intellectual disabilities before the session?

**Interviewee:** Yes.

**JM:** Did you do any prep work?

**Interviewee:** The only prep work that I do with all genetic counseling sessions is that when I know I’m going to be using guides like the Greenwood book, I put stickies on the pages that I want to flip to, or else I sit there and I’m flipping for like 20 minutes for the right page. So I often – and as I’m putting the stickies on the page, it does help me organize the order and what I want to say about each thing. With her, with people with
intellectual disabilities, and I can speak in generalities as well as specifics, using visual tools is very important. They often don’t words that you said. So, they may not remember the word chromosome, but they can see that this person has two of those things, and this person has one of those things. Just like someone without intellectual disabilities often remembers the pictures more than some of the terms, but I think it’s even more important with people with intellectual disabilities. It’s also I think important to have some parallels, when you talk with people with intellectual disabilities, to make things, like even to use the word “gene,” I often say something like “It’s sort of like a factory and it makes something that’s important for your body.” So once I say “factory,” I might even say to them “Have you ever been to a factory? Like a candy factory. And that candy factory’s job is to make something. And genes are inside your body and their job is to make something that’s important for your body.” So to use parallels in everyday life to help them understand something that you feel it’s important for them to understand, even though it may be a pretty complex topic. But sometimes you have to talk about complex topics.

**JM:** How were you able to assess how much the patient understood the information that you giving to her during the session?

**Interviewee:** If I remember correctly, I asked her a lot before I even got started. So, I asked her questions like “Do you know why you’re here?” or “What do you want to get out of this?” or “What are you [?]” Some things you just [?] understanding of why she was even referred. I asked her what she knows about getting periods, and why some girls do and some girls don’t, and why some girls are short and some girls aren’t. Just some basic information so that I had an understanding of where we were starting. And then as we go, I often do ask people in their own words kind of thing. Like “What does this mean to you?” I might say something to her – this is a couple of years ago. I don’t know why I picked her, but I do so much fragile X that some of the other ones stand out more, but I may have asked her something like “As you were growing up, did you think of yourself ever having a baby, or ever being a mother?” Or something [?] “Do you understand that by having this problem that you won’t be able to do that?” So, I try to ask them something about themself, and then by using that to reiterate something that I’ve already addressed. But some of it is more counseling. When I talk with women who have a full mutation with fragile X, for some of them it’s more counseling than education, in that you’re also dealing with some losses for them, or you’re dealing with them feeling different. You’re dealing with them knowing boys with fragile X who are more severe than them and their fear about that. Or the inability to have relationships. So there’s a lot more counseling than education in some of these sessions. So, I think that unfortunately some people go with them as an education thing. “I just want you to understand, you have a full mutation, you have a 50% risk of passing it on, blah blah blah.” But I have found over the years that there’s a lot more talking about feelings, and talking about feeling different, and talking about looking different, and people don’t know, I hide it. They don’t know I can’t handle money. Some of the shame. But most people with, females with fragile X syndrome are not in the moderate MR range. Some of them are, but more are in the mild MR range, so they are able to articulate some of their feelings better than people with moderate MR who aren’t often to articulate
their feelings. You have to get really concrete. Like “Does it make you sad?” And you might even say “Does it make you sad or happy that what we’re talking about is that you won’t be able to have a baby when you’re grown up?” So that they actually have to sit there and say “Does it make you sad or happy?” They have to – you have to feed them a little bit, because sometimes that just isn’t something that’s going to come out.

**JM:** When you counsel anyone with an intellectual disabilities, what does it feel like to you as a counselor? Is it frustrating, rewarding, challenging?

**Interviewee:** What it feels like for me is it’s actually sort of maternal, because you’re talking to someone who in certain ways is childlike and in certain ways is adult-like. And they sometimes have adult-like problems and decision, like if they do find themselves pregnant, or they’re not using birth control and they don’t really understand. So they can have adult-like problems, but you’re talking to them often at a level that you might talk to a child. So I actually find myself feeling sort of maternal towards them in that I care about them and I want the best for them, and I do understand their limitations. And I don’t have too much of an agenda. With a child, you often just want to bring something up. “So, did anything happen at school today?” You just want to bring things up. And you just want to give them a chance to articulate something in a safe place, and there aren’t as many safe places for people with intellectual disabilities. And so that’s why I say I think that you do, if you look at the different modes of genetic counseling, and you look into the model, educational versus counseling versus, I don’t know, I have all these various modes now in training. I think it’s more counseling than educational. And generally I don’t feel frustrated, and I generally do feel rewarded, because this person often doesn’t get an opportunity to talk like an adult, to talk about adult stuff, even though they may not have the cognitive abilities of adults. So, and if I found it frustrating, I probably wouldn’t be in this, in my niche in terms of fragile X. It wouldn’t be a good fit for me.

**JM:** What would you say is the most challenging thing when counseling this population?

**Interviewee:** I think the most challenging thing is – that’s a good question. The most challenging thing is that I guess I feel like number one I wish they understood more, or I wish that I had the secure feeling that they were taking something away that’s going to last, as opposed to being forgotten. And I also think that there are times that I work with people with intellectual disabilities and I know that somebody is either going to contradict or counteract whatever work I’ve done with that person. So in other words reproductive freedom, or self-esteem issues, or things like that. So when I know that there are forces going against me, that can be frustrating when I think that the material that we go over is probably going to be forgotten, so why do…it’s like one of those things where it’s like, you gotta talk about chromosomes if you’re talking to a girl about Turner’s, but so much of it’s going to be forgotten that you wonder why you’re doing it. And that’s why I think the pictorial thing is so important, because images do stick with you longer than language, I think, when you have disabilities.

**JM:** Would you say that you’re comfortable counseling this population?
Interviewee: I would say that for the most part I’m comfortable with counseling the population. I occasionally, someone gets referred to me and it’s sort of like someone has an agenda and they want me to “Talk to her about birth control!” or “Talk to her about fragile X and genetics.” And I feel a little forced. And there’s times I don’t connect with the client, or there are times that I do have a little bit of an agenda, and I’m concerned about them, some of the behaviors or them being taken advantage of, and I don’t feel effective. So there are times that it’s frustrating and I’m uncomfortable. So I would say that I’m probably comfortable 75 to 90% of the time.

JM: Do you have any other suggestions for other counselors who are preparing for a session with someone with an intellectual disabilities?

Interviewee: They should read Brenda Finucane's book. And it’s really short and – have you read it?

JM: I have.

Interviewee: It’s really short and really quick. I know it’s like 80 pages, but it’s big type, and it’s quick. And she’s also written, the same year she published that book she wrote a nice paper in the Journal of Genetic Counseling I think. So that’s one thing. I think they may want to talk to other people that have interacted with the client, whether it’s psychologists or social workers or such. And I think when possible, when it’s appropriate, they should consider or try to work with the client one on one. I think when you have a third party there – and that’s why when you asked me about if the social worker had come with that young woman with Turner’s syndrome – I think when there’s a third party there the person becomes even more a child. It’s almost like they brought their mother, even if they’re 20 years old. So I think if possible, if it seems reasonable to try to establish a relationship with the client and work with them without a third party, because a third party will often say things like “Do you understand what she’s saying? Do you hear what she’s…” You know, like, the third party kind of have their agenda, and their agenda is to get some kind of result out of this, a change often. Because they’re a social worker. They have to deal with the person running, or the person doing whatever they’re doing. So but as a genetic counselor, I’m always feeling like I want to establish a relationship and make this a safe place and talk about some pretty sensitive and charged issues. So the person has a…sometimes they’ll just walk away and they’ll “Gee, I know Leanne’s my friend.” And that’s great. I like that. That’s what I like. I like them to feel like I’m their friend. I am aligned with them. Versus “She taught me a lot.” That’s just not going to happen. So I would advise that if people can work with them without a third party, if it’s reasonable. Sometimes it’s not.

JM: I don’t have any other questions for you. Do you have any questions for me?

Interviewee: Well, I’m wondering why you chose this – this is your research project for graduation, correct? You’re a second year student?

JM: yes.
Interviewee: So why did you choose this topic?

JM: So when I was thinking about my thesis topic, I was trying to think about what a session as a new genetic counselor might be difficult for me. And I thought that this situation, counseling somebody with an intellectual disabilities would be a difficult situation for myself in particular, because one, I haven’t had too much experience with this population. And certainly in my graduate school training we don’t get too much experience with the population either, or during the rotations the chance that you might counsel someone who has an intellectual disabilities is very slim. So as a new counselor, I wanted to see if I could talk to other counselors who have had these experiences and see what it’s like, and see what the challenges are, and hopefully help myself when I am a new counselor when I am faced with this situation.

Interviewee: Good. I think the places you’re going to be faced with it the most are going to be in prenatal, particularly if you’re working in prenatal in an urban setting, and if you do do general genetics for pediatrics in terms of often when you see kids who have developmental disabilities, one of the parents has developmental disabilities. So certainly I was at Children’s Hospital in Oakland California for about 15 years, and probably a couple of times a year we saw kid who had a chromosome abnormality and one of the parents did, or VCFS and one of the parents had it. Or even just undiagnosed developmental disability and one of the parents had it. Or both parents had it. So it’s either of those two settings. In adult medicine you probably won’t see it as much, unless you’re dealing with a neurodegenerative clinic, and then the developmental disability is acquired later in life. We see people – do you know [?] [19:20] is? [?] syndrome? So males who have [?] often have some cognitive decline, so sometimes when you’re talking with them, particularly if they’re in their 70’s or so and they’ve had the disease for a little while, you’re talking to people who are at the other end but have lost cognitive skills. It’s a different population and probably not something you’re going to jump right into as a new grad.

JM: Thank you for participating on such short notice.
Interview 7

INT: Hi!

JM: Is now a good time?

INT: Yeah, sorry, the time just snuck on me.

JM: I have a couple of questions to ask you about your experience counseling someone with an intellectual disabilities. But before I do that, I just have some background information that I wanted to get. You’re a prenatal counselor?

INT: Yeah. I thought I’d emailed that back to you, but yes.

JM: And how many years of experience do you have?

INT: Four and a half.

JM: Do you have a family member or close relative with an intellectual disabilities?

INT: No.

JM: So my first question would be a particular situation where you counseled someone with an intellectual disabilities, what the session was like, what the challenges were, what you learned from it, that kind of thing.

INT: Sure. The case that first came to mind when I first read your original email was a case where the patient was referred because she had myotonic dystrophy. And so I went and got her from the waiting room, and I could tell she had, she kind of had a look about her, a little bit of a dysmorphic look. And unfortunately I didn’t have any medical records on her. All I had was the referral that said that she had myotonic dystrophy. So I was kind of winging it. She had her mother with her and also the father of the baby’s mother, and so all three of them came back to the session. And it was a challenging session, because she was obviously, obviously had mental retardation, and she was able to speak with me, but her mother really took over, and it was hard to, both the mothers did. The patient’s mother and then the father of the baby’s mother kept trying to answer all of the questions. And they weren’t even concerned about myotonic dystrophy. They had a lot of other issues that they wanted to talk about. And so it was challenging to me to – I tried to figure out if the patient’s mother had guardianship over her, or if she, who was making medical decisions, because I really felt it was important for the patient’s needs to be heard, but she kept getting overshadowed by the other two people there, so it was difficult.

JM: Did you ask the mother who had power of attorney?

INT: I did. And she claimed that she did, that she had guardianship over her daughter. So at least I was able to find that out. But at the same time I was concerned that the
patient herself wasn’t able to talk as much as I think she might have wanted to. Because she started to engage at the beginning of the session, but then when her mother and the father of the baby’s mother started talking, she really just shut down, and sat there and didn’t really participate any more.

JM: Was she referred because of her myotonic dystrophy?

INT: Yes. That was the reason for referral. And we never even, we talked about it a little bit, but that ended up not being, not even really getting addressed that much, because her own mother, the patient’s mother was not concerned about that at all. Did not want to talk about the genetics of it. She even questioned the diagnosis and didn’t know if that’s what her daughter had. And again, that was another challenge of the case that there were no medical records, but she had a, she had the face. She looked like the myotonic face with the droopy eyes and the mid-face hyperplasia. So I was really suspicious of it, and someone had put that on her referral, so. I really wanted to go over it and talk some more about it and get a medical record released and try to get more information, and they just, basically I don’t think they really, they really didn’t want to be at the appointment. And I was able to talk with them for a while about different things, but I think they had no interest in getting genetic counseling.

JM: What are some of the things you did talk about during the session?

INT: I started, I was able to take a family history, and [?] able to get some information about the patient’s symptoms and just her own medical history. It did come out that the father of the baby was slow as well, so we did talk about, even without a diagnosis of myotonic dystrophy, we did talk about the increased risk for the baby to have delay and possibly mental retardation. And I tried to talk about the inheritance of myotonic dystrophy and prenatal testing, and they just were not interested in that at all. And there were just a lot of other psychosocial issues about the care they were getting, and they weren’t happy with the OB and referring provider, and they wanted to switch providers. And then there was, if I remember correctly the patient was a smoker, so that was brought up as well, and they were trying to get her to quit smoking, and they were talking about that. And then they actually brought up a question about paternity, and so there were a lot of other things that were brought up.

JM: When the patient was engaged, were you able to assess whether she was understanding what you were talking about?

INT: Yeah. I remember she did, she would chime in here and there. Even after she stopped really talking that much, when something was said about her, like about the smoking, she said “I’ve cut down.” So I could tell from certain comments that she made that she was listening and I think she did understand what – obviously she understood what was being said about her. But she, I never got that much of an idea of what her concerns were, because she just didn’t – except for the beginning when she talked a little bit about what she knew about why she was here, she never really said anything else about – I tried to ask her if she was worried about anything, and she just said she was
worried about the process of giving birth, about having the baby, but she never really said that she was concerned about anything else.

**JM:** If there was anything that you could change about this session if you could go back and do it again, is there anything you would do differently?

**INT:** Yeah. I do. Sometimes I wish that I, I understand that her Mom had guardianship, but looking back I wish that I would have asked if it was okay if both the mothers would’ve left the room for a little while, and just ask permission to speak alone, to speak to the patient alone, and just be able to talk with her without someone else chiming in. And really I don’t know why I didn’t do that. I think the case was a little bit overwhelming for me, and it wasn’t what I expected, so I got kind of a little bit flustered, and there was a lot of stuff to address. But I definitely would have done that. I would have asked to have a few minutes alone with the patient and let her be able to talk and see if I could answer any questions for her and see if I could find out more what she was actually thinking.

**JM:** Have you had any other sessions with anybody else with an intellectual disabilities that you’ve talked to about genetic information?

**INT:** Uh-huh. Yeah. Sometimes I’ll, just in general, sometimes I’ll have patients and just from, because I usually take the medical and family history at the end, and so for example if I have someone that’s referred for advanced [maternal[9:00] age or an abnormal screen, we’ll go over that information first, and then I’ll take a history at the end. And sometimes when I’m talking to someone, I may think that perhaps they’re not quite understanding it and I’ll try to break it down to something more simple. And I’ve asked people before during the family history if I think they seem like they might have some delay or be a little slow, “Were you in special classes in school?” and try to assess that, because a lot of times people won’t just come out and say “I’m slow” or “I have some delay,” but they’ll be able to tell you that they were in special classes and things like that.

**JM:** I’ve heard that from a few of the other counselors that I’ve spoken to as well, that they’re so forthcoming admitting that they are a little slower.

**INT:** Yeah. And it’s hard – and you want to address it, I feel like you want to be sensitive about it and you don’t want them to feel any sort of shame or feel bad about anything, but I also think it’s important to know that if someone’s understanding, obviously you want to make sure someone’s understanding what you’re saying, and I do like to talk about the risks for the baby if one or both of the parents are slow.

**JM:** As a genetic counselor, how do you feel when you counsel people with an intellectual disabilities? Is it frustrating, is it rewarding? How would you describe it?

**INT:** I think to me most of the time it’s actually rewarding, because I like feeling like, well, I like feeling like I have, that’s one of the biggest things I like about my job is when I feel like I’ve taught someone something or if I’ve helped them understand something.
And when you feel like you do make that connection with someone, and that you’ve been able to successfully explain age or a quad screen to them, that can feel really good, because I think a lot of the times no one else has been able to take the time to really sit down with them and explain things. And that’s job and that’s what we specialize in, but a lot of time doctors or the nurses they see at prenatal visits don’t really have much time explain what this test is or what they’re getting done, and they may be confused about things, and it’s nice for me to feel like I have the time to help them understand something. So in general I’d say it’s rewarding. It definitely can be frustrating. That case I was telling you about at the beginning where the patient I think had myotonic dystrophy. That was just, to me that was just overwhelming and I don’t think I handled it the best way that I could have. But it’s a learning experience, and I’ll know what to do, hopefully I’ll know what to do next time.

JM: Would you say you’re comfortable counseling this population?

INT: I guess I’d say on the whole yes, but I think my experience has mostly been with people that have mild mental retardation, and I don’t have, the patient with myotonic dystrophy was definitely I think would be in the moderate category. So I don’t have much experience with moderate or even people with severe mental retardation. I think I might be a little more uncomfortable with those cases, just because I don’t have as much practice. But I think people that are on the milder end of the spectrum I feel pretty comfortable with.

JM: Do you have any suggestions for other counselors who are either preparing for a session with someone with an intellectual disabilities for the first time, or what to do before to prepare, or during the session of how to explain things to them or talk to them?

INT: One thing I’ve learned, especially when you’re trying to describe a risk, or a risk figure or number, that a lot of times having something tangible can really help. So one of my co-workers actually has a thing of marbles, and it’s full of blue marbles, and there’s one red marble. So she’s used that sometimes to show that you have one chance out of fifty, so what’s the chance you’ll pull this red marble out of the blue marbles? And we also have a lot of patients that English isn’t their first language, a really high Spanish speaking population, and so we’ve used that to help with language issues too. But I think sometimes it’s hard when you get into more abstract concepts it’s harder for people that have learning disabilities or mental retardation to understand that as well, so I think, or even a visual aid where there’s little outlines of people and one person is filled in. To me it seems helpful to have a visual aid or something like that to help explain a number like that. And then another thing I think is important is figuring out who has, like you were saying, power of attorney. Can they consent themselves? Can they sign to have an amnio? Things like that. Because that’s not an issue you deal with every day.

JM: Did your patient have an amnio?

INT: No. She had an ultrasound. She didn’t do any testing.
JM: I don’t have any more particular questions for you unless you have anything else to add.

INT: No, I don’t think so.

JM: Do you have any questions for me?

INT: Is this audiotaped?

JM: Yes.

INT: And you got my consent?

JM: I did.

INT: So it’ll be, I’m sure it’ll be anonymous when you publish [?][15:20]. Just like quotes and stuff.

JM: Yeah. And anything that I have, any of your information that I have is password protected on my computer as well.

INT: I think this is a great project. And thank you for asking me to participate.

JM: Thank you for participating. I really appreciate it.

INT: I’m excited to read what you come with.
Interview 8

**Int:** Hi, how are you?

**JM:** Is now a good time?

**Int:** That’s great.

**JM:** So thank you for participating. I really appreciate it. I just have a couple questions to ask you about your experiences counseling someone with an intellectual disabilities. My first question is that if you could tell me a little bit about a situation that sticks in your mind that involved a patient with an intellectual disabilities. What the nature of the session was, any of the challenges you encountered, things like that.

**Int:** Okay. Even though you told me to prepare for this I…so I guess the one that sticks out in my mind is one that I run into especially, I guess it depends upon, based, I’m trying to speak actual words and it’s just not coming. Sorry about that. I run into a similar problem between any level of learning difficulties slash also adolescent boys. And this is a younger man, I think he was in his twenties, and pretty significantly delayed. He’s the worse off of the two brothers. He’s the younger of the two brothers. They both have some pretty significant MR, which Mom was calling autism because I think it’s more socially acceptable to have autism than mental retardation at this point. So it was kind of trying to be sensitive of his needs while figuring out what he needed, but being sensitive about his disabilities, while also having him ask me out on a date multiple times. So kind of a socially incorrect situation combined with obviously it wouldn’t be performed, or that wouldn’t happen to a person who is not otherwise intellectually delayed. So that one of the more uncomfortable situations that just stands out as compared to others.

**JM:** What was he referred for?

**Int:** He was referred for autism, and I think there was a question of could he have [?][2:30] syndrome.

**JM:** Did you talk to him directly about his mental retardation or counsel him in anything particular?

**Int:** Honestly, no. To him, his level of understanding wasn’t…he can work in a warehouse. Before the economy went down the tubes he was employed in a very sheltered environment and was able to do some tasks, but he was not completely to the point where he understood that he even had an intellectual disabilities. So we talked about him being there. And I always will talk to either children or younger adults who are functioning on the level of children trying to explain that this isn’t going to hurt, we’re going to start by doing a lot of talking. We’re going to try to, I know you have a few concerns, whether it’s belly pain or it’s this or that, and we’re really here to try to figure out if we can find an answer for you, and maybe better how to treat those things. So I think I talked to him more in that sense, and then, but as far as counseling, we A never
really got an answer for this family as to what’s going on, but I think he was kind of left out of the end discussion. Other than “You’re healthy. You’re doing well. I’m glad that you’re helping your Mom around the house.” Those kinds of things.

**JM:** Do you think that he was able to understand what you were telling him in that respect?

**Int:** On that level, yes.

**JM:** Have you ever counseled anyone who maybe is on the more mild spectrum?

**Int:** Certainly.

**JM:** And what was that situation like?

**Int:** That one is even more touchy because I feel like when people are understanding of what you’re asking them, it’s embarrassing. It’s a source of embarrassment. A particular time isn’t standing out. But it’s interesting to see the delineation between different people. There are some people that say “Yup, I had learning problems in school. Yeah, I’m not so good at this, that or the other thing, but I get by.” And then there’s other people that are certainly more sheltered in what they’re, “No, I didn’t graduate, but it’s because I didn’t like school.” Or “No, I didn’t do this because I didn’t…” You know, kind of making excuses for it. And in those situations I feel like I really have to counsel them in part for their children. But trying to explain that the reason that we’re asking such invasive questions and such sensitive questions is really to learn the most about their child, and trying not to downplay all the things that they have done, and trying to make a super, make it a big point of, I don’t know, just trying to use it as a way to find information out about their child, and that I’m not grilling them on their inabilities in school and things like that.

**JM:** During these sessions, how are you able to assess the level of understanding of the patient?

**Int:** For the patient themselves, we usually have a pretty good feeling based on the parent. Our first question is “Do you know why you’re here? What are you main concerns? Are there any questions that you want to make sure are answered today?” And a lot of the time it is “My child has problems in school,” and we want to [?][6:20]. So that certainly helps, but when things are a little bit more indecisive, just not as clear, we try to get a handwriting sample. The kids are usually bored, so “Can you write on the paper for us? So we can see how well you can write your name. How well you can write first name and last name.” Sometimes we’ll ask them to do a really detailed picture. So “Let me see your house. Can you show us…How much detail can you put into a house?” Trying to ask them what their favorite book is, what they’re reading in school. Asking about what level of math they’re doing, what level…kinds of things like that. And then finally asking “What grade are you in? What school do you go to? How many kids are in your classroom?” And then if it’s still kind of unclear – I mean, every time I do ask parents
“Does your child have an IEP? Is there anything that your child needs extra help with in school?” Are they getting therapy, physical therapy, speech therapy, stuff like that.

**JM:** During a session, when you’re talking about particular issues, is there any way that you are able to assess how much of what you’re saying they understand?

**Int:** A lot of the time I try to…well, it depends. It depends whether it’s a child that I’m talking about and I think the parents are pretty parent, or I’m concerned about the competency of the parents. So asking to be, things to be repeated back. “Okay, so we talked about this, how would you put it into words?” Asking…and I haven’t used this as much as I thought I would when I was in counseling school, and I think it works a little bit better, or probably would use it more in a prenatal setting, but “If you’re going to go home and tell this to your husband, or tell this to your Mom or brother or sister, how would you tell them about this?” And that’s one that just talking to you I realize that I haven’t really used in a really long time, and I really like that technique, at least as a student. So I’m glad that you asked me, because I might try it [?][8:34]. But really trying to have them verbalize back to me at least a little bit of what I’ve said. But a lot of the time with a parent, based on the questions they ask you, you can really determine, you have a pretty good idea of where they’re coming from. If their questions totally don’t make any sense from what you just told them, then you know that you need to go back to the drawing board. If they seem to be appropriate questions for the conversation setting, that’s a pretty good gauge for me.

**JM:** As a counselor, what would you say it’s like to counsel this population? Is it frustrating, rewarding?

**Int:** It’s definitely all of those things. You’re frustrated – I’m not frustrated with the patient themselves. I’m usually frustrated with the social situation that they’re in, because that social situation makes it so much more difficult to get things done. It’s rewarding when you do finally get an answer for them or get them to complete a task, whether it be going to the blood station on the right day at the right time. It is like a little feat, but it’s frustrating that I have to call a family fifteen times to get them to do it before their authorization runs out. Those things make it a little bit difficult. But when you do have a family that you know just genuinely couldn’t do something on their own and then accomplishes that task, it’s nice to see that through from the beginning [?][10:15].

**JM:** Do you have any suggestions for other counselors who are preparing for a first session with a patient with an intellectual disabilities, either what to do beforehand or how to approach the session during the time?

**Int:** I think a lot of it has to just do with assessment of…and I think a lot of assessment comes from experience, but just general intuition. If you’ve been good at assessing situations since you were a little kid, you’re probably going to be good at assessing situations once you’re a counselor. And just assessing in what’s the tone of the parents? Do they come up as savvy with anything? Or do they come off as “We don’t really know much, but we’re really willing to learn.” Just keeping an open mind about what you have
in front of you, and what might be there but you can’t see. The underlying, the culture, the language. Just really having an open mind to what they actually might be there for. That really isn’t helpful at all other than just trying to be open to any possibility that could come across. And being sensitive and really treating the patient and their family how you would want somebody to treat you. You wouldn’t want somebody to talk down to you. You would want somebody to talk on your level, but still have enough control over the situation to be able to give you information. So I do water down what I’m saying based on my assessment of the patient and their family. That might always be completely correct, but talking at their level for what they’re going to understand is obviously important. I think assessment is a big part of that.

JM: Would you say that you’re comfortable counseling this population?

Int: Yes and no. I think I’m comfortable for the most part, but I think anytime you get too comfortable you miss things. So I do like to prepare a lot, a lot, a lot for my cases in general. I like to have background on people. I don’t like to go in cold, because I feel like they’ve been through so much and they’ve told their story so many times that it’s kind of rude not to. So on that level, I make myself as comfortable as I can be, but knowing that anything can change at any moment, so just not getting complacent with what I’m doing.

JM: I don’t have any other particular questions for you, unless you have anything else to add?

Int: Not particularly.

JM: Do you have any questions for me?

Int: How’s the thesis and everything going?

JM: So far so good. I’ve had a lot of counselors give me a lot of great feedback, so I’m not really sure what I was expecting when I started, but I’ve definitely been pleasantly surprised. So it’s going very well.

Int: And how many more interviews do you have to get?

JM: Two more.

Int: So the end is near. I had a classmate who did interviews and it was so hard to get the people to actually call her. She was doing patient population, so I think they are a little less reliable than the counselors maybe are. But you might be able to tell me something different.

JM: No, the counselors have been great that I’ve talked to. I think that a lot of the counselors that I’ve spoken to are very interested in this topic, and they wouldn’t have participated if they didn’t want to. That’s definitely nice to be able to do that.
**Int:** I think it’s a really great project idea. And it’s hard when you’re in that situation, when it’s you and say a 14-year-old kid who does have learning disabilities. And it’s you and them and Mom in the room. And either Mom’s downplaying their problems, or making more of them, and it’s an uncomfortable situation. You’re trying to deal with that 14 year old the best way that you can, but still take Mom’s concerns into account. I think it’s important, and I think it’s a really great project idea.
Interview 9

INT: It was a young woman who was referred in because of her intellectual disabilities, and the concern for risk to the pregnancy. And she had never really had a genetic evaluation herself, and so they wanted to get that taken care of and help her understand what risks may be present in the pregnancy. And she came to the appointment with her mother, and the woman herself that was disabled was probably I want to say in her mid 20’s at the time that I was seeing her, with her first pregnancy. And she was in a relationship with a gentleman who also was intellectually disabled. She, this patient was currently living at home with her mother, but had intended to leave and go live with her partner at some point in time. It was a difficult situation, because while I was with her and her mother, the patient – let me preface this by saying the patient had met with one of our social workers from our high risk clinic prior to coming to see me, and had sort of been made aware that because of her cognitive deficits there was a concern with how well she would be able to care for her child, and how well she could really understand the information that was being presented to her. And she acknowledged that she had limitations, but that she really felt with her mother there she could do everything, and then if her mother wasn’t there, there would be no problems as long as her partner was with her as well. And when I started talking with her, she was very concerned that the social worker was going to contact the local child protective services for Department of Child and Family. And that really became the focus of our discussion. She was not at all concerned about her child having any physical or intellectual deficits. She didn’t really care to figure out why she had the problem she had. She was not interested in having a genetics evaluation done by one of our geneticists. Her focus, and really just took over the session, was “Is DCS going to come in and take my baby away from me? I’m a competent person. I can do this. I can get help. And I promise, I promise, I promise I will.” And when we discussed the issue with her partner also having some limitations as well, she was very resistant to that, and felt that despite the fact that he had these limitations he still could be an adequate, together they could make adequate decisions for raising the child. And her mother pretty much sat quietly through the majority of this and didn’t add a lot to it. And then we spent some more time talking about some tests and recommendations, but she declined everything. She didn’t want to do anything. She was just very happy to be pregnant, very happy to be having a baby, very happy to be in a relationship, and didn’t want to do anything else. So I explained to her that I would be writing a letter to summarize our discussions and my concerns, and I made it very clear to her that I still, that myself and the social worker were still very concerned with her ability to be able to provide the appropriate care, and if she was going to be moving out of her mother’s home, we wanted to make sure that there was someone there to help her and her partner out, and that likely information would be passed on to our local Department of Child and Family Services. And she reluctantly agreed to that at first, as long as she was promised that they wouldn’t immediately take the child out of her care. Well, interestingly enough, the letter went home to her as well as to her physician and her partner. Got the letter and read it, and called me furious. Screaming at me, yelling, was very upset that we would pass any of this information on, that they were very competent people, they could handle the situation, they didn’t need anybody’s help, and they didn’t want the Department of Child and Family Services to be involved, and he wanted me to
rewrite the letter and take that information. He was very confrontational and very, very upset and very angry. I offered the option for him to come in to see me and meet with me, and he declined, and he made all kinds of threats that he was going to contact the institution and he was going to file charges against us and all these kinds of things. It was really interesting. And nothing ever came of it. She continued to follow up in the high-risk clinic. And I met with her one other time real briefly, again trying to just chat with her. She was very non-confrontational. But she didn’t want to pursue anything. Just didn’t want to do anything. And I, myself and her mother really just weren’t convinced that she truly understood what was going to be involved in raising a child and understood why we wanted her to consider having testing and what benefits that might have for her and how that could potentially be useful information for her child, once her child was born. We just never really were convinced she got it. But then after that I never had any more contact, and the Dad never again called us. It was really interesting. Because it was very unexpected when I got that phone call from him. Very unexpected. And he was so angry! Oh my!

**JM:** You’ll probably always remember that session.

**INT:** Yeah, definitely. Definitely. And then I had another, another very, I mean, two that really stand out. The other one also might be relevant to you. Another woman who herself was also intellectually disabled. Functioned probably in the mild MR range. And was married, and the pregnancy ended up being affected with spina bifida. And they, she and her husband came to see me, and I think her mother might also have been in attendance. I can’t remember. I think there was a third person there. So it was very clear that she did not really understand what spina bifida was, so I spent an extensive period of time discussing the impact of that, and I believe the fetus had hydrocephaly as well. We talked all about the details of that. And as we finished going through that, talking about options and how we would follow up. I think an amnio had been done and chromosomes were normal, I believe, or we were talking about an amnio, I can’t remember the details. But when we finished talking about all of the options — did they want to consider, would they consider adoption, would they consider termination, what did they want to do, she very clearly said that she was not comfortable with termination of pregnancy, that regardless of what was going on with the baby this was a child she wanted and she wanted to care for and would be there for and that would be fine. Her husband, however, felt completely opposite. And he made it very clear that he could not be in a position to have a child who had special medical needs. That he had a difficult enough time providing for his other child and taking care of his wife. And he was very I don’t know if confrontational but very upset with her. And he said “Look, I work full-time. I have to take care of you. You can’t drive on your own. You can’t go anywhere on your own. You can’t pay the bills on your own. You can’t go to the grocer on your own. I have to be the one to do all of that for you, and if you’re talking about bringing a child into this world who is going to require multiple follow-up visits and multiple specialists and have multiple handicaps, I can’t continue to do that, and we need to terminate this pregnancy.” And we had this discussion back and forth about what would be an option for them, and ultimately he said to her, because she was pretty, she held the line and pretty much said “I just can’t do that. I don’t believe in that. I can’t do it.” And he said to her, “If that’s the
choice you ultimately make I can’t stay in this relationship any longer, and I will leave.” And I was like “Maybe there’s some options,” and so we talked a little more about adoption. And she was open to hearing more about that, and so I explained that I needed some more time to try to research options for them. And I spent a lot of time doing that, trying to get information, and called her back multiple times to try to discuss what I found, and never heard back from them again. So I have no idea what ultimately happened. But it was a very difficult counseling session for me, because I’ve never had two people so divided on an issue and actually have a husband say to a wife “I will leave if you do not terminate this pregnancy.”

JM: That was during the session he said that?

INT: Absolutely during the session. I felt really inadequate in my counseling skills at that point. And I contacted a professional counselor. I tried to encourage them to go see a professional counselor who specializes in these types of situations, and I gave them the information. I don’t think they ever followed up, though.

JM: Was the husband intellectually disabled?

INT: No. He had, he was normal intelligence, he held a full-time job, had been previously married, had a child from a previous relationship, and was basically, the words he said was “I already have two children that I take care of,” meaning his wife and the other child. “I can’t bring another one into the situation who has physical and potentially intellectual handicaps.”

JM: It sounds like in the first situation you weren’t sure how much the patient understood what you were telling her. How were you able to assess how much the patient understood?

INT: Generally in those situations I try to ask them questions after we’ve gone through all the material, to have them try to reiterate back to me their level of understanding. And I also always start every session that I see in these scenarios by asking them “Can you tell me what you understand about why you’re here, what’s going on with your pregnancy, why it’s important for you to come see genetics?” So that’s how I start just about every session, because that allows me to try to assess what their level of understanding is, and ask them what questions. I also start by saying not only “Did you understand?” but “What questions do you have?” right away. And then I go from there. And then after we’ve gone through everything, I ask them to try to, “So tell me what you understand after our talking about all of this,” and see if they get it.

JM: Do you feel like that works well?

INT: Most of the time. I really do. I think most of the time it works well. It gives me a pretty good idea. And like I said, with that first session, I was pretty sure she didn’t understand the significance of everything, despite all the discussion.
JM: In either session, is there anything you would do differently if you could do the session again?

INT: Hindsight’s 20/20, right? With the first session, I just don’t know that there’s anything that I could have done differently. The second session, I, now, I couldn’t done it at the time because I wasn’t expecting it, but I’ve spent a lot more time now trying to figure out how to help people, how to work with people who are so divided on the issue of whether to continue a pregnancy or not. Because that really threw me for a loop in that session, with the gentleman sitting there basically telling his wife “I consider you to be so impaired that I have to treat you like a child, and if you don’t listen to what I tell you, I’m going to leave you and you’re going to be on your own.” So I don’t know at that time how I would have done it differently, and still at this moment I can’t tell you how I necessarily would do it.

JM: I don’t think anybody would expect that to come out of anyone’s mouth during a session.

INT: Yeah. It was really very difficult, and it was so awkward, and I just almost fell out of my chair when he said that.

JM: I’m sure I probably would have done the same thing.

INT: And I’m a pretty seasoned counselor. At the time, I guess I’ve been doing this now for seventeen years, so probably at the time I had been doing it for 14, 15 years. So I don’t know. I don’t think there’s much I could have done differently at the moment.

JM: Would you say you’re comfortable counseling this population?

INT: I would say as comfortable as you can be. I just had a, well, it was supposed to be a similar situation yesterday, an intellectually challenged patient. She was, but she was far more competent than I was led to believe from all the notes, so that was not at all challenging. She was much, much more competent than I was led to believe.

JM: One of the questions I asked in the background information is if you have a family member with an intellectual disabilities. Do you think that having the family member with an intellectual disabilities has made you more comfortable speaking with this population?

INT: Yeah, I would say it has, actually. Yeah. I definitely think so, because of just the interactions that I have on a daily basis. And I think also because when I’m doing most of this counseling I’m doing this mostly in my prenatal arena, but I do pediatric genetics as well, so I’m constantly involved with individuals who have intellectual disabilities in my pediatric environment too.

JM: Did you grow up with this relative?
INT: Yeah.

JM: If you had any suggestions for other counselors who are preparing for a session with patient with an intellectual disabilities, what would you suggest to do either to prepare for a session or during the session how to go about that?

INT: I think to prepare for the session, the main thing to keep in mind is to try to keep it as simple as possible, as basic as possible. Be prepared to use multiple analogies that are real-life everyday analogies that people can relate to, as simplistic as can be. And to be very flexible in your counseling agenda, because it’s often not going to go the way you hope it’s going to go. I think those are the main things.

JM: I don’t have any other particular questions for you, unless you have anything you want to add?

INT: No, I don’t think so.

JM: Okay. Do you have any questions for me?

INT: This is for your thesis? Is that right?

JM: Yes.

INT: And are you going to be doing any kind of presentation afterwards?

JM: Yeah. We do a presentation at the end of the year, so it’s mid-May I think is our presentation. It’s for our program.

INT: At your program. Okay. So no posters or anything, at this point, or plans?

JM: At this point, no, but most of the counselors that I have spoken to have been very interested in hearing the results of my study, so if I do anything formal like that I will certainly let you know.

INT: Particularly if there’s any kind of suggestion list, like you were just asking, ways for people to prepare. Yeah, I would definitely be interested. I thought you said this was like 30 minutes long!

JM: Almost. Well, I said 30 minutes, so you expect more time, because sometimes I don’t want to say it’ll be ten minutes and it runs for half an hour.

INT: How many people have you spoken with?

JM: So far I’ve spoken with about eight.

INT: And what was your goal? How many did you want to have?
JM: The goal was 12, but that’s not going to happen. I think I’m going to have about ten.

INT: Is that because you didn’t get enough people who responded to you?

JM: I got enough people, but something happened with some of the interviews that they got lost, so I didn’t use them.

INT: Oh, no!

JM: So yeah, I’m only going to have about ten. Originally when I first started we’d only planned on eight, but they were like “No, let’s make it more,” so ten I guess is a good compromise.

INT: So you’re somewhere in the middle.

JM: Yeah.

INT: Well, good luck with it. I hope you’re able to get some interesting information. It’s a great topic. It can be a difficult counseling session, but at the same time a pretty interesting one.
Interview 10

JM: Okay. It was only recording my voice and not yours. But I think I fixed it. Could you repeat what you just said about [?].

INT: Yeah, that’s fine. I guess I’m just, I’m remembering a genetic counseling session I had that was with a woman who, the chart had said that she had mild mental retardation, and she had 22Q [?] deletion syndrome. Her mother was there for part of the session, and I remember her not wanting her mother to be there. But I think I was hesitant at first, because even though I knew she had mild mental retardation, I didn’t really know what that translated into as far as what she could and could not understand. So I remember initially being somewhat uncomfortable and trying to cover up that discomfort, and increasing realizing that she was really funny, had a good sense of humor, and that, I think what I learned in general from the session was that working with people with intellectual disabilities can actually be very enjoyable. I found myself laughing during the session, and not at her, but with her.

JM: Did you find that she was able to grasp the information that you were giving her?

INT: She did. But had this been an initial genetic counseling session where I was telling her about her condition, I certainly would have had to give the information in a different way than I would if it was a genetic condition for somebody who did not have any intellectual impairment. But I did feel like she understood what I was saying. It wasn’t on a very scientific level, though, because this was a follow-up visit.

JM: How were you able to assess that she was actually understanding?

INT: I would usually ask, I would say “Did what I just say make sense?” And she might say “Yes” or “No” but if she said “Yes” I’d say “Tell me in your words what you think I just said.” And she would say back to me what her understanding was. And if it was a little bit off, I might say “That’s basically what I meant, but I also meant this. Do you understand what I’m saying now? And what do you think I mean now?” So to keep, sort of keep asking her that question. “What do you think I said in your words?”

JM: Do you feel comfortable counseling this population?

INT: Do I feel comfortable counseling with which population?

JM: Anybody with intellectual disabilities.

INT: I think I feel somewhat comfortable, but I wouldn’t say that I feel, I don’t know that I would use the word “comfortable.” I feel, I’d say I feel more comfortable counseling somebody who has an intellectually disability than I do, say, somebody who has a PhD in molecular biology, in some ways. But I wouldn’t say that I’m entirely comfortable. Certainly comfortable, but not as comfortable as I am say counseling somebody who I feel is the same intellectual ability as I am, say.
JM: If you were to go back and do that session again, is there anything you would do differently?

INT: I’m trying to think. I don’t think that I, I don’t think that I would have changed anything, no. I feel like I had a good rapport with her and like she understood what I was saying. So no. I mean, I think if there was anything I would have changed it would just be myself knowing ahead of time that I didn’t need to be nervous. I think that the discomfort comes with not so much that the person has an intellectual disabilities, but just not knowing where they lie on that spectrum. Because somebody with major mental retardation is very different moderate mental retardation, which is very different from mild. And even within the mild mental retardation there’s a spectrum. So I think the discomfort for me lies more with not quite knowing what footing I’m on when I step into the session.

JM: Do you have any suggestions for other counselors who are preparing for a session with a person with an intellectual disabilities, or things they could do during the session?

INT: So suggestions for counselors who are going to be working with people with intellectual disabilities?

JM: Yes.

INT: I think there’s something to be said for engaging in more small talk in the beginning to [?][5:08] out where they are on an intellectual level, both emotionally and cognitively. That’s one thing I would say. And what else? I think if there’s any way ahead of time to get a sense maybe from the parents what, not even necessarily what age level somebody is at, because even that doesn’t necessarily mean a lot to me. Like sometimes I’ll read in the chart “The patient is at a 4th grade level.” I’m like, I don’t even know any fourth graders. So if there’s a way to say to the parent ahead of time “What…” I don’t know how I’d even phrase the question, but maybe say “What types of things can Johnny understand and what types of things does he have more difficulty understanding? What level is he at gradewise?” Something like that.

JM: In general, from all of your experiences counseling people with intellectual disabilities, do you have anything to add about how to talk to them, or anything else that I haven’t asked about that you’d like to share?

INT: How to talk to them?

JM: Yeah.

INT: I think in a lot of ways you’d be doing the same things as you would be for anybody else, so with kindness, with compassion. And I think sometimes we have a tendency to worry too much that we’re either going to be talking above their level or below their level and they will somehow get offended. But I think that in general people
with intellectual disabilities, say they feel like they’re being condescended to, they’ll probably let you know in no uncertain terms, and it won’t necessarily be in a hostile way. So maybe just be as relaxed as you can be. And what else would I say as far as advice, talking to people, counseling them. I think a big one is to just really make it clear that you respect them as a person, and just because they have this intellectual disabilities per se doesn’t mean that they don’t have hopes, dreams, questions, thoughts. And to just make sure that comes across in your counseling, that you respect them.

**JM:** Do you have any questions for me?

**INT:** I’m just curious. The study, is it a qualitative study?

**JM:** Yeah. So I’m doing twelve interviews. Counselors in prenatal, adult, pediatric, all of those [specialties?]?

**INT:** Okay. It’s an interesting topic. And I guess the reason also why I was interested in this study is because I’m now doing research, so I’m working at a clinic that mostly sees kids with autism. Kids and adults. But also other developmental delay, just general developmental delay, but mostly autism, and so I’m consenting family members for this research study, which is just trying to find more genes associated with autism. And I find it really interesting in that, because autism, talk about a spectrum, not only is there Asperger’s, [?] [9:12] autism, but even within Asperger’s, there’s a spectrum within the spectrum. So I find myself thinking about this a lot when I’m about to meet with a family, because it’s not enough to say “This child has autism.” You have no idea. If somebody says a child has autism, whether or not they are five years old or 14 years old, you have no idea where they are on that spectrum intellectually. So I find it really interesting trying to [?] [9:40] during the consenting process, is this somebody who can give assent, or is it just going to be the parent signing for the child? And I guess that’s why I was interested in the study, because I find myself thinking about this topic a lot, and how it’s hard sometimes to figure that out. So I think it’s a worthwhile topic to pursue. I’m glad you’re doing the study.

**JM:** Most of the counselors that I’ve talked to have been very interested in the results of my study as well.

**INT:** Yeah. I would love…I don’t know if you have any plans to get back to anybody about the results, but I’d love to either read the study, or read the abstract, or…

**JM:** From all the feedback that I’ve gotten and all the counselors that are interested in hearing the results of my study, I definitely will put that in my plan to let all of you guys know [?].

**INT:** I’m sure you’ll be really stressed out towards the end trying to get your whole project together, so even if it’s after you give your final presentation at Brandeis, even if it’s after that, that’s fine, too. But someday I’d like to hear about it. Certainly give me a call or email if you think of any other questions to ask me.
APPENDIX E: GRANT PROPOSAL

Patients with Intellectual disabilities:
The Genetic Counseling Perspective

Principal Investigator: Jacquelyn Magner

Committee Members: Beth Rosen Sheidley, MS, LGC, Sharyn Lincoln, MS, CGC, Jonathan Picker MBChB, PhD, Marisa Likhite, MS, CGC

Statement of the problem

The definition of intellectual disabilities has long been debated, but most would agree that there is not a clear-cut answer because it is difficult to determine whether someone has a mild form of intellectual disabilities or instead has cognitive abilities on the lower end of the “normal” spectrum. Intellectual disabilities is defined as “cognitive functioning that is significantly below average and begins in the developmental stage” (Accardo et. al. 1998). In order to be diagnosed with intellectual disabilities, an individual must have significant delay in at least two areas of adaptive functioning, which include communication, self-direction, health and safety, functional academics, leisure, and work (The American Association of Intellectual and Developmental Disabilities (AAIDD). The number of people with intellectual disabilities who are making reproductive decisions each year is on the rise because many women with intellectual disabilities devote a lot of their time to downplaying their intellectual disabilities and trying to pass as “normal” in society. Many of these women feel that the ultimate way to acculturate themselves is by giving birth and mothering a child (Finucane, 1998).

In recent years, mental health professionals have shifted toward focusing on the strengths of people with intellectual disabilities (Greenspan, 2006). There has been an increased recognition that people who have intellectual disabilities are capable of taking part in many activities and making decisions on their own behalf. There has been considerable research focused on counseling individuals with intellectual disabilities regarding mental health issues. However, there is a dearth of research regarding genetic counseling for patients with intellectual disabilities.

In her 1998 book, Working With Women Who Have Mental Retardation: A Genetic Counselor’s Guide, Finucane addressed key strategies that could be helpful for genetic counselors when counseling a patient with intellectual disabilities. One strategy is to provide a discussion of important psychosocial issues rather than following the more traditional genetic counseling model, because many of the strategies that are used in a traditional counseling session are not effective when working with women who have intellectual disabilities. Other strategies suggested by Finucane include the use of visual aids to discuss risks, and assessment of how the patient views his/her intellectual disabilities. Information giving is often the focus of a genetic counseling session, but
shifting that focus to the patients thoughts and feelings has proven to be a more successful approach.

Unfortunately, Finucane does not present specific situations or scenarios, which might be helpful to genetic counselors struggling with how to best explain important concepts to a patient with intellectual disabilities. The purpose of this research project is to explore the experiences of genetic counselors who have counseled patients with intellectual disabilities. Through qualitative interviews with practicing genetic counselors, I plan to gather information about specific patient interactions, challenges faced, and strategies used.

Methodology:

Sampling Methods: I plan to recruit genetic counselors via the National Society of Genetic Counselors’ (NSGC) listserv to take part in a 30-minute audiotaped phone interview. Participants must have counseled at least one patient with a diagnosis of intellectual disabilities, and must live/work in the U.S. I plan to conduct a maximum of 12 interviews and provide each participant with a $25 gift certificate upon completion of study participation.

Data Collection Plan: The interviews will take place via phone and will be audiotaped. I will use a semi-structured interview guide, which will focus on the counselors’ direct experiences with patients.

Data Analysis Plan: The interviews will be transcribed and then coded using Atlas qualitative analysis software. I plan to use comparative and descriptive methods to analyze the data.

Funding:

The funding required for this thesis project involves transcribing the phone interviews, consultation with a qualitative analysis expert, gift certificates for the participants, and refreshments for 2 committee meetings.

The purpose of transcribing the interviews is to have documented data that I will be able to code and analyze using Atlas qualitative analysis software. The purpose of meeting with a qualitative analysis consultant is to help me code my interviews and analyze the data. The gift certificates are a token of appreciation for the genetic counselors who offered their time and experiences to participate in an interview. The food and drinks at committee meetings is a way to thank the committee members for taking the time to be on my committee and providing me with their expertise, insights, and suggestions.

References
APPENDIX F: IRB PROPOSAL

Patients with Intellectual disabilities: The Genetic Counseling Perspective

STUDY PURPOSE
The purpose of this study is to explore genetic counselors’ experiences with counseling patients who have intellectual disabilities. I anticipate that the counseling experiences shared by the genetic counselors participating in this study will be a useful and guiding tool for other genetic counselors.

STUDY SPONSOR
Brandeis University Genetic Counseling Program. The principal investigator has also applied for funding from the Brandeis University Graduate Students of Arts and Sciences Master’s Research Fund (Appendix D). The decision regarding this fund is pending at this time.

PRINCIPAL INVESTIGATOR’S QUALIFICATIONS TO DO THE RESEARCH
The Principal Investigator is a student in the Brandeis University Genetic Counseling Master’s Program and is conducting this research project as a requirement for the Master’s Thesis. The faculty advisor is Beth Rosen-Sheidley MS, LGC. Professor Sheidley is a Professor of the Practice and Co-Director of Research and Professional Development for the Brandeis University Genetic Counseling Graduate Program.

RESULTS OF PREVIOUS RELATED RESEARCH
The definition of intellectual disabilities (ID) has long been debated, but most would agree that there is not a clear-cut answer because it is difficult to determine whether someone has a mild form of intellectual disabilities or instead has cognitive abilities on the lower end of the “normal” spectrum. Intellectual disabilities is defined as “cognitive functioning that is significantly below average and begins in the developmental stage” (Accardo et. al. 1998). Intellectual disabilities is diagnosed when an individual has significant delay in at least two areas of adaptive functioning, which include communication, self-direction, health and safety, functional academics, leisure, and work (The American Association of Intellectual and Developmental Disabilities (AAIDD). The number of people with intellectual disabilities who are making reproductive decisions each year is on the rise because many women with intellectual disabilities devote a lot of their time to downplaying their intellectual disabilities and trying to pass as “normal” in society. Many of these women feel that the ultimate way to acculturate themselves is by giving birth and mothering a child (Finucane, 1998).

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strengths of people with intellectual disabilities (Greenspan, 2006). There has been an increased recognition that people who have intellectual disabilities are capable of taking part in many activities and making decisions on their own behalf. There has been considerable research focused on counseling individuals with intellectual disabilities regarding mental health issues. However, there is a dearth of research regarding genetic counseling for patients with intellectual disabilities.

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Unfortunately, Finucane does not present specific situations or scenarios, which might be helpful to genetic counselors struggling with how to best explain important concepts to a patient with intellectual disabilities. The purpose of this research project is to explore the experiences of genetic counselors who have counseled patients with intellectual disabilities. Through qualitative interviews with practicing genetic counselors, I plan to gather information about specific patient interactions, challenges faced, and strategies used.

SUBJECT CHARACTERISTICS
Study participants will be genetic counselors who have experience counseling patients with intellectual disabilities. I plan to recruit a maximum of 12 participants for this study.

SUBJECT INCLUSION CRITERIA
- Required to be genetic counselors who have counseled at least one patient with intellectual disabilities
- Male or female
- Can have any number of years experience as a genetic counselor
- Must be at least 18 years of age
- Must speak fluent English

SUBJECT EXCLUSION CRITERIA
- Counselors who have not counseled at least one patient with ID

STUDY DESIGN
I plan to recruit genetic counselors via the National Society of Genetic Counseling (NSGC) listserv to take part in a 30-minute audiotaped phone interview exploring their experiences with counseling patients with intellectual disabilities. This will be a qualitative study in which I plan to conduct a maximum of 12 interviews. Participants will receive a $25 gift certificate to Amazon.com upon completion of their interview. The phone interviews will follow a semi-structured interview guide (Appendix B), but will
focus on the genetic counselors’ experiences with patients who have intellectual disabilities. The interviews will be transcribed and then coded using Atlas qualitative analysis software. I plan to use comparative and descriptive methods to analyze the data.

STUDY PROCEDURES
Recruitment procedures and materials
I will have my faculty supervisor, Beth Rosen-Sheidley, post a recruitment notice (Appendix A) on the NSGC listserv.

Data Collection
I will be conducting a maximum of 12, audiotaped phone interviews. I will use a semi-structured interview guide (Appendix B), which will focus on the counselors’ direct experiences with patients.

Data Analysis
The interviews will be transcribed and then coded using Atlas qualitative analysis software. I plan to use comparative and descriptive methods to analyze the data.

ANTICIPATED RISKS AND BENEFITS TO PARTICIPATION IN THE STUDY
There will be no direct benefit to the participants in this study. We hope that in the future, information obtained from this study will help us gain a better understanding of the experiences that genetic counselors have had when counseling individuals with intellectual disabilities.

ADVERSE EVENTS
Participation in this study presents no more than minimal risk. However, it is possible that by taking part in this interview, that participants may experience thoughts or feelings that may be upsetting. Should that occur, participants may contact Beth Rosen-Sheidley at 781-736-2336.

INFORMED CONSENT
Following the recruitment period, I will obtain informed consent (Appendix C), after first having provided the participants with the form to read, reviewing the study details and informed consent with them, and answering any questions they have. Participants may fax the signed/dated form back to me and I will then sign/date as the PI and provide a copy back to the participant.

DOCUMENT STORAGE
All records containing identifying information, such as names, email addresses, telephone numbers, and home or work addresses will be kept strictly confidential during the study. All study related documents and materials (including consent forms, interview transcripts and audiotapes) will be kept in a locked, file cabinet drawer in the Genetic Counseling Program’s secure space and will only be accessible only to the Principal Investigator and Beth Rosen-Sheidley. Any databases containing identifiers will be password protected using a password known only to the Principal Investigator. Upon completion of this study, all data including audiotapes, transcripts, and databases will be destroyed. The
Brandeis Genetic Counseling Program will keep informed consent documents for 5 years in a locked filing cabinet.

**COMPENSATION**
Upon completion of an interview, participants will receive a $25 gift certificate to Amazon.com in the mail.

**PRIVACY/CONFIDENTIALITY**
All records containing identifying information, such as names, email addresses, telephone numbers, and home or work addresses will be kept strictly confidential during the study. All study related documents and materials (including consent forms, interview transcripts and audiotapes) will be kept in a secure location accessible only to the Principal Investigator, and any databases containing identifiers will be password protected using a password known only to the Principal Investigator. Transcripts, interview notes, and audiotapes will be labeled with a coded ID number, which will be assigned to participants upon enrollment into the study. If participants are quoted or referred to in any written or oral reports of the study, they will be given an alternate name. Participants will never be referred to by their real name or any other identifying information in any written or oral reports based on the interview.

**COSTS**
There will be no costs to study participants, other than the time they invest to participate in the study.