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## Exploring Perceptions of What Genetic Counseling is Amongst Families Affected by Genetic Conditions, Who Have Not Yet Had Genetic Counseling Themselves

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Exploring perceptions of what genetic counseling is amongst families affected by genetic conditions, who have not yet had genetic counseling themselves

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## PERCEPTIONS OF GENETIC COUNSELING

### **Abstract**

This study aimed to understand the perceptions and expectations that individuals who are affected or who have a genetic condition have about genetic counseling. This was a qualitative study that aimed to improve understanding of the layperson's perception of genetic counseling using interviews with participants. Interviews were conducted at Cardiff University in Cardiff, Wales. Interview transcripts were analyzed using thematic analysis to identify common themes amongst the participants' perceptions and to assess where these perceptions differ from professional definitions of genetic counseling. Themes were analyzed using the five dimensions of empowerment. The omnipresent theme among the study was the expectation of achieving emotional regulation from the genetic counseling session. The majority of participants indicated that they expect support from their health care providers and this expectation is even more-so in regards to their genetic counselors. The second pervasive theme among participants is the expectation of achieving cognitive control. Patients seek information about their conditions and about general genetics, many because of lack of knowledge about the field. Data from this study could better shape communications between patients and providers and create realistic expectations about what they should expect when they are referred to genetic counseling. Results from this work could help to resolve misconceptions and stigma the layperson has about genetic counseling.

*Key words:* perceptions, expectations, empowerment framework, misconceptions.

# PERCEPTIONS OF GENETIC COUNSELING

## Introduction

Genetic counseling is a client-centered communication process that aims to help patients understand, adapt, and adjust to medical or psychosocial consequences of genetic contributions to disease (Resta, Biesecker, Bennett, & Blum, 2006). It has been recognized that clients are less likely to have clear ideas about what to expect from a genetic counseling session than other medical consultations (Mitchie, Marteau, & Bobrow, 1997).

There have been few published studies exploring patient expectations of genetic counseling. Patient expectations can vary widely, and past research has not reached a consensus on what the average patient expects from their consultation. For example, a common view of genetic counseling is that its educational purpose is more important than its counseling purpose (Kessler, Kessler, Ward, & Opitz, 1984). However, many experts believe the most important part of the genetic counseling session to be the therapeutic process of providing emotional support (Jay, Afifi, & Samter, 2000). In addition, some studies suggest that many patients who were about to undergo genetic counseling rank supportive or emotional care as one of the least important elements of the counseling session, depending on a variety of background variables (Hayat Roshanai, Lampic, Ingvaldstad, Askmal, & Bjorvatn, 2012). Providing therapeutic and supportive care is a very important part of genetic counseling profession's definition of their work and may be more helpful for many patients. However, there may be patients who would prefer this to be a lesser focus during the consultation and may be looking solely for specific information or assessment.

Furthermore, genetics is increasingly permeating everyday medicine. Direct-to-consumer (DTC) personal genomic and ancestry testing have made a huge step in exposing the general public to a form of genetic testing. However, research has found that there is low knowledge of

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the possible adverse social consequences or the benefits of such testing (Haga, et al., 2013). It has been predicted that the implementation of genomic medicine across the whole health care system will mean that hundreds of thousands more patients will have a genomic test as part of their diagnostic pathway (Patch & Middleton, 2018). More people receiving genetic information, there is likely to be an influx of genetic counseling patients. In addition, the public expectations may be changed or be shaped by these experiences. Understanding expectations as they are today and understanding possible predictors of these expectations are essential for genetic counselors to offer patients the best health care experience and to achieve the highest patient satisfaction and optimal care outcomes.

The aim in this study was to look at people from families affected by genetic conditions who have not had genetic counseling themselves to find out more about what they perceive genetic counseling to be. This was a qualitative study that aims to improve understanding of the layperson's perception of genetic counseling. The data from this study could help better tailor communications to patients about support they will receive if they are referred to genetic counseling and will be used to develop an online resource to support realistic expectations for patients. It is anticipated that this work will help genetic counselors to resolve misconceptions people may have about genetic counseling before patients attend the clinic.

### **Methods**

Qualitative methods were employed to collect data using semi-structured interviews. Qualitative research systematically explores social phenomena, including how people experience aspects of their lives (Teherani, Martimianakis, Stenfors-Hayes, Wadhwa, & Varpio, 2015). These methods are most useful in areas of research that are not yet well understood. Because the

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goal of this research was to explore people's expectations of genetic counseling, qualitative research is appropriate.

Patient empowerment provides a useful approach to conceptualizing and measuring patient-reported outcomes from clinical genetics services (McAllister & Dearing, 2015). Patient expectations of genetic counseling could be usefully conceptualized using the theoretical framework of 'empowerment.' In this framework 'empowerment' summarizes the patient benefits from using genetics services. All of these can shape a patient's expectations in anticipation of genetic services (McAllister, Dunn, & Todd, Empowerment: qualitative underpinning of a new clinical genetics-specific patient-reported outcome, 2011). Therefore, McAllister et al.'s 2011 definition of 'empowerment' was used as the theoretical framework for interpreting the qualitative data collected in this study. This framework comprises five dimensions (McAllister & Dearing, 2015, 116-117):

1. "Cognitive control: having sufficient knowledge and understanding about the condition, including the risks to oneself and other relatives.
2. Decisional control: having available options or feeling able to make informed decisions between these options for managing risk.
3. Behavioral control: feeling able to use the health and social care systems effectively to reduce harm/improve life for oneself and other relatives.
4. Emotional regulation: feeling able to effectively manage emotional consequences of genetic information.
5. Hope: for fulfilling family life for oneself, relatives, and future descendants."

Participant recruitment via advertisement to the constituent patient support groups through the Genetic Alliance UK. The Genetic Alliance UK is a national charity and an alliance of over 200

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patient organizations. It is a resource that provides information, support, and influence on issues that families and individuals with genetic conditions face (Genetic Alliance, 2018). Interested parties contacted author Marion McAllister to indicate interested in participating, and a face-to-face, telephone, or Skype interview was scheduled. All participants were then sent a Participant Information Sheet and Consent Form by email and returned written informed consent prior to the interview. Recruitment procedures were approved by the Cardiff University School of Medicine Research Ethics Committee. All participants were residents of the United Kingdom.

Data collection and analysis was conducted at Cardiff University. The interviewer utilized an interview guide designed by incorporating experience from a prior study. The interview mode (face-to-face, telephone, or Skype) was conducted according to the interviewee's preference. The interviewer made clear to the interviewee that they had the opportunity to decline any question or to stop the interview at any time. Participants were asked demographic questions but could decline to answer these. All interviews were audio-recorded and later transcribed in full. In accordance with research governance approval, the audio recordings were deleted from the audio-recorder following transcription.

Transcriptions were analyzed using a combination of framework analysis (Ritchie & Spencer, 1994), where the conceptual framework was relevant, as well as some of the tools of grounded theory methodology including: (1) open coding: labels or 'codes' indicate significant pieces of text as important and related to study goals, (2) constant comparison: each new instance of a code was compared with every other instance of that code, and the definition was defined accordingly, and (3) questioning the data in relation to the research question at hand (Glaser & Strauss, 1967).

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Sections of the interview transcripts which appeared to reflect expectations and understandings of what genetic counseling is were coded within the theoretical framework of empowerment using framework analysis, and other codes were developed using open coding. Disagreements on codes were resolved by discussion amongst the authors until eleven codes were agreed upon (See Table 2). Codes were then subdivided using the five dimensions described in McAllister et al.'s 2011 definition of empowerment: cognitive control, decisional control, behavioral control, emotional regulation, and hope. For the purposes of this study, codes were defined as a statement or a series of sentences spoken by the interviewee in response to a question or on their own that had to do with expectation, understanding, or knowledge they have about genetic counseling. The distribution of the 11 codes within the 5-dimension framework is described in Table I.

### **Results**

There were 9 total interviews conducted. All but one interview was conducted over the phone. One interview was conducted over Skype video chat by participant request. Of the nine participants, eight were female. Participants were between the ages of 30 and 60 years old (two participants did not disclose this information). Five of the nine participants had children of their own. All individuals disclosed the genetic syndrome they or their relative were affected with. Seven participants were affected themselves with the genetic condition and two had close family members who were affected. One participant disclosed that she herself was affected as well as her sisters and mother. Five participants chose to disclose their highest education level (secondary school-university). For confidentiality, participants are labeled and identified as P1 – P9 (See Table II).



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As each code corresponds to a major theme amongst the interviews, results are reported thematically and broken down into the five-dimension framework:

### **Cognitive control:**

Information seeking: Five participants expected to receive some sort of information when consulting with a genetic counselor. The desire to accumulate more information about the familial genetic condition and how it might affect quality-of-life was identified as a code among responses. This example is illustrative:

“I guess information, knowledge is what you’re after to start off with and how is it going to impact and how is it going to affect your [relative] and you know how things are going to turn out and what obstacles is [they] going to be looking at and how are things going to be and how different is [their] life going to be than how you maybe thought it might be. (P1)

Of those information-seekers, all (n=5) specifically indicated that they expect simplified explanations of genetic test results, medical terminology, or a more in-depth information and support relating to their own / their family condition:

“Yeah and have them properly explained because it’s like you know sometimes as a lay person, you can’t interpret results in a way that a medically trained person can. (P2)

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“A little bit more support, maybe explain in less medical terms. Because when you do go look up Wiedermann Steiner, there’s a lot of medical, it’s so medical out there. So, a little bit more simple sort of. (P3)

Unfamiliarity: The majority (n=6) indicated that had little idea of what genetic counseling and the genetics field in general might be. They were either simply unsure what a genetic counseling session entailed, or they had never heard of the profession prior to agreeing to participate in the study, as indicated by the following:

“I don’t know whether anybody knows what genetic counseling actually is (P6)

“I have no knowledge of [what] they do (P1)

Test-curious: One participant expressed that they wanted to have informative genetic testing done at their genetic counseling session. Genetic test results would contribute to their risk assessment and would play an important role in their decision-making. In addition, they expected that tests they’ve already had through other health care providers to be more thoroughly explained by the counselor, as detailed by this quotation:

“...other tests may be offered. Other testing you know possible tests available in future testing (P2)

**Decisional control:**

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Risk-refiners: Three participants wanted clear information about their future risk of developing symptoms or a full risk assessment for their children. Genetic risk information would help them make family-planning decisions, screening decisions, and/or decisions regarding their own future-planning. Risk-refiners were defined as those seeking genetic risk information. They were identified as respondents vocal about their expectations when seeing a genetic counselor to receive concrete risk figures for their own use. This risk information could come in the form of testing options that the counselee could choose from. Participants expected to receive options on genetic testing for them to choose from. Risk information could also give them the ability to choose between different treatments or choose some course of action regarding their care.

Examples from participants include the following:

“Well I think for future generations to you know make informed choices and whether or not they want to take the risk to have children or not (P2)

“Then of course, I know I’ve got this gene, and not really sure what it causes. Have I given it to my children? I don’t know. So that’s where I kind of am at the moment. (P6)

### **Behavioral control:**

Self-motivation: Of the nine participants, five indicated that they were interested in having a session with a genetic counselor due to their own strong-willed and hopeful nature. They were

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determined to obtain the best care available and to do as much as they could for their own health.

This sort of assertiveness is portrayed in the following statements:

“I sort of went out looking for answers and knowledge and information and just to try and understand my condition a bit more and maybe why I was having the issues I was having and maybe some of the behavior I was presenting. (P1)

Certain self-motivated participants expected that engaging with a genetic counselor meant being able to participate in research studies, as indicated by this participant:

“I’m in the agreement that we have to live by research and I’m really proactive when it comes to that. (P3)

### **Emotional regulation.**

Support-seekers: Participants overwhelmingly voiced the need for support. Six participants highlighted therapy, or emotional support, as their main goal for a genetic counseling session:

...[I need] just like support so that I can help her just to understand it just as much as she can. I would expect them just to, you know, help and give advice and things like that. And just be there. That if she’s having a bad day, you know I’d like to make an appointment to say can we come see you in two weeks or two months time... (P3)

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“I believe that you do need some kind of counseling. Because it’s all interlinked, you need the strength, you need the support, you need people to believe you and be behind you. If you don’t have that that, then I do think it affected you managing and understand and having support for you condition. (P6)

“Kind of like a psychologist, like a counselor. And just deal with any issues or how you feel or anything that may happen in your future, those kinds of things. (P7)

Losing the Joy: This code is exemplified by two participants who were seeking a professional to support them emotionally to deal with their condition and the practical challenges of the day-to-day take of living with or dealing with that condition. In contrast, some believed that a genetic counselor might actually have the opposite effect. Instead of making life easier, a genetic counseling session might actually make it more difficult to deal with the condition. The appointment itself along with the information received at that appointment might cause one to ‘lose the joy’ of living without that information, as exemplified by this participant:

“...getting all this information and the hospital appointments, reading all this stuff up on the internet and going to the hospital appointments, they take up a lot of your time and that takes them away from enjoying your daughter... I think you could possibly get too obsessed, you could sort of overthink it and instead of sort of enjoying your life, and just getting on with living... you would be thinking about that all the time and obsessing about that all the time and maybe wasting a bit of time rather than just getting on with things and just dealing with [them] as they come along. (P1)

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Evading sadness: Three participants indicated that they assumed people would avoid genetic counseling because it would give them information that would make them sad or depressed. These individuals who were described as evading sadness indicated that they felt that risk information or diagnostic information could be devastating for them.

“...if you found out you have something that most likely [will] kill you in the near future I think that could be very negative, depressive.... Everybody deals with things in different ways but I think for some people that could be really, you know they might feel like ‘what’s the point of going on?’ (P2)

“If someone’s got something that’s maybe life threatening or something, that might be difficult. This might make people feel worse, it might not help. (P7)

Isolation: Three interviewees expressed that they felt they were already isolated by the hardships of their condition and seeing a genetic counselor might help them to cope with this. This was especially true for those participants who describe having felt isolated or alone during their diagnostic process up until this point, as indicated by this participant:

“It’s not always ideal having a rare condition as well, it can be very misunderstood. And it can be very isolating, I have never found another adult with my condition. So, it’s very isolating. (P2)

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Dependence: Another theme among the participants was the expectation that they would find more help or direction from genetic counselors than the medical professionals they'd seen in the past. These participants (n=3) have taken on a more dependent role when it comes to their health and seek someone who will give them more direction. Sometimes this is due to a history of having felt pushed-aside by their health care system and expect a genetic counseling session to be different. They are hoping that a genetic counseling session might be a good point to find better care from providers. This is evidenced by these participants:

“No, [I've] not had any genetic counseling at all, no. Neither since we've had the diagnosis or before or after. Basically, we've been told 'this is what's wrong with [patient] from genetics' and that's it. We've not had any follow ups; we've not had anything. It's basically been pushed under the carpet a little bit really. (P3)

“[Patient] just doesn't seem very keen to find out what's wrong. So, I kind of had to force him into doing things. I don't think the doctors are that interested or bothered...(P7)

The expectation of getting support from genetic counselors is pervasive among the participants of this study. All participants expressed the expectation of achieving some sort of emotional regulation from their genetic counseling session.

### **Hope.**

Only one participant expressed that their main reason for seeking genetic counseling was to fulfill a hope for their own or for their family members' future.

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“Sometimes I have my doubts, but you know I still think you’ve got to hope, you’ve got to hope. (P2)

### **What is genetic counseling?**

Participants often offered their understanding of genetic counseling and what it is to them. Definitions varied based on their experiences and the expectations they amassed throughout the course of their disease. Some participants presumed genetic counseling would be an emotional and cognitive support system for patients who have had a long and difficult journey to a diagnosis, as in this participant’s case:

“...just somebody that actually understands the patient, gives them support, gets help and guidance...because obviously they’re going to have all these issues...someone to go to when they’re feeling like they just don’t have the answers. (P2)

Others assumed genetic counselors were genetic disease experts trained in mental and emotional therapy:

“I think it would be counseling... genetic counseling... probably how you feel about having a genetic condition. (P5)

“Erm, probably someone who like supports you if you have a genetic condition?...like a normal kind of counselor not just a genetic counselor. (P7)



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This participant description of genetic counseling described it as an explanation of a diagnosis and the ability to talk to an expert and find out their treatment, management, and testing options:

“I’d assume it would be the chance to sit and talk to somebody about what the diagnosis is and what that might mean? And then again, is there any signposting for support. Something like that. But to me, hearing counselling is to be able to talk it through and ask any questions. I mean, it must be really hard, there must be so many genetic conditions, you can’t expect the person you’re talking to know everything about that condition. I think it’s more the chance to talk that through and what are the options going forwards.  
(P8)

Participants had many definitions of genetic counseling, the most prevalent having to do with emotional support about the progress of their disease and the difficulties they’ve had as patients of genetic conditions.

### **Discussion**

This article presents the expectations of genetic counseling expressed by those who have not undergone the service themselves, but who are affected with or who have a family history of some genetic condition. The omnipresent theme among the study was the expectation of achieving emotional regulation from the genetic counseling session. This is in contrast to the research done by Hayat *et al* in 2012, a study which found that receiving risk information was very important. However, the current study’s participants plainly expressed the need for

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continued professional psychological support, something that is not a common practice for genetic counselors. Although a psychosocial assessment with mental or emotional support during the genetic counseling session is common, this is hardly ever ongoing. In general, genetic counselors only see patients once with at most two follow-ups – seeing a patient many times over the course of their disease is very rare. When an expectation does not align with actuality, strides can be taken to correct it.

The majority of participants indicated that they would like more support from their health care providers and greatly expect such support from their genetic counselors. This need for support stemmed from lack thereof in previous health care experiences and was perhaps related to a higher need because of their declining health and rising dependence. Participants described how a genetic diagnosis can cause a significant amount of emotional turmoil within a family and highlighted the need for their themselves or their affected relatives to be given emotional support. Some patients so assumed genetic counseling would have to do with emotional regulation, they wanted to avoid it. This was coded as ‘loss of joy’ because of the participant’s assumption that it would be depressing or take away from their happiness in some way. Previous research has suggested that patients often decline genetic counseling because of the potential emotional impact on themselves or on their families (Geer, Ropka, Cohn, Jones, & Miesfeldt, 2001). The perception that genetic counseling can lead to psychological damage could deter patients from scheduling their suggested appointments with a genetic counselor, which could in turn be costly to the profession overall.

Participants seemed to associate the term “counselor” within the professional title as meaning a strong psycho-social supporter that has a good knowledge of genetics and genetic conditions. Because of this association, they perceived a genetic counselor to be a health care

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provider that would provide counseling for quality-of-life issues having to do with the course of some condition that could be scheduled as these issues arise. They understood genetic counselors to be “someone to talk to” during the more difficult times of their lives. While genetic counselors do have the capacity to use psychosocial techniques during the session, the psychological assessment and counsel only a small proportion in most cases. Patients who perceive their genetic counselor to be a therapist could feel dissatisfied with their experience. In addition, patients making multiple appointments for mental health reasons could be time-consuming and profitless.

This expectation is also prevalent in the participants’ definitions of genetic counseling. Patients, especially those who felt disregarded by their health care providers in the past, expect more emotional support from their counselors. They have been given information from providers in the past and are now hoping for either more in-depth descriptions of the same information or support to be able to mentally and emotionally deal with it.

Austin *et al.* suggested in 2014 genetic counseling as a form of psychotherapy using the communication of genetic information as a therapeutic goal. This concept is similar to the expectations described by participants of this study. Not fulfilling such expectations can impede the effectiveness of genetic counseling. Genetic counselors need to find new ways to attend to psychotherapeutics in clinic encounters rather than just communicating information. By incorporating the psychotherapeutic techniques, Austin *et al.* suggests clients can achieve an emotional state where information communication is more productive (Austin, Semaka, & Hadjipavlou, 2014).

The second pervasive theme among participants was the expectation of achieving cognitive control. Patients overwhelmingly seek information about their conditions and about

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general genetics, due to either simple curiosity or from a history of lacking information about the field.

Some participants made it clear throughout their interviews that their journey to a diagnosis has been very difficult and that they have been given little information from their health care providers. Others seem to have had very negative experiences in the past. These participants anticipated that genetic counselors would be a more receptive professional that would listen to them at a deeper and more inclusive level than other providers. Participants indicated that they valued health care professionals who took them seriously, giving the overarching impression of comprehensive care. Previous research has found that patients rank being taken seriously by their doctors as very important (Hayat Roshanai, Lampic, Ingvaldstad, Askmal, & Bjorvatn, 2012). Hayat *et al*'s study found that variables impressed upon patient expectations, such as gender and socioeconomic status. Genetic counselors do have the competence to provide multiple forms of cognitive control, such as risk assessments, testing options, medical managements, treatments, and preventions, and research options. Most of the points during the genetic counseling session has to do with informed consent and assuring the patient has the information to make good cognitive decisions about themselves and their future.

In addition, while cognitive control by receiving genetic information is conducive to informed decision-making and reducing confusing or anxiety, it might introduce new uncertainties. Knowing clients will expect information is important for constructing a genetic counseling session that will give patient new information but addressing their fears and anxiety simultaneously. Incorporating a psychosocial approach can help patients adapt to these new uncertainties (Austin, Semaka, & Hadjipavlou, 2014).

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A main weakness of this research is that it is limited to mostly female perspectives. A gender-diverse sample would be more representative. Furthermore, all participants were individuals living in the United Kingdom who are members of patient support groups and may not be generalized beyond this. It is likely that expectations are impacted by culture, as well as educational background and beliefs, so further investigation of other cultures would be valuable. Additionally, the low participation rate means data saturation was not reached, and a larger sample size would increase the value and reliability of the findings.

### **Conclusion**

Before seeing a genetic counselor, people who have genetic conditions or have a family history of a genetic condition have many expectations about genetic counseling. Most importantly, these expectations revolve around psychosocial and cognitive support. Genetic conditions are often stressful diagnoses that require a lot of support. These patients, especially those who have had negative experiences in the past from health care professionals, seek emotional support from a person they can call a ‘counselor,’ that they know is an expert on their diagnosis. While genetic counseling does provide psychosocial support to patients in a more extensive way than most health care providers, it is important that patients understand that genetic counselors are not trained in psychotherapy. For lifelong mental health support, a licensed therapist or psychologist would be more suitable. Providing patients with cognitive control is support that genetic counselors can give to their patients by providing informed consent and by sharing the knowledge from our extensive training in genetics.

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Table I. Coded within the McAllister et al. Patient Empowerment Framework (McAllister, Dunn, & Todd, Empowerment: qualitative underpinning of a new clinical genetics-specific patient-reported outcome, 2011).

<i>Empowerment Framework</i>	<b>Cognitive Control</b>	<b>Decisional Control</b>	<b>Behavioral Control</b>	<b>Emotional Regulation</b>	<b>Hope</b>
<i>Codes</i>	information-seeker	risk-seeker	assertive	support-seeker	holding on to hope
	ignorant			loss of joy	
	test-curious			evading sadness	
				isolation	
				dependent	

PERCEPTIONS OF GENETIC COUNSELING

Table II. Patient Demographics.

<b>Participant Number</b>	<b>Gender</b>	<b>Age</b>	<b># of Children</b>	<b>Genetic Condition</b>	<b>Highest Education</b>	<b>Relationship to Affected</b>
<b>P1</b>	Female	49	0	Turner's syndrome	Secondary school	Self
<b>P2</b>	Female	59	3	Chronic mucocutaneous candidiasis	Higher education college	Self
<b>P3</b>	Female	47	3	Wiedemann Steiner Syndrome	Secondary school	Daughter
<b>P4</b>	Male	35	0	No formal dx, primary gonadogenesis	University	Self
<b>P5</b>	Female	30	2	Pseudoanthoma elasticum	University	Spouse
<b>P6</b>	Female	44	2	Hypermobile Ehlers-danlos syndrome	N/A	Self
<b>P7</b>	Female	30	0	Russell-Silver syndrome	N/A	Self
<b>P8</b>	Female	N/A	1	Wiedemann Steiner syndrome	N/A	Son
<b>P9</b>	Female	N/A	N/A	Familial partial lipodystrophy type 2	N/A	Self, sisters, mother