

Deaf Individuals' Attitudes and Perceptions of Genetic Counseling and Genetic Testing

Master's Thesis

Presented to

The Faculty of the Graduate School of Arts and Sciences

Brandeis University

Graduate Program in Genetic Counseling

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In Partial Fulfillment

of the Requirements for the Degree

Master of Science

in

Genetic Counseling

by

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May 2019

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ACKNOWLEDGEMENTS

First, I want to thank my thesis advisor, Gretchen Schneider, MS, CGC for her guidance, patience, support, and efforts to ensure that this project was a success. She believed in me and I could not have done it without her. I would also like to thank the other members of my thesis committee: Andrea Oza, MS, CGC, Karen Turley, BA, MED, Katie McCarthy, MBA, and Patrick Costello, MED, EDS. They volunteered their time, expertise, and unique perspectives throughout this process and I am so appreciative of their ideas and input.

I would also like to express my gratitude to the Deaf community, especially the individuals who participated in my study. Without their willingness to share their personal stories, this study would not have come to life. I thoroughly enjoyed my conversations with each and every one of them and their input has been invaluable.

Furthermore, I would like to thank Lauren Lichten, MS, CGC, Missy Goldberg, Gayun Chan-Smutko, MS, CGC, Judith Tsipis, PhD, and the entire Brandeis University Genetic Counseling Program for believing in me, supporting me, and giving me the opportunity to becoming a genetic counselor.

To my classmates: thank you so much for your encouragement, compassion, and joy throughout our time at Brandeis. There's no one else I would rather have by my side moving into our careers. To JR Vibandor: I can't put into words how much you have helped me through the past two years and I could not have made it through without you. And finally, to my family and friends: thank you for your love, support, and faith in me. You are the reason I am where I am today and you mean the world to me.

ABSTRACT

Deaf Individuals' Attitudes and Perceptions of Genetic Counseling and Genetic Testing

A thesis presented to the Graduate Program in Genetic Counseling

Graduate School of Arts and Sciences
Brandeis University
Waltham, Massachusetts

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Deaf Americans face linguistic and cultural barriers around access to public services and medical care, including genetic counseling and genetic testing. Healthcare providers' cultural competency and communication accommodation for Deaf patients directly impact patient care; however, most providers have little knowledge of how to interact with this population. As genetic testing increasingly becomes part of routine clinical care, more Deaf individuals will be referred for genetic counseling. To provide quality care, it is essential for genetic counselors to understand Deaf patients' perspectives.

The purpose of this qualitative study was to document attitudes and opinions of Deaf individuals regarding genetic counseling and genetic testing. Six semi-structured interviews were completed with culturally Deaf adults who primarily use American Sign Language (ASL) to explore personal experiences with medicine and genetics, opinions on genetic testing in different scenarios, and suggestions for providers. Interviews were coded based on emergent themes and analyzed with an inductive approach.

Identified themes included: personal experiences influence perception of all providers; participants have low expectations for providers and value provider familiarity with Deaf patients; participants had more exposure to non-medical genetic concepts than medical genetic testing; and Deaf individuals support the availability of genetic testing for all.

Participant recommendations included: accommodate for preferred communication methods by using interpreters and simplifying jargon; respect patient choices by presenting options without bias; and be open minded to Deaf culture by acknowledging the cultural model of deafness and advocating for positive change within the medical community.

This study supports and adds nuance to prior research, emphasizing the importance of cultural competency and communication accommodation for genetic counselors working with Deaf patients. Continued efforts to understand Deaf culture and perspectives are essential to improve relationships with and provide quality healthcare to this population.

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INTRODUCTION

Deaf Americans face unique barriers in accessing public services and medical care (Richardson, 2014; Steinberg, 2006). Not only is there a linguistic barrier for individuals whose primary language is American Sign Language (ASL), but also a cultural barrier about what it means to be deaf (medically deaf), Deaf (part of the Deaf cultural community), or hard of hearing. The Massachusetts Commission for the Deaf and Hard of Hearing (MCDHH) describes the difference between the “medical” and “cultural” models of deafness. The medical model, typically used by healthcare providers, defines a deaf individual as someone who lacks auditory capability and emphasizes the use of assistive devices. The cultural model, promoted by Deaf people, focuses on the abilities of Deaf individuals rather than their inability to hear, asserting that deafness is a gain of a visual language, community, and culture with a rich history. The dichotomy between these two models results in a unique combination of linguistic and cultural barriers, causing Deaf individuals to feel misunderstood by healthcare providers. This often results in inadequate care such as missed appointments and incorrect diagnoses or prescriptions (Scheier, 2009; Steinberg, 2002; Steinberg, 2006).

While many factors complicate relationships between Deaf patients and their healthcare providers, Deaf individuals see conflicting views on deafness as one of the most significant sources of discord (Iezzoni, 2004; Scheier, 2009). This disconnect is compounded by the fact that many healthcare providers are unaware of the conflict, likely due to inadequate training. A 2014 survey assessing Deaf awareness training in genetic counseling programs revealed that 77% of participants had no or limited Deaf awareness training and that one third of the respondents felt their Deaf awareness training was insufficient for clinical care (Nagakura,

2014). Similar results are found in surveys of medical residents and practicing providers, demonstrating that many healthcare providers lack training and understanding regarding Deaf individuals, practices, and culture (Hoang, 2011; Richardson, 2014).

Provider competency in communication and cultural understanding can positively affect Deaf individuals' healthcare. When healthcare providers practice accessible communication habits and attempt to use patient-preferred strategies such as using qualified interpreters and written communication, it encourages mutual trust between the Deaf patient and the provider (Preminger, 2014; Steinberg, 2002). Additional studies have supported and expanded upon this assertion, suggesting that improved outcomes over time result from clinical care customized to meet the cultural, linguistic, and communication needs of Deaf patients in both general and specialty settings (Baldwin, 2012; Boudreault, 2018; Palmer, 2014). In genetic counseling, a counselor's positive attitude towards deaf people along with culturally and linguistically appropriate counseling has been shown to increase patient-counselor connection along with improved genetics knowledge and comprehension in Deaf patients (Baldwin, 2012; Enns, 2010).

There is limited literature available to help healthcare providers understand Deaf patients' perspectives on genetic counseling and genetic testing as a whole. Previous surveys in English focused on Deaf attitudes toward genetic testing for deafness with conflicting results. One study found that the majority of participants had negative perceptions of genetic testing and its implications while others showed that participants had a strong interest in genetic testing and that participants have both positive and negative reactions to genetic testing results (Boudreault, 2010; Martinez, 2003; Palmer, 2013). It is unknown whether these studies reflect Deaf people's perspectives on genetic testing outside of genes for deafness.

Professional literature and anecdotal evidence show that Deaf individuals often experience difficulty being understood by healthcare providers on linguistic as well as cultural levels (Steinberg, 2006). For example, Deaf patients often report lacking health knowledge and even avoiding health services due to communication problems with healthcare providers (Iezzoni, 2004; Steinberg, 2002). These issues include difficulty accessing interpreters, staff and provider miscommunication, and staff reliance on phone calls (Scheier, 2009; Steinberg, 2006). In addition to communication barriers, different beliefs around what it means to be Deaf can create further personal and cultural obstacles to healthcare service for Deaf individuals. Previous studies identified that genetic counselors and other healthcare providers lack information about Deaf individuals and culture, suggesting a need for further inquiry (Nagakura, 2014; Richardson, 2014). As genetic counseling and testing increasingly becomes part of routine clinical care, it is even more important that genetic counselors understand their Deaf patients' unique attitudes and perspectives to provide quality clinical care to this population.

The goal of this exploratory qualitative study was to interview Deaf adults who use ASL as their primary method of communication and explore their personal experiences with medicine and genetics, their opinions on genetic testing in different scenarios, and their recommendations for healthcare providers and genetic counselors. By documenting the attitudes and opinions of Deaf individuals regarding genetic testing and genetic counseling, this study can provide genetic counselors, and other healthcare providers, insight into the perspectives of the Deaf community and strategies to improve relationships with and provide quality healthcare to Deaf patients.

METHODS

This was a qualitative study consisting of semi-structured interviews with Deaf individuals conducted using ASL. The Brandeis Committee for Protection of Human Subjects, deemed the protocol to be exempt from further Institutional Review Board (IRB) oversight.

Recruitment Procedures

Between December 2018-February 2019, study participants were recruited through The Voice of the Deaf Community in Massachusetts (VDCMA) Facebook group and the Florida Association of the Deaf, Inc. (FAD) Facebook page. The VDCMA is a public Facebook group for Deaf individuals in Massachusetts to express their thoughts on issues concerning Deaf language, culture, and rights. The FAD is a public Facebook page for Deaf, Hard of Hearing and Late Deafened individuals in Florida designed to advocate for and support these individuals as well as organizations and associations working with them. The administrators of each platform approved (Appendix A) the content of the recruitment notice (Appendix B) and accompanying American Sign Language video notice (available upon request) and agreed to post them once the study was approved by the Brandeis IRB.

After viewing the recruitment video and notice, interested individuals contacted the interviewer by email or text. All potential participants received a response with questions about the inclusion criteria to confirm that they met the eligibility requirements. To be eligible to participate, individuals were required to; be 18-40 years old, identify as culturally Deaf, communicate primarily through American Sign Language, and be willing to be video and audio recorded throughout the interview. Individuals were excluded from participation if they had

syndromic deafness or if they did not have access to a reliable phone and internet connection for video conferencing and video relay service.

Once confirmation of eligibility was received, interested participants were provided with an information sheet (Appendix C) which described the purpose, risks and benefits, and procedures of the study. The email or text containing the information sheet also asked the participant to confirm their continued interest in the study once they had read through the information sheet, and encouraged them to reach out to the interviewer for questions or concerns.

Interviews were scheduled on a rolling basis. Sorenson and Convo provided video relay services. At the beginning of the interview, participants had the option to review the information sheet with the interviewer and interpreter. After consent and agreement to be recorded was confirmed, the session video and audio was recorded using Zoom video conferencing.

Interview Procedure

A semi-structured interview guide (Appendix D) was used for data collection. The interviews consisted of a series of questions about the interviewee's experiences with healthcare providers, genetic counseling, and genetic testing. Additional questions explored the interviewee's opinions on genetic testing in different scenarios, carrier screening, and genetic counseling. Interviews lasted 45 to 75 minutes. The audio recording was transcribed verbatim by Mulberry Studios Transcription Services. The completed transcripts were reviewed by the interviewer and edited for accuracy.

Demographic information for each participant including their age, gender, family history of deafness, parental status, health status, and highest level of education was collected after the audio and video recording was turned off at the end of the interview.

Data Analysis

Comparative and descriptive analysis of interview responses were manually performed using an inductive approach to identify patterns and themes. ATLAS.ti8, a qualitative data analysis software program, was used to code and analyze responses as well as to identify themes that describe Deaf individuals' opinions in regards to healthcare providers, genetics and genetic counseling, and genetic testing in various situations.

RESULTS

Demographics

A total of six participants were interviewed. The demographic data of participants is presented in aggregate to protect their anonymity (Table 1). The current age of participants ranged from twenty four to forty years old. Five participants identified as female and one identified as male. Four participants grew up in families where other relatives were also Deaf and two participants grew up in families where they were the only Deaf individual. One participant had a child and five participants did not have children. Two participants had ongoing health concerns requiring follow up with specialists at least once a year and four had no ongoing health concerns requiring more than routine care. All participants had completed a four year bachelor's degree. In addition to a bachelor's degree, three participants had completed, or were in the process of completing, a master's degree and two participants had completed, or were in the process of completing, a doctorate degree.

Table 1: Participant Demographics

Age Range	24-40 years
Gender	5 female 1 male
Family History	4 from Deaf families 2 from hearing families
Parental Status	1 with children 5 without children
Health Status	2 with ongoing health concerns 4 without ongoing health concerns
Highest Level of Education	1 obtained bachelor's degree 3 obtained/obtaining master's degree 2 obtained/obtaining doctorate degree

Table 2: Discussion Topics, Themes, and Subthemes

Discussion Topics	Themes	Subthemes
I. Personal Experience with Medicine	1. Personal experiences influence perception of all healthcare providers	a. Positive experiences lead to positive perceptions
		b. Negative experiences lead to negative perceptions
	2. Expectations are low for healthcare providers and staff	a. Inadequate care and communication expected
		b. Adequate care and minimal effort greatly valued
	3. Improved care and positive emotions associated with providers familiar with Deaf patients	a. Medical facilities in locations with higher Deaf populations lead to positive experiences
		b. Long term relationships with providers lead to positive experiences
II. Experience with Genetics/ Genetic Counselors	4. Deaf individuals had variable knowledge of, or personal experience with, medical genetic testing	a. Genetic counselors are not viewed as significantly different from other medical providers
		b. Medical genetic testing is most frequently known in relation to deafness
	5. Deaf individuals are likely to have knowledge of, or personal experience with, genetic concepts, including testing, outside of the medical setting	a. Direct to consumer genetic testing is widely known and correlated with positive perceptions
		b. Genetic engineering is widely known and correlated with extremely negative perceptions
III. Opinions on Genetic Testing	6. All types of genetic testing should be available to everyone	a. Personal responses varied significantly (Table 3)

Upon interviewing this cohort, six major themes based on three topics of discussion were identified (Table 2). The first discussion topic was personal experience with medicine, and participants’ responses revealed the following themes: (1) Deaf individuals’ personal experiences influence their perception of all healthcare providers; (2) Deaf individuals’ expectations are low for healthcare providers and staff; and (3) Deaf individuals associate improved care and positive emotions with providers who are familiar with Deaf patients.

The second topic was experience with genetics/genetic counselors, with responses revealing two themes: (4) Deaf individuals had variable knowledge of or personal experience

with medical genetic testing and (5) Deaf individuals are likely to have knowledge of or personal experience with genetic concepts, including testing, outside of the medical setting.

The third discussion topic, opinions on genetic testing, revealed one major theme: (6) all types of genetic testing should be available to everyone. After completing discussion of the three topics, participants were invited to give recommendations for healthcare providers to improve care and better understand Deaf patients.

I. Personal Experience with Medicine

1. Personal experiences impact perception of medical providers

We asked participants about their personal experience with the medical community, including how their healthcare providers accommodate for them and if that strategy has met their needs. Preferred accommodation practices varied by participant. Four of the six participants felt their current providers were able to meet their needs and they were satisfied with the quality of care they received. One participant described that their opinion of healthcare providers has improved due to recent positive experiences:

“Massachusetts Health has been really good with [accommodations]. I was born and raised in Wisconsin. And their health system is weaker, it had been a challenge there. But Massachusetts has been great for me. Always had interpreters on the ready.”

However, three participants felt their needs were only sometime met by their current providers or that they had received inadequate care in the past. All participants had a personal experience or heard stories from a friend where a lack of accommodations led to inadequate care. One such participant explained that their previous negative experience was bad enough that they have avoided finding a new provider:

“I don’t have a primary care right now... I haven’t found a doctor who is culturally experienced with Deaf people. [Recently I had] pneumonia... the doctor came in and started talking. There was no interpreter or anything present. And as soon as he found out I was deaf he just left. He just turned around and left the room... then maybe 10, 15 minutes later he came back and he was shaking. I felt very awkward. And though we used the VRI, I was misdiagnosed and got... the wrong medication. And it was just something

basic. [The doctor] just seemed scared and overwhelmed and didn't know how to deal with the situation. And I suffered two weeks with pneumonia before I got on the right dosage, the right medication."

2. Expectations are low for healthcare providers and staff

We also asked about participants' expectations for healthcare providers. Five of the six participants felt their healthcare providers do not, or have never, understood their perspectives, culture, or needs as a Deaf individual. The participant who expressed that they felt understood by their providers qualified that it is a partial understanding:

"Well, I wouldn't say that [my providers] have an in-depth understanding, no. But I know they definitely have an understanding of ASL itself. So if they understand and accept ASL, I think that's the most important part. And if they're accepting and understanding the importance of ASL, I feel like if the situation shows up where the Deaf culture needs to be there versus hearing culture, they at least have a basic understanding. Or if the situation comes up where the doctor may not know deaf tendencies, they're open-minded and willing to learn more about that. So would I say that they have an in-depth understanding or concept of Deaf culture, maybe not, but yeah."

Partially because of this misunderstanding, half of the participants expressed that they expect difficulty in communication and potential inadequate care when interacting with providers. One participant noted they are unable to use their preferred communication method:

"Typically, unfortunately, we have to write back and forth. They're supposed to provide an interpreter but they don't. And sometimes they do VRI, video remote interpreting. Unfortunately, it's not successful. It's just so much trouble, it's just not worth it. It's just easier to write back and forth. So, I'm lucky, you know, that I can lip read a little bit, and we just write back and forth."

Another participant explained that even when doctors are accommodating, difficulty can arise when interacting with other healthcare employees such as office staff:

"I had a receptionist, she was at the front desk. She wasn't willing to work with me—I just had a really bad experience. And most of it's been through phone for a... appointment, just to set up checkups and everything. And so they just continued to cancel my appointment, and they don't tell me. And they reschedule me, and then you know, if I have to ask for an emergency or whatever, then nobody's available. And it's like, as a deaf individual, it's not very accessible because of the phone tag, when they try to call me, rather than emailing me. So that's been probably the worst."

Despite these difficulties, five of the six participants expressed that they highly valued their providers and were grateful when they attempted to understand and accommodate for them. One participant describes how appreciative they felt to have a provider who makes appointments easier for them:

“[My doctor] is willing to wait for the interpreter to translate from English to ASL if needed. I'm very blessed to have a great doctor who's not critical or aggressive and is willing to communicate with me.”

Another participant explained that their provider has gotten better over time and that they feel their connection is partially due to the provider's own experience as a minority:

“I think so far my doctor, every time I have met with her, I think it's been every time, she's definitely learned... I prefer to not have an interpreter there. The first time I met... she asked if I needed an interpreter, wanting to work it out. And I said I'd be willing to write. She said she'd be willing to write for me, and I was willing to do that... I got really fortunate that my doctor is a person of color, and then a female. And so she seems to get it, I think. I think that helps a lot.”

3. Improved care and positive emotions associated with providers familiar with Deaf patients

During the interviews, all six participants described preference for, and increased trust in, healthcare providers that have experience working with Deaf patients. For four participants, this preference was related to medical facilities in particular cities and states that have higher Deaf populations. Two participants described positive experiences in Massachusetts with one specifically highlighting a hospital for their familiarity with Deaf patients:

“[My] doctor's actually under UMass, University of Massachusetts. So I think they have a lot of experience related to Deaf individuals at the hospital and that location. Many Deaf people typically live within that area. So I typically will go to that hospital for that reason. But they have a lot of experience with interpreters as well, and I think my doctor's very experienced, all the doctors are very, very nice and friendly, very willing to wait for the interpreter to show up the office [if they're] not there on time.”

Additionally, three participants recounted positive experiences in Washington, D.C., sometimes related to Gallaudet University, the first university for deaf and hard of hearing students. One participant described a particular facility that was well known:

“Here in Washington, DC, there was one place that a lot of people in the deaf community go to. And they seem to know. It's a little bit better for the accommodations. I wouldn't say it's 100 percent but it's better than I've experienced in the past.”

Another participant described an interaction with a genetic counselor, attributing the counselor's skill in accommodating to their location and experience with Deaf patients:

“I think [the genetic counselor] was conscious of what it was like to work with the interpreter. They seemed knowledgeable... it didn't seem awkward at all... But then, again, that's in Washington, D.C., and that's where Gallaudet University is. So they have many, many deaf clients at the hospital. So they definitely would meet more deaf people there.”

However, prior experience with Deaf patients was not the only factor that led to positive provider interactions. Three participants conveyed that long relationships with the same provider were beneficial when providers attempted to improve accessibility, even when the provider was not accommodating at the start. One participant described how their interactions with their provider progressed over time:

“I’ve been with [my doctor] for about ten years. So I feel close to this one. In the start ... they wouldn’t get an interpreter, then there would be a laptop, and we’d type back and forth. It was really awful! My brother and my dad go to the same doctor too. My whole family’s deaf. So they accommodate pretty well now.”

II. Experience with Genetics/Genetic Counselors

4. Deaf individuals had variable knowledge of, or personal experience with, medical genetic testing

We asked participants about their prior knowledge of, and experience with, genetics in a medical setting, including if they knew about carrier screening or genetic counseling. Prior knowledge of genetics varied highly between participants. Three participants had personal experience with medical genetic testing and all of these participants had seen a genetic counselor. Participants did not view genetic counselors as significantly different from other health professionals in providing accommodations or understanding of Deaf individuals’ perspectives, culture, and needs. One participant describes their confusing experience receiving genetic testing results from who they believe was a genetic counselor:

“I didn’t have any sign language interpreter at the time [I got genetic testing], so I can tell you, I didn’t have a full understanding of what was going on. But from what I remember, there was a person, a doctor, and then there was someone else who I believe was the genetic specialist [genetic counselor]. But I’m not sure, honestly. They just kind of gave me the results... They just said it was a specialist to give the genetic results about deafness. But that was it.”

Another participant describes their frustration at not being provided an interpreter at a genetic counseling appointment in their past, and subsequently not knowing what happened:

“Based on my own personal experience... they should be providing interpreters [but] at that time the GC just didn’t. Now that’s the law. But they didn’t before. So... I was ignored. I’m curious as to why, in the past, the access—good patient access wasn’t provided. ... It affects me now, my past. I have strong feelings

about it.... And I'm the one who's deaf. I was there in the first place, why wasn't anybody sharing that information with me?"

Participants who had not seen a genetic counselor personally had widely varying exposure to, and understanding of, genetic counseling. One participant recounted a definition of genetic counseling related to screening and information giving:

"My understanding about genetic counseling is that they are counselors who recommend and provide information and results of screening to explain the risks and the approaches and the outcomes. Counselors would provide information. That's what my understanding is that they do."

Another participant described how their opinion on genetic counselors improved over time:

"My first exposure to genetic counseling was like baby designers. That's what I thought, designer babies. That's a common idea that the [Deaf] community has. But as time has moved along, I've gotten a little bit more information. I realized it's not about designer babies, it's just information that is passed on. You know, it's a place of learning about your history. It's a place to find out your history and what you carry. And I think it's something that could help families."

Conversely, other participants expressed concern that genetic counselors were the same as other healthcare providers in their lack of understanding of the Deaf community. This participant attributed the similarity to genetic counselor use of the medical model of deafness:

"Well, I think [with genetic counselors] it's just another medical model, like a diagnosis, like an illness.... and that's not our perspective in the Deaf community, you know, that there is something genetically wrong or it's an illness, that's a very different thing. Genetic counseling is one thing and I think that's totally fine. But when you talk about testing for deafness, I think that's something that's not necessary."

When discussing medical genetic testing, all participants had prior understanding of genetic testing related to deafness. Two participants specifically mentioned connexin 26 and its connection to hereditary deafness. One participant described their experience discussing concerns about genetic testing within the Deaf community and speculated about its utility:

"Most of the conversations [about genetic testing] tend to be of 'not being easy to trust' 'what it can find' or 'if they would even find the gene [for deafness], would we want that?' You're always interested about your family history and everything like that and what you possibly may or may not have. But talking about specific testing, I'm not sure if I would take those tests or what I would do with that information once I found out."

While genetic testing for deafness was well known, prior knowledge of carrier screening and other types of genetic testing was less common. Of the six participants, two identified that carrier screening was related to specific recessive conditions and three had never come across the term

carrier screening before. One participant primarily thought of carrier screening as related to testing for deafness in a family:

“Carrier Screening? I think it has something to do with connexin-26. Yes. Any deaf family would want to know if they have that genetic marker. I have heard of screening for that, yes.”

5. Deaf individuals are likely to have knowledge of, or personal experience with, genetic concepts, including testing, outside of the medical setting

During interviews, we discussed participants’ experience not only with medical genetics, but also genetic concepts, including testing, outside of the medical setting. Participants were most likely to introduce to topic of direct to consumer (DTC) genetic testing and genetic engineering. All six participants referred to 23andMe as an example of DTC genetic testing. Three of the participants had personally obtained a 23andMe kit for ancestry testing and two of these also received health information. All three participants described their experience with DTC genetic testing very positively, noting how interesting and helpful the results were:

“Yeah, I went out actually took that test [23andMe]. I'm curious, I wanted to know what kind of genes I had. It was really fascinating. I got my results and I noticed some things and I really thought that for myself to know about me, I just, I thought that was so cool.”

“[23andMe] was definitely helpful for me, I would say. I think that the genetics part was cool because I already knew my family [ethnic] background anyway... My parents, they're from Ireland and my great-grandmother was from Greece. I knew all that information, but just to see how everything was put into place... And seeing that I'm a percentage of Native American was jaw dropping to me. I thought, ‘wow, I didn't know that was there’ so that was a cool part. Another part was my, just to know the risk for, let's say... diabetes or a risk for a type of Alzheimer's. It would show me if it was negative or it was a positive. And it kind of helped me to know if I would need to worry about that in the future or not.”

Although all three participants who had personal experience with DTC testing viewed it positively, one expressed concern for how less educated peers may view their results:

“I [have a degree in] biomedical sciences. So I already have a very good genetics and science background. I think it's very sticky. I think if you want to know who you are as a person, where your ancestry comes from, that's great. But using... 23andMe, if you use that as a tool to see if you will gain weight, if you're at risk of breast cancer, to see if you maybe have deafness in your family, or blindness, or any of that, that's when I see it as a very dangerous thing. It is cool. But, at the same time, if you're not educated about that process, I think you can interpret everything incorrectly.”

In contrast to their favorable comments surrounding DTC genetic testing, genetic engineering was viewed extremely negatively by all participants. All six participants acknowledged concern or fear within the Deaf community regarding genetic engineering. One participant conveyed that they were apprehensive about the expanding capabilities of new technology:

“Going in and [editing] the DNA... all the technology nowadays is shocking. And more and more people are curious... and are doing genetic testing—and are you know, fearful... nowadays, it’s like we can play with the DNA, and make changes... it’s so different now.”

Another participant highlighted that their concerns are primarily related to genetic engineering rather than testing or screening:

“One thing I want to emphasize, and again my perspective, and even the Deaf community’s perspective, is that we are fine with screening and testing. The concerns would be about engineering itself. That’s where there’s fear and the trepidation in the community. There’s not enough bioethics conversations out there. If there was more of that, I think that would be a bridge between the medical community and the [Deaf] community at large.”

Multiple participants connected the Deaf community’s unease around genetics in general to be related to the potential consequence of genetic engineering if it is used to prevent deafness in future generations: erasure of Deaf culture. One participant described their fear that they could live to see the end of the Deaf cultural line:

“I am concerned about that, in my lifetime, that that could happen, that there could be no more deaf kids.”

III. Opinions on Genetic Testing

6. All types of genetic testing should be available to everyone

We asked participants their opinions on a variety of genetic testing scenarios in cancer, pediatric, prenatal, and preconception settings. Topics of discussion included whether genetic testing should be available to individuals in the described scenario and what each individual thought they would personally do in those settings and why. All six participants felt that genetic testing should be an option for patients in all presented scenarios. Even so, each participant’s reasons for supporting genetic testing and their personal responses regarding whether they would

utilize genetic testing in given situations varied significantly (Table 3). One participant summed up the Deaf community’s view of testing to be as variable as its members:

“The Deaf community’s view on screening: some are open to it, some are resistant, as a community to do [different kinds of] genetic testing.”

Table 3: Personal Responses to Genetic Testing Scenarios

	Positive	Negative	Conflicting
Cancer	Preparation Screening Prevention Family history	Fear for the future	
Pediatric	Parent’s right to know Preparation Family planning	Unnecessary testing	News delivery method
Prenatal	Preparation Parent’s right to know Patient autonomy	Unnecessary testing News delivery method	Termination decisions
Carrier Screening for Cystic Fibrosis	Preparation Patient autonomy Family planning	Fear for the future	Termination decisions
Carrier Screening for deafness in a d/Deaf couple	Family history Family planning Preparation Identity		Unnecessary Testing
Carrier Screening for deafness in a hearing couple	Preparation Deaf culture involvement Language acquisition (ASL)	Termination decisions Assistive devices Misconceptions	News delivery method

When discussing genetic testing in the cancer setting, participants described genetic testing as beneficial in general or for them personally for the purposes of preventative care, screening, and prevention as well as to learn about how their family history of cancer contributed to their own risk. Concern was expressed by participants regarding whether genetic testing would produce unnecessary anxiety about future cancer risk.

For genetic testing in a pediatric setting, participants felt that parents have a right to know if their child has a genetic condition and that the testing results could be useful for preparation

for the future and family planning decisions. Some participants felt that pediatric genetic testing was unnecessary since it might not change anything for the child's care. The topic of how results were communicated brought up both positive and negative reactions in participants. Some felt that providers had an opportunity to improve a situation with the way they explained results, and others felt that the more likely scenario was that providers would exacerbate or over-exaggerate the seriousness of a situation.

In the prenatal setting, genetic testing was perceived positively by participants in regards to preparation for a child's condition, the parents' right to know if the pregnancy is at risk for a genetic condition, and for a patient to be in charge of their own medical care. However, some participants felt prenatal genetic testing was unnecessary if the pregnancy was not high risk and other were concerned that the delivery of unexpected news would be detrimental to a pregnancy. There were conflicting opinions about how termination decisions play into the acceptability of prenatal genetic testing. Some participants supported testing so parents were able to choose termination in cases of serious conditions and others felt that testing should not be available if parents would terminate a pregnancy affected with a serious genetic condition.

During the interviews, carrier screening was discussed in three different contexts: screening for cystic fibrosis (CF), screening for deafness in a Deaf couple, and screening for deafness in a hearing couple. In all cases, participants supported the availability of carrier screening to help parents be prepared for their child's future.

Discussion of carrier screening for cystic fibrosis focused around its status as a serious condition with all participants supporting its availability to all parents. Participants felt that parents should have the right to choose to know about this information and that testing would be beneficial for family planning purposes. Concern arose from participants around whether genetic

testing would produce unnecessary anxiety about their child's disease course as well as the risk to future pregnancies. There were conflicting opinions about carrier screening for CF relating to termination decisions. Some participants supported screening so parents have the option to terminate an affected pregnancy and others felt that screening should not be available if parents would terminate an affected pregnancy.

Carrier screening for deafness in a Deaf couple was viewed positively overall. Participant perceived benefits included knowing about the family history, being able to plan for a hearing or deaf child, and learning more about their Deaf identity. One participant felt that carrier screening in Deaf couples was unnecessary if parents would be accepting of a deaf or hearing child.

Carrier screening for deafness in hearing couples was the most polarizing topic. Some participants strongly supported its availability, citing the opportunity for parents to get involved with Deaf culture and learn ASL, which would help the child acquire language early. Other participants strongly opposed carrier screening for deafness in hearing couples, citing concerns that parents would terminate deaf pregnancies, providers would only present the options related to assistive devices, and that parents would only see negative deaf stereotypes rather than the positive potential opportunities. All participants believed that the way that healthcare providers delivered news to hearing parents would have a large impact on the parents' perspective. Some felt this was a valuable opportunity to educate new parents while others felt it would lead to perpetuation of the medical model of deafness.

III. Recommendations for Genetic Counselors and Healthcare Providers

At the end of the interview, participants were asked what recommendations they had for genetic counselors and other healthcare providers. Their suggestions fit into the following three

categories: accommodate for communication differences, respect patient autonomy, and be open minded (Table 4).

Table 4: Recommendations for Genetic Counselors and Healthcare Providers

Accommodate for communication differences	<ol style="list-style-type: none"> 1. Use only certified interpreters 2. Maintain eye contact 3. Use computer for writing 4. Explain things before/ after exam 5. Use less jargon
Respect patient autonomy	<ol style="list-style-type: none"> 1. Present all options equally (without bias) 2. Educate hearing families about Deaf options (ASL, Deaf culture) 3. Don't push assistive devices
Be open minded	<ol style="list-style-type: none"> 1. Recognize Deaf culture 2. Do not assume 3. Include Deaf people 4. Recognize the provider's role in making positive change

Accommodate for communication differences

The most common participant recommendation was for health providers was to adapt to the preferred communication style of the Deaf patient. For many Deaf patients, that involves using certified interpreters. One participant put it simply saying:

“[Healthcare providers] should always be providing interpreters. That’s the number one thing.”

Participants emphasized that using a certified interpreter is essential for adequate care. One participant described how not having an interpreter was a barrier to her health care choices:

“I never had an interpreter. I always had my mother that took care of everything for me. So I had no knowledge of what was available.”

Two participants mentioned that another important aspect of providers using interpreters is appropriate etiquette such as maintaining eye contact with the patient during conversation. One participant described how valuable eye contact is for them to connect to their provider:

“I would definitely want the doctor to know about my eye contact. That’s very important. For example, the nurse should look at me when they’re talking to me and don’t look at the interpreter. They’re typically looking straight at the interpreter and not looking directly to me... But that’s something, I would really want them to understand because if I don’t have eye contact from the doctor, then I don’t feel connected to them. I feel very disconnected, like the conversation is between the interpreter and the doctor.”

However, interpreters are not always present at appointments and not all Deaf individuals prefer to use them. Four of the six participants described writing back and forth to doctors at some point during their care by preference or out of necessity. One participant described the difficulty of this process and the benefits of using computers for improving reliable communication:

“You know, they commonly say that doctors have bad handwriting. Sometimes interpreters don’t show up. A lot of times the doctor’s office doesn’t even request an interpreter. You’re stuck with the doctor writing. If the doctor has horrible handwriting, and they don’t have the patience, then you’ve got to get a nurse to write for the doctor... But as of late, using the laptop instead of writing is a little bit better, because it forces it all to be clear. Then the doctor can speak, and the computer can translate it as text.”

In addition to different communication strategies, participants suggested ways to make the appointment process more easily understandable for Deaf patients. One participant noted that healthcare providers often speak during exams and recommended providing explanations before and after procedures so Deaf patients can be aware of what’s happening throughout:

“As long as they make sure ... during the examination, [to] not talk, and then wait until it’s done to explain whatever is going on...and also explain everything before you actually do it, and then do the examination, a lot of doctors don’t do that very well.”

Finally, participants brought up the fact that the language barrier between English and ASL can complicate communication of already complex medical terms. One participant described how even when information is provided, it’s not always understandable to patients:

“It would be nice if they understood that medical literacy is not a strong suit of deaf people in general, because English is a second language for them. For example, if they show you medical papers, they have a lot of complicated words and vocabulary that are not part of the current vernacular of the Deaf community. So it’s hard to catch. But it’s been like that. So if they could make accommodations--that would be easier.”

Respect patient autonomy

Five of the six participants noted a need for healthcare providers to be nondirective in their counseling and care. One participant connected this idea specifically to the impact of word choices and the language used to describe deafness or medical conditions:

“Just in general, as a counselor in the medical profession, just to be aware of how it is you’re communicating, your word choice and language.”

Half of the participants felt that this nondirective approach was most important when a genetic counselor was presenting options to hearing parent of a deaf child. One participant emphasized to start with acceptance of deafness rather than concern:

“Everything should be presented thoughtfully. The presentation is what is important [and] that they not be too judgmental about people's choices... If a family comes in, and there's a high chance that they'll have a deaf child, [the genetic counselor] starts with the thought of “that's fine” not to look at that situation and say, ‘that's a medical concern having a deaf child.’ They have to take a lot more things into consideration and not to be judgmental and to push their opinions. Be compassionate when they deal with us.”

Five participants recommended that providers should educate hearing families about Deaf cultural options along with medical options such as assistive devices and speech therapy. One participant described what Deaf options they would want providers to know about:

“I'd talk to [the provider] about all the resources, about sign language, how it's beneficial, Deaf schools, organizations, different supports that deaf people have [like] LEAD-K [Language Equality and Acquisition for Deaf Kids] and advocate for language acquisition.”

Another participant mentioned how providers could also provide Deaf cultural information by connecting families to the Deaf community:

“If [providers] ever meet hearing parents with deaf children or meet hearing parents that are pregnant with a deaf child, I would want them to suggest to them, for them to be able to contact and meet deaf people first and just see what the community is like, instead of...giving them just the medical perspective and saying ‘Oh, well, your baby can have a cochlear implant or your baby can have speech therapy’, and all that. That's only one way to look at it. I want the doctors to also realize that, it's not just cochlear implants, deaf people, they have a whole lot of things that they've actually adjusted to their deafness.”

Additionally, three participants discussed the possibility of getting genetic counseling for their own family planning. This potential often came with concern about how a genetic counselor would address the topic of deafness with Deaf patients that have extensive personal experience. One participant described how their Deaf identity and culture has given them expertise in deafness and accommodations that a genetic counselor could not provide:

“I think if the genetic counselor, if they were to explain to me, ‘Hey, your child could possibly be deaf’, or ‘whatever other thing that could be related to deafness’, I would definitely want them to know and hopefully see that I'm a Deaf individual, first and foremost. Deafness is not really, the same as a medical condition, it has a language, and it has a culture. And it's really a lot more than just being a medical condition... And I would really want a [provider] to be able to see that, see that I come from a Deaf community... And I wouldn't want the [provider] to be trying to tell me stuff about getting the baby a cochlear implant or doing speech therapy or trying to be able to talk and stuff. I really would not want their influence in regards to that. Just because being Deaf, I already know everything related to that... that was

part of my upbringing. And in my community, I've seen everyone. I've seen people with a cochlear implant. I've seen people that have speech backgrounds. I've seen people that have actually gone to the hearing schools and stuff like that. And honestly, the doctor would be an outsider. They're not deaf, they're not involved with the Deaf community and Deaf culture. So they are honestly kind of clueless. I really wouldn't want them to try and influence my decision relating to that. I would honestly be the best person to make that decision."

Be open minded

All six participants suggested that the most impactful things a healthcare provider can do for a Deaf patient are to accept them for who they are and be open minded. Four participants described this recognition of Deaf culture as an essential part of multicultural counseling:

"[Healthcare providers need] cultural accommodation, and cultural competency, and cultural sensitivity"

"[Genetic counselors should] look at being Deaf as, you know, the culture, compared to another illness that a person has... So for me, it's up to the genetic counselor... [It's a part of] cultural competency."

One participant emphasized how demeaning negative comments from providers can be, especially since they feel Deaf individuals are treated differently than other cultural groups:

"[Providers] need to be culturally sensitive as well. I don't want to hear the doctor say, "Oh, you're deaf" as a negative... It's like telling black people, "Oh, what a terrible experience," something similar to that."

In addition to the acknowledgement of Deaf culture, three participants also pointed out the importance of not making broad assumptions. Two participants described how providers often overlook the diverse experiences of individuals in the Deaf community with different educations, preferences, and needs:

"I would want a doctor to recognize that deaf people typically are educated, you know, very educated, and that's oftentimes judged and [they assume] that we're not educated... Often I'll say something and they'll look surprised at me, at the terminology that I know or the phrases that I know. Like of course, I'm educated. I grew up in the school system. You know, so maybe I'd want doctors to be more open-minded to something like that... Definitely have more awareness that there's a big spectrum with deaf education. Some were born and raised and have no exposure to ASL"

"[I would want my providers to have] knowledge that there are different deaf individuals. All deaf people are not culturally Deaf or culturally included. Some doctors make the assumption that all deaf people come from the same part of the culture. So they lump us all together instead of saying that we're different and we have various accommodation needs."

Two other participants highlighted the need for providers to be aware of their assumptions and willing to ask questions when they interact with a deaf patient:

“Doctors will look at me as a disabled person, but I want them to know how to ask me things, not have those preconceived ideas.”

“I think the medical community should recognize their biases, and know that deaf people should have options, that we live as other people do, and we have situations other people do. But they make assumptions about all deaf people. [Providers] should check their internal biases when they are going to have a deaf patient.”

Three participants suggested that the lack of awareness may be due to lack of experience and recommended that providers find ways to involve Deaf individuals in provider education and experiences. One participant suggested that training programs would benefit from further education about, and interaction with, Deaf patients:

“When I’ve talked to the doctors before, they only get a short, maybe 10-minute education, presentation, on dealing with someone who is deaf [in medical school]. And was it the right information? Was it accurate? Was it beneficial? Probably not. It needs to be given to them by someone in that community who has those experiences. In the residency maybe they should take an opportunity to work with deaf people and maybe they would know in future how to deal with the situation.”

Another participant recommended that providers could build understanding of the Deaf perspective by interacting with Deaf individuals not as patients, but in their home communities:

“I suggest that [providers] should try and get involved with the Deaf community, try and look into the Deaf culture and Deaf language, and see exactly what [deaf people] can do first before they try and just offer them “solutions” and just one way to kind of try and “correct” it. Because it doesn’t have to be corrected.”

Finally, all six participant expressed that it was important to them for providers to recognize their role in making positive change in the medical community by accepting Deaf culture and understanding Deaf perspectives. One participant felt that positive change would act as reparation and called attention to how the medical perspective has harmed the Deaf community:

“I would want to motivate [providers] and try and encourage them to look at exactly how oppressed deaf people are compared to hearing people, and why. They’re discriminated against. That’s what the medical perspective has given to the Deaf community... The medical opinion really does have a big influence on the entire general population and their perspective. And I want to say, ‘Hey, will everything be equal for deaf people and hearing people? Do you guys understand?’ That’s something I want them to look at.”

Another participant highlighted the significance of a provider’s impact—either positively or negatively—to individual patients and the larger community.

“I know [the provider’s] role in the medical community is very important... But people in the medical community will say, you know, ‘Don’t teach your child American Sign Language, only English,’ that kind of thing. So I really just think that while it’s important to focus on the family and the patient, you also have

to consider how it is you can change things within the medical community and change that [medical] perspective.”

Similarly, another participant shared a personal experience of a provider’s impact and emphasized that providers have the opportunity to affect the quality of people’s lives:

“Having to grow up speaking, and not having American Sign Language until an early teen, I just feel like I missed out on a lot. I was bullied a lot. I wish I had access to that language earlier on. I was picked on, couldn’t make friends, because I couldn’t communicate. But you know, it’s something that we just keep emphasizing. It hurts deaf people to not have that. I mean genetic testing is great. And it’s good for people to have that information. But it just seems to me, as a provider who provides results, you have a good chance, or a higher chance of being able to have an impact on someone’s life.”

Because of this ability to make a difference, one participant asserted that healthcare providers have not only the opportunity, but also an obligation to push for positive change:

“I would really want to just make sure that the doctors understand and notice what their responsibilities are. And if there’s like a possibility... that they may have hearing families with deaf children, I just hope that you guys are able to have that conversation and you can explain to them and just say, ‘Hey, if you just kind of push all this information to the side, you already have evidence, you already have research, you have interviews, if you just push all this to the side, and you still just go ahead with your medical perspective to the hearing parents, just know that you are responsible and you’re liable for the impact that you’re going to have on those families.’”

DISCUSSION

Through this study, Deaf individuals' stories and perspectives were collected, allowing for a better characterization of the Deaf community's attitudes and opinions towards genetic counseling and genetic testing. This study found that participants' experiences with members of the medical community—either positive or negative—influenced the way they view other providers, Deaf individuals often have low expectations for providers, and they prefer providers who have experience with Deaf patients. The study also found that interviewees had variable familiarity with medical genetic testing, were more likely to know of genetic testing relating to deafness, and were more likely to be familiar with genetic concepts outside of the medical settings such as direct to consumer genetic testing and genetic engineering. Finally, all participants supported the availability of genetic testing options in different scenarios presented to them. These themes both replicated and expanded upon the current literature on this topic.

Overall, this study supported the findings of previous research. Similar to prior studies, this study's participants described feeling misunderstood by their healthcare providers, sometimes resulting in inadequate care and negative perceptions of other providers (Iezzoni, 2004; Steinberg, 2002; Steinberg, 2006). Participants also highly valued the rare providers that had experience with Deaf patients, bolstering the assertion that most providers lack training and understanding regarding Deaf individuals, practices, and culture (Hoang, 2011; Nagakura, 2014). Moreover, participants viewed provider efforts to improve communication and understanding of Deaf patients very positively, further emphasizing the importance of cultural competency and communication accommodation to the patient-provider relationship as well as health outcomes (Baldwin, 2012; Enns, 2010; Palmer, 2017; Preminger, 2014). This verification is not

consistently found in qualitative research and is particularly rare for studies with small cohorts or understudied groups of individuals, making this study's replication of prior themes critically important.

In addition to supporting the findings of previous research, this study also found novel themes, adding nuance to the existing research about the Deaf community. One surprising finding was that, while genetic counselors often perceive themselves to have advanced skills in patient interaction and understanding, the Deaf individuals in this study perceived genetic counselors to have the same strengths and weaknesses as other healthcare providers. This is interesting considering Nagakura's 2014 study showing that half of recent genetic counseling graduates felt that their Deaf Awareness Training was sufficient to interact with Deaf individuals. This discrepancy may imply that Deaf individuals have a hard time finding and connecting with genetic counselors who have sufficient Deaf awareness training or that Deaf individuals have higher standards for cultural awareness than expected by genetic counselors. If the latter explanation proves true, it is possible that existing Deaf awareness training of genetic counselors is not sufficient to meet the needs of Deaf individuals.

Another novel finding was how interested participants in this study were in genetics. Previous research focused on Deaf attitudes toward genetic testing for deafness has conflicting conclusions showing positive, negative, and mixed perceptions (Martinez, 2003; Palmer, 2013). Three of this study's participants had previously undergone medical genetic testing including genetic counseling. In this study, all participants supported the availability of genetic testing in all situations discussed: cancer, pediatric, and prenatal settings as well as carrier screening for cystic fibrosis, carrier screening for deafness in a Deaf couple, and carrier screening for deafness in a hearing couple. Though personal reasons to pursue genetic testing varied, all participants

said that they would pursue genetic testing in three or more of the six scenarios discussed. Additionally, three participants had previously undergone direct to consumer genetic testing. These responses provide support to assertions that Deaf individuals have a strong interest in genetic testing (Boudreault, 2010). Additionally, prior studies only discussed genetic testing for deafness, making this study the first to assess Deaf individuals' opinions on genetic testing outside of genes for deafness.

Finally, Deaf individuals shared new stories of their personal experiences with genetic counseling and genetic testing for the first time in this study. The Deaf community is historically understudied, particularly in regards to medicine, making any contribution to existing knowledge about this group incredibly valuable.

Practice Implications

Through the participant recommendations, genetic counselors and other healthcare providers can gain insight on concrete ways to improve clinical care of Deaf patients. The most common participant advice for healthcare providers was to adapt to the preferred communication style of the Deaf patient. Providers can accommodate by first asking what strategy works best for that person. For patients who prefer ASL interpreters, providers can use certified interpreters at every interaction to ensure accuracy of information exchanged, even if bilingual family members are present. For patients who prefer to write back and forth, providers can use computers to type to ensure efficiency and legibility. For all Deaf patients, providers can make sure to maintain eye contact with their patient during conversation to build a connection with them, especially when there is an interpreter present. For appointments in which there is a physical exam, providers can explain what they will do before and after the exam rather than during it so Deaf patients can be aware of what is happening throughout. In all interactions and in patient materials, providers can

use less medical jargon to assure that information is understandable to Deaf patients, for whom English may be a second language.

Another common participant recommendation was for healthcare providers to respect patient choices by being nondirective in their counseling and care. Specific suggestions for providers working with hearing parents of deaf children included using neutral or positive language to describe deafness, providing information about Deaf culture to hearing parents of deaf children along with medical options like assistive devices, and connecting families to the Deaf community. Participants also recommended that providers working with Deaf adults respect the personal experience of the Deaf individual when addressing the topic of deafness by acknowledging the patient as the expert on their own situation.

Finally, all participants suggested that the most impactful things a provider can do to improve relationships with their Deaf patients is to accept Deaf individuals for who they are and be open minded to Deaf culture. Providers can build cultural competency by learning about and acknowledging the cultural model of deafness. To be accepting of Deaf individuals with different educations, preferences, and needs, providers can build self-awareness about their assumptions and ask their patients questions about what works best. If they do not have much interaction with Deaf individuals, providers can build understanding of the Deaf perspective by reaching out to Deaf communities and including Deaf people in education of future providers. Furthermore, all participants expressed that it was important for providers to recognize their role in making positive change in the medical community. Participants suggested that providers can learn about Deaf oppression to understand their patients' experiences and the importance of change. To maximize the impact to patients, providers can start these adjustments now by deliberately changing the way they talk about deafness around hearing and Deaf individuals. Additionally,

participants recommended that providers look for opportunities to shift the perspective of the medical community by encouraging other providers to adopt these practices.

Study Limitations

This study has several limitations, including small sample size, selection bias, diversity in the participants, use of interpreters, and potential effects of the interviewer on the responses and interpretations. Only six individuals were interviewed due to time and participant interest constraints. Since this is a qualitative study with a small sample size, findings are not generalizable to all Deaf individuals who use ASL to communicate or the Deaf community as a whole.

Participants were recruited through the Facebook group “The Voice of the Deaf Community in Massachusetts” and the “Florida Association of the Deaf, Inc.” Facebook page. Hence, there is a possibility for selection bias as it is unknown what factors motivated their participation. It may be that Deaf individuals who are more trusting of the medical community or more interested in genetics were more likely to view the recruitment video and be willing to discuss their experiences. This prospect is more likely since multiple individuals left negative Facebook comments regarding genetics or the medical community on the recruitment posts and none of these commenters participated in the study.

Additionally, the participants differed from each other in age, gender, family history, parental status, health status, location, and level of education. Hence, the diversity in the participant characteristics could have influenced their experiences and responses. For example, Deaf individuals who grew up in hearing families may have different opinions of the utility of genetic information about deafness than those who grew up in Deaf families. Despite this diversity, there are still groups within the Deaf community that were excluded from this study

such as individuals with syndromic deafness, those who primarily speak English, and those who communicate using other methods such as tactile signing, Cued Speech, or Manually-Coded English systems.

Furthermore, VRS interpreters were used during the interviews due to the language barrier between the English-speaking interviewer and the ASL-using interviewee. Different interpreters were used for each interview due to VRS company policy. Because of these factors, there is a possibility that questions or answers in the interview were translated inaccurately. The notes taken during interviews along with video recordings and audio transcripts were reviewed, compared, and edited multiple times to mitigate this risk.

Lastly, the same interviewer conducted all the semi-structured video interviews. However, given the qualitative nature of the research method, the participants' responses may have been influenced by the interviewer. Additionally, there is a potential for a personal bias as the editing, analysis, and interpretation of the responses relied on the interviewer alone.

Research Recommendations

This was the first study to conduct individual interviews assessing Deaf individuals' attitudes and perceptions of genetic counseling and genetic testing. Given this study's goal to expand understanding of the Deaf community's perspective on genetic counseling and genetic testing, conducting a study with a larger sample size would help improve the depth of knowledge available on the subject and provide support for recurrent themes. Future studies including all individuals in the Deaf community, regardless of communication preference, would complement existing information and allow for development of a more robust and comprehensive knowledge base. It would be interesting to compare cohorts who did and did not have experience with genetic testing and/or genetic counseling to see how perceptions differed with personal

experience. Finally, to continue building a trusting relationship between the medical and Deaf communities, it is essential to involve Deaf individuals in all steps of future research of the Deaf community: from idea development to publication and presentation.

CONCLUSION

This qualitative study documented the attitudes and opinions of Deaf individuals who primarily use American Sign Language (ASL) regarding genetic counseling and genetic testing. Themes that emerged from the study included: (1) Participants' experiences with members of the medical community—either positive or negative—impacted the way they view other healthcare providers. (2) Deaf individuals typically have low expectations for their providers and staff. (3) Deaf individuals prefer providers that have experience with Deaf patients. (4) Interviewees had variable familiarity with medical genetic testing and were more likely to know of genetic testing relating to deafness. (5) Participants were more likely to be familiar with genetic concepts outside of the medical settings such as direct to consumer genetic testing and genetic engineering. (6) All interviewees supported the availability of genetic testing options in different scenarios presented to them.

Through the participant recommendations, genetic counselors and other healthcare providers can gain insight on concrete ways to improve clinical care of Deaf patients. Suggestions fell into three categories: (1) Adapt to the preferred communication style of the Deaf patient by asking what works best, using interpreters or computer typing, maintaining eye contact, explaining before and after exams, and simplifying jargon. (2) Respect patient choices with nondirective counseling and care by avoiding negative descriptions of deafness, providing information about Deaf culture and communities to hearing parents of deaf children, and acknowledging Deaf patients as the expert on their own situation. (3) Be open minded to Deaf culture by learning about Deaf perspectives, acknowledging the cultural model of deafness,

building self-awareness about prior assumptions, including Deaf people, and recognizing the provider role in making positive change in the medical community.

This study supports and adds nuance to prior research, emphasizing the importance of cultural competency and communication accommodation for genetic counselors and other healthcare providers working with Deaf patients. As genetic testing increasingly becomes part of routine clinical care, more Deaf individuals will be referred for genetic counseling. Continued efforts to understand Deaf culture and perspectives are essential to improve relationships with and provide quality healthcare to this population.

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APPENDIX A: PERMISSION DOCUMENTATION

Permission from The Voice of the Deaf Community in Massachusetts Administrator

The image shows two screenshots of a Facebook Messenger conversation. The top screenshot shows the start of the conversation on October 15th at 5:24 PM. Julie Howell sends a message to John Pirone, who is the administrator of 'The Voice of the Deaf Community in Massachusetts'. Julie explains her thesis project, which aims to provide a space for Deaf individuals to share their perspectives on genetic counseling and carrier screening. She mentions that she is a second-year genetic counseling student at Brandeis University and is currently obtaining her Master's degree in Genetic Counseling. She also notes that she is hearing but has become involved with the Deaf community through friends, ASL classes, and a placement at The Learning Center for the Deaf in Framingham, MA.

The bottom screenshot shows the continuation of the conversation. Julie asks John for permission to post a recruitment blurb for her study on the Facebook page. She includes a written statement and a video with the statement in ASL. The recruitment blurb asks if anyone is interested in sharing their perspective on genetic testing and counseling. Julie expresses her hope that the study will act as a resource for genetics professionals and help identify areas of potential concern. She concludes with a thank you and her name, Julie Howell.

Message 1 (OCT 15TH, 5:24PM):

Hello John!

I'm contacting you about posting on the Voice of the Deaf Community in MA Facebook page as you are the admin. My name is Julie Howell and I'm a second year genetic counseling student at Brandeis University in Waltham, MA. I am obtaining my Master's degree in Genetic Counseling.

I am hearing, but I have been lucky enough to get involved with the Deaf community through a few Deaf friends, ASL classes, and most recently through a community placement with The Learning Center for the Deaf in Framingham, MA. Through my friends' experiences as well as my classes, I've quickly realized that medical communities and the Deaf community have very different perspectives on deafness and medical management for Deaf individuals. Because of this disparity, I hope to use my thesis as an opportunity to provide Deaf individuals with a space within the medical community to share their perspectives and give genetic counselors and other genetic and medical professionals an opportunity to learn from them.

For my thesis project, I will be asking the opinions of culturally Deaf individuals on genetic counseling and carrier screening. I will be conducting individual video interviews with an ASL interpreter with adults (18-35) who are culturally Deaf and

Message 2:

within the medical community to share their perspectives and give genetic counselors and other genetic and medical professionals an opportunity to learn from them.

For my thesis project, I will be asking the opinions of culturally Deaf individuals on genetic counseling and carrier screening. I will be conducting individual video interviews with an ASL interpreter with adults (18-35) who are culturally Deaf and communicate primarily through ASL. The study aims to identify themes among Deaf individuals with regards to genetic testing and counseling. It is my hope that this study will act as a resource to genetics professionals looking to understand the perspectives of Deaf individuals and help identify areas of potential concern that may arise when providing care to these patients.

I'd like to ask if you would allow me to post a recruitment blurb for my study on the Voice of the Deaf Community in MA page? I've included the written statement below, but the post will also contain a video with the statement in ASL.

Please let me know what you think or if I can provide you with any additional information. Thank you for your time and consideration!
Best Wishes,
Julie Howell

Recruitment Blurb:

"Are you interested in sharing your perspective on genetic testing and genetic counseling? We want to learn from you! My name is Julie Howell and I am a second year graduate student at the Brandeis University Master's Program in Genetic

Julie Home Create

John Pirone
Active 33m ago

Recruitment Blurbs:

"Are you interested in sharing your perspective on genetic testing and genetic counseling? We want to learn from you! My name is Julie Howell and I am a second year graduate student at the Brandeis University Master's Program in Genetic Counseling. For my thesis project, I am looking to interview Deaf individuals age 18-35 that communicate primarily with ASL and learn about their thoughts on genetic testing and counseling. Interviews will last about one hour and participants will receive a \$25 Amazon gift card! I hope my research project will give Deaf individuals a space to share their opinions and genetic counselors and other genetic health professionals an opportunity to learn from them. The interviews will be conducted over Zoom video conferencing with an ASL interpreter. Please pass along this message to anyone who may be interested and contact Julie via email, text, or VRS if you're interested in participating or learning more!
Email: jhowell@brandeis.edu
Text or VRS Call: 937-239-5320"

OCT 20TH, 7:31PM

You can now call each other and see information like Active Status and when you've read messages.

Yes, please. You can post your study on TVDCM and I will approve it. Thank you!

OCT 20TH, 10:05PM

Thank you so much! Ill let you know any updates before I post in late November.

Type a message...

Permission from the Florida Association of the Deaf, Inc. Administrator

Julie Home Create

Florida Association of the Deaf, Inc.

Florida Association of the Deaf, Inc.
3.6K people like this
Nonprofit Organization

OCT 22ND, 9:34PM

Hello FAD Team,

My name is Julie Howell and I'm a second year genetic counseling student at Brandeis University in Waltham, MA. I am obtaining my Master's degree in Genetic Counseling.

I am hearing, but I have been lucky enough to get involved with the Deaf community through a few Deaf friends, ASL classes, and most recently through a community placement with The Learning Center for the Deaf in Framingham, MA. Through my friends' experiences as well as my classes, I've quickly realized that medical communities and the Deaf community have very different perspectives on deafness and medical management for Deaf individuals. Because of this disparity, I hope to use my thesis as an opportunity to provide Deaf individuals with a space within the medical community to share their perspectives and give genetic counselors and other genetic and medical professionals an opportunity to learn from them.

For my thesis project, I will be asking the opinions of culturally Deaf individuals on genetic counseling and carrier screening. I will be conducting individual video interviews with an ASL interpreter with adults (18-35) who are culturally Deaf and communicate primarily through ASL. The study aims to identify themes among Deaf

Type a message...

Florida Association of the Deaf, Inc.

communicate primarily through ASL. The study aims to identify themes among Deaf individuals with regards to genetic testing and counseling. It is my hope that this study will act as a resource to genetics professionals looking to understand the perspectives of Deaf individuals and help identify areas of potential concern that may arise when providing care to these patients.

I'd like to ask if you would allow me to post a recruitment blurb for my study on the Voice of the Deaf Community in MA page? I've included the written statement below, but the post will also contain a video with the statement in ASL.

Please let me know what you think or if I can provide you with any additional information. Thank you for your time and consideration!
Best Wishes,
Julie Howell

Recruitment Blurb:

*Are you interested in sharing your perspective on genetic testing and genetic counseling? We want to learn from you! My name is Julie Howell and I am a second year graduate student at the Brandeis University Master's Program in Genetic Counseling. For my thesis project, I am looking to interview Deaf individuals age 18-35 that communicate primarily with ASL and learn about their thoughts on genetic testing and counseling. Interviews will last about one hour and participants will receive a \$25 Amazon gift card! I hope my research project will give Deaf individuals a space to share their opinions and genetic counselors and other genetic health professionals an opportunity to learn from them. The interviews will be conducted over Zoom video conferencing with an ASL interpreter. Please pass along this

Type a message...

Florida Association of the Deaf, Inc.

over Zoom video conferencing with an ASL interpreter. Please pass along this message to anyone who may be interested and contact Julie via email, text, or VRS if you're interested in participating or learning more!
Email: jhowell@brandeis.edu
Text or VRS Call: 937-239-5320*

Thanks for messaging us. We try to be as responsive as possible. We'll get back to you soon.

WED 10:35 AM

If you want us to post your recruitment blurb, we strongly suggest you to do a signing video to help our DHH community members to understand what it is about. Thanks.

Thank you so much for your response! I will absolutely have a signed video of the message. Unfortunately it is not complete yet. I am not fluent in ASL and I was lucky to have a Deaf individual on my thesis committee volunteer to sign in the video. Would you allow me to post if I have a video in ASL along with the written English? Thank you again for your time and consideration!

Yes we will post the video in ASL with the captions in English.

Thank you so much for the opportunity! I will work on getting the video ready and I'll get back to you when it is complete and ready to post.

Type a message...

APPENDIX B: RECRUITMENT NOTICE

Recruitment Notice for VDCMA and FAD (ASL video translation available upon request)

Are you interested in sharing your perspective on genetic testing and genetic counseling? We want to learn from you! Brandeis University student Julie Howell wants to gain an understanding of the Deaf community's perspective on genetic counseling and genetic testing. The goal of her research project is to give Deaf individuals a space to share their opinions. She hopes this will also give genetic counselors and other health professionals an opportunity to learn from the Deaf community. Julie Howell is a second year graduate student at the Brandeis University Master's Program in Genetic Counseling. For her thesis project, she will interview Deaf individuals age 18-40 that communicate primarily through ASL and learn about their thoughts on genetic testing and genetic counseling. Interviews will last about one hour and participants will receive a \$25 Amazon gift card! The interviews will be conducted over Zoom video conferencing with an ASL interpreter over VRS. Thank you so much for considering this study! This research would not be possible without the generosity of individuals in the Deaf community who are willing to share their stories. Feel free to share this information with anyone you think would be interested in participating or learning more! Please contact Julie via email, text, or VRS for more details:

Email: jhowell@brandeis.edu

Text or VRS Call: 937-239-5320

APPENDIX C: INFORMATION SHEET

BRANDEIS UNIVERSITY GENETIC COUNSELING GRADUATE PROGRAM

Deaf Individuals' Attitudes and Perceptions of Genetic Counseling and Genetic Testing

You are invited to participate in a research study being conducted by Julie Howell, second year student in the Genetic Counseling Master's Program at Brandeis University. The study is being conducted under the supervision of Gretchen Schneider, MS, CGC, who is the Director of the Brandeis Genetic Counseling Program.

Please read this information sheet carefully. Participating in this research study is voluntary. We encourage you to ask questions if you want more information about any part of the sheet or the study. Please keep this information sheet for your records. It has important information, including whom to contact if you have questions in the future.

If you decide to participate in this study, we will review this information sheet and you will be asked confirm that you want to participate in the study before we begin the video interview with an American Sign Language (ASL) interpreter. You can decide to withdraw from this study at any time and for any reason.

What is this study about?

We are conducting this research study because we are trying to learn more about the opinions of culturally Deaf individuals and the Deaf community as a whole on genetic counseling and different types of genetic testing, including carrier screening. We aim to learn about what influences these attitudes and perspectives. We hope that this study will be a resource for genetic counselors and other health professionals who want to understand the perspectives of Deaf individuals.

Why have we asked you to participate?

We are asking you to take part in this research study because you are between the ages of 18 and 40, your preferred language is ASL, you identify as culturally Deaf, you have no health problems requiring a specialist outside of deafness, and you have reliable internet and phone connection.

What will you be asked to do if you participate?

If you decide to participate in this study, a video interview will be scheduled at your convenience. The video interview will occur via Zoom: Video Conferencing with an ASL interpreter available through Sorenson: Video Relay Service. The interview will be audio and video recorded using Zoom and is expected to last between 45-60 minutes. Notes will be taken during the interview. The interview will consist of a series of questions about your experience with health providers, genetic counseling, and carrier screening. It will also have questions about your opinions on genetic testing in different scenarios, carrier screening, and genetic counseling. At the end of the interview, additional questions will be asked to gather demographic and social information.

Are there any possible risks to you?

This study poses minimal risks to you. You may feel uncomfortable answering some of the questions. If a certain question or topic makes you feel uncomfortable, you are free to skip the question or ask the interviewer to move on to another topic. You can also withdraw from the study completely at any time.

There is always the risk that your information could be accidentally disclosed to people not connected with this study; however, we will do our utmost to secure your information so your identity and information remains confidential.

Will you benefit from participating in the study?

People often experience benefits, such as reduction of stress, by having the opportunity to talk about their experiences. Otherwise, you will not benefit directly by participating in this study. However, we hope the study will result in a better understanding of the Deaf community's opinions on genetic counseling and different types of genetic testing, including carrier screening. We also hope this information can provide information to genetic counselors and other health professionals serving Deaf individuals and their families.

Will it cost you anything to participate in the study?

Other than the time you spend answering questions during the phone interview, there will be no cost to you to participate in this study.

Will you be compensated or receive anything for participating in the study?

If you choose to participate in this research study, you will receive a \$25 Amazon.com gift card via e-mail to thank you for your time and valuable insight.

How will your information be kept private?

All data obtained during this study will be kept confidential. After the video interviews are conducted, your name will be removed from the interview transcript and only the student researcher will know which participants provided which interview responses. Pseudonyms will be used when reporting data from the interviews – no identifying information (name, race/ethnicity, etc.) will be used. All data will be uploaded to box.com, an encrypted site, to keep the data confidential and maintain privacy for all participants.

After your name has been removed from the interview transcript, data from this study will be reviewed by the research committee. The research committee includes Gretchen Schneider, who is the Director of the Brandeis Genetic Counseling program and a genetic counselor; Andrea Oza, a genetic counselor at Boston Children's Hospital; Karen Turley, a high school English teacher at The Learning Center for the Deaf in Framingham, MA; Katie Prins McCarthy, the Coordinator of Outreach and Support Services at the Boston Children's Hospital Deaf and Hard of Hearing Program; and Patrick Costello, Director of the Deaf Cultural Center at The Learning Center for the Deaf in Framingham, MA.

There is a possibility that data obtained from this study will be published in a peer-reviewed journal, but data will not have identifying information on it in order to protect the participants. Data will be kept until December 31st, 2020 for presentation and publishing purposes and will then be destroyed.

What if you don't want to participate or change your mind partway through?

Participating in this study is completely voluntary. You have the right to refuse to participate in all or a part of this study. Even if you decide to participate now, you may change your mind and withdraw from the study at any time without penalty. You may also refuse to answer specific questions at any time without penalty – simply skip them on the questionnaire or ask the interviewer to move on to another topic.

Who can you contact if you have more questions?

If you have any questions about the research being conducted or your participation in the research, please contact the student researcher, Julie Howell, by email at jhowell@brandeis.edu.

If you have any questions about your rights as a subject in this research, would like to speak with someone other than the researchers about concerns you have about the study, or in the event the researchers cannot be reached, please contact the Brandeis Committee for Protection of Human Subjects at irb@brandeis.edu.

APPENDIX D: INTERVIEW GUIDE

INITIAL CONTACT QUESTIONS TO DETERMINE ELIGIBILITY

Thank you so much for your interest in participating in my study! For this study I will interview Deaf individuals about their attitudes and perceptions of genetic counseling and testing. The study aims to identify themes among Deaf individuals with regards to genetic testing and counseling. I hope to use my thesis as an opportunity to provide Deaf individuals with a space within the medical community to share their perspectives and give genetic counselors and other genetic and medical professionals an opportunity to learn from them. Now I'm going to ask you a few questions to see if you are eligible for an interview:

1. What is your age?
2. What is your preferred language?
3. Do you identify as culturally Deaf?
4. Were you born with any medical problems, or had any problems as a child or now that required you to see any special healthcare providers?
 - a. If yes; what were they?
5. Do you have reliable internet and phone connection for video calling?
6. Would you be okay with being video and audio recorded for the full interview? The only people who will see the videos are me and my research supervisor at Brandeis University.

If meets criteria for the study:

1. How do you prefer to be contacted for scheduling? Via text, email, video call?

If does not meet criteria for the study:

2. Unfortunately, you do not meet the criteria for this study. Thank you so much for your interest in participating!

INTRODUCTION

[Interviewer will sign the following herself]

First I want to thank you for working with me and being willing to share your story. I got involved with the Deaf community through Deaf friends, ASL classes, and most recently through an internship with The Learning Center for the Deaf in Framingham, MA. Through others stories, I came to understand that the medical community and the Deaf community have very different perspectives on deafness. I hope my thesis can be an opportunity for Deaf individuals to share their perspectives with the medical community and for genetic counselors and other medical professionals to learn from them. Thank you again! Now I will use the interpreter for the rest of the interview.

[Interviewer will now switch to speaking and the VRS interpreter will translate]

For my study I will interview culturally Deaf individuals, asking about their opinions and thoughts about genetic counseling and testing. The goal is to identify themes among Deaf individuals with regards to genetic testing and counseling. I hope this study can assist genetic counselors and medical professionals in understanding the perspectives of Deaf individuals and identify areas of potential concern that may arise when providing care to deaf/Deaf patients.

INFORMED CONSENT

We will now review the information sheet that was sent to you via email:

1. Do you have any questions about the information sheet?
2. Would you like me to review what's in the information sheet since we have an ASL Interpreter?
 - a. If yes: The information sheet reviews the purpose of the study, the criteria of participation, the risks and benefits of participation, the cost and compensation of

participation, and how information is kept private. It also says that participation is completely voluntary, that you can withdraw from the study at any time, and that you can skip any questions during the interview. The sheet also has contact information for the researchers and other people you can contact about the research. Do you have any questions about this?

- b. Do you consent to be a part of this study? [Interviewer documents consent]
3. Is it okay for me to turn on audio and video recording of this interview? [Interviewer turns on audio and video recording.]

QUESTIONS

First I would like to ask you some questions about your own healthcare.

1. Do you currently see any healthcare providers on a regular basis?
 - a. Yes: What type? What for? How often do you go? Do you see other doctors?
 - b. No: What circumstances influence your decision? (Healthy, dislike doctor, busy, difficulty for you/doctor getting interpreter, dislike VRI interpreter?)
2. How have your healthcare providers accommodated for you as a Deaf patient?
 - a. Do you feel that strategy has met your needs as a Deaf patient?
3. Do you feel your healthcare providers have understood your perspectives, culture, and needs as a Deaf individual and patient?
 - a. Yes: What have they done that worked well for you?
 - b. No: What could they have done better? Is there anything specific that you would have liked them to understand before your visit?

Now I'm going to ask you some questions about genetics and health.

4. At any of your appointments have you ever discussed genetics or genetic testing in any context? If yes, can you explain to me the reason for the discussion?
5. What information do you know about carrier screening for genetic conditions?
 - a. Carrier screening is a type of genetic test that can tell a person if they inherited a broken gene from one parent. It looks for broken genes that cause certain traits or genetic conditions if a child inherits a broken gene from both their parents.

Carriers are usually healthy and do not have the genetic condition.
 - b. An example that you may be familiar with is the condition Usher syndrome. For example: A child has Usher syndrome. Neither of the child's parents have Usher syndrome, but both parents have one broken copy of the Usher syndrome gene. Since the child inherited one broken copy of the Usher syndrome gene from mom and inherited one broken copy of the Usher syndrome gene from dad, the child has two broken copies and has Usher syndrome.
 - c. Carrier screening allows a couple to find out the chances of having a child with a genetic condition such as cystic fibrosis, sickle cell anemia, or Usher syndrome. It can also determine whether a couple, hearing or Deaf, may have a deaf child.

Now I'm going to ask about your thoughts about genetic testing in different situations. I'll give six different scenarios.

6. For each scenario, I'll ask about two things. 1) If people in general should have the option to pursue genetic testing in this situation and 2) if you were in this situation what would you do?
 - a. Test an adult for a broken gene that can increase their risk for a type of cancer present in family members

- b. Test a one year old who was born with a heart defect, extra fingers, and is delayed in their developmental milestones to identify the symptoms' cause
- c. Test to determine if a pregnancy is affected with Down Syndrome
- d. Carrier screening for cystic fibrosis (serious life limiting lung condition)
- e. *Only ask if not discussed previously: Test to see if broken genes are the cause of a person's deafness. This test will also tell if you can pass on a gene for deafness.*

The next scenario is the last one:

- f. In general, do you think that hearing couples should have the option to pursue carrier screening for deafness?
7. There are healthcare providers who provide GC services. Have you ever seen a GC?
- Yes: In what context?
- No: What information do you know about genetic counseling?
- a. Genetic counseling is a process to evaluate and understand a family's chance of an inherited medical condition. The purpose of genetic counseling is to provide this information to patients so they can make informed decisions about their health and medical care. Genetic counselors provide options but do not tell patients how to use this information.
8. What are your thoughts about genetic counseling?
9. Do you think you would ever seek out genetic counseling?
- Yes: In what context?
- No: Why not? What factors influence that decision?
10. If you had an appointment for genetic counseling, what would you want the genetic counselor to know or understand before you met with them?

At the end of the interview: My goal is to share perspectives of the Deaf community.

1. Is there anything you would want to say to genetic counselors or health professionals?
2. Is there anything you wanted to discuss that I haven't asked about?

Thank you so much for sharing your experiences and opinions. My research would not be possible without the generosity of individuals in the Deaf community, such as yourself, who are willing to share their stories.

DEMOGRAPHICS

Now I'm going to turn off the recording and ask a few demographic questions. This information will be kept confidential and separate from your answers to questions in the interview.

1. How old are you?
2. What gender do you identify as?
3. Is anyone else in your family deaf/Deaf?
4. Do you have any children?
5. What is your highest level of education?