

**Prenatal Genetic Counselors' Perceptions of their Disability Training and Preparedness for  
Addressing Disability in Practice: A Thematic Analysis**

by

**Nicole Danielle Weise**

BS Biological Sciences, University of California, Santa Barbara, 2016

Submitted to the Graduate Faculty of the  
School of Public Health in partial fulfillment  
of the requirements for the degree of  
Master of Science

University of Pittsburgh

2024

UNIVERSITY OF PITTSBURGH  
SCHOOL OF PUBLIC HEALTH

This thesis was presented  
by

**Nicole Danielle Weise**

It was defended on  
April 9, 2024

and approved by

Michael J. Deem, PhD, Associate Professor, Department of Human Genetics, School of Public Health

Amy Houtrow, MD, PhD, MPH, Professor and Vice Chair, Department of Physical Medicine & Rehabilitation, School of Medicine

Thesis Advisor: Robin E. Grubs, PhD, MS, LCGC, Associate Professor, Department of Human Genetics, School of Public Health

Copyright © by Nicole Danielle Weise

2024

# **Prenatal Genetic Counselors' Perceptions of their Disability Training and Preparedness for Addressing Disability in Practice: A Thematic Analysis**

Nicole Danielle Weise, MS

University of Pittsburgh, 2024

Prenatal genetic counselors often engage families in discussions about what it would mean for their child to be born with a genetic condition and, potentially, live with a disability. They are responsible for providing adequate, value-neutral information about the impact of genetic disease and facilitating informed decision-making based on their patients' beliefs and values. Without sufficient disability training aimed at improving the knowledge, confidence, and competence necessary to support people with disabilities, genetic counseling students may feel unprepared or even uncomfortable having such discussions when they enter the profession. Prior studies have found that genetic counselors are more comfortable discussing the medical aspects of genetic disease, as opposed to social aspects and lived experiences, in part due to their graduate training. To our knowledge, there are no published qualitative studies exploring the extent and impact of disability training in genetic counseling programs. Through nine semi-structured interviews, this qualitative study explored prenatal genetic counselors' comfort level with, and preparedness for, addressing disability in practice, as well as examined the perceived adequacy and impact of their disability training. Using semantic inductive thematic analysis, we identified six themes: 1) experiences outside of genetic counseling training contribute to disability competency, 2) theoretical and experiential learning about disability outside of a medical context are critical for disability training, 3) limitations of disability training in genetic counseling programs, 4) disability training differences and inconsistencies between and within genetic counseling programs, 5)

challenges in discussing disability with patients, and 6) variability in genetic counseling approaches to and strategies for discussing disability with patients. The results captured by this study may be used to inform and improve disability curricula within genetic counseling programs. Disability training that incorporates the diverse perspectives of individuals with disabilities is necessary for genetic counseling curricula and the promotion of public health.

## Table of Contents

<b>Acknowledgements .....</b>	<b>x</b>
<b>Preface.....</b>	<b>xii</b>
<b>1.0 Introduction.....</b>	<b>1</b>
<b>1.1 Specific Aims.....</b>	<b>3</b>
<b>2.0 Manuscript.....</b>	<b>4</b>
<b>2.1 Background.....</b>	<b>4</b>
<b>2.2 Methods .....</b>	<b>9</b>
<b>2.2.1 Study Design .....</b>	<b>9</b>
<b>2.2.2 Participants and Recruitment.....</b>	<b>10</b>
<b>2.2.3 Interviews.....</b>	<b>11</b>
<b>2.2.4 Data Analysis .....</b>	<b>13</b>
<b>2.3 Results.....</b>	<b>14</b>
<b>2.3.1 Participant Demographics.....</b>	<b>14</b>
<b>2.3.2 Identified Themes.....</b>	<b>15</b>
<b>2.3.2.1 Experiences Outside of Genetic Counseling Training Contribute to Disability Competency.....</b>	<b>17</b>
<b>2.3.2.2 Theoretical and Experiential Learning about Disability Outside of a Medical Context are Critical for Disability Training.....</b>	<b>20</b>
<b>2.3.2.3 Limitations of Disability Training in Genetic Counseling Programs</b>	<b>22</b>
<b>2.3.2.4 Disability Training Differences and Inconsistencies Between and Within Genetic Counseling Programs .....</b>	<b>23</b>

2.3.2.5 Challenges in Discussing Disability with Patients.....	26
2.3.2.6 Variability in Genetic Counseling Approaches to and Strategies for Discussing Disability with Patients.....	27
2.4 Discussion .....	29
2.4.1 Experiences Outside of Genetic Counseling Training Contribute to Disability Competency .....	29
2.4.2 Theoretical and Experiential Learning About Disability Outside of a Medical Context are Critical for Disability Training.....	32
2.4.3 Limitations of Disability Training in Genetic Counseling Programs.....	35
2.4.4 Disability Training Differences and Inconsistencies Between and Within Genetic Counseling Programs.....	37
2.4.5 Challenges in Discussing Disability with Patients .....	40
2.4.6 Variability in Genetic Counseling Approaches to and Strategies for Discussing Disability with Patients.....	42
2.4.7 Study Limitations .....	44
2.4.7.1 Self-Selection Bias .....	44
2.4.7.2 Theoretical Saturation.....	45
2.5 Conclusion .....	46
2.6 Research Significance to Genetic Counseling and Public Health .....	49
2.6.1 Research Significance to Genetic Counseling.....	49
2.6.2 Research Significance to Public Health .....	50
Appendix A IRB Approval.....	52
Appendix A.1 IRB Modification .....	53

<b>Appendix B Recruitment Flyer.....</b>	<b>54</b>
<b>Appendix C Study Introduction .....</b>	<b>55</b>
<b>Appendix D Semi-Structured Interview Guide.....</b>	<b>56</b>
<b>Bibliography .....</b>	<b>59</b>



## List of Tables

<b>Table 1. Participant Demographic Information .....</b>	<b>15</b>
<b>Table 2. Identified Themes.....</b>	<b>16</b>

## Acknowledgements

First and foremost, I would like to express my gratitude to the nine genetic counselors that took part in this study. I deeply appreciate your willingness to share your experiences and viewpoints with me. This study would not have been possible without you.

I also would like to thank the members of my thesis committee. My thesis advisor, Robin E. Grubs, PhD, MS, LCGC, was instrumental in the development of this project. Thank you for enabling me to work through challenges so that I could learn and grow, while also providing me with guidance when I came to a standstill. I would not be here without your advice and support. Thank you to Michael J. Deem, PhD for inspiring this work. Our discussions about autonomy, nondirectiveness, and the expressivist objection have fundamentally changed the lens through which I view genetic counseling, and I will be a more thoughtful genetic counselor thanks to you. Thank you to Amy Houtrow, MD, PhD, MPH for making my LEND journey possible. My growth as a leader, communicator, interdisciplinary team member, community partner, and advocate are due to my LEND training, which I will take with me wherever I go. Thank you for supporting LEND trainees so that we may, in turn, support children and families in the ways they need.

Thank you to my program advisor, Jodie Vento, MGC, LCGC for your unwavering support and guidance. You have always seen the best in me, even when I can't see it in myself. Thank you to Meredith Jones, MS, LCGC for piloting our interview guide and providing invaluable feedback towards its development, as well as supporting my professional growth as a future genetic counselor. Lastly, thank you to each and every clinical supervisor at University of Pittsburgh Medical Center (UPMC) Magee-Women's Hospital, UPMC Children's Hospital of

Pittsburgh, UPMC Primary Care Precision Medicine Clinic, and Allegheny Health Network (AHN) for providing me with the tools necessary to be the best genetic counselor I can be.

## Preface

Language has the ability to both empower and stigmatize. We recognize that terminology related to disability is not merely a matter of semantics – the ways in which we communicate about disability have a profound impact on how individuals and communities are perceived and treated. Throughout this manuscript, we aimed to be thoughtful, respectful, and inclusive in our language when communicating about disability. In so doing, we felt it was important to acknowledge the diverse perspectives regarding language use around disability – particularly, person-first language and identity-first language.

Person-first language (for example, “people with Down syndrome” or “individuals with disabilities”) is a linguistic prescription that places the person before a disability, disease, or condition. This type of language aims to describe what a person *has*, as opposed to who a person *is*. Individuals who do not feel that their disability, disease, or condition is an aspect of their identity may prefer person-first language, which instead emphasizes their “unique combination of strengths, needs, and experiences (both related and unrelated to their disability), and the acknowledgement that these might be different from those of others who happen to have the same diagnosis” (Vivanti, 2019, p. 691).

However, many people do not use person-first language when referring to themselves or others. Instead, they may use identity-first language (for example, “disabled,” “autistic,” or “D/deaf”). Advocates for identity-first language argue that these identities are essential aspects of who they are – one cannot separate themselves from these identities any more than they can separate themselves from other identities, such as race, ethnicity, gender, or sexuality. Further, some feel that person-first language further marginalizes disability – it has been argued that

person-first language communicates that disability is inherently negative and denies the significant role disability can play in shaping a person's life, identity, and community (Vivanti, 2019).

Ultimately, there is no one-size-fits-all approach to language use surrounding disability. Though this manuscript often, but not always, utilized person-first language, we recognize that the choice between person-first language and identity-first language is a matter of preference among individuals and communities themselves. Our use of person-first language and identity-first language throughout this manuscript are not meant to endorse one approach over the other, but rather, to respect the diverse viewpoints on this matter and those who hold them.

## 1.0 Introduction

*Disability* is a broad term that encompasses physical, cognitive, psychological, communicative, sensory, behavioral, and neurodevelopmental disabilities. The frameworks through which disability is conceptualized are typically referred to as models, with the most well-known being the medical model and the social model. The medical model views disability as an impairment of a body system or function that needs treatment, therapy, or correction. This model suggests that healthcare professionals are the experts on disability, as opposed to the individuals with disabilities themselves. In contrast, the social model argues that disability is socially constructed and arises when there is discordance between a person's needs and their environment; it is the environment, not the impairment, that creates barriers for the individual and results in disability (Olkin, 2002). It is important for healthcare professionals, including genetic counselors, to be aware of how disability is conceptualized by themselves and others, as viewing disability solely through a medicalized lens dismisses the diverse lived experiences of those with disabilities. In the words of disability rights advocate Emily Ladau, "there is no singular disability experience" (Ladau, 2021, p. 4).

Because genetic counselors often speak with patients about genetic conditions that may be associated with one or more disabilities, it is imperative that they understand not only how a genetic diagnosis can impact an individual beyond its medical implications, but also that the experiences of people with such conditions can vary. Genetic counselors can work to gain such an understanding and strengthen their disability competencies through disability training. Graduate training is an optimal time for genetic counseling students to learn from and about these lived

experiences – didactic education, clinical exposure, volunteer work, interdisciplinary training, and graduate research projects all have the potential to incorporate disability competencies.

Calls to formally incorporate disability training into healthcare professional education have resulted in the development of the *Core Competencies on Disability for Health Care Education*, which were established by the Alliance for Disability in Health Care Education in June 2019 (Havercamp et al., 2021). To date, these competencies have not been formally integrated into the Accreditation Council for Genetic Counseling (ACGC)'s practice-based competencies for genetic counselors (ACGC, 2023a).

Studies aimed at exploring the extent to which disability training is included in genetic counseling programs have found that, while disability training is incorporated, there is considerable variability in the type and amount of training offered (Sanborn and Patterson, 2014). More recently, Douglas et al. (2023) explored the impact of various types of disability training. They found that the most commonly reported disability training experiences were class lectures and assigned readings, despite these being the least valuable experiences for genetic counselors. The most impactful experiences were those that provided exposure to individuals with disabilities and insights into their everyday lives. This same study also explored genetic counselors' perceived comfort level with disability. The results revealed that genetic counselors are more comfortable discussing the medical aspects of disability as opposed to the social and lifestyle aspects (Douglas et al., 2023). This is consistent with prior studies (Farrelly et al., 2012; Madeo et al., 2011).

There are few, if any, published qualitative studies exploring how genetic counselors perceive the adequacy of their disability training and what factors contribute to their comfort level with discussing disability. This study utilized semi-structured interviews of prenatal genetic counselors who graduated from an ACGC-accredited genetic counseling program between 2019

and 2022. Participants were interviewed to learn more about the extent and impact of their disability training, their personal experiences with disability, what experiences prepared them to address topics related to disability in practice, and how comfortable they feel in doing so.

### **1.1 Specific Aims**

Specific Aim 1: To examine recently graduated prenatal genetic counselors' perceptions of the adequacy of their graduate training related to disability.

Specific Aim 2: To explore recently graduated prenatal genetic counselor's approach to, comfort level with, and preparedness for, addressing disability with patients and working with individuals with disabilities.



## **2.0 Manuscript**

### **2.1 Background**

Prenatal genetic counselors often engage in discussions related to disability. They may deliver diagnoses of conditions associated with disabilities, provide information about the medical and social realities of living with these conditions, or engage patients in the decision-making process with regard to pregnancy termination and family planning. Such discussions reveal a challenge in genetic counseling practice: genetic counselors ought to be advocates for the rights and interests of disabled patients while simultaneously offering prenatal genetic screening and testing, technologies which have been criticized as tools used to avoid the birth of children with disabilities (Madeo et al., 2011).

The manner in which genetic counselors present information about genetic disease is important because it influences patients' perceptions of their ability to cope with and care for a child with a genetic diagnosis. This, in turn, influences decisions regarding the management of the pregnancy (Lenihan, 2012). Farrelly et al. (2012) explored how prenatal genetic counselors delivered information related to disability. Overall, genetic counselors were found to focus more heavily on the medical aspects of genetic diagnoses associated with disability, such as physical and cognitive differences, than on the more social aspects of disability, such as social support and the ability to go to school, drive a car, or live independently. Genetic counselors largely characterized their role as "providing information," as opposed to facilitating informed decision-making. When genetic counselors did ask questions to elicit a patient's experiences, feelings, and

beliefs about disability, many did not acknowledge or explore the patient's response, opting instead to provide more information (Farrelly et al., 2012).

This focus on the medical aspects of disability has been characterized elsewhere in the literature. Madeo et al. reviewed multiple studies which found that genetic counseling students and genetic counselors tend to focus on medical aspects of disability as opposed to social aspects. While these aspects alone are not inaccurate, they are inadequate, as they do not fully represent the experiences of those with disabilities, who have been found to relate their quality of life to personal and social aspects more than medical aspects (Madeo et al., 2011).

In 2012, Hodgson and Weil explored the reasons why genetic counselors do not routinely discuss the broader social implications of living with genetic conditions resulting in disability. Genetic counselors in this study felt that their lack of existing counseling skills, limited understanding of what it is like to parent a child with a disability, fear of upsetting or overwhelming patients, and personal biases were barriers to discussing disability more deeply with patients. Some felt that they were not sufficiently trained or experienced to address issues related to disability in the prenatal setting (Hodgson and Weil, 2012).

With regard to barriers such as personal biases, genetic counseling students and genetic counselors have shown implicit bias against disability. While factors such as genetic counseling training, personal experience working with individuals with disabilities, and experience as a genetic counselor have not been shown to impact these implicit attitudes, appropriate training may prevent implicit biases from adversely affecting genetic counselors' ability to provide balanced, patient-centered, nondirective counseling (Gould et al., 2019). Subsequent studies have supported this conclusion. For example, a study by Vaimberg et al. (2021) found that genetic healthcare professionals and trainees across multiple disciplines, including medicine, nursing, and genetic

counseling, have shown implicit bias against individuals with physical disabilities. However, despite having *implicit* bias, those that had experience working with individuals with disabilities showed less *explicit* bias in their clinical recommendations and counseling. This underscores the value of working with individuals with disabilities during professional training (Vaimberg et al., 2021). In addition to improving the ability to provide unbiased counseling, experience working with individuals with disabilities may also lessen counselors' own discomfort and uncertainty with counseling about disability (Brasington, 2007).

Negative attitudes and misconceptions among healthcare providers regarding disability are significant barriers to care (Nocon and Sayce, 2008). Further, limited knowledge and understanding of disability among healthcare providers is a barrier to timely and effective coordination of healthcare services, resulting in inequitable experiences for those with disabilities (Kroll and Neri, 2003; Scheer et al., 2003). These barriers underscore the necessity of disability training for all healthcare providers, including genetic counselors. Disability training is believed to help healthcare providers see disability as a functional limitation that may or may not impact a person's quality of life, as well as an important part of a person's identity, as opposed to a negative health outcome that should be treated or prevented. Training in patient-centered care, in which the person with the disability plays a role in their own medical care and drives decision-making, is thought to improve the quality of their care (Bowen et al., 2020).

Despite calls for improved and expanded disability training within healthcare professional training programs, there have not been clear standards for what should be taught. In response, the Alliance for Disability in Health Care Education developed its *Core Competencies on Disability for Health Care Education* in 2019. These competencies include 1) acquire "a conceptual framework of disability in the context of human diversity, the lifespan, wellness, injury, and social

and cultural environments,” 2) demonstrate a “mastery of general principles of professionalism, communication, respect for patients, and recognizes optimal health and quality of life from the patient’s perspective,” 3) “understand and identify legal requirements for providing health care... to meet the individual needs of people with disabilities,” 4) “engage and collaborate with team members within and outside their own discipline to provide high-quality, interprofessional team-based health care to people with disabilities,” 5) “collect and interpret relevant information about the health and function of patients with disabilities and engage patients in creating a plan of care that includes essential and optimal services and supports,” and 6) “demonstrate knowledge of effective strategies to engage patients with disabilities in creating a coordinated plan of care with needed services and supports” (Havercamp et al., 2021, p. 5).

To date, these competencies have not been adopted by the Accreditation Council for Genetic Counseling (ACGC), the accrediting body for genetic counseling programs in the United States and Canada. ACGC has developed seven practice-based competencies for the profession to guide didactic and experiential training. Disability is briefly mentioned in the definition of the term *diverse* in Appendix 1 of the *Practice-Based Competencies for Genetic Counselors*. The term *diverse* is mentioned in competency 6a, in which genetic counselors are expected to be trained to “demonstrate how disparities, inequities, and systemic bias affect access to healthcare for diverse populations” (ACGC, 2023a). ACGC also sets standards for genetic counseling program accreditation, which are described in the *Standards of Accreditation for Graduate Programs in Genetic Counseling*. These standards list “awareness and appreciation of multiple forms of identity, including... disability” as a required component of a program’s diversity, equity, inclusion, and justice content area (ACGC, 2023b). However, these standards do not outline the

quantity and quality of disability training, leaving individual programs little guidance in developing their curriculum.

There are a limited number of studies exploring how disability competencies are included in genetic counseling training. A 2012 survey of ACGC-accredited genetic counseling program directors found that, while each program incorporated disability training into their curriculum, there was considerable variability in the type and amount of training offered. Most programs surveyed offered opportunities to attend a support group for individuals with disabilities (60%) and/or for family members and caretakers of individuals with disabilities (70%), a workshop on appropriate language surrounding disability (80%), a workshop with parents of a child with a disability (70%), and a workshop on the nature and history of disability (65%). The researchers suggested that the lack of standardized disability training is likely to result in a growing genetic counseling workforce that has varying awareness of disability issues and, as a result, will vary in their ability to navigate complex topics related to disability (Sanborn and Patterson, 2014).

More recently, a 2023 survey of genetic counselors found that the majority reported receiving adequate disability training while in graduate school. However, while 89% reported feeling comfortable discussing the *medical* aspects of disability, 65% reported feeling comfortable discussing the *social and lifestyle* aspects of disability. The most impactful educational experiences were those that provided exposure to individuals with disabilities and insights into their everyday lives. These included speaking to individuals with disabilities, speaking with family members or caregivers, visiting specialized education facilities, and visiting community centers for people with disabilities. The least impactful educational experiences were class lectures and assigned readings, which were paradoxically the experiences that were most consistently offered across programs. In the same study, members of the disability community who received genetic counseling reported

that their genetic counselor was knowledgeable about the medical aspects of disability, but less knowledgeable about the social and lifestyle aspects of disability, if it even came up. Together, this suggests that disability training does not fully meet the needs of practicing genetic counselors (Douglas et al., 2023).

There are few, if any, published qualitative studies exploring how prenatal genetic counselors perceive the adequacy of their disability training and what factors contribute to their approach to, and comfort level with, discussing disability. This study utilized one-on-one, semi-structured interviews of prenatal genetic counselors who graduated between 2019 and 2022, as these individuals would be more likely to recall details of their graduate training than those who graduated less recently. The specific aims of this study were to 1) examine recently graduated prenatal genetic counselors' perceptions of the adequacy of their graduate training related to disability and 2) to explore recently graduated prenatal genetic counselor's approach to, comfort level with, and preparedness for, addressing disability with patients and working with individuals with disabilities. It is our hope that the perspectives gathered through this study may be used to help inform the development of disability training and curriculum within genetic counseling programs.

## **2.2 Methods**

### **2.2.1 Study Design**

This qualitative study utilized one-on-one semi-structured interviews to explore how graduate training and lived experiences contributed to recently graduated prenatal genetic

counselor's comfort level with, and preparedness for, addressing disability with patients and working with individuals with disabilities.

The Institutional Review Board (IRB) of the University of Pittsburgh reviewed this study and determined that it met the regulatory requirements for exempt research under 45 CFR 46.104(d) on September 7, 2023 (Appendix A). A modification to the IRB protocol was approved on September 22, 2023 (Appendix A.1).

## **2.2.2 Participants and Recruitment**

Eligible participants were prenatal genetic counselors working in patient-facing roles in the United States or Canada that graduated from an ACGC-accredited genetic counseling program between 2019 and 2022. Participants were excluded from this study if they worked full-time in specialty areas other than prenatal (e.g. oncology, pediatrics), did not work in the United States or Canada, or graduated from an ACGC-accredited genetic counseling program prior to 2019 or after 2022.

In order to facilitate recruitment, a flyer (Appendix B) was distributed to several email listservs, including the National Society of Genetic Counselors (NSGC) Research Survey listserv on September 27, 2023, October 25, 2023, and January 24, 2024. These NSGC Research Survey emails were then forwarded to members of the NSGC Prenatal Special Interest Group listserv in order to enhance visibility. The recruitment flyer was also distributed to the Genetic Counselor Educators Association (GCEA) listserv on September 11, 2023 and October 30, 2023. As part of this email distribution, ACGC-accredited genetic counseling program directors and/or program leaders were asked to distribute the flyer to alumni who graduated between the years 2019 and 2022. The recruitment flyer was posted to several social media platforms, including the University

of Pittsburgh Genetic Counseling Program’s Instagram account, as well as the Principal Investigator’s personal Instagram, Twitter, LinkedIn, and Reddit accounts. Lastly, this flyer was posted in the research recruitment channels of two community Discord servers: “GC Chat” and “GenePool: A Genetic Counseling Community.”

The recruitment flyer included a brief description of the study aims, inclusion criteria, and participation requirements. A QR code was also included on the flyer, which directed participants to a Qualtrics survey that was designed to confirm participants met inclusion criteria and gather participants’ contact information in order to arrange an interview.

Eleven participants completed the Qualtrics survey, with ten meeting inclusion criteria – one participant was excluded from this study because they were not a prenatal genetic counselor. Nine interviews were conducted via Zoom in total, as one participant did not respond to contact after completing the survey.

### **2.2.3 Interviews**

Following completion of the survey, eligible participants were contacted via their preferred mode of contact (as indicated in the survey) in order to arrange a date and time for an interview. The Principal Investigator provided participants with the option of sending their availability via email or choosing a date and time on NeedToMeet, an online meeting and appointment scheduler. Once the Principal Investigator finalized a date and time for each interview, a Zoom invitation was created and sent to each participant.

Participants were interviewed via Zoom between October 2023 and February 2024. The duration of interviews ranged from 50 minutes to 92 minutes. The average duration of interviews was 64 minutes. A verbal introduction to the study (Appendix C), which included an overview of



the study goals, protection of confidentiality, and study team contact information, was read to each participant. Each participant consented to participate in an interview and for the audio of the interview to be recorded.

A semi-structured interview guide was used in this study (Appendix D). This guide was initially developed by the Principal Investigator and then reviewed and revised by the study team. The Principal Investigator and one member of the study team piloted the interview with a prenatal genetic counselor not otherwise involved in the study in order to re-word and re-order questions, add clarifying prompts, and estimate the expected length of an interview. Following this pilot, the guide was refined and finalized by the study team. Interview questions included those relating to participants' day-to-day work as a genetic counselor, challenges faced in their job related to discussing disability, the types of disability training they received (including didactic, clinical, and other related training), their perspectives on their disability training, and their personal experiences with disability or individuals with disabilities. Interview questions also included those intended to gather demographic information, including the genetic counseling program they attended, the amount of time they have been working as a genetic counselor, the number of positions they have held since graduating, and the location in which they currently practice (location being defined as one of six NSGC regions within the US or Canada).

All participants consented to being re-contacted after their interview in case clarification was needed. Additional questions arose after one interview: this participant was recontacted via email to clarify those questions. Audio from each interview was recorded for the purposes of transcription. All audio recordings and transcripts were de-identified. No video recordings from the interviews were saved. Following each interview, audio recordings were transcribed via an

automated transcription service (Otter.ai) and reviewed to ensure accurate transcription and de-identification. Once transcription was complete, audio recordings were destroyed.

#### **2.2.4 Data Analysis**

Each transcript was uploaded to the software program MAXQDA 2022, which was utilized for code annotation, organization, comparison, and review. Semantic inductive thematic analysis, the phases of which are described by Braun and Clarke (Braun and Clark, 2006), was carried out by the Principal Investigator and study team. The steps are outlined as follows:

1. **Familiarize yourself with the data:** Transcribe data (if necessary), read, and re-read the data, noting down initial ideas.
2. **Generate initial codes:** Code interesting features of the data in a systematic fashion across the entire data set, collating data relevant to each code.
3. **Search for themes:** Collate codes into potential themes, gathering all data relevant to each potential theme.
4. **Review themes:** Check if the themes work in relation to the coded extract (Level 1) and the entire data set (Level 2), generating a thematic ‘map’ of the analysis.
5. **Define and name themes:** Ongoing analysis to refine the specifics of each theme, and the overall story the analysis tells, generating clear definitions and names for each theme.
6. **Produce the report:** Select vivid, compelling extract examples, final analysis of selected extracts, relating back of the analysis to the research question and literature, producing a scholarly report of the analysis.

*Semantic* thematic analysis involves identifying themes within the explicit or surface meanings of the data. Data are organized to show patterns in semantic content, and the significance

of these patterns and their broader meanings and implications are theorized. Semantic thematic analysis differs from a *latent* approach, which identifies the underlying ideas, assumptions, and conceptualizations and interprets them (Braun and Clark, 2006).

Initial, line-by-line codes for each interview were manually generated by the Principal Investigator and reviewed by one member of the study team. Discrepancies in codes were resolved. A codebook was generated to define each of these codes, and memos were created for each interview to aid in the process of coding. These codes were then collated into potential themes – each theme was reviewed to ensure it adequately captured its associated coded extracts and was distinct from other codes. Preliminary themes were analyzed, and this analysis informed the creation of additional interview questions to ask subsequent participants. Themes, subthemes, and associated codes were refined by the study team.

## **2.3 Results**

### **2.3.1 Participant Demographics**

Nine prenatal genetic counselors were interviewed for this study. Demographic information about the participants is listed in Table 1. The nine participants attended nine different ACGC-accredited genetic counseling programs. Due to small cohort size, the names of these programs have been excluded from Table 1 in order to maintain de-identification of all participants.

**Table 1. Participant Demographic Information**

<b>Participant ID</b>	<b>Year of Graduation</b>	<b>Length of Time as a Genetic Counselor</b>	<b>Positions Held Since Graduating</b>	<b>Current Region*</b>
001	2022	1.5 years	1	2
002	2022	1.5 years	1	2
003	2019	4.5 years	3	5
004	2022	1.5 years	1	4
005	2021	2.5 years	1	5
006	2022	1.5 years	1	4
007	2020	3.5 years	1	2
008	2019	4.75 years	2	3
009	2022	1 year	1	6

\*Region 1: CT, MA, ME, NH, RI, VT, CN, Maritime Provinces. Region 2: DC, DE, MD, NJ, NY, PA, VA, WV, PR, VI, Quebec. Region 3: AL, FL, GA, KY, LA, MS, NC, SC, TN. Region 4: AR, IA, IL, IN, KS, MI, MN, MO, ND, NE, OH, OK, SD, WI, Ontario. Region 5: AZ, CO, MT, NM, TX, UT, WY, Alberta, Manitoba, Saskatchewan. Region 6: AK, CA, HI, ID, NV, OR, WA, British Columbia, Yukon.

### **2.3.2 Identified Themes**

A codebook comprised of 1,623 codes was developed. Through semantic inductive thematic analysis, we developed six themes: 1) experiences outside of genetic counseling training contribute to disability competency, 2) theoretical and experiential learning about disability outside of a medical context are critical for disability training, 3) limitations of disability training in genetic counseling programs, 4) disability training differences and inconsistencies between and within genetic counseling programs, 5) challenges in discussing disability with patients, and 6) variability in genetic counseling approaches to and strategies for discussing disability with patients. These themes are summarized in Table 2.

**Table 2. Identified Themes**

<b>Theme</b>	<b>Description</b>
<b>Experiences outside of genetic counseling training contribute to disability competency.</b>	Participants described their experiences outside of their graduate training, particularly personal experiences with disability, work or volunteer experience prior to their graduate training, and work experience after their training, that contributed to their disability awareness and competency.
<b>Theoretical and experiential learning about disability outside of a medical context are critical for disability training.</b>	Participants reflected on how disability training was incorporated into their graduate training and which types of experiences contributed the most to their comfort with disability and disability competency as practicing prenatal genetic counselors.
<b>Limitations of disability training in genetic counseling programs.</b>	Participants shared that they would have liked additional disability training during graduate school. Some participants felt their training did not fully prepare them for every patient encounter, including disability-related training. However, they did acknowledge potential challenges programs face in incorporating more disability training.
<b>Disability training differences and inconsistencies between and within genetic counseling programs.</b>	Participants discussed a wide variety of disability training offered by their respective programs. Students within the same program also experienced differences in their disability training, leading to variability in disability competencies even within a single cohort.
<b>Challenges in discussing disability with patients.</b>	Participants face challenges when communicating the mental, physical, developmental, and social impacts of genetic conditions associated with disabilities.
<b>Variability in genetic counseling approaches to and strategies for discussing disability with patients.</b>	Participants described similarities and differences in how they counsel about disability, including the quality, quantity, and timing of information conveyed to patients.

### **2.3.2.1 Experiences Outside of Genetic Counseling Training Contribute to Disability**

#### **Competency**

Participants commented on experiences outside of their graduate training that they felt contributed to their disability competency and comfort with discussing disability in practice. These included previous work and volunteer experiences, personal experience with disability, personal relationships with individuals with disabilities, and work experience as a patient-facing prenatal genetic counselor.

#### Previous Work And Volunteer Experiences

Three participants had experience working or volunteering with individuals with disabilities in a professional context prior to graduate school. When asked how they felt their graduate training influenced how they talk about disability in practice, one participant who worked as a services coordinator for people with developmental disabilities felt their prior work experience had a stronger impact:

Honestly, I don't know that they really influenced it other than giving me the medical background... And I think a large part of that is that I have a strong background in disability advocacy. And I was very, I feel I was much more comfortable with disability before school than a lot of my classmates. (Participant 007)

Another participant expressed a similar sentiment regarding their prior volunteer work:

I honestly think more of my comfort and exposure to [disability] has come prior to grad school... I was very involved in different organizations like the Muscular Dystrophy Association. (Participant 009)

#### Personal Experience with Disability and Relationships with Individuals with Disabilities

Personal experiences with disability, as well as personal relationships with individuals with disabilities, promoted a greater sense of ease in working with individuals with disabilities and talking about disability with patients. One participant stated they sometimes identify as having a disability due to their anxiety. They felt that their personal experience:

helps [them] connect a lot with [their] patients that [they're] counseling, especially if they have anxiety, or just any sort of neurodivergent situation. You know, everyone's a little bit different. But having an understanding of or finding that the way that your brain works isn't necessarily the same as everybody else's. Whether they have a difference that's similar to the way mine is, is a connection point. (Participant 008)

The same participant also reflected on their friendship with an individual with a disability and the impact of that relationship:

My best friend has a physical disability... seeing her experience has, I think, passively, but also kind of actively, shaped my understanding of what it means to be disabled in the world... I think it probably super shapes my perception of what it means to be disabled. And probably a little bit how I counsel about it. (Participant 008)

Having a relationship with an individual with a disability influenced another participant's comfort level with individuals with disabilities during their rotations: They shared:

When I was doing more of my pediatric rotations in school, I had more comfort being in the room and engaging with people with intellectual disabilities than some of my classmates who might not have known anyone personally or have that experience I did. (Participant 005)

Participants also acknowledge the limitations of personal experiences and relationships.

One participant commented that tapping into such experiences during counseling runs the risk of countertransference, which can have a detrimental effect on the counselor-client relationship. Another shared that their clinical supervisor's ability to provide balanced information was adversely impacted by her personal relationship with a neighbor's child who had Down syndrome: because this child had significant behavioral challenges that complicated their family's ability to arrange care, the participant felt that the supervisor tended to focus on negative aspects of the condition when counseling patients.

One participant that did not have such personal experiences or relationships reflected on how this impacted their comfort level with disability:

I don't identify as having any disabilities. I truthfully do not have a lot of experience with disability, and I think that that's why it was something that was like, kind of scary. (Participant 002)

## Work Experiences as a Patient-Facing Prenatal Genetic Counselor

Participants felt that their experience as a patient-facing prenatal genetic counselor was a factor in their overall comfort level with discussing disability or counseling patients with disabilities. One participant felt that work experience had a larger impact on their approach to discussing disability than their graduate training:

Most of my approach has just been built off the year of experience that I have... I don't think our prenatal class instructor really even tied in disability and how to talk about that. (Participant 004)

Another participant expressed that work experience was the most significant influence in their counseling approach. They stated:

Just having the experience of being able to listen to my patients and hear how they handle diagnoses... I think has informed my practice a lot more than any sort of personal experience. (Participant 006)

Work experience also improved one participant's comfort with disclosing a diagnosis and discussing disability. They shared:

I've definitely gotten more and more comfortable with it. I think the other thing is, over time, I've seen how patients respond to talking about these conditions... I've met more patients who have family members with these conditions... I've gotten more experience with patients modeling how they talk about their own conditions. And that's made me feel more comfortable with addressing it and building off that. (Participant 007)

Some participants rarely see patients with genetic conditions themselves, and this lack of experience impacts their comfort with counseling these patients: As one participant mentioned:

I just find myself more nervous... because I think I have less practice... I think I'm more worried about saying the wrong thing because it is so personal to that person who has the condition themselves. (Participant 005)

Some participants feel discomfort counseling patients with intellectual disability when compared to other types of disabilities. One participant noted:

Intellectual disability is probably the toughest one for me. I feel like I do have a lot of trouble bringing the information down to a level where I feel like the patient can understand what's going on. (Participant 001)



### **2.3.2.2 Theoretical and Experiential Learning about Disability Outside of a Medical Context are Critical for Disability Training**

Participants reflected on the graduate-level education, training, and experiences that contributed to their disability competency. Participants noted that graduate training is the time for students to build a disability competency foundation. This is, in part, because not all students have experience with individuals with disabilities prior to their graduate training.

Every participant described that their most impactful graduate experiences were speaking to people with genetic conditions and their families and learning about their lived experiences, including their experiences with receiving a diagnosis, day-to-day lives, feelings of dismissal by providers, and self-advocacy. These graduate experiences included guest or panel speakers, clinical rotations, and volunteer work, particularly with disability advocacy organizations or support groups for those with genetic conditions. One participant stated:

The best way to learn about a genetic condition is from the person who has it. The medical journals and all of that can be really helpful, But, actually hearing it from a person... can also be really helpful. (Participant 002)

Several participants said that their most impactful clinical exposure to disability occurred in the pediatric setting. Upon reflecting on their pediatric rotation, where they had the opportunity to interact with children and people with disabilities, one participant said:

I don't know that I was very comfortable working with people with disabilities until that point in my training. (Participant 004)

Another participant noted that observing other providers in this setting provided them with examples of how to effectively work with these patients:

Hearing some of those conversations where it is babies and kids with disabilities and how doctors are working with them...I think is really helpful. (Participant 006)

Participants also described their experiences observing clinical supervisors model how to discuss disability. One participant stated:

I had some GCs who were my supervisors who did a really fantastic job of talking about disability... they just really presented that balanced perspective and I think that's why I tried to model my counseling off of that. (Participant 004)

Others recalled negative examples of supervisors modeling how to talk about disability and reflected on their experience. One participant observed a geneticist present a rather medicalized and negative picture of a genetic condition to a patient. This experience led the participant to be more conscious of the way they discuss genetic diagnoses.

With regard to coursework, some participants explicitly stated that information about disability and the lived/social aspects of genetic conditions were woven into the curriculum. One participant mentioned:

You can't separate out the condition and the disability... I think entwining it into conversations about the conditions was a good approach. Because they really are part of one another, in a lot of ways. (Participant 007)

However, most participants felt that their program placed a larger emphasis on the medical aspects of genetic conditions (physical features, symptoms, testing modalities, inheritance, and treatment) in their didactic training. One participant shared why learning about genetic conditions beyond the medical aspects is critical:

All [the families] see is their happy, healthy child in front of them. Where we see a list of problems. That really puts things in perspective for me... ultimately, they have to take their child's development day by day, and that child will show them what they need. (Participant 003)

This sentiment was echoed by another participant:

I think it's just important to be introduced to views that are not your own. I think you're in such a silo. There's a diversity of views within genetic counseling, but not the full range...seeing it from a view that's more socially focused, or... seeing it as something along a spectrum of diversity... I think you just don't hear those things when you're in biomedical training. (Participant 005)

### 2.3.2.3 Limitations of Disability Training in Genetic Counseling Programs

Participants commented on deficits in their disability training while in graduate school. Although every participant's training included learning about the unique perspectives of people with disabilities, some would have liked to have been exposed to a wider variety of perspectives. One participant stated,

It could have probably been more comprehensive in terms of having more targeted experiences, rather than guest speakers here and there. I feel like we talked a lot about disability and how to talk about it with people, but we didn't actually hear from a lot of different people with disabilities... We heard from the instructors. (Participant 003)

Another participant echoed this when asked what specific experiences would have enhanced their disability training:

I think more activities or simulations to provide some of those maybe more rare circumstances... I had one prenatal rotation and didn't get any opportunities to talk about disability in that context... maybe having a standardized patient, or some sort of activity or role play. (Participant 004)

Several participants commented that they wanted more opportunities for continued care or long-term follow-up with a patient that had a genetic diagnosis so that they could learn more about their life with a disability. One participant wished they had received more tangible training on how to communicate with individuals with intellectual disability: They noted:

I wish we had maybe spent a little bit more time on some of the different ways that we can communicate with individuals that have learning disability... we did that, but I think we did it at a higher level than what people really, really need. (Participant 001)

Participants acknowledged that genetic counseling training cannot cover everything a genetic counselor needs to know for practice. One participant stated:

I think it's hard because you only learn like 5% of what you need to know about genetic counseling and actually being a genetic counselor in grad school. And so, I think during grad school, I felt like everything was really great and I did feel really good about my disability education... But you don't know really what it's going to be like until you start practicing. (Participant 002)

#### **2.3.2.4 Disability Training Differences and Inconsistencies Between and Within Genetic Counseling Programs**

All nine participants in this study attended different ACGC-accredited genetic counseling programs. Participants commented on both the quantity and quality of their disability training, which differed across programs. One participant felt strongly that their program was dedicated to incorporating disabled perspectives and disability competency into all areas of their graduate training. When discussing disability training, they stated:

I can't speak enough to the importance of having that foundation of understanding disability and recognition that it looks different for everybody, in that it looks different than your experience of talking about disability as a baseline for genetic counseling, as well as from your own perspective about disability, which is something that people may or may not have experienced themselves. (Participant 008)

Conversely, multiple participants felt that their program did not incorporate disability training nearly as much as they wanted or needed. In reflecting on their training, multiple participants felt that their disability training could have been more extensive. One commented on the dearth of disabled perspectives shared during their training:

I wish that my program would have done more on the disability perspectives. We had that a little bit. So, we had a journal club type of class, and each semester had a different theme. And one of the themes was about that. But I just wished that there would have been more didactic training because I don't know that I necessarily felt prepared. (Participant 004)

In addition to the differences in disability training between programs, participants commented on the variability within their individual programs. One of the most impactful yet least tangible aspects of training was learning from one's peers. Multiple participants talked about peer learning as a means of learning about disability. One participant talked about having a classmate who had a disability and whose thesis project focused on disabled individuals' views on genetic testing. They shared:

I think just learning from both the design of her project and her findings in that project, as well as what she contributed in class, or the views she'd share, I think that was really

valuable. Which is just complete luck that that's someone who was in the program around the same time as I was. (Participant 005)

Peer learning also amplified the formal disability training participants received from their programs. A participant whose disability training included attending support groups for individuals or families with genetic conditions noted:

Students talk. So, I got the benefit of my support group, but then I got to hear about the other support groups. (Participant 007)

Participants discussed opportunities for disability training that were optional, not available to all students, or occurred by chance. Two participants commented on their thesis projects, for which they chose to focus on genetic conditions that can cause a person to experience disability. One of these participants, whose thesis project focused on Retinitis Pigmentosa, learned how to work with individuals with vision loss more effectively:

The mentor that I worked with for that thesis research had a good amount of experience working with folks that also had vision loss. And she gave me, I think, some wonderful tips for working with folks that have vision loss. You know, she was like, if you try to contact one of your study participants, and they don't pick up right away, it's probably just that they couldn't get to their phone in time. So just like, wait a minute, give them a call back, instead of leaving a voicemail. She had a lot of really insightful tips for working with folks of that population, that I don't think I ever really would have thought of. (Participant 001)

Multiple participants discussed ethics courses offered by their respective programs. One participant had a personal interest in ethics and elected to take an optional ethics course. When reflecting on the impact of their didactic disability training, they shared:

The largest part was really probably through the optional class that I took. And I was the only person in my cohort who took that class. (Participant 001)

Another participant's program offered a mandatory ethics course, but the curriculum was based on student interest, which did not necessarily include topics related to disability. They talked about how each student was able to choose an ethics topic that was of personal interest to them, rather than learn from a standardized curriculum set by their program.

Participants also commented on volunteer work within their programs. For those whose programs required volunteer work, several mentioned that students could choose where they dedicated their time. Some participants chose to volunteer with disability advocacy organizations such as the Down Syndrome Congress. Most participants communicated that their programs didn't require volunteer work or outside opportunities related to disability. One mentioned:

I felt like it was more like, this is something you should consider doing, find your own opportunities. And it could have been more closely incorporated in our training. I feel like some people got more involved in that kind of stuff that others, because it was really dependent on how much somebody was prioritizing doing that. (Participant 003)

Even when volunteer work with individuals and families with a genetic condition were assigned by their program, participants noted that they did not get the most out of those experiences, depending on the families to whom they were assigned. As one participant shared:

Some of my classmates had a really, really great experience. Other classmates didn't. (Participant 006)

With regard to clinical training, multiple participants indicated that their clinical exposure to disability depended on what patients they were assigned during their rotations. One participant stated:

I think that some people, depending on what their rotations were, were probably more directly exposed to [disability] than others. (Participant 003)

Another participant noted that they were negatively impacted by such gaps in their clinical training:

You just don't get to practice those skills as much. So, I feel like my personal ability to practice things and talk about disability, and maybe see people who are disabled and have me be the one talking to them rather than me observing. I think that was lacking. (Participant 002)

### 2.3.2.5 Challenges in Discussing Disability with Patients

Every participant acknowledged that there are challenges associated with communicating about the variability of features and effects of a genetic diagnosis. Participants pointed out that no two people, even if they have the same diagnosis, will have the same experience. One participant stated:

We can't tell them how severe it's going to be. We can't tell them how significant intellectual disability is going to be...we see children born with Down syndrome all the time who have [ventricular septal defects] that weren't able to be identified on fetal echo. (Participant 003)

Even among family members, a genetic condition with the same underlying cause can present in different ways. Such challenges can be addressed, at least in part, by disability training. One participant noted:

Coming out of school I felt really, really prepared, and had, not a deep level of understanding, but perhaps a really good breadth of understanding of what disabilities can look like, and what it can mean in different ways to different families, and how people can react super differently. (Participant 008)

Participants also found the variability in patients' reactions to a diagnosis and values surrounding disability to be challenging. Multiple participants found that disability means something different to every patient, underscoring the importance of exploring their beliefs and values with them. One participant commented:

Even with the same type of condition and the same type of disability, everyone's going to have a different experience. And I think some of that comes from how the healthcare providers approach it and talk about it, which is a really important factor. Some of it comes from the family support, or whatever other supports they had in place in their school or in their workplace, just in their social circles. (Participant 009)

### **2.3.2.6 Variability in Genetic Counseling Approaches to and Strategies for Discussing**

#### **Disability with Patients**

When discussing their disability training, participants talked about approaches and strategies they learned, and continue to use, when speaking with patients about disability. For example, participants mentioned using neutral language, avoiding value-laden words like “good” and “bad,” mirroring patient language, assessing patients’ information needs, and exploring their values and experiences regarding disease, disability, and pregnancy termination. One participant pointed out that this type of approach allows patients the opportunity to consider the possible impact of a genetic diagnosis on their family, including whether or not such a diagnosis would result in disability for their child and how this would impact their lives. Another participant discussed the importance of language use when talking to parents and families:

It matters how you talk about these things, especially with parents who are like, well, this is my child, I love my child, stop trying to tell me that something's wrong with my child or stop trying to tell me that like, everything is bad. (Participant 002)

In an effort to meet the information needs of families, some participants discussed that they do not always provide detailed information about a possible genetic diagnosis. In talking about Down syndrome with patients in the pre-test setting, one participant stated:

I oftentimes actually don't talk about it in a whole lot of detail. I usually will check in to see if patients are familiar. And if they say yes, I don't really go into a ton of detail. (Participant 006).

Another participant discussed that they do not always provide parents with balanced information about genetic conditions such as Down syndrome because they don’t view every aspect of this condition as influential in parents’ decision-making process:

They don't need me to tell them how happy children with Down syndrome can be. They need me to tell them, like, these are potential problems that these children can or may encounter. (Participant 003)



Participants also differed in the timing of the delivery of this information. For example, some provide the most information about a genetic condition during pre-test counseling, whereas other participants provide this information after a test has returned a positive result. Participants suggested that they strive to balance the need to provide relevant information for the purposes of informed decision-making with their patients' preferences. Several participants expressed that pre-test counseling sessions are the ideal time to discuss genetic conditions, disability, and patients' values. One stated:

In the in the pretest, it's all so hypothetical. I mean, they're there in the moment, they have the pregnancy, they have this decision in front of them. But you're still thinking about, 'Okay, what if this comes back positive? What if it comes back with this type of syndrome'... I think people are a little bit in that space, better able to express their values, get into a slightly more complex or deeper conversation. In the post-test, you've sort of just dropped a bomb, you know, you're giving the result and you're immediately discussing it. So, I find in those post-test conversations, it's a lot more rare that I'm able to have that sort of values-based or more complex conversation. People just really aren't biting at that as much or they aren't engaging that often. The questions at that point are very logistical for people. (Participant 005)

Other participants mentioned that they take a different approach by discussing genetic conditions and their potential impact on their patients during post-test counseling, once a diagnosis is delivered. For some, this is because they may be testing for many potential conditions, without a single diagnosis in mind:

I usually talk about disability very briefly in pretest, because usually, there's a significant amount of content that we are going over. (Participant 008)

For others, they may prefer to defer a conversation about a patients' values related to disability until after results have returned:

I also in the pre-test setting typically focus more on the pregnancy outcomes, rather than necessarily the long-term outcomes. I really focus on, if this result comes back positive for this condition, how might your pregnancy look different? Versus, how might your life look different? Because I don't necessarily know how what their long-term goals are. So, I really can focus on the immediate, and then, you know, based on those results, we can have a further conversation about further goals and how this result fits into what they're wanting for the pregnancy and things like that. (Participant 006)

## **2.4 Discussion**

Using semantic inductive thematic analysis, we identified six themes: 1) experiences outside of genetic counseling training contribute to disability competency, 2) theoretical and experiential learning about disability outside of a medical context are critical for disability training, 3) limitations of disability training in genetic counseling programs, 4) disability training differences and inconsistencies between and within genetic counseling programs, 5) challenges in discussing disability with patients, and 6) variability in genetic counseling approaches to and strategies for discussing disability with patients.

### **2.4.1 Experiences Outside of Genetic Counseling Training Contribute to Disability**

#### **Competency**

Multiple participants commented that their personal and professional experiences with individuals with disabilities prior to their graduate training improved their competency and comfort with addressing disability in practice. For some, these experiences had a greater impact than their graduate-level disability training. One participant, who worked full-time as a direct support professional for a person with intellectual and physical disabilities and as a services coordinator for people with developmental disabilities, discussed how these experiences provided unique insights into their clients' daily lives. Through these roles, this participant gained a better understanding of how some people with disabilities interact with the world around them, complete activities of daily living, achieve educational goals through an Individualized Education Program, attain physical safety, and secure resources for themselves and their families. While one person's lived experience with disability does not necessarily reflect the lived experience of another person,

understanding the ways in which disability can fit into a person's life can help genetic counselors feel more comfortable broaching topics related to disability and having targeted discussions about how disability may or may not impact patients and their families.

Those that identified as having a disability, as well as those with personal relationships with family members and friends with disabilities, felt that these close relationships shaped their views on disability and what it is like to be disabled. Several participants described how people with disabilities are more similar to people without disabilities than they are different. Participants shared how this perspective enhances their ability to humanize individuals with disabilities, and empathize with patients who have disabilities. One participant, who identifies as disabled, stated that their personal experience increases their patience and compassion and allows them to better anticipate the needs of their neurodivergent patients. These findings are consistent with the results of a 2023 qualitative study that explored the experiences of recent genetic counseling graduates who identified as having a disability or chronic illness. Participants in this study reported that their own lived experiences with disability motivated them to provide high-quality, patient-centered care. These participants also felt that their experiences contributed to increased empathy and understanding when working with patients with disabilities (Epstein et al., 2023). In the same year, Darr et al. found that genetic counselors believe that their disability enhances their ability to empathize with patients (Darr et al., 2023) and a study by Winchester et al. revealed that exposure to individuals with disabilities provides students with a broader scope of knowledge, deeper understanding of their lived experience, increased knowledge of resources, and increased comfort level with disability (Winchester et al., 2023).

Participants that face challenges when working with people with disabilities note that this is, in part, because of their lack of experience. For one participant, their dearth of experience

interacting with people with intellectual disability makes counseling more difficult because they are less sure of how to adjust their language and communication style to meet the needs of this population. In contrast, one participant who had a close family friend with intellectual disability felt more comfortable engaging with patients with disabilities specifically because of their experience. Given the impact of such experiences, including disability-related work and volunteer experiences prior to graduate training, ACGC-accredited genetic counseling programs may consider recommending or requiring such professional experiences as a prerequisite for admission, as this is currently not required or even recommended by all programs.

Experience working with individuals with disabilities and broaching topics related to disability as a patient-facing prenatal genetic counselor also contributed to participants' comfort and competence. Participants discussed how their work experience contributed to increased flexibility in their counseling. This flexibility allows them to better recognize the needs of their patients so that they may provide tailored information, but also enables them to be a more active listener and source of support for patients and families. The impact of work experience on genetic counselors' comfort with discussing disability has previously been discussed in the literature. Brasington (2007) discussed how her initial, medicalized perceptions of Down syndrome shifted to be more patient- and family-centered over the course of her career (Brasington, 2007), underscoring the impact of genetic counseling work experience on disability related competencies.

## **2.4.2 Theoretical and Experiential Learning About Disability Outside of a Medical Context are Critical for Disability Training**

Participants commented that hearing directly from individuals with genetic conditions about their lived experiences with disability, as opposed to instructors who did not themselves have a genetic condition or experience disability, was the most impactful component of their disability training. These opportunities most commonly included guest speakers and panelists that spoke to participants' classes, but also included volunteer experiences with advocacy organizations and meeting one-on-one with a person with a genetic condition. When hearing from these individuals, participants received insight into their lives beyond the medical characteristics associated with their diagnosis.

Medical information about genetic conditions is foundational knowledge for genetic counselors – they must be able to understand and communicate aspects of genetic conditions, including features, symptoms, treatment, management, prognosis, inheritance, and genetic etiology. However, in order to better understand the lived experiences of those with a genetic condition and be able to communicate what living with such a condition could look like for another person, it is pivotal for genetic counselors to hear a diverse array of perspectives from those living with such conditions. Beyond knowing how a person can be impacted from a medical standpoint, genetic counselors also need to have a sense of the social and lifestyle impacts of a genetic condition, including supports needed at different stages of life, the ability to work, go to school, and navigate various systems effectively or independently, what advocacy and community look like, and how people secure affordable and attainable insurance coverage, benefits and services, and legal rights and protections.

Most participants reported that, while their graduate training did incorporate these social and lifestyle aspects, they primarily learned about the medical aspects of genetic conditions. This focus on communicating the medical impact of genetic disease has been described among genetic counselors and genetic counseling students previously (Douglas et al., 2023; Farrelly et al., 2012; Madeo et al., 2011; Winchester et al., 2023). Importantly, patients and disability advocates have expressed negative perceptions of this approach (Douglas et al., 2023; Madeo et al., 2011; Winchester et al., 2023), underscoring the need for genetic counseling programs to prioritize disability education beyond the medical aspects of genetic disease.

Surprisingly, clinical rotations had a lesser impact on some participants' disability-related comfort and competency than other disability training opportunities. Clinical rotations provide students with opportunities to interact directly with individuals who may have a genetic condition and, as a result, one or more disabilities; however, some participants did not feel that they had many of these opportunities during their graduate training. This may be due to the brief nature of individual rotation opportunities, variability in which patients are scheduled day-to-day, and a lack of long-term follow up care for patients during graduate school, which limit opportunities to establish relationships with patients and families. These opportunities are important components of disability training, as they have been shown to reduce the effect of implicit biases against disability. Vaimberg et al. (2021) examined the effect of implicit and explicit biases on genetic healthcare professionals' clinical recommendations for prenatal testing. Most participants in this study showed both implicit and explicit bias against individuals with physical disabilities, which contributed to biased clinical recommendations for disabled patients. Notably, those with experience working with individuals with disabilities had fewer biased clinical recommendations,

underscoring the importance of working with individuals with disabilities during professional training (Vaimberg et al., 2021).

Several participants did discuss instances when clinical supervisors modeled how to interact with individuals with disabilities. This most often occurred in the pediatric setting, where children and young adults are seen by a genetic counselor and a medical geneticist in order to investigate possible genetic etiologies for a person's clinical presentation or receive follow-up care after a diagnosis is made. Participants also discussed the importance of taking a patient- and family-centered approach in this setting: while the collection of detailed pregnancy, birth, developmental, and medical histories is important in narrowing down a list of differential diagnoses, providers must remember that they are working with families who view their child as a person, not simply a collection of symptoms and features. When providers modeled a patient-centered approach in the clinic, where patients were engaged in and in charge of their own care, participants noticed the impact it had on their patients and families. These, in turn, were impactful learning experiences for participants. Because individuals with disabilities face barriers to genetic healthcare services, including feelings of exclusion from the informed decision-making process, there have been calls for more respectful and inclusive genetic healthcare services in which educational resources are co-developed with individuals with disabilities (Strnadová et al., 2023). Overall, patient-centered counseling training, recognition of one's own implicit biases, and experience working with individuals with disabilities positively impacted genetic healthcare professionals' ability to provide patient-centered care (Vaimberg et al., 2021).

### 2.4.3 Limitations of Disability Training in Genetic Counseling Programs

While all participants' graduate training included hearing directly from individuals with genetic conditions, multiple participants mentioned wanting to hear more perspectives. In understanding that every person has a unique experience, participants felt it was important to hear a wide range of experiences, even from those with the same genetic condition. Winchester et al. (2023) discussed the importance of students having multiple opportunities to learn about disability so that their understanding of disability is not confined to a single representation (Winchester et al., 2023). Some participants acknowledged that this type of training is foundational for genetic counselors, especially for those who do not have experience with individuals with disabilities prior to graduate school – as previously discussed, genetic counseling programs do not routinely require that applicants have experience working with people with disabilities as a prerequisite for applications, meaning some students have never had such experiences.

Participants also commented that they wanted more targeted experiences to learn about disability. For some, these would have included role plays or standardized patient experiences where they could practice counseling individuals with or about disability without the risk of adversely impacting real patients. Other participants commented that they would have benefited from participating in an interdisciplinary clinic or engaging in long-term follow up care to better understand how a genetic diagnosis can impact a person over time. Understanding how a person adapts to a diagnosis and navigates various systems in order to identify and secure supports for themselves and their families would have been valuable. Such experiences also may have allowed participants to establish long-term relationships with patients, which can be difficult for students due to the brief nature of clinical rotations. The ability to engage in long-term follow up care aligns with one of the Alliance for Disability in Health Care Education's six *Core Competencies on*



*Disability for Health Care Education*: “Competency 6: Clinical Care Over the Lifespan and During Transitions.” Meeting this competency requires that learners “demonstrate knowledge of effective strategies to engage patients with disabilities in creating a coordinated plan of care with needed services and supports” (Havercamp et al., 2021, p. 5). To date, the *Core Competencies on Disability for Health Care Education* have not been formally integrated into ACGC’s Practice-Based Competencies for Genetic Counselors (ACGC, 2023a).

Disability advocates themselves have provided recommendations for effective disability education and training within genetic counseling programs. These include inviting advocates to speak about their experiences in classrooms, organizing interviews between students and advocates, implementing shadowing and buddy programs to expose students to the day-to-day experiences of advocates, having students attend advocacy organization conferences, and allowing students to explore different healthcare environments (including nursing homes, rehabilitation centers, and neonatal intensive care units) (Winchester et al., 2023). These recommendations align with the experiences participants felt were most impactful for their disability training.

An inherent limitation for genetic counseling training is the time with which programs have to train students. Training typically spans between 18 and 30 months, with most programs graduating genetic counselors after 21 months of training. Multiple participants commented that programs can’t possibly prepare students for every clinical scenario they may encounter. As our collective knowledge of genetics and the areas in which genetic counselors practice continue to expand, there will be more information for training programs to cover in the same amount of time (Murphy et al., 2023; Riconda et al., 2018; Waltman et al., 2016). As one participant discussed, this can result in programs de-prioritizing disability training. However, by overlooking the importance of disability training, training programs risk graduating genetic counselors who are ill-

equipped to tackle complex topics and scenarios related to disability (Sanborn and Patterson, 2014).

#### **2.4.4 Disability Training Differences and Inconsistencies Between and Within Genetic Counseling Programs**

Participants from nine different ACGC-accredited genetic counseling programs were interviewed for this study. While each program incorporated at least some form of disability training, they differed in the quantity, quality, timing, and consistency. Participants whose training included conversations about disability, early and often, felt that they had a valuable framework through which they could learn about genetic conditions. Their ability to consider the impact of genetic disease through a lens of disability, as well as to recognize the influence of other factors (e.g., race, ethnicity, culture, and socio-economic background), allowed them to have a richer understanding of the potential experiences of living with a genetic condition. As acknowledged by some participants, this emphasis on viewing genetic disease through a disability lens was not adopted by all participants' respective programs.

Participants also commented on disability training elements that are not within a program's control, leading to inconsistencies in disability training among students within a single program. One such element was peer learning, whereby participants learned about disability through discussions with their classmates. Those that had a personal interest in learning about disability, either through volunteer work or their thesis project, contributed to the learning of their classmates through formal and informal conversations about their experiences. Student's experiences prior to graduate school also contribute to peer learning. Genetic counseling programs often have broad admissions requirements, including crisis counseling and advocacy experience, which enable

students with diverse backgrounds and experiences to be equally competitive for admission. Participants that had prior personal and professional experience with individuals with disabilities shared what they learned from these experiences with their classmates, contributing to the disability training of their peers. Conversely, participants without such experiences who had classmates that did were able to hear and learn from them.

Participants that had classmates with disabilities themselves discussed how talking to these peers promoted their understanding of disability in ways they would not have otherwise had. This underscores the importance of diverse student cohorts, inclusive of students with disabilities and/or a background in disability advocacy, who can contribute to peer learning within and across cohorts. However, genetic counseling programs must be equipped to facilitate accommodations and mentorship for their disabled students while also taking steps to prevent them from experiencing discrimination, isolation, alienation, and tokenism, as these can adversely impact prospective and practicing genetic counselors (Darr et al., 2023).

Programs are less able to control clinical rotation opportunities for students, as these experiences are largely dependent on which patients are on a clinic's schedule. As previously discussed, pediatric rotations offered the most opportunities for students to learn about disability and interact with individuals with disabilities. However, if a student doesn't have a patient with a disability or doesn't work with a provider that models how to talk about disability, then they may not receive the same level of disability training as their peers. This, in part, contributes to a lack of standardized disability training for all students within a single program.

Even when programs aim to standardize disability training opportunities outside of the classroom and clinic, students may not have the same or similar opportunities to learn about people's lived experience with disability. In particular, participants discussed their volunteer

experiences during their graduate training. Multiple participants noted that, while volunteer work was required by their program, students were able to select the organizations with which they volunteered their time. Several participants chose to volunteer with organizations whose mission was related to disability awareness or advocacy. Meanwhile, some of their classmates chose to volunteer their time in other ways, including supporting prospective genetic counseling students or assisting with silent auctions for charity. When programs did require participants to volunteer with individuals and families with disabilities, participants reflected that they and their classmates did not always get the most out of these experiences. For some, this was because they were not sure what questions to ask to gain a deeper understanding of their experiences or the families with whom they were paired did not know what insights to offer. Another participant discussed that because students in their cohort were assigned to different families, those who were assigned to families with experience talking to students about their lived experiences benefited more than those who were assigned to families that were adjusting to a new diagnosis or did not know what elements of their lives to share. These also contribute to inconsistent disability training among students from the same program.

Several of these inconsistencies in graduate-level disability training can be addressed by genetic counseling programs. If prior experience working or volunteering with individuals with disabilities is not a prerequisite for genetic counseling program admission, students with minimal exposure prior to matriculation can receive supplementary graduate training tailored to improving their disability competencies. Further, programs that provide opportunities for students to speak with patients and families with genetic conditions can provide their students with a list of generative questions to ask so that they may gain a better understanding of their diagnostic experience, supports, strengths, challenges, and lived experiences. Similarly, they can also provide

patients and families with talking points aimed at helping students gain a better understanding of their experience with genetic disease and disability.

#### **2.4.5 Challenges in Discussing Disability with Patients**

In providing information about genetic disease to patients and families, prenatal genetic counselors aim to communicate what to expect from a genetic diagnosis, as such information is important for informed decision-making. However, participants commented that it is challenging to provide information about genetic conditions for which the features, symptoms, and outcomes exist on a spectrum. This challenge was described by Hodgson and Weil in 2012: through a series of interactive workshops, they found that prenatal genetic counselors experienced difficulty in providing information about Down syndrome because of the condition's variability in physical, intellectual, and developmental features (Hodgson and Weil, 2012). Multiple participants in our study used Down syndrome as an example when discussing this challenge, as individuals with this condition have varying degrees of intellectual disability, developmental delay, and medical complications. Parents themselves have recounted the experience of receiving a Down syndrome diagnosis on behalf of their child – providers have been found to discuss this condition through a medicalized lens, often providing too much information all at once and using unfamiliar medical terminology (Grane et al., 2023). Over time, practice guidelines for communicating a diagnosis of Down syndrome have been developed in order to improve experiences for parents and families. In discussing what families can expect from having a child with Down syndrome, genetic counselors can provide general expectations based on what most children with the condition experience, but such experiences can vary person-to-person (Sheets et al., 2011). For parents who want to understand how a genetic diagnosis will impact their child's life, and especially for those who may

choose to end a pregnancy because of this impact, this level of uncertainty can be difficult to navigate. In communicating such variability, genetic counselors have been found to be more comfortable discussing the medical aspects of genetic conditions as opposed to the social and lifestyle aspects (Douglas et al., 2023; Farrelly et al., 2012; Madeo et al., 2011). This focus on the medical aspects of genetic disease mirrors the medical focus of participants' graduate training.

Participants face additional challenges when counseling patients about a genetic condition that is present in other family members; these patients may understand how the condition impacts one family member, but not necessarily how it could have a different impact on their child. For some participants, this requires a fine balance between honoring the lived experience of families while simultaneously upholding their responsibility to deliver complete information about a genetic condition to families. In 2014, Boardman interviewed individuals with at least one person in their family (living or deceased) with a diagnosis of spinal muscular atrophy (SMA) to explore how their experiences with SMA influence their reproductive decision-making and attitudes towards having children with the genetic condition. As Boardman reported, multiple participants in this study “entirely align[ed] the identity of a (hypothetical) foetus diagnosed with SMA with their existing family member with SMA, so much so that they were often conceptualized as the same person” (Boardman, 2014, p. 21). Participants in our study found situations such as this challenging, as it can be difficult to convey to families that such conceptualizations may not be realistic. These challenges underscore the importance of hearing a wide variety of perspectives from individuals with genetic conditions, including different perspectives from people with the same genetic diagnosis, even within the same family.

Further, it is important that genetic counselors consider how their patients define disability, as disability can mean something different to everyone. One participant discussed how, for one

patient, needing extra help in school would be considered a disability, but for another patient, it may not. This emphasizes the importance of describing the impact of a condition beyond words such as *intellectual disability* or *developmental delay*, as these terms may not always have the same meaning to patients and families. People's experiences may be shaped by where they grew up and where they currently live, their family's and social circle's cultural beliefs about genetic disease and disability, and what support, community, and accommodations look like for people living with impairments and/or disabilities. Therefore, it is important for genetic counselors to explore what disability means to each and every patient.

## **2.4.6 Variability in Genetic Counseling Approaches to and Strategies for Discussing**

### **Disability with Patients**

Participants shared that their primary approach to genetic counseling is following the patient's lead, which allows them to employ tailored counseling strategies depending on the needs of their patients. Some of these strategies relate to the quality, quantity, and timing of the information they convey about genetic conditions. With regard to quality, many participants discussed how they use neutral, matter-of-fact language when discussing genetic disease in order to give patients the opportunity to express their reactions and values in an unbiased setting. In doing so, these participants enable patients to determine and communicate their informational needs. This approach exemplifies nondirectiveness, or the provision of balanced information in order to support autonomous decision-making, which has been described as a central tenet of genetic counseling (Uhlmann et al., 2009).

However, in following the patient's lead, some participants communicated that they do not always provide balanced or complete information about a genetic condition. Patients may express

that they are uninterested in learning more about a genetic condition, often because they feel that they already have the information they need, or they don't feel such information would change their decision regarding their pregnancy. When faced with these scenarios, participants differed in their approach. One participant stated they still provide complete information about a genetic condition because the patient may have incomplete or incorrect information, or because new information may still be important for their decision-making. Other participants discussed that, when a patient does not want additional information, they honor that request and move on. While the latter approach reflects participants' desire to maintain rapport with patients and meet their needs, avoiding the provision of balanced information about the impact of genetic disease may impede informed decision making (Farrelly et al., 2012).

Multiple participants noted that they provide more information about a genetic condition when there is a particular differential diagnosis in mind. Most often, this occurs when there is a prenatal screening result directing the conversation. Participants expressed that when they do not provide a lot of information about a genetic condition, it may be because they are screening or testing for many conditions that cannot be covered in great detail individually. This is particularly salient for expanded carrier screening, as it is not possible to provide in-depth pre-test counseling about each of the conditions included on expanded panels (Benn et al., 2014). This is also the case for certain diagnostic genetic tests, including chromosomal microarrays, which can return unexpected or uncertain results such as rare, *de novo* microdeletions/microduplications or variants of uncertain significance.

Participants also differed in the timing in which they provide information about genetic disease during a genetic counseling session. Some commented that they often deliver most of this information in the pre-test setting (before a person has had diagnostic genetic testing), while others



do so in the post-test setting (after results have been returned). Those that provide more information during pre-test counseling do so because their patients typically want the information at that time. Additionally, discussing this information in the pre-test setting allows patients to have “lead-time” to consider what decision they might make regarding their pregnancy based on their personal values, should their results come back positive. These participants also expressed that the post-test setting concentrates on decision-making because patients have made their decision and are not interested in discussing their values further once a diagnosis is delivered. Other participants provide more information about genetic disease once a diagnosis has been delivered because the results are known, allowing them to review more fully what these results mean for their patients.

Farrelly et al. (2012) suggested that prenatal genetic counselors should engage patients in in-depth discussions about genetic testing, possible results of such testing, disability, and what options are available to patients (pregnancy termination or pregnancy continuation followed by raising the child or adopting the child out) *before* deciding to undergo prenatal screening. Genetic counselors that have such discussions after a definitive diagnosis may do so in order to avoid increasing anxieties for patients. However, this approach eliminates the opportunity to provide anticipatory guidance for patients in the pre-test setting, which may ultimately impact the genetic counseling process (Farrelly et al., 2012).

## **2.4.7 Study Limitations**

### **2.4.7.1 Self-Selection Bias**

Self-selection bias is a potential limitation of this study. Participants were recruited through the use of a recruitment flyer, through which individuals could express interest in participating in the study and sign up for an interview. This allowed individuals to choose whether or not to

participate in this study. People who have a personal interest in disability may have been more likely to participate in this study than those who do not. These individuals may be more thoughtful about, or even critical of, their disability training when compared to those who did not choose to participate. In order to reduce such bias in future studies, it may be helpful to ask participants what made them interested in participating in the study. In this way, investigators can assess the degree to which personal interest drew them to participate.

#### **2.4.7.2 Theoretical Saturation**

Existing literature has defined theoretical saturation in a variety of ways. Saunders et al. (2018) suggest multiple models, including inductive thematic saturation. In this model, theoretical saturation is the point at which new codes or themes are no longer identified within the data, and/or new theoretical insights are no longer gained from the data (Saunders et al., 2018). While we developed novel codes from each interview, the preliminary themes generated throughout the data collection process did not substantially change following our ninth and final interview. However, because our final interview did result in some minor modifications to our themes, it is possible that theoretical saturation may not have been fully achieved. Additional interviews could be performed to assess the themes developed in this study and to further assess theoretical saturation.

Notably, conceptualizations of theoretical saturation have been critiqued in the literature. In 2019, Braun and Clarke provided a commentary on the usefulness of saturation in thematic analysis. They argue that meaning is generated through interpretation of the data, which will always yield new insights and, as a result, novel themes (Braun and Clarke, 2019). The concept of *theoretical sufficiency* was proposed by Dey as an alternative to theoretical saturation; sufficiency is the point at which the investigator(s) has reached a level of understanding adequate enough to build a theory (Dey, 1999). The themes developed in this study began to take form after the fourth

interview and were nearly fully developed after the seventh interview. While the last two interviews yielded novel codes, they did not substantially alter the nascent themes. Therefore, theoretical sufficiency was achieved in this study.

The concept of *information power* was developed as another alternative to saturation. Information power argues that the more information a sample holds, the fewer samples are needed. Factors that increase information power include a narrow study aim, specificity of participant experiences, pre-existing theory, high-quality dialogue between the interviewer and participants, and the chosen analysis strategy (for example, in-depth narrative analysis requires fewer participants than exploratory analysis) (Malterud et al., 2016). While this study has relatively broad study aims, we feel the specificity of participant experiences (all participants were recently graduated prenatal genetic counselors in the United States and Canada), high-quality dialogue, and in-depth analysis of the data contribute to information power in this study. However, future studies can increase information power by addressing these factors. For example, exploring the impact of a particular disability training experience, such as lectures from disabled guest speakers, may narrow study aims and subsequently necessitate fewer study participants in order to reach information power.

## **2.5 Conclusion**

This study aimed to examine recently graduated prenatal genetic counselors' perceptions of the adequacy of their graduate training related to disability and explore their approach to, comfort level with, and preparedness for addressing disability with patients and working with individuals with disabilities. The perspectives shared by the nine participants in this study provide

a unique insight into how genetic counselors perceive their disability training and what adjustments could be made to improve such training for future genetic counseling trainees.

Participants varied in their opinions and perceptions of their disability training: while some felt that they had robust and impactful training that prepared them to discuss disability as a patient-facing prenatal genetic counselor, others felt that their disability training was deficient or even deprioritized by their training program. Participants who felt they had robust disability training highlighted the components of their training that prepared them the most for practice. One such component was didactic curriculum that integrated the medical, social, and lifestyle factors that impact people with a variety of genetic conditions: these factors are not siloed for patients and therefore should not be siloed in genetic counseling education. Additionally, impactful disability training included opportunities to hear directly from individuals with genetic conditions and their families, including guest speakers, panelists, members of advocacy organizations, patients from clinical rotations, and people in the community who have a genetic diagnosis.

However, there still were limitations to these types of training: some participants felt their didactic training was *primarily* focused on the medical aspects of genetic disease. Additionally, most participants who heard directly from those with genetic conditions felt that did not hear a diverse enough array of perspectives; no two people will have the same experience, even with the same genetic diagnosis. Participants who expressed a desire for additional disability training proposed specific experiences they felt would have been impactful, including interacting with simulated patients with disabilities or having opportunities to discuss disability with a simulated patient, learning how to communicate with people with intellectual disability from someone with such expertise, working with people with disabilities in an interdisciplinary clinic, and participating in the long-term follow-up care of patients following a diagnosis.

Learning about the lived experiences and perspectives of people with disabilities increased participant's comfort and competency with disability. Given variability in disability training across and even within genetic counseling programs, learning experiences outside of graduate training can positively influence prenatal genetic counselors' comfort and competency with individuals with disabilities. The participants who felt the most comfortable broaching topics related to disability in practice had a combination of learning experiences related to disability, including those gained through personal, work, volunteer, and graduate experiences. Those with these experiences felt comfortable and prepared to address challenges faced during their day-to-day work as a prenatal genetic counselor.

The results of this study highlight the impact of consistent, high-quality disability training for genetic counselors. Genetic counseling programs that aim to graduate reflective, culturally competent genetic counselors must ensure that their didactic curriculum, clinical fieldwork, and volunteer experiences provide ample opportunity for students to learn the diverse perspectives of people with genetic conditions who experience disability. This requires program leaders to acknowledge when they themselves are not the expert and seek out the expertise of those with such lived expertise.

## **2.6 Research Significance to Genetic Counseling and Public Health**

### **2.6.1 Research Significance to Genetic Counseling**

This study has implications for the development of genetic counseling curricula. Although there have been calls to improve and expand disability training across healthcare professional training programs, genetic counseling programs have differed in their development and implementation of such training. As Sanborn and Patterson suggested ten years ago, these inconsistencies may result in a growing genetic counseling workforce that has varying awareness of disability issues and, as a result, will vary in their ability to navigate complex topics related to disability (Sanborn and Patterson, 2014). The results of this study are consistent with that claim: participants' graduate training contributed to differences in their understanding of, and ability to navigate discussions about, how genetic disease can impact patients and their families.

This underscores the importance of efforts to improve the quality, quantity, and consistency of disability training across genetic counseling programs. By better understanding the strengths and limitations of disability training, as perceived by those that receive this training and have entered the genetic counseling workforce, genetic counseling programs can improve their disability-related curriculum. As discussed by participants, impactful disability-related curriculum includes continued opportunities to learn about the diverse array of lived experiences of individuals with genetic conditions who experience disability. These opportunities include guest lectures and immersive volunteer and clinical experiences and should provide students with insights into people's experiences beyond their medical care.

Efforts aimed at improving curriculum can be made by not only program leaders, but also the Accreditation Council for Genetic Counseling (ACGC), which sets the standards for genetic

counseling curriculum development. ACGC can consider expanding their practice-based competencies to incorporate disability competencies as defined in the *Core Competencies on Disability for Health Care Education*. Through these improvements, individuals with disabilities stand to benefit from the increased understanding, empathy, and competency of their genetic counselors.

### **2.6.2 Research Significance to Public Health**

This study also has relevance to public health. The Public Health National Center for Innovations (PHNCI) established their 10 Essential Public Health Services, which provide a framework for providers “to protect and promote the health of people in all communities,” including those in the disability community. These services include “Communicate effectively to inform and educate people about health, factors that influence it, and how to improve it” and “Strengthen, support, and mobilize communities and partnerships to improve health.” (PHNCI, 2020).

The way genetic counselors frame disability when discussing genetic disease with their patients has important implications for public health. Genetic counselors receive training on how to communicate with a variety of populations and audiences, as well as educate patients and the public about how genetics can influence their health and the health of their family members. If genetic counselors communicate the medical impact of genetic disease without also discussing what support and social factors influence the experiences of individuals with these conditions, they are not adequately educating their patients.

Genetic counselors are responsible for advocating for the rights and interests of disabled patients while simultaneously offering prenatal genetic screening and testing, tools which may be

utilized to avoid the birth of a child with a disability (Madeo et al., 2011). Members of the disability community have argued that such tools may result in selective termination and lead to a decrease in, or elimination of, the number of people born and living with certain genetic conditions associated with disability. This is thought to send a message to individuals living with disabilities that their lives are not worth living, resulting in harm (Edwards, 2004). If genetic counselors are not educated about the disability community's concerns over prenatal genetic testing technologies, their ability to strengthen and support the disability community is diminished. As advocates for individuals with disabilities, genetic counselors must strive to better understand their lived experiences while acknowledging that they are not a homogenous group. Graduate training is one of a number of opportunities for genetic counselors to work towards this understanding. By attaining and employing disability cultural humility in their practice, genetic counselors can strengthen trust between themselves and the disability community.



## Appendix A IRB Approval



### EXEMPT DETERMINATION

Date:	September 7, 2023
IRB:	STUDY23070057
PI:	Nicole Weise
Title:	Recently Graduated Genetic Counselors' Perceptions of their Disability Training and Preparedness for Addressing Disability in Practice
Funding:	None

The Institutional Review Board reviewed and determined the above referenced study meets the regulatory requirements for exempt research under 45 CFR 46.104(d).

#### Determination Documentation

Determination Date:	9/7/2023
Exempt Category:	(2)(ii) Tests, surveys, interviews, or observation (low risk)
Determinations:	
Approved Documents:	<ul style="list-style-type: none"> <li>• Weise Nicole - Interview Guide_v2023.08.28_FOR_IRB.docx, Category: Data Collection;</li> <li>• Nicole Weise Study Recruitment.pdf, Category: Recruitment Materials;</li> <li>• University_of_Pittsburgh_Interview_Participation_Survey_Version_0.02(1).docx, Category: Recruitment Materials;</li> <li>• Weise Nicole - Study Introductory Script_v2023.08.28_FOR_IRB.dotx, Category: Recruitment Materials;</li> </ul>

If you have any questions, please contact the University of Pittsburgh IRB Coordinator, [Stacy Eckstein](#).  
**NOTE:** Modifications are only required if they will affect the exempt determination. It is important to **close your study when finished** by submitting a Continuing Review.

*Please take a moment to complete our [Satisfaction Survey](#) as we appreciate your feedback.*

## Appendix A.1 IRB Modification



### EXEMPT DETERMINATION

Date:	September 22, 2023
IRB:	MOD23070057-001
PI:	Nicole Weise
Title:	Recently Graduated Genetic Counselors' Perceptions of their Disability Training and Preparedness for Addressing Disability in Practice
Funding:	None

The Institutional Review Board reviewed and determined the above referenced study meets the regulatory requirements for exempt research under 45 CFR 46.104(d).

#### Determination Documentation

Determination Date:	9/22/2023
Exempt Category:	(2)(ii) Tests, surveys, interviews, or observation (low risk)
Determinations:	
Approved Documents:	None

If you have any questions, please contact the University of Pittsburgh IRB Coordinator, [Stacy Eckstein](#).  
**NOTE:** Modifications are only required if they will affect the exempt determination. It is important to close your study when finished by submitting a Continuing Review.

Please take a moment to complete our [Satisfaction Survey](#) as we appreciate your feedback.

# Are you a prenatal genetic counselor?

*We are conducting a research study to understand how your genetic counseling training prepared you to address disability in practice.*

You are eligible if you:

- Are currently working as a prenatal genetic counselor in the United States or Canada
- Graduated from an ACGC accredited genetic counseling program between 2019 and 2022

Participation includes a 1-hour interview, during which we will discuss:

- Your day-to-day as a prenatal GC
- How disability was incorporated into your coursework, clinical experiences, and other graduate training
- What experiences prepared you to address disability in practice

To participate, please scan the QR code below:



Questions?  
Please contact:  
Nicole Weise  
[ndw37@pitt.edu](mailto:ndw37@pitt.edu)



## Appendix C Study Introduction

Thank you for your participation in this study. My name is Nicole Weise. I am a second-year graduate student in the University of Pittsburgh's Master of Science in Genetic Counseling program. I am conducting this study as part of my thesis project. The purpose of this study is to understand genetic counselors' perspectives on how their training and experiences influence their approach to disability in the context of prenatal genetic counseling. *Disability* is a broad term that encompasses physical, cognitive, psychological, communicative, sensory, behavioral, and neurodevelopmental disabilities.

The goals of this research study are two-fold: first, to learn from recently graduated genetic counselors about their views on the adequacy of their graduate training related to disability. Second, to understand how comfortable recently graduated genetic counselors are with addressing disability with patients and families and working with individuals with disabilities.

I anticipate this virtual interview will last approximately one hour.

Your participation in this study is completely voluntary. You may withdraw from the study at any time. If you choose not to participate, or if you do not complete the interview, this will have no effect on your relationship with the University of Pittsburgh.

Your confidentiality is important, and as such, we have taken steps to protect it. The audio from this interview will be recorded for the purposes of transcription. Audio recordings will be transcribed and deidentified, meaning that they will not be connected to your name or any other identifying information, although we will collect basic information for aggregate reporting. Deidentified interview transcripts will be stored on a personal, password-protected computer and will be shared with other members of the study team for the purpose of data analysis. The original audio recordings will be deleted after transcription. No video recordings from the interview will be saved. Although every reasonable effort has been taken, confidentiality during Internet communication activities cannot be guaranteed and it is possible that additional information beyond that collected for research purposes may be captured and used by others not associated with this study. There are no additional, foreseeable risks associated with this project. There are also no direct benefits to you.

You may contact me with any questions or concerns by calling (XXX)XXX-XXXX or emailing [ndw37@pitt.edu](mailto:ndw37@pitt.edu). You may contact my faculty mentor, Dr. Robin Grubs, MS, PhD, LCGC at [rgrubs@pitt.edu](mailto:rgrubs@pitt.edu). Finally, you may contact the Human Subjects Protection Advocate of the Human Research Protection office, University of Pittsburgh at 1(866)212-2668 to discuss any concerns.

Are you willing to participate in this study?  
Do you have any questions before we begin?

## Appendix D Semi-Structured Interview Guide

Date: \_\_\_\_\_  
Study Participant ID: \_\_\_\_\_  
Confirmation of Verbal Consent: \_\_\_\_\_

I'd like to learn a little bit about you in terms of your professional goals.

1. What drew you to genetic counseling?

I'd like to hear more about your position as a patient-facing prenatal genetic counselor, including your day-to-day work.

1. How long have you been working as a prenatal genetic counselor?
2. Can you tell me about the different patient populations and indications you see as a prenatal genetic counselor?

Prenatal genetic counselors often meet with individuals to discuss how genetic factors can impact their pregnancy and family planning. They may then discuss a prenatal diagnosis of a genetic condition associated with a disability, such as Down syndrome or spinal muscular atrophy (SMA).

1. In what ways do you discuss disability with patients?
  - a. How do you talk to patients about disability during pre-test counseling?
  - b. Does your counseling change in post-test counseling?
    - i. If so, how does your counseling change?
2. Do you find these discussions to be challenging in any way?
  - a. If so, can you describe how they are challenging?
3. For some genetic counselors, the way they approach these conversations changes over time. Have you changed how you discuss disability during your years of practice?
  - a. What influenced you to change how you talk about disability?
  - b. In what ways did you change how you discuss disability?
4. How do you feel your graduate training prepared you to have these discussions?

Prenatal genetic counselors may meet with disabled individuals themselves to help them in their family planning and care.

1. In what ways do you interact with disabled individuals in your current position?
2. Do you find these interactions to be challenging in any way?
  - a. If so, how do you feel they are challenging?
3. How do you feel your graduate training prepared you to have these discussions?

Genetic counseling programs vary in their disability training. They may incorporate disability training into their didactic coursework, clinical rotations, or supplemental academic experiences for their students. I'd like to start by talking about what didactic training you might have received about disability. This might include medical education about genetic conditions that result in disability, disability rights, disabled perspectives, ethics, and social determinants of health.

1. When you consider your didactic training, how were disability training and disabled perspectives incorporated into your coursework?
  - a. Were there lectures that talked about disability from a theoretical lens or an ethical lens?
  - b. Were disabled individuals invited to your classes to speak?
  - c. Was the disability critique of prenatal genetic testing discussed during your training?
  - d. How were disabled perspectives otherwise shared?
2. How did your coursework on disability influence how you approach disability in practice?
  - a. Were there any lessons or lectures that you draw on today in practice?

Next, I'd like to talk about how your training program incorporated disability education into clinical observations and rotations. Clinical supervisors may have modeled how to discuss disability or work with disabled patients.

1. Tell me about a time when a provider talked about disability with a patient. What aspects of that conversation did you think were effective or not effective?
2. Were there any lessons you learned during your rotations that you use now in practice, with regard to disability?

Some programs have opportunities for disability training outside of didactic and clinical training provided by their program. For example, some programs encourage students to attend social or support groups for disabled individuals. Additionally, some programs allow students to participate in their university's LEND program. LEND is an interdisciplinary program that aims to educate graduate students in the field of disabilities and improve the lives of individuals with disabilities and their families.

1. Did you participate in any supplemental academic experiences that contributed to your disability-related knowledge and skills?
  - a. How did these experiences impact your training?
2. Have you engaged in any continuing education around the topic of disability?

We are interested in knowing your perspective on your disability training.

1. What do you feel were areas of strength in your disability training?
2. In what ways do you feel your disability training was insufficient, inaccurate, or incomplete?
3. As you reflect on your own training, what would meaningful disability training include?
  - a. If you could have changed your disability education and training, what would you have changed?
  - b. What graduate experiences would have improved your skills in working with disabled patients?
  - c. What graduate experiences would have improved your skills in discussing genetic diagnoses associated with disabilities?

We recognize that personal experiences influence how genetic counselors approach their practice. If you're comfortable sharing, we are also interested in knowing if you have your own personal experience with disability, or if you have known or interacted with a disabled individual outside of your graduate training, that influenced how you approach disability as a genetic counselor.

1. What kind of personal experiences, outside of your graduate training, have you had with disability?
2. How have these experiences influenced how you approach disability in practice?

What else would you like for me to know about your training? What have I not asked that you think is important to ask about this topic?

Lastly, we have a few questions aimed at gathering some demographic information.

1. We want to ensure that we are hearing from genetic counselors that graduated from a range of programs. Which genetic counseling program did you attend?
2. How long have you been a genetic counselor?
3. How many genetic counseling positions have you held since graduating?
4. In which region do you currently work as a genetic counselor?
  - a. Region 1: CT, MA, ME, NH, RI, VT, CN, Maritime Provinces
  - b. Region 2: DC, DE, MD, NJ, NY, PA, VA, WV, PR, VI, Quebec
  - c. Region 3: AL, FL, GA, KY, LA, MS, NC, SC, TN
  - d. Region 4: AR, IA, IL, IN, KS, MI, MN, MO, ND, NE, OH, OK, SD, WI, Ontario
  - e. Region 5: AZ, CO, MT, NM, TX, UT, WY, Alberta, Manitoba, Saskatchewan
  - f. Region 6: AK, CA, HI, ID, NV, OR, WA, British Columbia, Yukon

If I have any questions that arise after our interview, would it be okay for me to contact you in the future for clarification?

## Bibliography

- Accreditation Council for Genetic Counseling (ACGC). (2023a). *Practice-Based Competencies for Genetic Counselors*.
- Accreditation Council for Genetic Counseling (ACGC). (2023b). *Standards of Accreditation for Graduate Programs in Genetic Counseling*.
- Benn, P., Chapman, A. R., Erickson, K., Defrancesco, M. S., Wilkins-Haug, L., Egan, J. F. X., & Schulkin, J. (2014). Obstetricians and gynecologists' practice and opinions of expanded carrier testing and noninvasive prenatal testing. *Prenatal Diagnosis, 34*(2), 145–152.
- Boardman, F. K. (2014). The expressivist objection to prenatal testing: The experiences of families living with genetic disease. *Social Science & Medicine (1982), 107*, 18–25.
- Bowen, C. N., Havercamp, S. M., Karpiak Bowen, S., & Nye, G. (2020). A call to action: Preparing a disability-competent health care workforce. *Disability and Health Journal, 13*(4), 100941.
- Brasington, C. K. (2007). What I wish I knew then...reflections from personal experiences in counseling about Down syndrome. *Journal of Genetic Counseling, 16*(6), 731–734.
- Braun, V., & Clarke, V. (2006). Using thematic analysis in psychology. *Qualitative Research in Psychology, 3*(2), 77–101.
- Braun, V., & Clarke, V. (2021). To saturate or not to saturate? Questioning data saturation as a useful concept for thematic analysis and sample-size rationales. *Qualitative Research in Sport, Exercise and Health, 13*(2), 201–216.
- Darr, K., McCarthy Veach, P., Wurtmann, E., & LeRoy, B. (2023). Effects of genetic counselor disabilities on their professional experiences: A qualitative investigation of North American counselors' perceptions. *Journal of Genetic Counseling, 32*(1), 235–249.
- Dey, I. (1999). *Grounding grounded theory: Guidelines for qualitative inquiry*. Academic Press.
- Douglas, L., Czerwinski, J., Vukovic, R., Ashfaq, M., Lunstroth, R., & Wagner, C. (2023). Disability education and implications for genetic counselor training. *Journal of Genetic Counseling, 32*(6), 1131–1143.
- Farrelly, E., Cho, M. K., Erby, L., Roter, D., Stenzel, A., & Ormond, K. (2012). Genetic counseling for prenatal testing: Where is the discussion about disability? *Journal of Genetic Counseling, 21*(6), 814–824.
- Edwards, S. D. (2004). Disability, identity and the “expressivist objection.” *Journal of Medical Ethics, 30*(4), 418–420.



- Epstein, R., Fuerstman, L., Heidlebaugh, A., Schiller, J., & Zayhowski, K. (2023). Experiences of genetic counseling students with disabilities and chronic illnesses: A qualitative study. *Journal of Genetic Counseling, 32*(6), 1161–1173.
- Gould, H., Hashmi, S. S., Wagner, V. F., Stoll, K., Ostermaier, K., & Czerwinski, J. (2019). Examining genetic counselors' implicit attitudes toward disability. *Journal of Genetic Counseling, 28*(6), 1098–1106.
- Grane, F. M., Lynn, F., Balfe, J., Molloy, E., & Marsh, L. (2023). Down syndrome: Parental experiences of a postnatal diagnosis. *Journal of Intellectual Disabilities: JOID, 27*(4), 1032–1044.
- Havercamp, S. M., Barnhart, W. R., Robinson, A. C., & Whalen Smith, C. N. (2021). What should we teach about disability? National consensus on disability competencies for health care education. *Disability and Health Journal, 14*(2), 100989.
- Hodgson, J., & Weil, J. (2012). Talking about disability in prenatal genetic counseling: A report of two interactive workshops. *Journal of Genetic Counseling, 21*(1), 17–23.
- Kroll, T., & Neri, M. T. (2003). Experiences with care co-ordination among people with cerebral palsy, multiple sclerosis, or spinal cord injury. *Disability and Rehabilitation, 25*(19), 1106–1114.
- Ladau, E. (2021). *Demystifying disability: What to know, what to say, and how to be an ally* (First edition). Ten Speed Press.
- Lenihan, M. (2012). The road less traveled: One genetic counselor's reflections on disability and the genetic counseling profession. *Journal of Genetic Counseling, 21*(6), 784–786.
- Madeo, A. C., Biesecker, B. B., Brasington, C., Erby, L. H., & Peters, K. F. (2011). The relationship between the genetic counseling profession and the disability community: A commentary. *American Journal of Medical Genetics. Part A, 155A*(8), 1777–1785.
- Malterud, K., Siersma, V. D., & Guassora, A. D. (2016). Sample Size in Qualitative Interview Studies: Guided by Information Power. *Qualitative Health Research, 26*(13), 1753–1760.
- Murphy, E. E., Ordal, L., Niendorf, K., Ketelsen, K., Eichmeyer, J. N., & Mills, R. (2023). Genetic counseling graduate program faculty perspectives on sharing education materials among programs. *Journal of Genetic Counseling, 32*(6), 1174–1183.
- Nocon, A., & Sayce, L. (2008). Primary healthcare for people with mental health problems or learning disabilities. *Health Policy (Amsterdam, Netherlands), 86*(2–3), 325–334.
- Olkin, R. (2002). Could you hold the door for me? Including disability in diversity. *Cultural Diversity & Ethnic Minority Psychology, 8*(2), 130–137.
- Public Health National Center for Innovations (PHNCI). (2020). *The 10 Essential Public Health Services*.

- Riconda, D., Grubs, R. E., Champion, M. W., & Cragun, D. (2018). Genetic counselor training for the next generation: Where do we go from here? *American Journal of Medical Genetics. Part C, Seminars in Medical Genetics*, 178(1), 38–45.
- Sanborn, E., & Patterson, A. R. (2014). Disability training in the genetic counseling curricula: Bridging the gap between genetic counselors and the disability community. *American Journal of Medical Genetics. Part A*, 164A(8), 1909–1915.
- Saunders, B., Sim, J., Kingstone, T., Baker, S., Waterfield, J., Bartlam, B., Burroughs, H., & Jinks, C. (2018). Saturation in qualitative research: Exploring its conceptualization and operationalization. *Quality & Quantity*, 52(4), 1893–1907.
- Scheer, J., Kroll, T., Neri, M. T., & Beatty, P. (2003). Access barriers for persons with disabilities: The consumer's perspective. *Journal of Disability Policy Studies*, 13(4), 221–230.
- Sheets, K. B., Crissman, B. G., Feist, C. D., Sell, S. L., Johnson, L. R., Donahue, K. C., Masser-Frye, D., Brookshire, G. S., Carre, A. M., Lagrave, D., & Brasington, C. K. (2011). Practice guidelines for communicating a prenatal or postnatal diagnosis of Down syndrome: Recommendations of the national society of genetic counselors. *Journal of Genetic Counseling*, 20(5), 432–441.
- Strnadová, I., Loblinzk, J., Scully, J. L., Danker, J., Tso, M., Jackaman, K.-M., Dunn, M., Willow, S. A., Sarfaraz, S., Fitzgerald, V., Boyle, J., & Palmer, E. E. (2023). “I am not a number!” Opinions and preferences of people with intellectual disability about genetic healthcare. *European Journal of Human Genetics*, 31, 1057–1065.
- Uhlmann, W. R., Schuette, J. L., & Yashar, B. M. (Eds.). (2009). *A guide to genetic counseling* (Second edition). John Wiley & Sons.
- Vaimberg, E., Demers, L., Ford, E., Sabatello, M., Stevens, B., & Dasgupta, S. (2021). Project Inclusive Genetics: Exploring the impact of patient-centered counseling training on physical disability bias in the prenatal setting. *PloS One*, 16(8), e0255722.
- Vivanti, G. (2020). Ask the Editor: What is the Most Appropriate Way to Talk About Individuals with a Diagnosis of Autism? *Journal of Autism and Developmental Disorders*, 50(2), 691–693.
- Waltman, L., Runke, C., Balcom, J., Riley, J. D., Lilley, M., Christian, S., Zetsche, L., & Goodenberger, M. L. (2016). Further Defining the Role of the Laboratory Genetic Counselor. *Journal of Genetic Counseling*, 25(4), 786–798.
- Winchester, D., Karger, L., Wasserman, D., & Naik, H. (2023). Disability training for genetic counseling students: The advocates' perspective. *Journal of Genetic Counseling*.