Healthcare Providers' Perspectives on Genetic Counselors

by

Lucy Maree Eleanor Galea

Bachelor of Science, University of Southern Queensland, 2013

Bachelor of Science (Honors), University of Southern Queensland, 2014

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

2023

UNIVERSITY OF PITTSBURGH

SCHOOL OF PUBLIC HEALTH

This thesis was presented

by

Lucy Maree Eleanor Galea

It was defended on

April 5, 2023

and approved by

Jodie Vento, MGC, CGC, Assistant Professor, Human Genetics;
Director, Genetic Counseling Program,
Human Genetics, School of Public Health, University of Pittsburgh

Robin Grubs, MS, PhD, CGC, Associate Professor
Human Genetics, School of Public Health, University of Pittsburgh

Cynthia Salter, MPH, PhD, Assistant Professor
Behavioral and Community Health Sciences, School of Public Health, University of Pittsburgh

Thesis Advisor: Charlotte Skinner, MS, MPH, CGC, Adjunct Professor
Human Genetics, School of Public Health, University of Pittsburgh

Healthcare Providers’ Perspectives of Genetic Counselors

Lucy Maree Eleanor Galea, MS

University of Pittsburgh, 2023

Background: As the genetic counseling profession expands, it is increasingly important for healthcare providers to understand genetic counselors’ scope of practice, and their role in patient care and within the multidisciplinary team. Little is currently known about the level of knowledge healthcare providers have about genetic counselors’ scope of practice, role, and skill set. This study aims to address this gap in the literature.

Methods: Healthcare providers in direct patient care roles at UPMC Magee Womens Hospital and UPMC Children’s Hospital of Pittsburgh were surveyed through a twenty-question survey that covered scope of practice, facilitation of patient care, reasons for referring patients for genetic counseling and the demographics of the respondents. For the second part of the study providers were interviewed to further explore these areas. At the end of the survey, respondents could opt into a thirty-minute interview. The survey results were analyzed by Fisher’s exact test, and the interview transcripts were analyzed using thematic analysis.

Results: Forty-nine surveys were utilized for data analysis, and two interviews were conducted with three total participants. Providers had a good understanding of genetic counselors’ scope of practice and were significantly more likely to know the degree held by a genetic counselor if they themselves had a medical degree (p-value: 0.004). However, providers did not have a clear understanding of whether Pennsylvanian genetic counselors are permitted by their licensure to order genetic testing. Most respondents referred frequently to genetic counselors, with 68% referring at least monthly.

Interview participants reported high levels of positive regard for genetic counselors and their understanding of scope of practice was dependent on the interactions they had with genetic counselors.

Conclusion: This study shows that providers in Pennsylvania need to be educated about Pennsylvanian genetic counselor scope of practice, particularly the provision related to who can serve as the ordering provider of genetic testing. This research is critical to understanding how genetic counselors are viewed by healthcare providers and is significant for public health as a referral to a genetic counselor is often part of the appropriate application of many national guidelines for individuals with genetic conditions.

Table of Contents

List of Tables

List of Figures

Acknowledgements

I would like to first and foremost acknowledge and thank my committee for their mentorship, wisdom, and guidance throughout this process. Thank you especially to Charlotte and Jodie for trusting me to bring your idea to life. And an extra thanks to Jodie for helping to get my survey to the right people for distribution.

To the staff at the Children's Hospital of Pittsburgh and UPMC Magee Womens Hospital who helped to distribute my survey to providers, and filled out my survey, thank you, this research would not have been as successful as it was without you. A huge thank you to my three interview participants for taking time out of their day to speak with me about genetic counselors.

To the people who encouraged me to apply to a genetic counseling program in the early days of the pandemic, I'm not sure I would've believed it was possible to go back to university, apart from all your support and motivation. And thank you to all those who have continued to support me through this journey, it has meant a lot to have you by my side, both near and far.

Thank you to my husband, Ashton, for believing in me for the past three years, even when I didn't believe in myself. Your understanding and care, late night cups of tea and laughs, and constant reminders of the light at the end of the tunnel, have made my graduate school experience so much easier.

And finally, to the Class of 2023, how do I begin to thank eleven people who have made such an impact on my life in the space of two short years. When I applied to Pitt, I could never have imagined a better group of people to have been on this journey with. I feel honored to call you all my friends and I cannot wait to see what incredible genetic counselors you all become.

List of Abbreviations

ACGC – Accreditation Council of Genetic Counselors

CCC-SLP – Certificate of Clinical Competence in Speech-Language Pathology

UPMC CHP – Children’s Hospital of Pittsburgh

GAO – Government Accountability Office

NSGC – National Society of Genetic Counselors

Ob/Gyn – Obstetrician/Gynecologist

PAGC – Pennsylvania Association of Genetic Counselors

PCPs – Primary Care Physicians

UPMC – University of Pittsburgh Medical Center

1.0 Introduction

1.1 Background and Specific Aims

Genetic counseling is still a relatively young profession among healthcare workers, and the profession is currently growing rapidly. According to the National Society of Genetic Counselors (NSGC), the number of genetic counselors in the United States has grown from 1,155 certified genetic counselors in 1999 to 5,629 in 2021 (NSGC, 2021a). This growth is expected to continue with the number of available graduate student placements expanding each year to aid in meeting demand within the field. As the profession continues to expand, it is important for the healthcare providers who are interacting with and referring to genetic counselors to understand the scope of practice and role of the genetic counselor on a multidisciplinary team. Little to no information currently is available about the level of knowledge healthcare providers have about genetic counselors' scope of practice, role and skill set.

Many healthcare providers have limited training in the new and evolving field of genetics, and are therefore often unable or underprepared to talk to patients about genetic risk, to order appropriate genetic testing, and to counsel about test results (Mikat-Stevens et al., 2015). However, even an increase in genetics knowledge has not been shown to translate into a higher utilization of genetic counseling services (Clyman et al., 2007).

Research about physician referrals to genetic counselors has found differing patterns (Diamonstein et al., 2018; Truong et al., 2021). The Diamonstein et al. (2018) study shows that one in five physicians do not understand the vital role that a genetic counselor can play in patient care, and suggests that they may not understand the scope of practice for genetic counselors. As physicians are the providers who most frequently make referrals, it is important for them to understand the role that a genetic counselor can play in patient care so that they can communicate this with patients and refer appropriately.

The National Society of Genetic Counselors (NSGC) has model language for genetic counselor scope of practice, which is part of the drafted model language for genetic counselor licensure (NSGC, n.d.). Through state licensure, individual states set their own scope of practice. For example, legislation passed for genetic counselors' scope of practice in Pennsylvania is largely in alignment with the NSGC model legislation, but the word 'order' has been omitted from the third provision for scope of practice (Medical Practice Act of 1985 - Regulation of Genetic Counselors, 2011), which NSGC suggests as 'identify, order, and coordinate genetic laboratory tests and other diagnostic studies as appropriate for genetic assessment' (NSGC, n.d.). This means that genetic counselors in Pennsylvania have the ability to identify and coordinate genetic testing, but cannot act as the ordering provider (Medical Practice Act of 1985 - Regulation of Genetic Counselors, 2011). This clause along with an explicit statement about genetic counselors not ordering any kind of testing (Medical Practice Act of 1985 - Regulation of Genetic Counselors, 2011) makes Pennsylvania different from other states where licensed genetic counselors can order testing. Healthcare providers who are not genetic counselors or do not work closely with them may not realize this subtle difference.

To our knowledge there has been no research in Pennsylvania or the United States that has examined health care providers' knowledge of genetic counselors' scope of practice, and little research has investigated the perception of genetic counselors' function or role in a multidisciplinary team (Hudson et al., 2019; Vito et al., 2022).

Through this project, healthcare providers involved in direct patient care in western Pennsylvania were surveyed to elicit their understanding of genetic counselors’ scope of practice, and what kind of role they play within the medical team for patients. Healthcare providers who identified themselves as being willing to participate, were then offered an interview, to allow for the opportunity to gain a deeper understanding of their knowledge and perceptions of genetic counselors.

1.1.1 Specific Aim I

Create and distribute a survey to healthcare providers in western Pennsylvania, to elicit:

a. Their understanding/perception of genetic counselors' function in facilitating care for patients;
b. Their understanding/perception of genetic counselors' scope of practice in Pennsylvania.

1.1.2 Specific Aim II

Conduct interviews with healthcare providers to gain a deeper understanding of their knowledge of:

a. Genetic counselors’ training, competencies, and function in providing care for patients;
b. The scope of practice for genetic counselors in Pennsylvania;
c. The perceived role and utility of genetic counselors within a multidisciplinary team.

2.0 Manuscript

2.1 Background

2.1.1 Genetic Counseling Profession

Genetic counseling is ‘the process of helping people understand and adapt to the medical, psychological and familial implications of the genetic contributions to disease’ (Resta et al., 2006). The profession of genetic counseling has been growing since the first master’s level program was established in 1969 (Marks & Richter, 1976), with 54 programs now established across the United States and four accredited programs in Canada (ACGC, 2022). A 100% increase in the number of certified American genetic counselors was reported in the last ten years, and this number is expected to expand another 100% over the next decade (NSGC, 2022a), signifying a rapid expansion of the profession. The genetic counseling profession has extended beyond the United States, with graduate genetic counseling training programs in 24 countries (University of South Carolina, n.d.).

As the depth of knowledge of genetics and gene function has grown, so too has the ability of genetic counselors to become key providers in patient healthcare. According to the latest NSGC Professional Status Survey, the leading ‘primary areas of practice’ in 2021 were adult cancer genetics (39%), prenatal genetics (23%), and pediatric genetics (18%) (NSGC, 2022b). Genetic counselors also have an abundance of roles including in specialty clinics and laboratory-based positions, above and beyond the primary areas of practice (NSGC, 2022b). Due to this expansion beyond the traditional clinics that genetic counselors have been involved with, it is increasingly important that healthcare providers who interact with genetic counselors understand a genetic counselor’s scope of practice.

2.1.2 Genetic Counselors’ Scope of Practice

Genetic counselors’ scope of practice is set by the state issuing licensure, allowing a genetic counselor to legally practice in the state within which they see patients. According to the NSGC website, as of July 2022, there are 31 states that currently issue state licensure for genetic counseling, with another 17 that have legislative bills passed or that are in the rule making process (NSGC, 2022c).

A nuance of the scope of practice for Pennsylvanian genetic counselors, is that they are licensed to ‘identify and coordinate genetic testing’ and other diagnostic testing as required, but are not permitted to act as the sole ordering provider for tests (Medical Practice Act of 1985 - Regulation of Genetic Counselors, 2011). The NSGC model legislation wording is for genetic counselors to be able to ‘identify, coordinate and order’ genetic testing (NSGC, n.d.). All other wording in the Pennsylvania licensure bill is the same as the model NSGC wording. However, Pennsylvania’s legislation explicitly calls out that genetic counselors cannot order a test for ‘genetic disease or condition’ in Section 13.4c(2) (Medical Practice Act of 1985 - Regulation of Genetic Counselors, 2011). Many states with licensure include the language ‘identify and coordinate genetic testing’ (NSGC, 2021b), with 11 states explicitly allowing genetic counselors to order testing and three states allow genetic counselors to order genetic testing in agreement with a physician. There have been ongoing efforts to amend the Pennsylvanian legislation, sponsored by the Pennsylvania Association of Genetic Counselors (PAGC) to allow genetic counselors to be the ordering provider of genetic tests. These efforts continued with the introduction of H.B.1233 and H.B.1236 in the 2021-2022 session of the Pennsylvania House of Representatives (Quinn, 2021).

To date, there has been no research exploring this explicit nuance in scope of practice for Pennsylvania genetic counselors, and no research into healthcare providers' understanding of genetic counselors' scope of practice.

2.1.3 Utilization of Genetic Counselors

2.1.3.1 Trends in Referrals to Genetic Counselors

Patients most commonly hear about genetic counseling from their healthcare provider, (Liang et al., 2018) and providers are more likely to refer to genetic counseling if they understand the value and purpose of genetic counseling (Prochniak et al., 2012). A genetic counseling referral can be made for several reasons. Research has found that physicians were motivated to refer to genetic counseling due to a need for diagnostic or predictive genetic testing (Czekalski et al., 2022; Liang et al., 2018; Linfield et al., 2022), patient's desire for further risk information including recurrence risk, a lack of specific knowledge that the PCP identified within themselves, and for specialist care coordination (Truong et al., 2021).

The data on referral to genetic counselors from physicians has changed significantly over the past five years. In a 2018 survey of Texas physicians of varying specialties, 72% of respondents had 'never or rarely' referred their patients for genetic counseling services (Diamonstein et al., 2018), compared to a 2021 study that found that 89% of United States Primary Care Physician (PCP) respondents had referred to genetic counseling at least once, with participants reporting a median of three referrals per year (Truong et al., 2021). This difference could be due to several factors, such as an increase in the awareness of genetic counselors in the three years between these studies, and the types of physicians surveyed.

Rates of referral to genetic counselors may also differ depending on the state within which a patient is located, and the number of genetic counselors available. Harding and colleagues (2019) found that healthcare providers who did not have easily accessible genetic counselors in their region, were more likely to triage who to send to a genetics expert compared with patients they could manage on their own. This study showed that providers were aware of the importance of a genetic counseling referral, but that these providers were also acutely aware of how strained a system can be without an adequate number of genetic counselors to provide the necessary patient services.

2.1.3.2 Genetic Counselors as Part of the Patient Team

Genetic counselors are increasingly becoming part of multidisciplinary teams to help facilitate better patient experiences and outcomes. A study from Europe showed that the roles genetic counselors provided on a multidisciplinary team included collecting a family history and constructing a pedigree, ensuring that patients understood the material being presented and answering their questions, and informing patients of their options for genetic testing (Catapano et al., 2022). Healthcare providers have reported appreciating genetic counselors' knowledge of genetics, their drive to share genetic information with other providers, and the way they are able to interact and share their knowledge with patients (Vito et al., 2022). Overall, a general positive regard for genetic counselors on multidisciplinary teams has been reported, with 77% of participants not reporting any negative aspects of their work with a genetic counselor (Vito et al., 2022), and genetic counselors have been rated as a highly important provider and member of the multidisciplinary team (Catapano et al., 2022).

Interestingly, several studies have shown that psychosocial counseling is not perceived to be the sole responsibility of the genetic counselor, but is seen as a shared responsibility among providers (Catapano et al., 2022; Cordier et al., 2016; Prochniak et al., 2012), despite genetic counselors' specialized training in this area and having this role as part of their scope of practice.

Multidisciplinary care can take many forms, with novel delivery models appearing in the literature. Published models include multidisciplinary clinics in pediatric oncology (Hudson et al., 2019; Shah et al., 2017), a primary care and precision medicine clinic (Massart et al., 2022), and a dermatology and clinical genetics clinic (Parker et al., 2021). Primary care clinics that have a genetic counselor embedded within the practice have been found to have significantly higher referral rates to genetic counselors (Truong et al., 2021).

2.1.4 Barriers to Genetic Counseling

There are many identified barriers to genetic counseling that come from patients, healthcare systems, and providers.

Patients don't have a high level of awareness about the genetic counseling profession (Liang et al., 2018; Pasca et al., 2021; Riesgraf et al., 2015), but do understand that genetic counseling could be beneficial after being given a description of genetic counseling (Liang et al., 2018; Riesgraf et al., 2015). The most commonly found patient barrier to genetic counselor referral is patient refusal or disinterest (Czekalski et al., 2022; Truong et al., 2021), with other barriers including not understanding the referral reason, patient concern for ethical or legal implications, uncertainty around insurance coverage and financial burden of genetic testing, priority given to other care, or a lack of or inaccurate knowledge of family history (Czekalski et al., 2022; Hyatt et al., 2019; Liang et al., 2018; Seibel et al., 2022; Venier et al., 2022).

Providers, although the most appropriate person to refer a patient, often lack the knowledge regarding genetics including the ability to assess personal and family medical history for genetic conditions (Baldwin et al., 2014; Prochniak et al., 2012; Soller, 2022; Truong et al., 2021). PCPs have also identified that they feel ill-equipped to share specifics about a genetic counseling appointment and to be able to adequately convey why their patient needs to follow through with a referral (Seibel et al., 2022). Other provider-cited barriers that have been found include fear of patient discrimination by society and insurance companies, and concern for patient anxiety regarding genetic testing results (Hauser et al., 2018; Mikat-Stevens et al., 2015).

Another barrier worth noting is the implicit bias of providers not referring ethnic minorities for genetic counseling and/or genetic testing as frequently as the non-Hispanic white population (Choi et al., 2022; Ademuyiwa et al., 2021; Peterson et al., 2020; Olsabeck et al., 2019; Manrriquez et al., 2018), even as programs such as universal tumor screening are implemented (Ademuyiwa et al., 2021; Muller et al., 2018; Peterson et al., 2020).

2.1.4.1 The Importance of the ‘Access to Genetic Counselor Services’ Act

Another barrier to genetic counseling that affects both patients and providers, is that genetic counselors are not formally recognized as healthcare providers by the Centers for Medicare and Medicaid Services (CMS). This current standing means that genetic counselors cannot bill for appointments with patients, including some circumstances involving private health insurance. This leads to inequality in accessing appropriate genetic care (Campbell, 2022). The ‘Access to Genetic Counselors Act’ (*H.R.2144 - 117th Congress (2021-2022)*, 2021) would allow genetic counselors to bill for services provided, without the genetic testing being signed off on by a physician. If the federal government passes this bill, and Medicare recognizes genetic counselors as providers, it paves the way for private insurance companies to also cover the cost of a genetic counseling appointment, which is currently often absorbed by the healthcare system employing the genetic counselor (Campbell, 2022).

2.1.5 Genetics and Healthcare Providers

2.1.5.1 Comfort Levels of Healthcare Providers Ordering and Understanding Genetic Testing

Most healthcare providers will have to care for patients with a genetic condition or hereditary predisposition to disease. Collectively, there are 30 million individuals, or 1 in 10, living with rare disease in the United States (GARD, n.d.), with 80% of these having a genetic component (NHGRI, 2018). These statistics illustrate why it is important that providers have the resources and ability to care for these patients appropriately, which includes knowing when to refer to a genetics expert such as a genetic counselor, feeling comfortable with genetic testing and results, and having up-to-date education about genetics to help inform their practice.

However, many healthcare providers feel ill-equipped to discuss genetic topics with their patients due to limited genetics curriculum in their undergraduate and postgraduate training, lack of detailed knowledge about genetic conditions and the fast pace at which the genetics field moves (Harding et al., 2019a). Due to many of the same concerns providers often feel uncomfortable ordering or interpreting genetic testing reports (Evenson et al., 2016; Hauser et al., 2018; Liang et al., 2018; Pet et al., 2019; Selkirk et al., 2013). In one study of PCPs, only 14% of participants said they would feel comfortable interpreting genetic testing results (Hauser et al., 2018), and another found that only 16% of physicians from multiple specialties were confident discussing genetic testing with patients (Selkirk et al., 2013).

Medical specialty has been found to play a part in how often practitioners discuss genetic testing and results, and how confident they feel doing so, with PCPs feeling less certain about how and when to incorporate genetic testing compared to their specialist colleagues (Selkirk et al., 2013). Selkirk et al. (2013) found that 44% of PCPs reported being not being comfortable with genetic testing. It is understandable that research has identified that some specialists have a better understanding of and confidence in navigating and discussing genetic testing compared to PCPs, as they are more likely to utilize this technology and interpret results more frequently. For example, gynecological oncologists have been found to feel comfortable in discussing negative (88%) and positive (91%) genetic testing results, but are less confident in their ability to discuss variants of uncertain significance (63%) (Liang et al., 2018). Genetic counselors are a resource available to other providers to better ensure that patients with genetic conditions receive appropriate counseling regarding genetic results and recurrence risks, and that physicians incorporate genetic information into patient management in the most beneficial way.

2.1.5.2 Genetics Education of the Healthcare Provider

It was found that in a study of PCPs, nurse practitioners and residents, 78% of respondents had received formal genetics education, however almost 70% of participants answered that they would benefit from information regarding how to discuss genetics and genetic testing with patients (Hauser et al., 2018). Over 80% of participants in the same study indicated that they would like an easier way to order genetic testing and identify resources, for both the patient and the provider, on managing a patient when they receive a positive genetic test result (Hauser et al., 2018).

Continuing education has been effective in increasing provider knowledge and comfort, with several pilot studies showing improvement in these metrics after lectures or online modules (Clyman et al., 2007; Harding et al., 2019a; East et al., 2022; Hajek et al., 2022; Tri, 2022). PCPs have also identified that continuing education can occur in numerous other ways such as email communication about genomics updates, face-to-face seminars, inclusion of a genetic professional presenting in case rounds, pamphlets and online genetic databases, and timely access to a genetics expert for professional conversations and questions (Harding et al., 2019a).

Unfortunately, even though PCPs are interested in increasing their genetics knowledge, there still appears to be inadequate ‘preparation and support’ for providers in these roles (Harding et al., 2019a). Ultimately, it is important to remember that PCPs are not genetics experts, and those in the genetics field should not expect them to be (Harding et al., 2019b). This knowledge gap, which for generalists may never be filled, is further reason to integrate genetic counselors into primary care clinics and to continue their integration into multidisciplinary clinics, to help bridge the gap in genetic care for patients. Published literature highlights primary care and multidisciplinary models that have been successful (Massart et al., 2022; Truong et al., 2021), and the continued collaboration between healthcare providers and genetic counselors will likely enhance patient accessibility to genetic care and genetic counselors.

2.2 Methods

2.2.1 Recruitment

IRB exemption approval was obtained from the University of Pittsburgh prior to distribution of the survey (see Appendix A).

Participants for the survey and subsequent interviews were recruited through a number of methods, from December 14th, 2022 to February 8th, 2023. The online Qualtrics survey was distributed via email through Division Chiefs at the University of Pittsburgh Medical Center Magee-Womens Hospital (UPMC MWH) and at UPMC Children's Hospital of Pittsburgh (UPMC CHP) (Appendix B). The survey was also advertised through the 'Physician's Flash' newsletter at UPMC CHP on January 9th, 2023, through the 'Extra', the internal UPMC newsletter on January 12th, 2023 and on InfoNet, the UPMC Intranet, beginning January 12th, 2023 (Appendix C).

Any healthcare providers currently in direct patient care roles, that were not genetic counselors, within the UPMC healthcare system were eligible for this study.

2.2.2 Survey and Interview Design

A 20-question survey was designed for this study, with input from thesis committee members. Prior to IRB approval, the survey was piloted with a local Obstetrician/Gynecologist, thesis committee members, and students of the 2023 Pitt genetic counseling class. No major changes were made to the survey after piloting. It was estimated that the average time to take the survey would be approximately five to seven minutes. Questions included reason for referral and referral rates to genetic counselors, interactions with genetic counselors, facilitation of care for patients by genetic counselors, genetic counselors' scope of practice, and demographics of the survey participant (Appendix D).

The final survey question elicited respondents' interest in participating in a thirty-minute interview. For respondents who answered yes, they were taken to a new, separate survey that asked for their name and email address before asking them to use a hyperlink to the primary researcher's Calendly link to set a time for the interview (Appendix E).

Interview questions were designed in conjunction with the thesis committee, to expand on the questions asked in the survey (Appendix F). Participants were queried about their background, interactions with genetic counselors and understanding of a genetic counselors' day-to-day work. Knowledge of scope of practice was further investigated by using specific genetic counselor scope of practice examples and asking if these were beneficial to the participant's practice and patient care.

Participants were explicitly asked if they believed that genetic counselors in Pennsylvania were able to be the ordering provider on a genetic test. They were then asked what they thought about Pennsylvanian licensure which does not permit genetic counselors to order genetic testing. Questions then inquired about how participants see a genetic counselor's role in multidisciplinary teams and what they bring to patient care, before being asked to identify anything that was surprising or unexpected in their interactions with a genetic counselor.

2.2.3 Data and Statistical Analysis

2.2.3.1 Survey Data

Survey responses were analyzed utilizing Microsoft Excel and Stata v17.0 (StataCorp, 2021). Descriptive statistics were generated in Stata and visualized in Microsoft Excel to determine how often a survey response was chosen and the demographics of the respondents. Due to small cell values ($n < 5$), the Fisher's Exact Test was utilized in Stata to calculate statistical significance in each test run. The p-value cut off utilized for statistical significance was 0.05. Odds ratios were calculated from contingency tables generated by Stata, by the primary investigator.

For statistical analysis in Stata, some data categories were combined to increase power of the tests. Those who selected MD, DO, and/or PhD as their response for respondent degree, were grouped together in a category called 'Doctor'; Those who selected PA-C, CRNP or Other were grouped together in a second category, called 'Advanced Practice Providers (APP)/Other'. For the questions of time practicing in total, and time practicing in Pennsylvania, the year categories were collapsed into two groups: equal to or less than 10 years, or 11 years and greater.

2.2.3.2 Interview Data

Two interviews, with three participants were conducted via Microsoft Teams. The automatically generated transcripts from Microsoft Teams were used as the base transcripts before interviews were then watched at least twice by the primary researcher to make amendments to the transcripts and to ensure accuracy of the documents. This also allowed the primary researcher to gain familiarity with the interviews and the context of the transcripts.

Interviews were analyzed utilizing thematic analysis, as per Braun and Clark (2006). Transcripts were coded to help identify common threads throughout the interviews. A codebook with the meaning units (codes), definitions and examples was constructed (see Appendix G). The transcripts and codes were reviewed by a committee member with expertise in qualitative research. Codes were then organized into two themes, with memos written to help organize thoughts and patterns in the development and finalization of the themes.

2.3 Results

2.3.1 Survey Results

There were 65 responses recorded by Qualtrics after the survey opened, with 49 responses included in the final analysis; 16 surveys were excluded because respondents completed 17% or less of the survey. Three respondents did not provide their demographic information but completed the remainder of the survey and therefore were included in the analysis.

2.3.1.1 Demographics

Of the respondents, 63.83% were doctors (MD or MD/PhD or DO), 12.77% were physician assistants (PA-C), and 10.64% were certified registered nurse practitioners (CRNP). Another 10.64% of respondents chose ‘Other’ and provided their degree in the text box. There were no statistical associations found between type of provider (degree) or specialty and their interaction with a genetic counselor.

Respondents practiced in a wide range of patient care, with 39.13% choosing some form of pediatric specialty care, responding with either ‘Pediatrics’ or ‘Other’ and then writing their pediatric specialty in the box provided (e.g., pediatric ophthalmology). For the purposes of analyses, if a respondent chose ‘Other’ they were kept in that category, even when they indicated pediatrics. See Figure 1 for the range of specialties that respondents practiced in. All respondents except for one answered that their primary institutional setting was a university medical center.

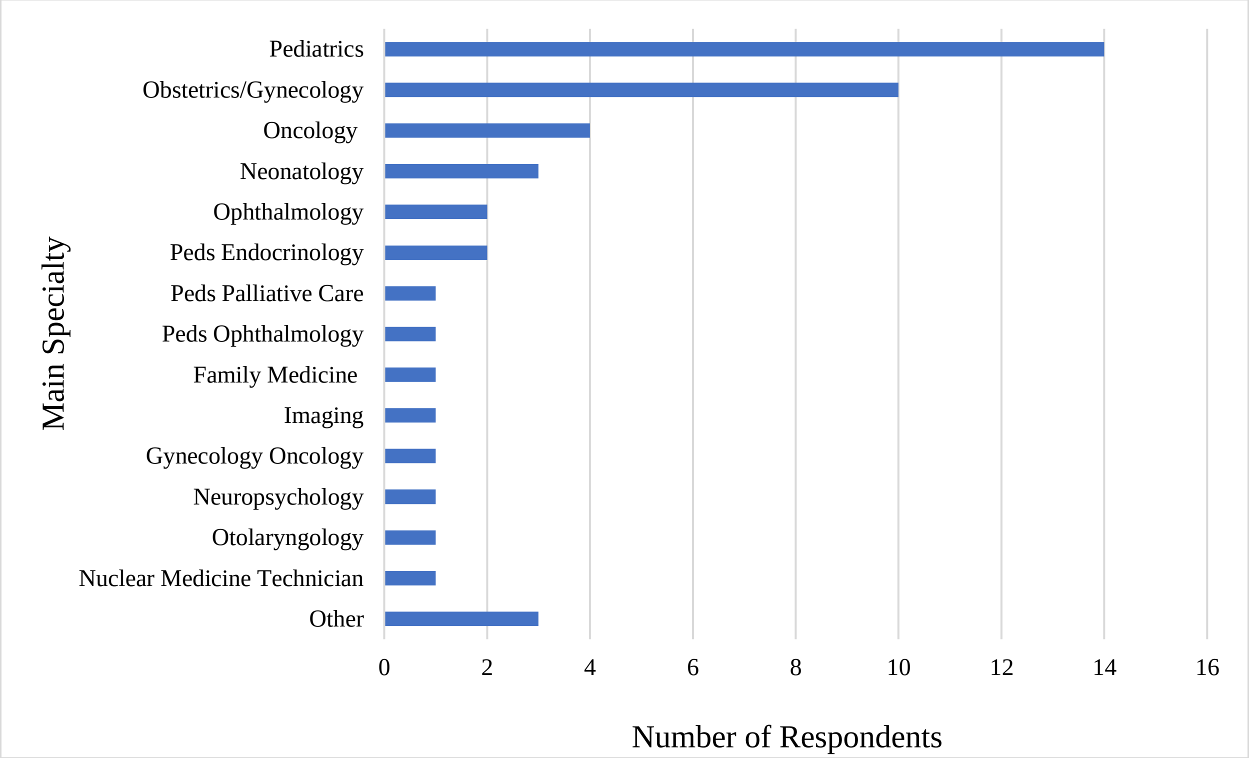


Figure 1: Main specialty of respondents (n=46)

Respondents were asked how long they had been practicing in their chosen field, and how long they had been practicing in Pennsylvania. The median total practicing time among respondents was 11 to 20 years, and the median time spent practicing in Pennsylvania was 6 to 10 years. See Figure 2 for the range of time practicing in total and in Pennsylvania among respondents.

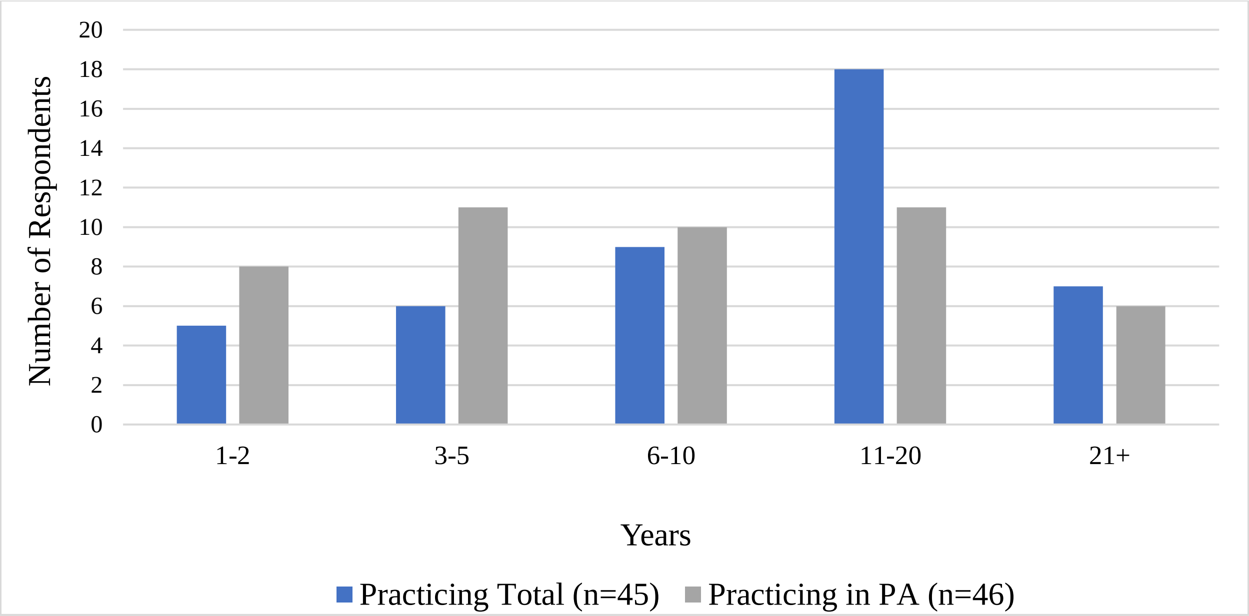


Figure 2: Time spent practicing in total and in Pennsylvania by respondents

2.3.1.2 Interactions of Respondents with Genetic Counselors

Most survey respondents (83.67%) had interacted with a genetic counselor at some point in their career; 14.29% had not interacted or worked with a genetic counselor, and 2.03% were unsure. As shown in Figure 3, the most common type of interaction had with genetic counselors was a referral to a genetic counselor. Providers were allowed to choose all options that applied to them.

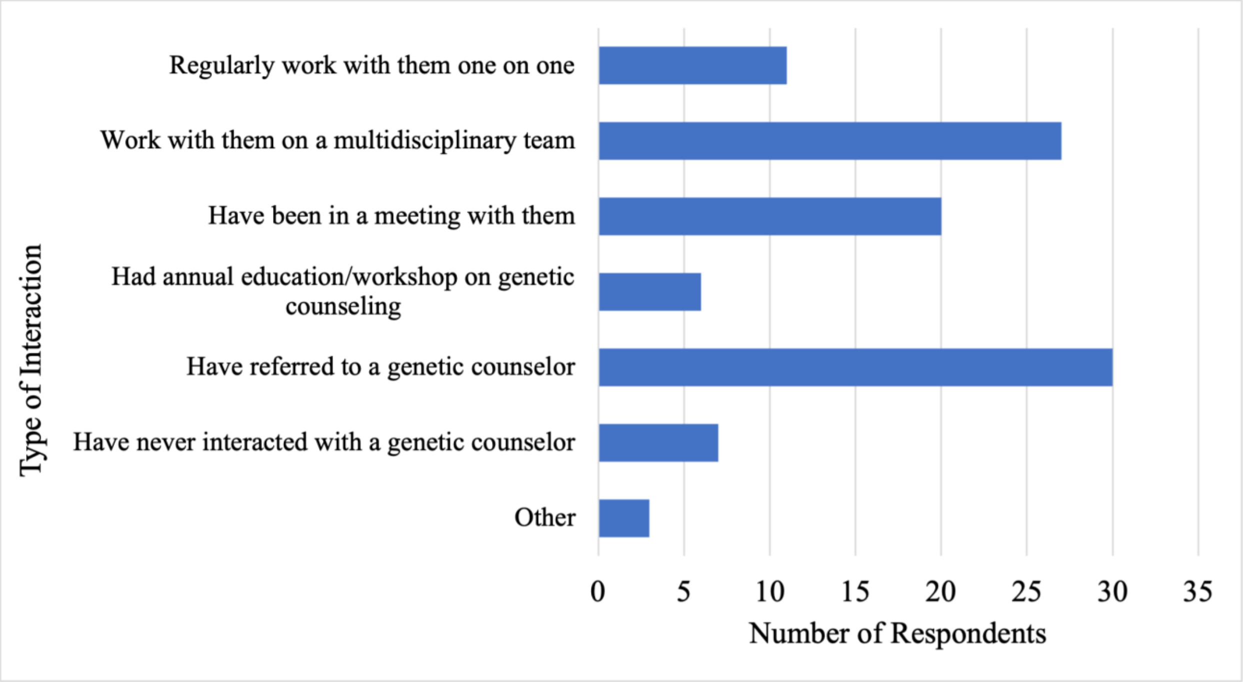


Figure 3: Types of interactions with genetic counselors

For those respondents who chose ‘other’, some common responses included reading patient notes, discussing or emailing patient questions, and one provider stated that they had shadowed a genetic counselor.

Respondents who had interacted with a genetic counselor were significantly more likely to have interacted with them in multidisciplinary clinic (p-value: 0.001), have referred patients to a genetic counselor (p-value: 0.010) or have been in a meeting with a genetic counselor (p-value: 0.032). Respondents who indicated that they had interacted with a genetic counselor in any way, were significantly more likely to choose that genetic counselors need a master’s degree to practice (p-value: 0.030).

A significant association was also found between time spent practicing as a provider and whether the individual had worked with a genetic counselor one-on-one (p-value 0.027; 1-sided Fisher’s exact p-value: 0.014). Providers who had been practicing for 11 years or more were 10.69

times more likely to have worked with a genetic counselor one on one, compared to providers who had been practicing for 10 years or less. It was also found that a significant association existed between providers who had been practicing 11 years or longer, and those who had attended an annual education conference or workshop on genetic counseling (p-value: 0.027).

2.3.1.3 Referral Patterns of Respondents

Respondents were asked two questions about their referral patterns to genetic counselors. When asked how frequently they refer to genetic counselors, 30.61% said they referred weekly, 28.57% referred monthly, and 8.16% referred to genetic counselors daily. See Figure 4 for full results.

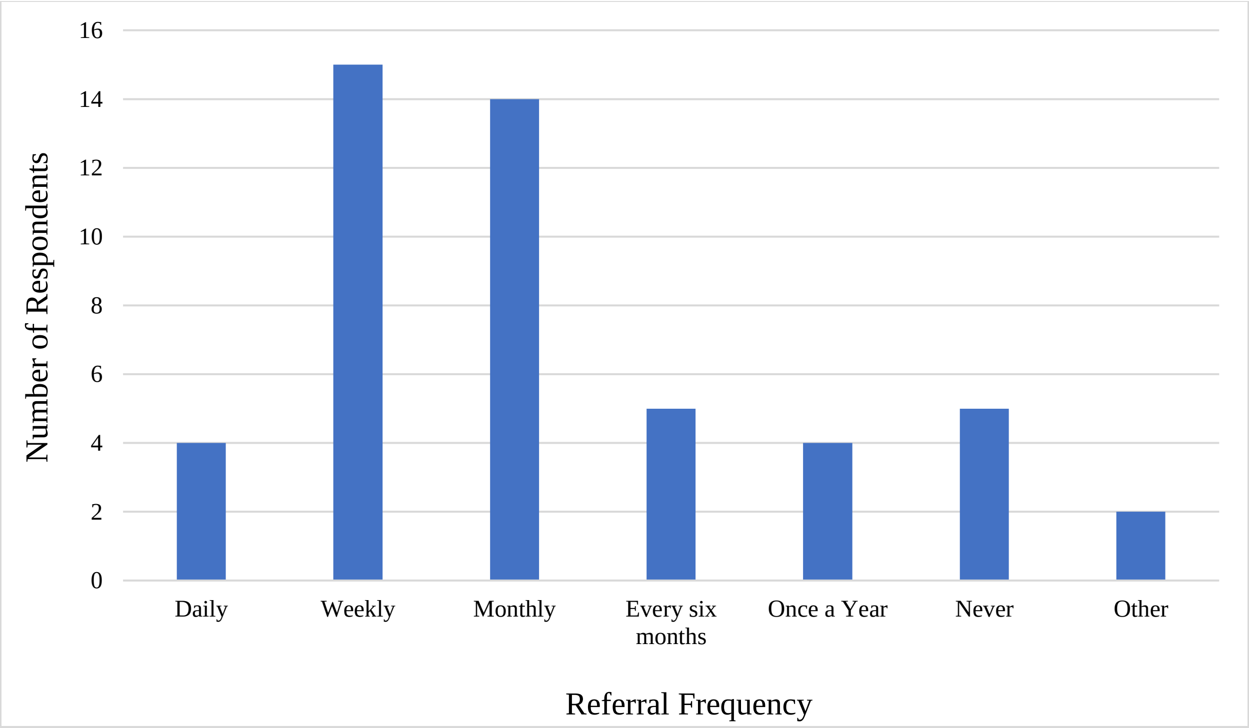


Figure 4: Respondents referral patterns to genetic counselors

(n=49)

For respondents who answered that they refer ‘once a year’ or ‘never’, skip logic was used to provide them with an additional question about reasons or barriers as to why they don’t refer to genetic counselors, with eight response options including a text box for free text responses. Nine respondents received this question. Respondents could choose all options that applied. The most common response was ‘other’, with those who utilized the text box most often citing that it was not within their scope of practice to refer to genetic counselors. Three individuals chose from the listed options; one indicated they were ‘unsure if my patient population would be suitable for genetic testing’ (Pediatric CCC-SLP), one chose the former response plus that the ‘information would be too complicated for my patients’ (Ob/Gyn MD), and one chose that they were ‘unsure if it would be beneficial for my patients’ (Ob/Gyn PA-C).

The second question regarding referral patterns for all respondents found that most respondents refer due to patient or family medical history. For full responses given, see Figure 5. The most common ‘other’ text response was related to prenatal indications including ultrasound findings. Due to the oversight of not including prenatal findings as a separate option, this was included as its own category in Figure 5 below. However, for analyses, ultrasound/prenatal findings were included with ‘other’.

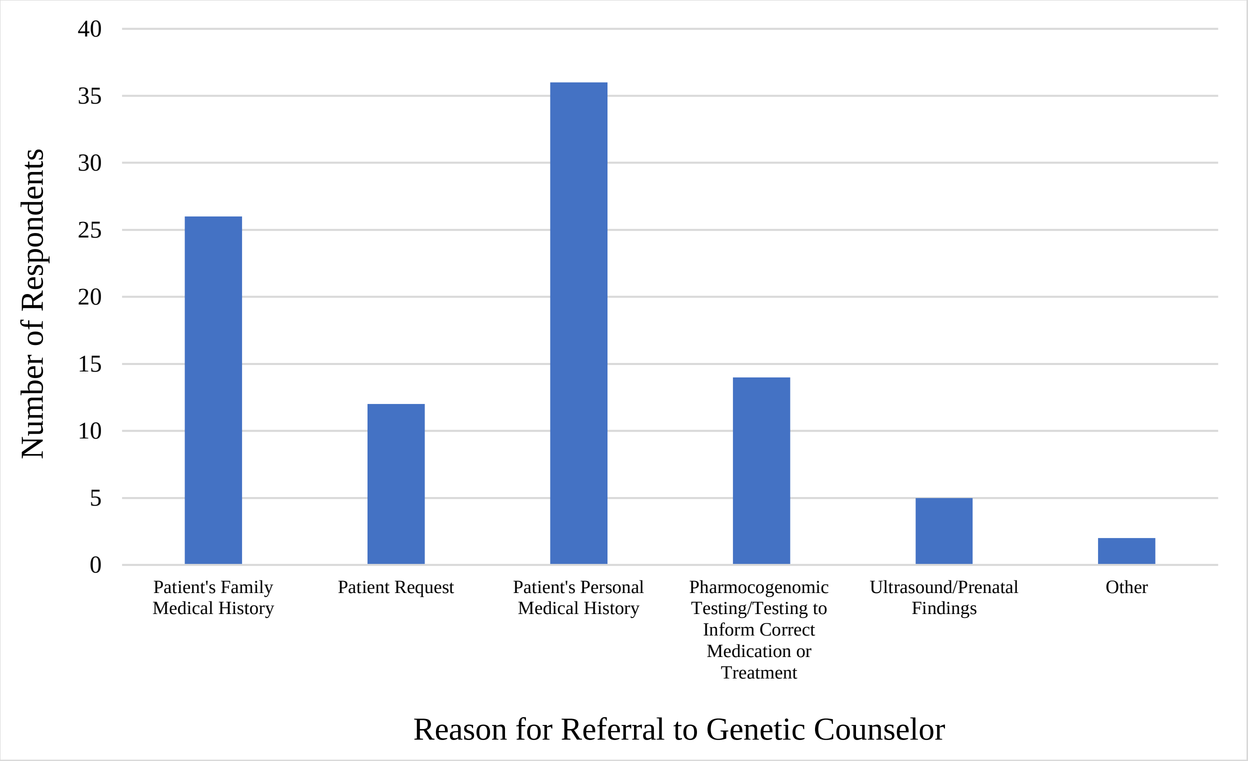


Figure 5: Reason for referral to genetic counseling

2.3.1.4 Facilitation of Care by Genetic Counselors

Respondents were asked how genetic counselors facilitate or could facilitate care for their patients, with answers for this question directly relating to the Pennsylvania licensure for a genetic counselor's scope of practice. The most frequently chosen response was 'explaining genetic test results to patients and their families' and 'explaining the risk of a genetic condition and recurrence risk for other family members', as seen in Table 1. Providers could choose multiple options for this question.

Table 1: Respondents choices for facilitation of care provided by genetic counselors

(Providers could choose multiple options; percentages do not add to 100%)

Facilitation of Care	Percentage (n=49)
Providing Psychosocial Care	51.02%
Providing accurate family history assessment to determine genetic risk factors	89.80%
Applying their expert knowledge of genetics and genomics	89.80%
Providing patients with information to make the best decision for themselves	85.71%
Helping coordinate multidiscplinary care	59.18%
Explaining the risk of a genetic condition and recurrence risk for other family members	93.88%
Explaining management options after an abnormal result	89.90%
Explaining genetic test results to patients and their families	93.88%
Other	4.08%

Some of the ‘other’ responses provided included ‘coordinating cascade testing’, ‘NBS’ (newborn screening), ‘helping with health insurance approval/coverage for genetic testing’ and ‘I don’t know what genetic counselors do’.

Providers who chose ‘genetic counselors can help coordinate multidisciplinary care’, were more likely to have worked one-on-one with a genetic counselor (p-value: 0.001). There was also a significant association between providers who had referred to a genetic counselor anytime in the last six months and those providers who thought that genetic counselors explain genetic and recurrence risk to patients (p-value: 0.016).

Respondents who had interacted with a genetic counselor in a meeting, were significantly more likely to say that genetic counselors ‘provide patients with information to make the best decision for themselves’ (p-value: 0.032). There is also a significant association between the specialty of the provider and the respondent choosing that genetic counselors help coordinate multidisciplinary care (p-value: 0.045). Providers who chose that a patient's personal medical history is a reason that they refer patients to genetic counselors, were likely to choose that genetic counselors explain genetic testing results (p-value: 0.012).

2.3.1.5 Education Requirements for Practicing Genetic Counselors

Respondents were asked what they believed the required minimum education level is for practicing genetic counselors. 81.63% of respondents answered that genetic counselors needed a master's degree to practice, with 10.20% responding that it is a bachelor's degree, 4.08% chose a medical degree (MD) and 4.08% chose a PhD.

A significant association was found between the required education level for a genetic counselor and those who had interacted with a genetic counselor by referring to them (p-value: 0.046). Respondents who indicated that they were a doctor (had an MD, DO and/or PhD degree) were significantly more likely to choose the correct education level for a genetic counselor (p-value: 0.004). As can be seen in Figure 6, these individuals were 12.89 times more likely to choose that a genetic counselor's required level of education is a master's degree, compared to those respondents who indicated they were not a doctor. A significant association was also found between providers who correctly chose a genetic counselor's degree and those who believed that genetic counselors could facilitate care for patients through explanation of genetic test results to patients and families (p-value: 0.018), and between those who correctly chose degree and who chose 'genetic counselors explain risk of a genetic condition and recurrence risk' (p-value: 0.018).

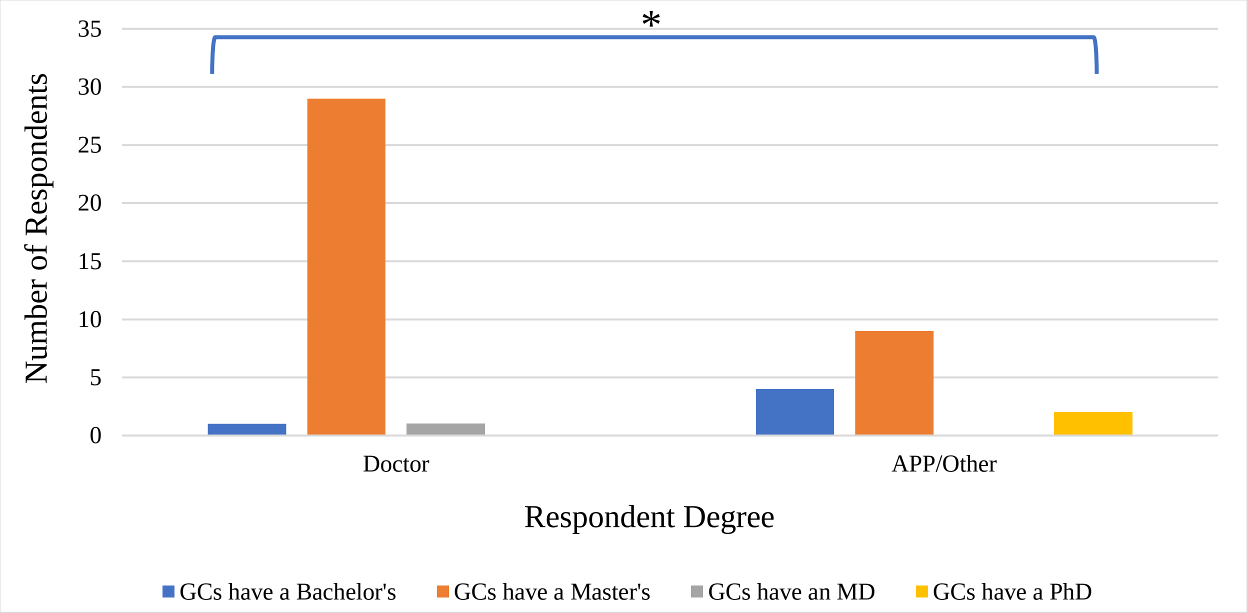


Figure 6: Respondent degree by type of degree a genetic counselor holds

(n=49; p-value: 0.004)

2.3.1.6 Respondents Understanding of Genetic Counselors' Scope of Practice

Understanding of genetic counselors' scope of practice was evaluated through eight questions. Six questions came directly from the Pennsylvania genetic counseling legislation (Medical Practice Act of 1985 - Regulation of Genetic Counselors, 2011), and two were incorrect statements. The first incorrect statement asked about the genetic counselor's ability to perform physical examinations, diagnose and treat patients, with the second asking about a genetic counselor's ability to be the ordering provider of genetic testing. The researchers did not want the statement about the ordering provider of genetic testing to be the only incorrect statement, and therefore thought it was important to have a second incorrect statement in the response set.

All respondents answered that they believed that the explanation of genetics, genetic testing and risk is part of genetic counselors' scope of practice, as can be seen in Figure 7.

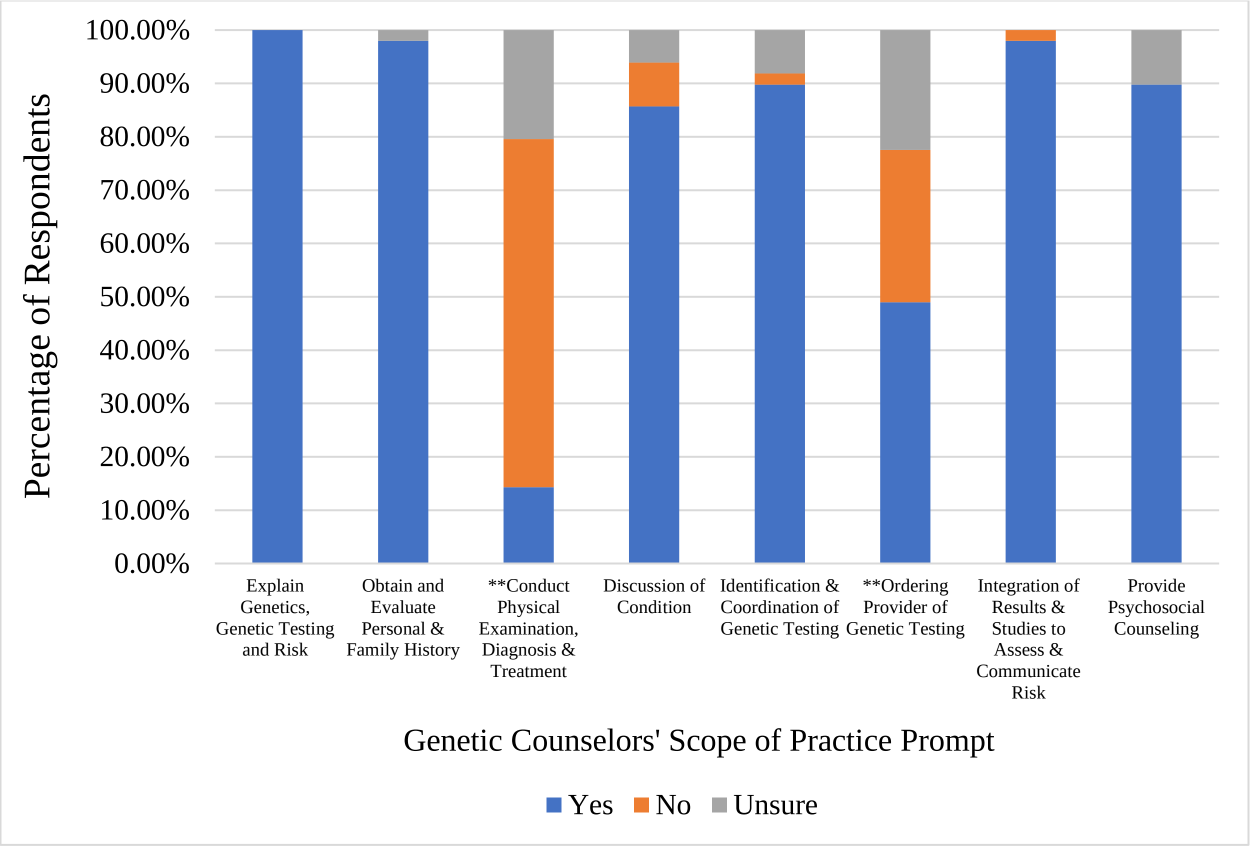


Figure 7: Responses to genetic counseling scope of practice questions

Questions denoted with ** are the incorrect statements. (n=49)

There was a significant association between how frequently providers refer to genetic counselors and their understanding that genetic counselors can identify and coordinate genetic testing (p-value: 0.005), as well as between referral frequency and respondents understanding of whether genetic counselors can order genetic testing (p-value: 0.027). Figure 8 shows that providers who referred weekly to genetic counselors were seven times more likely to say that genetic counselors cannot be the ordering provider, when compared to those who refer monthly.

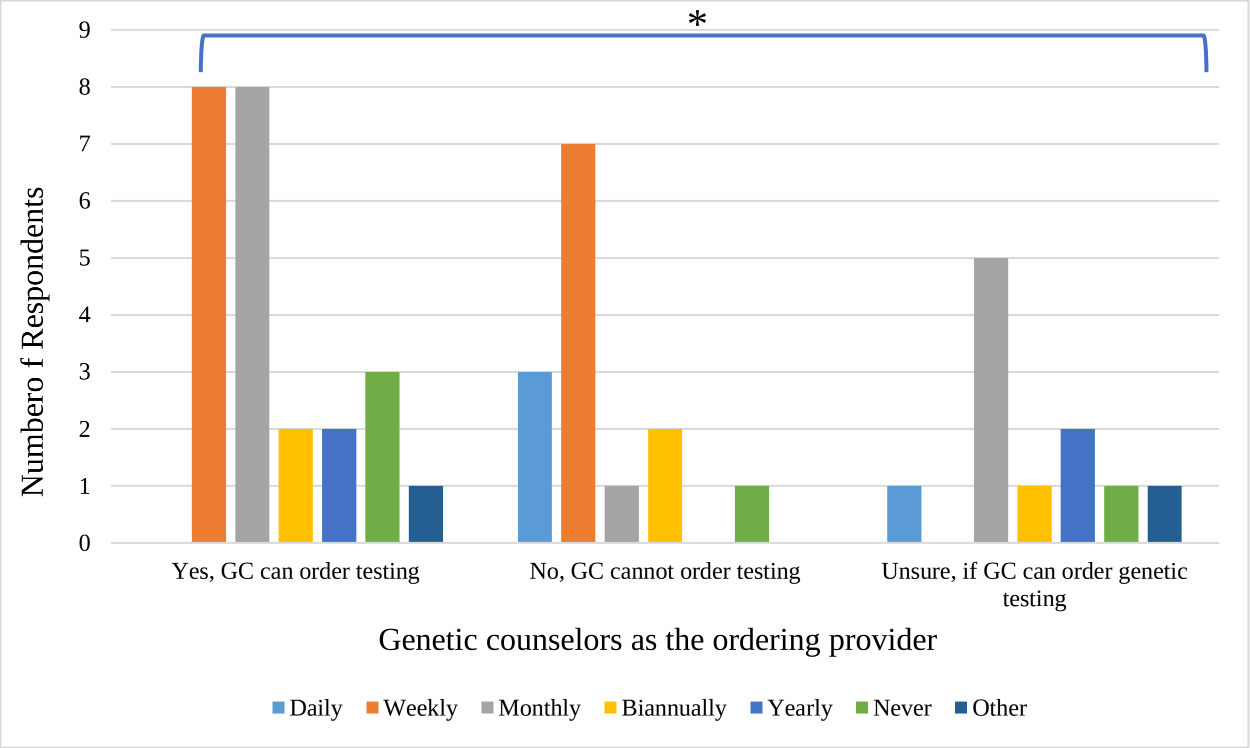


Figure 8: Genetic counselors as the ordering provider by referral frequency (n=49)

There was no significant association between time spent practicing in Pennsylvania and knowing whether genetic counselors can be the ordering provider of genetic testing. However, a significant association was found between total length of time practicing and knowing if genetic counselors can be the ordering provider for testing (p-value: 0.042). As seen in Table 2, the odds of a provider who had been practicing for 11 years or more answering that genetic counselors cannot order genetic testing, was 4.375 times greater than those who had been practicing for 10 years or less.

Table 2: Respondent practicing time by rdering provider of genetic testing

(p-value: 0.042; n=45)

Practicing Time for Respondent	Can Genetic Counselors Order Genetic Testing as the Ordering Provider?		
	Yes	No	Unsure
≤ 10 Years	14	4	2
11+ Years	8	10	7

A significant association was found between time spent practicing in Pennsylvania and whether respondents thought genetic counselors' scope of practice includes conducting physical examinations, diagnosing, and treating patients (p-value: 0.016). Figure 10 shows that respondents who had been practicing for 10 years or less, were 4.67 times more likely to answer 'No' compared to those who had been practicing for 11 years or more.

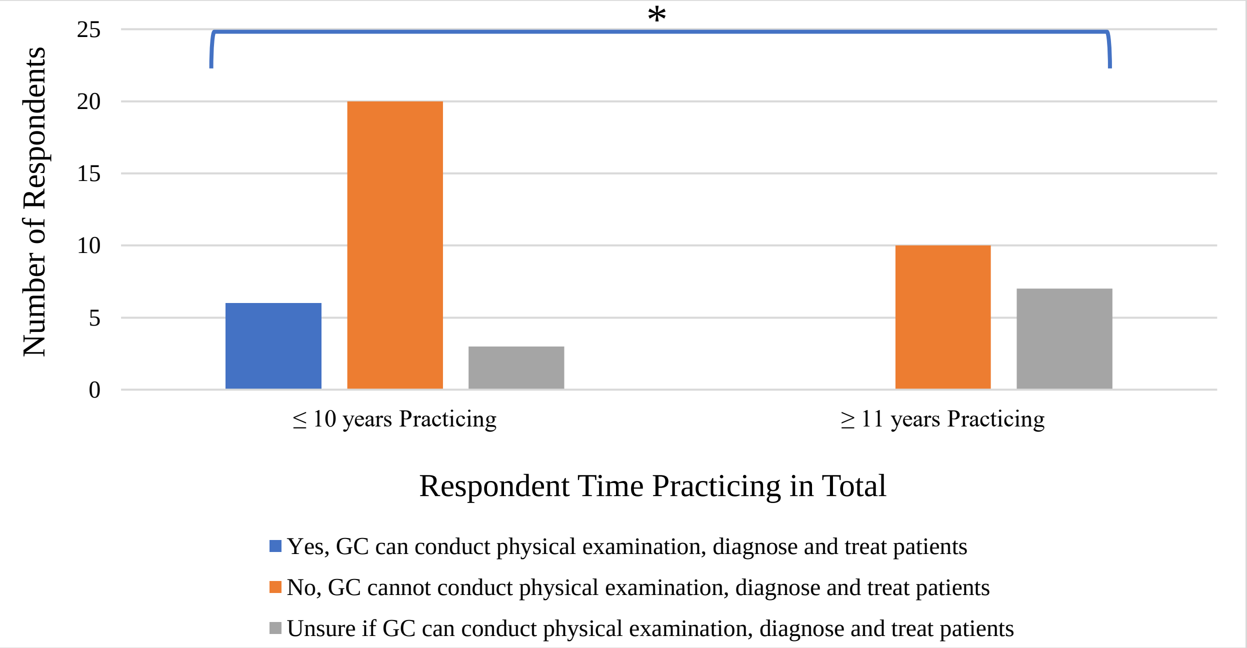


Figure 9: Time spent practicing in Pennsylvania by if genetic counselors can conduct physical examinations, diagnose and treat patients

(n=45)

When comparing respondent choices within the scope of practice questions, a significant association was found between those who chose that a genetic counselor's scope of practice includes both 'evaluating a patient or family's response to the risk of a genetic condition or the risk of recurrence and provide patient-centered psychosocial counseling' and 'discussing the features, natural history, diagnosis means and management of genetic conditions' (p-value: 0.035). The odds of providers answering yes to both questions was 26 times greater than not answering yes to both questions.

The only association found between scope of practice and genetic counselors' facilitation of care for patients, was between the variables 'discuss the features, natural history, diagnosis means and management of genetic conditions' and 'providing patients with the information to make the best decision for themselves' (p-value: 0.027). The odds of a respondent choosing both these answers was 3.167 times greater than choosing 'No' for either question, as seen in Figure 10.

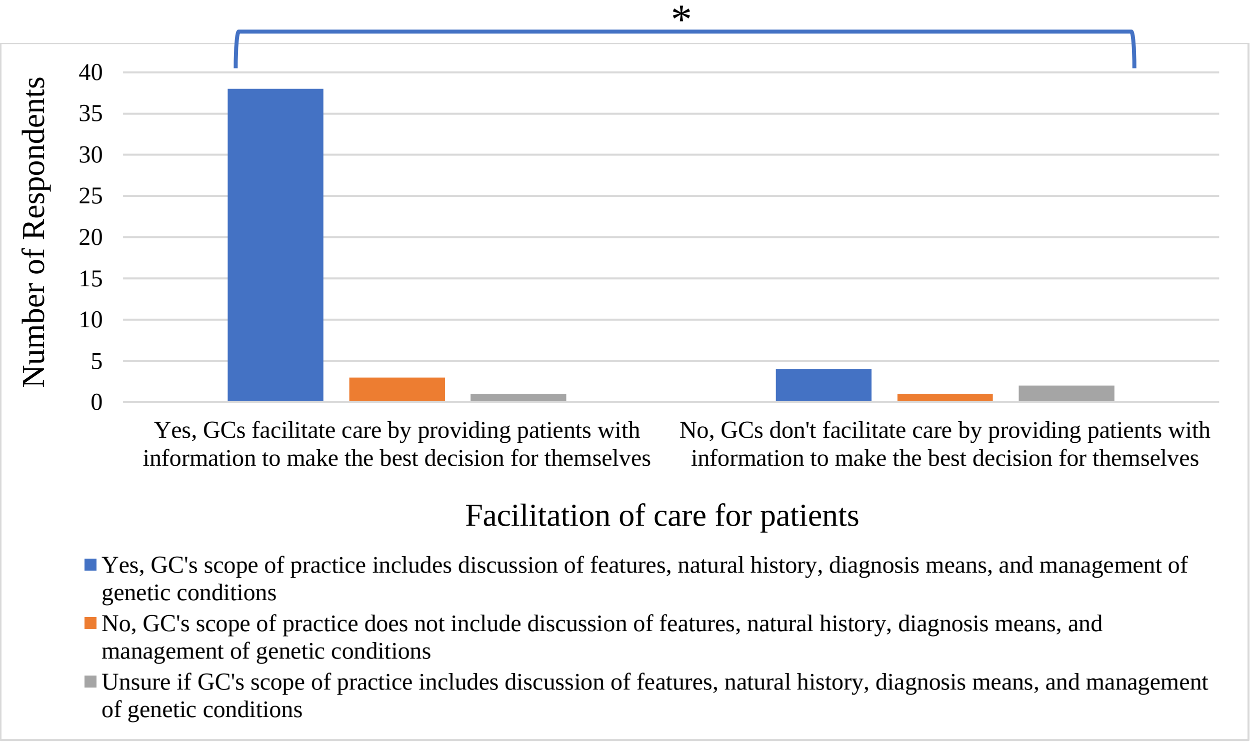


Figure 10: Scope of practice includes 'discussion of features, natural history, diagnosis means, and management of genetic conditions' by facilitation of care for patients includes' providing patients with information to make the best decision for themselves'

(n=49)

2.3.2 Interview Results

Out of the 49 survey respondents, 17 indicated they would be interested in participating in a thirty-minute interview, and seven individuals opened or filled out the secondary survey. Three interviews were scheduled and two were completed. The four individuals who expressed interest but did not book an appointment were contacted via email by the primary researcher, but none replied. There was one participant in interview one and two participants in interview two, for three interviewees in total. They will be referred to as Participant One, Two and Three. For interview two, Participant Two answered most of the questions, with Participant Three adding to the conversation when asked if they had anything additional to contribute.

Through thematic analysis, two themes were created from the eighteen codes generated in the analysis of the interview transcripts. Each theme with its definition is provided in Table 2 and are fully described in the following sections. The codebook is in Appendix G.

Table 3: Themes and their definitions

Engagement with genetic counselors' drives understanding, awareness, and impact of genetic counseling
Definition: Engaging with genetic counselors increases providers' awareness and understanding of the genetic counseling process, which in turn increases providers' understanding of the importance of genetic counseling.
Interaction with genetic counselors creates confidence and positive regard
Definition: Participants' confidence and positive regard toward genetic counselors increases with interaction.

2.3.2.1 Interview Participant Demographics

Interview participants were asked about their background and current professional role. Participant One has been a CRNP in a Gynecology clinic at UPMC MWH for the past four years, and Participants Two and Three have been working in sonography and sonography leadership positions for approximately 25 years each at UPMC MWH. Participant Three also had approximately nine years of radiology technology experience prior to becoming a sonographer.

When asked about how often they refer to genetic counselors, Participant One said they referred at least once or twice a week, but that the frequency could be variable depending on the patients they saw. This is the main way that Participant One interacts with genetic counselors, on referral basis and reading notes from genetic counselors after a patient has followed through on a referral. Participants Two and Three don't personally refer, but the ultrasound department refers to the UPMC prenatal genetic counselors when an anomaly is found on prenatal ultrasound, and they are the individuals who help patients to schedule these appointments. Participant Two and Three's interaction with genetic counselors are mostly to do with scheduling. However, Participant Two goes to the UPMC MWH prenatal genetic counselors' clinical case meetings on a weekly basis.

Regardless of referral frequency, all participants demonstrated awareness and knowledge about the kinds of reasons to refer to genetic counselors. Participant One showed a thorough understanding of the types of reproductive cancers that they should refer patients for, including those with early age of onset, ovarian cancer, and male breast cancer. This participant said that they mostly gained this knowledge from a previous mentor in their first CRNP position and that they had continued asking 'red flag' questions during patient intakes since that position. When asked about prenatal referrals, Participant One said that they do not see obstetric patients but would feel comfortable referring for prenatal reasons if it came up during an appointment. Participants Two and Three were in the unique position that all patients they interacted with were being referred to genetic counseling due to an anomaly found on a prenatal ultrasound.

2.3.2.2 Participants' Understanding of Genetic Counselors' Multi-Faceted Scope of Practice

Genetic counselors' scope of practice extends across a wide range of topics, most of which were included in the survey. In the interview, target questions and examples were utilized to elicit participants' understanding of a genetic counselor's scope of practice and how important they believed this was to the provider's practice. When given four specific examples, all participants agreed that these examples were beneficial to their practice and for patients (see Appendix G for full examples given). All participants also spoke organically about these topics, as well as other topics about genetic counselor scope of practice throughout the interviews.

Day-to-day genetic counseling scope of practice was frequently mentioned by all three participants, with their detail and understanding varying depending on their level of interactions with genetic counselors, and the field in which they were practicing. When asked about what they saw genetic counselors doing on a daily basis, Participant One said:

> *'Recommending screening, giving people probably risks and benefits of screening, and then probably getting the results back, interpreting those results to say 'hey this is what you need to do next'.'* (Participant One)

Participant Two's understanding was broader and included scope of practice work beyond direct clinical interactions with patients:

> *'They're either counseling prior to, you know, ultrasound or testing or additional testing or they're counseling after the ultrasound results have been given... give them additional resources... do some pre-authorizations and they talk about different options with them'* (Participant Two)

Psychosocial support was also independently mentioned by both Participants One and Two, with Participant One focusing on validation of family history:

> *'They're also on the end of reassurance too...Yes, this cancer is in your family. And yes, it made an impact and it doesn't increase your risk of any of that kind of cancer'* (Participant One)

Participant Two spoke about genetic counselors getting to know their patients:

> *'They're also doing a little bit of social like psychology with them too... they'll say, 'AB presented with J, her husband, who they've been together for however long'...'* (Participant Two)

The utilization of genetic information and genetic counselors was spoken about by both Participants One and Two. Participant Two spoke about the utilization of genetic counseling for prenatal diagnosis and helping patients to understand and conceptualize all the information they have been given. Participant One had a broader view in terms of follow up and using genetic information to help guide a patient's future management:

> *'We're making sure we're seeing people appropriately with a little closer follow up. ...it's all part of making sure that we're keeping people healthy and well with the information we have, right, like we didn't have all this genetic stuff a longtime ago. But we have it now. So, we should utilize it for people.'* (Participant One)

The involvement of a genetic counselor on multidisciplinary teams helps to ensure that patients have access to genetic information that can impact the management of their health. Participant One mentioned referral to the high-risk breast cancer clinic at the Hillman Cancer Center:

> *'So, some people... already come to me with the BRCA gene [pathogenic variant]. And so, then I like send them to the high risk cancer clinic, which also includes genetic counselors I think.'* (Participant One)

Participant Two talked about the interdisciplinary work that occurs between the genetics and obstetrical ultrasound departments at MWH:

> *‘Our doctors refer that explanation to the counselors. They’ll explain the ultrasound findings and then say, ‘the genetic counselors will help you put all this together’.’* (Participant Two)

Participant One outlined how a multidisciplinary approach can help to ensure that patients are receiving care appropriately according to clinical practice guidelines:

> *‘Yeah, I think, I mean obviously it's another piece of the puzzle of like how we're making sure that we're screening people appropriately. ...I'm the one physically collecting the pap smear, and doing the breast exam, and ordering the mammogram. But from the genetic side of things where I don't have that expertise and training and knowledge to say, hey, if this is part of it, allowing that to be part of it too and I will say, yeah, like I get, when I get feedback from the higher risk folks or like I'm sometimes the one like I'm ordering this six month mammogram and then high risk is ordering the MRI on the other side.’* (Participant One)

Continuing education is also a vital part of a genetic counselor’s scope of practice due to the ever-evolving field of genetics. Participant Two explicitly talked about this during their interview:

> *‘I think it’s also continuing education with them, because they’re constantly, you know, genetics is changing... I know they have content like CME credits too, but it’s really you know, their expertise is just growing rapidly. They constantly have to stay on top and they also comment on articles they’ve read most recently.’* (Participant Two)

When asked about formal training that genetic counselors must receive to be able to practice, all three participants answered with a master's degree. However, there was hesitation in how the question was answered:

> *'From listening to them give their presentation, I feel like they almost have to have a master's degree, but I don't know that for sure.'* (Participant Two)

> *'I think they do [have a master's degree].'* (Participant Three)

In fact, part of Participant Two's motivation for scheduling an interview, was so they could learn about genetic counselors' training.

Participant One had some prior exposure to the kind of degree that a genetic counselor needs to practice, but still expressed uncertainty when they gave their answer.

> *'I know this because I had a roommate or a hallmate in college who went on to be a genetic counselor. So, I think it's a master's, right?'* (Participant One)

Regardless of exposure to, or interaction or engagement with genetic counselors, all three participants showed uncertainty about the entry level degree for genetic counselors.

2.3.2.2.1 Ordering Provider of Genetic Testing as a Genetic Counselor in Pennsylvania

Interview participants were explicitly asked if they thought that genetic counselors could order genetic testing in Pennsylvania. However, Participant Two brought up the topic prior to the question being asked:

> *'The only piece that I'm really familiar with, with scope of practice, is that they, from my understanding, they cannot write an order for a patient. I*

> *mean they can create the order, but an ordering physician has to sign it.'* (Participant Two)

Participant Three also mentioned that they knew about this aspect of the Pennsylvania licensure scope of practice. When Participant One was asked about who can order genetic testing in Pennsylvania, they considered their professional interactions with genetic counselors to answer the question:

> *'I think, I'm just going back on like the visit summaries I've gotten. I think it's like a counselor that order the test, right? I know they probably work with a physician, but can the counselor actually order the test? I don't know.'* (Participant One)

When pressed for an answer, they said:

> *'I'll say my final answer will be that the genetic counselor orders the test.'* (Participant One)

With Pennsylvania being the only State to officially have this provision, participants were asked if they believe genetic testing should be able to be ordered by Pennsylvanian genetic counselors. All participants agreed that genetic counselors are the appropriate providers to be ordering genetic testing:

> *'It sounds dumb to me, to be honest. ...I think we should really rely on the provider with the level of expertise and training to order appropriate testing... I would expect the genetic counselor to be able to order that [genetic testing] and I think that would be appropriate.'* (Participant One)

Participant Two also believed that genetic counselors are the appropriate provider to order genetic testing:

> *'I think they should be able to order genetic testing'* (Participant Two)

When Participant One was asked how comfortable they are with interpreting genetic testing, they said:

> *'Not super comfortable at all. This is why I typically tell patients that I defer to genetics for this reason... Obviously there's certain ones like Lynch syndrome stuff that I'm aware of, but at baseline, like if someone came to me and said they had some obscure genetic mutation, I would say, 'hey it's worth your time to talk to a genetic counselor'.'* (Participant One)

This quote shows the confidence that Participant One has in referring to genetic counselors and their recognition that genetic counselors can help patients understand complex genetic variations.

2.3.2.3 Theme: Engagement with Genetic Counselors Drives Understanding, Awareness, and Impact of Genetic Counseling

The types of engagement with genetic counselors appeared to shape participants' understanding and awareness of genetic counselors and the impact that genetic counselors can have on patients. Participants were directly asked about their interactions with genetic counselors, which often elicited examples of not only engagement and interaction, but also participants' awareness and understanding of different facets of a genetic counselor's practice. Participant One's engagement with genetic counselors was usually unidirectional and indirect:

> *‘Honestly, my interaction with genetic counselors is always on the back end... I’ll put the referral in, have them see somebody, and some of the ones at UPMC, I get, like a little, like, summary when someone has seen them. So, it’s usually like a nice little note about like what they talked about, what they’re planning as far as testing and sometimes for people it even includes like... oh this is what we recommend for like future screening’* (Participant One)

Participant Two had more direct, bi-directional exposure to genetic counselors and shared her experiences:

> *‘I do attend their clinical meetings, but not their business ones, just to listen to the students, and you know they do presentations... Fellows and the students do presentations that I find really interesting...I would say I interact with the genetics department in some way close to a daily basis. But often that’s just a matter of scheduling. You know, trying to accommodate and schedule patients. More from a management side and less from individual cases...’* (Participant Two)

Participant Three had similar interactions with genetic counseling:

> *‘I see them all on the same interactions as well [as Participant Two], as well as like scheduling patients, rescheduling patients, like doing that side of the clinic as well sometimes’* (Participant Three)

Later in the interview, Participant Three said that they do not attend the clinical meetings that Participant Two does.

However, since Participant Two attends clinical meetings with prenatal genetic counselors and has frequent professional interactions with genetic counselors, they demonstrated a deeper understanding and awareness of what occurred during an appointment including the educational aspect of genetic counseling:

> *'I mostly think they educate...they're letting the couple know what all the options are, talking them through, answering their questions. They're also doing a little bit of social like psychology with them too... I hear them present in their presentations... Here's what their pedigree looks and based on all of these other things, this is what our recommendation like to get blood work or whatever. ...They're kind of empowering, just letting people understand what all their options are.'* (Participant Two)

This understanding for Participant Two also extended to the psychosocial elements of a genetic counseling appointment:

> *'...Somehow, they sit there, and they ask the probing questions in a way that causes that patient to reflect... or to do some testing to say, you know, they can recognize these things and say we're gonna test for something because we think there may be a genetic thing going on here.'* (Participant Two)

Participant One had a good understanding of the importance of a family history for not only referral reasons, but also for accuracy during the genetic counseling appointment. They shared:

> *'I have my patients like, do a little homework on the front end before they see you guys, to say 'hey you know, talk to people in your family. If you want Mom on the Zoom call or whatever, like that's fine'.'* (Participant One)

Both Participants One and Two talked about the importance of genetic counseling for patients. When giving examples of why genetic counseling is important, both Participants One and Two described the critical aspect of anticipatory guidance provided by genetic counselors.

> *'...I had a patient more recently who knew she had BRCA [pathogenic variant], and I believe she got incidentally diagnosed with like DCIS, but like, so she's already had, you know, genetics follow through. So, I think it made her experience with pursuing kind of the oncology side of things a little bit better because she had kind of been preparing for it'* (Participant One)

Participant Two spoke of the importance of the patients understanding the testing and the importance of the consent process:

> *'Our patients don't understand what testing they're having done. If it's invasive, you know, like CVS or amniocentesis, or noninvasive with the first trimester screen. They need to understand the test before we do their exam. So, I do think it's important for them to have counseling, and it's important afterward too. So, they understand that, you know, ultrasound is not... doesn't diagnose anything and they need to understand the difference.'* (Participant Two)

Participant Two also spoke about the genetic counselor's role in tying together all the important medical information, including family history, ultrasound results, and genetic testing:

> *'I don't think that's something that their ordering physician is capable of doing. It's not their specialty. You know that they depend on it [genetic counseling] too. So, we need that person in between to put it all together'* (Participant Two)

Overall, Participants One and Two's comments suggest they have a solid understanding of the genetic counseling process and what occurs in appointments as well as a belief that genetic counseling is beneficial for patients.

2.3.2.4 Theme: Interaction with Genetic Counselors Creates Confidence and Positive Regard

Overwhelming, all participants were extremely pleased with their interactions with genetic counselors and the positive experiences that they have had with the profession. These interactions, although different, led to similar feelings toward the profession overall. For example, Participant One said:

> *'I very much appreciate the circling back and the 'hey this is what we're planning on doing', because in some scenarios it has made a big difference... I definitely see that there's benefit to you guys in more than that [referring to reproductive cancers]... So yeah, I appreciate you guys. You're wonderful.'* (Participant One)

Participant Two had made many positive statements about genetic counselors. This included when they were speaking about the amount of knowledge that a genetic counselor has:

> *'They really have a lot of knowledge, so, I mean it seems like it's as strong as at a doctor's level... Their expertise is just growing rapidly'* (Participant Two)

> *'I really do respect their knowledge, and am blown away most of the time by their presentations'* (Participant Two)

When asked to share why they participated in an interview, Participant One said:

> *'I've had great experience with genetic counselors, particularly in this job. So, I wanted to support you guys too and share my positive experience because they, I mean, they honestly just have been positive, like there's never been a time where a genetic counselor has wronged me or anything... I also wanted to share the positive feedback too.'* (Participant One)

Overall from these interviews, it can be seen that genetic counselors appear to foster a positive work environment and culture.

2.4 Discussion

2.4.1 Provider Interactions with and Referrals to Genetic Counselors

This research assessed the frequency and types of interactions healthcare providers had with genetic counselors. Findings showed that 84% of respondents had interacted with a genetic counselor at some point in their career, with 76% having referred to a genetic counselor at least once every six months. These findings are different to previous literature that showed that 72% of

Texas physicians had never or rarely referred to genetic counselors (Diamonstein et al., 2018). However, the referral frequency from this study is similar to rates published by Truong et al. (2021), in which 89% of PCPs surveyed said that they had referred to a genetic counselor at least once, with a median frequency of three referrals per year.

Based on a small sample size, these findings include encouragingly high rates of referral to genetic counselors, with the main reasons for referral being personal and/or family histories. Although previous research has found that PCPs are unclear on when family history is an indication for a genetic counselor referral (Baldwin et al., 2014; Truong et al., 2021), survey respondents in this study primarily were specialists who could be more likely to understand the familial reasons to refer a patient, compared to a generalist PCP (Selkirk et al., 2013). This makes it difficult to compare this research to previous literature.

The high rates of genetic counselor interaction and referral in this study could also be influenced by the fact that this research was conducted at two hospitals with long-standing on-site genetic counseling programs, and that providers surveyed may be more likely to see patients with an indication for genetic counseling referral. Interview Participant One's and Two's answers reflected this as both worked in specialist fields. Participant One, who works in a gynecology office, was asking patients daily for family history of cancers and referring as required based on the patients' responses. Participant Two, who worked in the obstetrical and gynecological ultrasound department knew that finding anomalies on prenatal ultrasound prompted referral to a genetic counselor.

The nine respondents who had rarely or never referred to a genetic counselor mostly cited that it was not within their scope of practice to do so. Three respondents chose from the listed options, which included patients not understanding the information received during a genetic counseling appointment, and their patient population not being suitable for genetic testing. It is reassuring that only three of the nine respondents chose these options, as the options were derived from well-documented provider biases that pose barriers to patients receiving care that they require (Hauser et al., 2018; Mikat-Stevens et al., 2015). Genetic counselors are trained to educate patients of all levels of health literacy and obtain informed consent for any genetic testing. Therefore, concern regarding patient understanding should not be a barrier to a healthcare provider referring to genetic counselors. Patients have also demonstrated interest in genetic counseling and testing when given information about why they might be referred (Desrosiers et al., 2019; Evenson et al., 2016). Given that Participant Two reported that they often interfacing directly with patients to make genetic counseling appointments, this experience seemed to allow them to provide some anticipatory guidance about what the visit would look like. This kind of anticipatory guidance has been shown to help patient understanding of what genetic counseling may involve and how it could help patients and families (Riesgraf et al., 2015).

Almost two thirds of respondents had interacted with a genetic counselor by referring to them, and over half had worked on a multidisciplinary team that included a genetic counselor. A significant association was found between provider specialty and interaction with a genetic counselor in a multidisciplinary clinic. These data suggest that specialty providers are perhaps more likely to interact with a genetic counselor in a multidisciplinary clinic, potentially due to the higher likelihood of genetic diagnosis in these specialties. Catapano and colleagues (2022) showed that working closely with genetic counselors increases the providers' awareness of and collaboration with genetic counselors. This research may overrepresent providers who interact with genetic counselors in a multidisciplinary team environment, due to the high level of Pediatric, Obstetrician/Gynecologist and Neonatology providers that responded to the survey and the setting of the study.

The significance of the association between those respondents who had interacted with a genetic counselor in a meeting, and those who chose that genetic counselors can help to provide patients with information to make the best decision for themselves, a deeper understanding of genetic counselor scope may be related to the types of interactions respondents had with genetic counselors. Participants One and Two both demonstrated a good understanding of genetic counseling, but they had different types of interactions with genetic counselors, and each showed differing levels of understanding of a genetic counselor's scope of practice. Participant Two had more direct interactions with genetic counselors, attending weekly clinical meetings with genetic counselors, and their comments reflected a more nuanced understanding of what genetic counselors do beyond the activities outlined in the scope of practice. This is compared with Participant One, whose knowledge of genetic counselors was based on referrals and reading patient notes, which is a more indirect form of interaction with genetic counselors. Participant One's comments suggest that they did not interact with a genetic counselor in a direct way.

Although based on only one interview, Participant Two's experience might suggest that direct contact with genetic counselors, such as attending genetic counseling clinical meetings, is a valuable educational tool for healthcare providers to gain a deeper understanding and appreciation of genetic counselors' expertise and the work they do with patients. Providers who have not been in a meeting with a genetic counselor may not be as aware of these nuances of a genetic counselor's scope of practice, especially if they are only referring to genetic counselors and may therefore have no direct conversations with them.

Interestingly, there were no statistical associations found between the types of interactions that providers had with genetic counselors, and the reasons why providers referred patients. This result suggests that the type of interaction that a provider has with a genetic counselor does not influence the reason why healthcare providers are referring patients to genetic counselors.

Overall, Participants One and Two were both willing to share the positive experiences they have had with genetic counselors, which is consistent with the overwhelmingly positive experience of genetic counselor integration into the multidisciplinary team that previous research has found (Vito et al., 2022).

2.4.2 Facilitation of Care for Patients

Encouragingly, the most frequently chosen responses for facilitation of care for patients were 'explaining the risk of a genetic condition and recurrence risk for other family members' and 'explaining genetic test results to patients and their families' (both 94%). Participants One and Two also spoke about these aspects of patient care, without being prompted, showing that providers understand these services provided by genetic counselors. These findings are concordant with previous research from Europe that ranked similar options highly (Catapano et al., 2022). Both this study and the European study (Catapano et al., 2022) found that coordination of care was not ranked highly.

Facilitation of psychosocial care was the least chosen option (51%), which may indicate that this is not a primary reason that providers refer to genetic counselors, or that perhaps providers see this as part of their own practice (Prochniak et al. 2012). This is in agreement with several studies that showed that psychosocial support was not a highly important consideration for providers when referring to a genetic counselor (Catapano et al., 2022; Cordier et al., 2016), or that it was a shared role for providers in a multidisciplinary team (Hudson et al., 2019).

When Participant One was asked about genetic counselors providing psychosocial care, they said:

> *'It's kind of a lot on someone to go for genetic counseling and what that means. So, I'm sure there is a decent amount of psychosocial stuff going on there'* (Participant One)

It is possible that psychosocial counseling was something this participant had not considered in relation to genetic counseling previously. Previous research has shown that physicians who refer to genetic counselors are more likely to refer to a genetic counselor when they understand the value of a genetic counseling appointment, which includes the unique ability of genetic counselors to address psychosocial issues (Prochniak et al., 2012).

The association between referring patients for personal history and facilitation of care through the explanation of test results is unsurprising. Participant One spoke about this in their interview and outlined that they feel much more comfortable referring to a genetic counselor for the interpretation of results for patients. Research has shown that providers are concerned about interpreting genetic test results themselves (Evenson et al., 2016; Pet et al., 2019; Selkirk et al., 2013), thus Participant One is not unique in feeling uncomfortable in interpreting results. Certain specialties have shown more confidence in their level of interpreting negative and positive results, but are still uncertain as to how to interpret variants of uncertain significance (Liang et al., 2018).

2.4.3 Understanding and Perceptions of Scope of Practice

This research study predominately focused on genetic counselor scope of practice, with a particular interest in whether providers were aware of the Pennsylvanian licensure provision which does not allow genetic counselors to order genetic testing (Medical Practice Act of 1985 - Regulation of Genetic Counselors, 2011). From the primary researcher's understanding, this is the first study of Pennsylvanian providers' understanding about genetic counselors' scope of practice within the state.

Among the scope of practice questions, the question about ordering tests had the least agreement between respondents. It does not appear that providers have a clear understanding as to whether Pennsylvanian genetic counselors can order genetic testing as part of their scope of practice; only two factors from the survey were found to be significantly associated with the correct answer: frequency of referral and providers who had been practicing in Pennsylvania for 11 years or longer.

Providers who had been practicing in Pennsylvania 11 years or longer, were approximately four times more likely to know that genetic counselors cannot order genetic testing. This could be attributed to the fact that these providers were in Pennsylvania when the licensure legislation was passed in 2011, and therefore remember this nuance from the time of the legislation. It could also be that they have been working in Pennsylvania long enough to have encountered and learned about this provision within their own practice. This was encountered during the interviews; Participants Two and Three, who have been practicing in Pennsylvania for over 25 years, understood this limitation of the scope of practice for genetic counselors, compared to Participant One, a CRNP of four years, who incorrectly said that genetic counselors could order genetic testing. Participant two spoke about processes that were created in the light of the new legislation in 2011, which shows that each hospital has had its own way of dealing with the fact that Pennsylvanian genetic counselors cannot order genetic testing.

The frequency of referral to genetic counselors also appears to be associated with understanding that genetic counselors in Pennsylvania cannot be ordering providers of genetic testing. This is perhaps not surprising as these providers are likely receiving referral information back from genetic counselors and, therefore, may have a better understanding of this issue. There may also be a greater chance for the referring provider to view the order for genetic testing in the electronic medical record, noting who signed off on it.

Survey respondents who identified as doctors were more likely than APPs and other providers to correctly identify a genetic counselor's level of formal training. However, all three interview participants used the qualifier 'I think' when speaking about the entry level degree for genetic counselors. It could be that doctors were more likely to correctly identify the level of education that a genetic counselor had, knowing that genetic counseling is not a specialty that medical doctors can pursue in residency. Given this, it shows that there is still work to be done in promoting the field and educating other providers about the training that is required to become a genetic counselor, especially to those who do not have much exposure to the profession.

For all other questions regarding scope of practice, there appears to be a good understanding among study participants and there were no associations across the provider's degree type, specialty, and total time spent in practice.

Although there were no associations found between scope of practice and interactions of providers with genetic counselors, the interviews provided insight into the kind of understanding that providers have about genetic counselors and how they develop this understanding. It was evident from the interviews, that a provider can gain a good understanding of the genetic counseling scope of practice even from reading referral notes and interacting in indirect ways with a genetic counselor, such as the experience of Participant One. However, Participant Two's direct contact and engagement with genetic counselors shows that there are effective ways in which to gain deeper understanding of a genetic counselor's scope of practice.

The significant association found between the identification and coordination of genetic testing, and frequency of referral to genetic counselors, shows that providers value the ability of genetic counselors to undertake this task when referring.

When comparing scope of practice answers with facilitation of care responses, one significant association was found between 'providing patients with the information to make the best decision for themselves' and 'discussing the features, natural history, diagnosis means, and management of genetic conditions'. This significant association may highlight understanding that respondents have about the work that genetic counselors do to ensure that patients have all the information they need to be able to make the best decision for themselves and their family. Interview Participant Two also pointed out the relationship between these two variables, when she spoke about genetic counselors educating patients and giving them all their options to make the best decision for themselves.

2.4.4 Study Limitations and Future Directions

This study provided new and encouraging evidence regarding healthcare providers' perspectives on genetic counselors facilitating patient care and scope of practice in Pennsylvania. However, due to the small number of the survey responses and interviews conducted, there are some limitations that need to be considered. The small size of the survey and interview responses limits the statistical power and generalizability of the study. Additionally, although the survey captured a wide range of specialties, the sample was not representative of the entire healthcare community. The small distribution footprint across UPMC MWH and UPMC CHP meant that only certain types of providers would receive the survey invitation. In future studies, it would be helpful to have a broader distribution across hospitals in the UPMC system and beyond, including providers in family medicine clinics and private practice to increase response rates to a survey and the variety of participants' practice setting.

Based on answers provided, it seems that selection bias is possible since approximately 84% of participants had interacted with a genetic counselor in the past. Those who had not interacted with or referred to a genetic counselor previously, or perhaps did not know what a genetic counselor is, may have been less inclined to fill out the survey compared to those who had. Both UPMC MWH and UPMC CHP have longstanding genetic counseling units embedded within the hospital, which means that there could have been a sample bias by surveying and interviewing providers that have direct access to genetic counselors within the hospital which they work. To help overcome this limitation in the future, survey recruitment could be broadened to target those who have not referred previously, or to have two recruiting arms targeting both those who have and have not referred to genetic counselors. Expanding beyond UPMC MWH and UPMC CHP would also help to increase the diversity of healthcare providers among the respondents.

These limitations extend to the interviews conducted, whereby all three participants have had positive experiences with genetic counselors in their practice, which likely led to a selection bias of those who were interested in participating in an interview. Due to the primary researcher's role as a genetic counseling student, it may have been likely that participants did not feel comfortable sharing negative experiences with genetic counselors. In the future, additional interviews, focus groups, or a neutral interviewer could help to remove this limitation. It is also recognized that patient-facing healthcare providers are busy professionals, who may not have time to complete an interview.

Due to the brief nature of the survey, there were limitations in assessing why providers chose the answers that they did. Utilizing a Likert scale in future studies to assess the confidence level of the provider responding could be an informative addition, particularly in eliciting provider knowledge about genetic counselor educational degree, in which providers showed significant uncertainty in the interview portion of this research. Asking more nuanced questions with additional text boxes to allow respondents to explain why answers were chosen could also allow a deeper view into providers' perspectives about genetic counselors' scope of practice and facilitation of care for patients, providing supplementation for future interviews.

A future direction from this research could be the incorporation of healthcare providers into genetic counseling clinical meetings and case conferences or having genetic counselors attend multidisciplinary meetings to present cases (e.g., tumor boards, department meetings). Interview Participant Two's experience with attending these meetings appeared to significantly enhance their understanding of and respect for genetic counselors. And although it is not generalizable due to only being one data point, it would be worth investigating this kind of exposure to the genetic counseling field in the future. A question on a future survey could be to ask if respondents have been able to access these types of meetings to help enhance their understanding of the genetic counselor's profession. This type of engagement with genetic counselors could also be applied in future studies to assess understanding of a genetic counselor's scope of practice before and after attending a certain number of genetic counselor clinical meetings.

2.5 Conclusion

This study aimed to assess healthcare providers' perspectives about genetic counselors' ability to facilitate care for patients, and their understanding of Pennsylvanian scope of practice for genetic counselors. Data from 49 survey respondents and three interview participants found that most providers have a good understanding of the types of care that genetic counselors can provide for a patient, and of what is included in the scope of practice for genetic counselors in Pennsylvania. Analysis showed that there was no significant association between specialty or degree of the provider, and their understanding of genetic counseling scope of practice. The analysis of a provider's understanding of whether genetic counselors can order genetic testing in Pennsylvania found that those who refer more frequently are more likely to understand that genetic counselors cannot order genetic testing. Interviews with three participants showed that there is support for genetic counselors to be able to order their own genetic testing, as the provider with the expertise in the field. Providers appear to value and understand the care that genetic counselors can facilitate for patients. However, this understanding appears to be dependent on the type of interaction had with a genetic counselor. The novel data in this research is valuable to understanding how the genetic counseling practice is viewed by our colleagues and is important to understand as the genetic counseling field continues to expand.

3.0 Research Significance to Genetic Counseling and Public Health

To the knowledge of the author, there is no published research that has examined other healthcare providers' perceptions of genetic counselors' explicit scope of practice. This study was not meant to be a comprehensive assessment of this topic, but rather a starting point in beginning to understand how healthcare providers view genetic counselors, their scope of practice and their facilitation of care for patients.

The aim of integrating interviews into the study design was to gain a deeper understanding of the answers provided in the survey; as was hoped, the interviews did provide greater insight, particularly into the nuances of providers' understanding of scope of practice and interactions they have with genetic counselors, including how this can vary from provider to provider. Previously, much of this information has been understood anecdotally, talked about among genetic counselors at work or a professional conference, or maybe genetic counselors had a hunch about providers' perception of scope of practice and facilitation of care. To have these topics formally documented is the first step in being able to further understand them.

It is important for genetic counselors to recognize that not every provider is going to know what services genetic counselors can provide, or what expertise they bring to a multidisciplinary team, as highlighted by those survey respondents who did not refer to or know about genetic counselors. This is one of the challenges of being a relatively new healthcare profession. These survey responses suggest that genetic counselors and those who do understand the role a genetic counselor can play in patient care, should strive to advocate for the profession. Participant Two's direct exposure to genetic counselors through attending weekly clinical case meetings, highlights one way of exposing those outside the genetics field is an opportunity to showcase to the multifaceted work of a genetic counselor. The same interview participant also highlighted utilizing presentations by genetic counselors for the sonography department to help with multidisciplinary knowledge sharing between departments, which could also be a way to spread accurate information about genetic counselors.

Understanding what healthcare providers' perceptions of genetic counselors are, and what they can do within their scope of practice, is especially important for being able to accurately educate healthcare providers about genetic counseling services. This research showed that providers are unaware or unsure whether genetic counselors in Pennsylvania can order genetic testing. Interview Participant One thought that genetic counselors could order genetic testing, whereas Participants Two and Three definitively knew that they could not, due to processes created to work around this provision, thus reflecting the confusion demonstrated by survey respondents. Anecdotally we know that these processes exist in numerous forms around the state of Pennsylvania, which can lead to patients being lost to follow up, time being added to the genetic testing process and creation of disparity in who is easily able to access genetic testing which is a public health concern. Enabling equitable access to healthcare for all is one of the ten essential public health services (EPHS) according to the Center for Disease Control and Prevention (CDC), and utilizing legal and regulatory actions, another EPHS, is key to changing this issue.

Amending the Pennsylvanian genetic counselor licensure is the only way to allow genetic counselors to order genetic testing and to address this barrier in access to genetic testing. The Pennsylvania Association of Genetic Counselors (PAGC) is currently deciding whether to introduce another bill to the Pennsylvanian senate, after having a number of bills expire for this amendment, with the latest bill expiration occurring in the 2021-2022 session. This research, although limited, shows that there is support from healthcare providers for genetic counselors to be an ordering provider of genetic testing. Both Participants One and Two outlined that the genetic counselor is the appropriate provider to be ordering genetic testing.

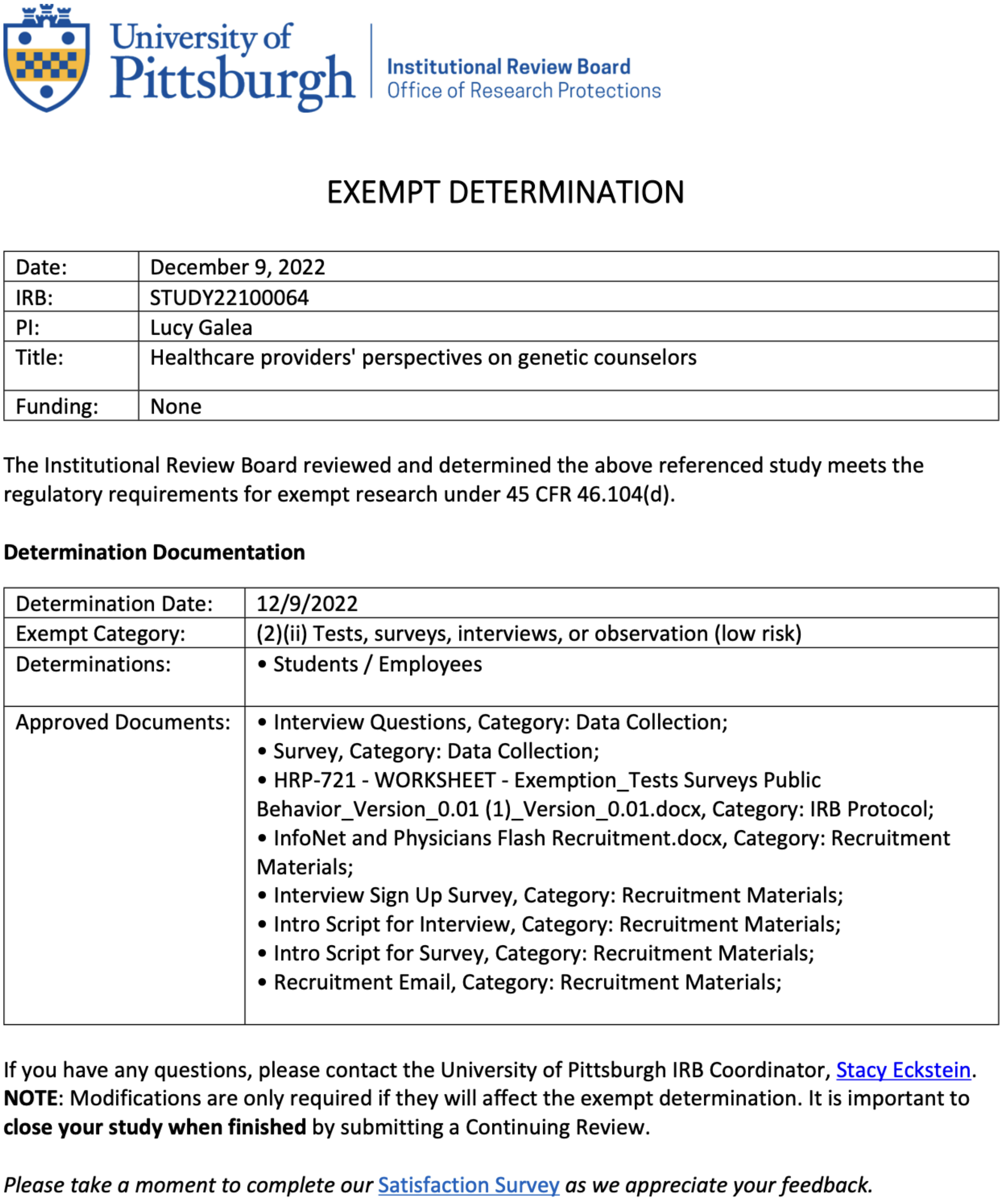
Until changes to licensure happens, it is important to educate healthcare providers about who can order genetic testing and reinforce the importance of genetic counselors as part of a patient's medical team. As part of educational efforts, it would be valuable to emphasize the Pennsylvanian genetic counselor scope of practice, including the ability to identify and coordinate genetic testing, and what kinds of care a genetic counselor can bring, to ensure that providers do not get discouraged from referring patients to genetic counselors due to the limitation of test ordering.

The survey and interview responses in this study provide an encouraging view into the integration of genetic counselors, with a demonstrated high rate of interaction through multidisciplinary teams. By having better communication between providers and expanding unified, multidisciplinary care for patients, we can better support patients over the long term, especially those with rare disease. Offering this type of care for patients is important considering that 1 in 10 individuals in the United States live with a rare disease (GARD, n.d.), and 80% of these conditions have a genetic component to them (NHGRI, 2018).

These types of teams directly contribute to public health by helping to show innovative ways to improve, strengthen, and support healthcare partnerships. This type of care typically involves fewer individual appointments for the patient by seeing all their providers in one multi-hour appointment, thus reducing travel and necessary lost workdays, and enables equitable care for all patients, thus meeting the core function of the EPHS. Other ways that this can impact public health include more unified cancer screening for patients with a cancer predisposition syndrome (such as the National Cancer Consortium Network guidelines for cancer predisposition syndromes), and appropriate guidelines for providers to follow when a genetic cause of disease may be suspected for pediatric and prenatal patients.

It is encouraging to find that most providers understand genetic counselors' scope of practice. It is reasonable to expect that the greater the number of healthcare providers that know about genetic counselors, the greater the number of patients who will be referred to a genetic counselor or who will not miss out on critical genetics care due to a healthcare provider not knowing that these services exist. Being knowledgeable about genetic counselors and their scope of practice is especially important when serving diverse populations, as it has been well documented that there are implicit biases against minority populations of patients that disproportionately affect the rates of referral for genetic counseling (Ademuyiwa et al., 2021; Peterson et al., 2020; Muller et al., 2018). Understanding what a genetic counselor can do for a patient is the first step in being able to equitably refer patients to a genetic counselor.

Appendix A IRB Approval



University of Pittsburgh
Institutional Review Board
Office of Research Protections

EXEMPT DETERMINATION

Date:	December 9, 2022
IRB:	STUDY22100064
PI:	Lucy Galea
Title:	Healthcare providers' perspectives on genetic counselors
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:	12/9/2022
Exempt Category:	(2)(ii) Tests, surveys, interviews, or observation (low risk)
Determinations:	• Students / Employees
Approved Documents:	• Interview Questions, Category: Data Collection; • Survey, Category: Data Collection; • HRP-721 - WORKSHEET - Exemption_Tests Surveys Public Behavior_Version_0.01 (1)_Version_0.01.docx, Category: IRB Protocol; • InfoNet and Physicians Flash Recruitment.docx, Category: Recruitment Materials; • Interview Sign Up Survey, Category: Recruitment Materials; • Intro Script for Interview, Category: Recruitment Materials; • Intro Script for Survey, Category: Recruitment Materials; • Recruitment Email, Category: Recruitment Materials;

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 B Recruitment Email

Hello,
You are invited to participate in the following research study:

Healthcare providers perspectives on genetic counselors.

All healthcare providers *who are not genetic counselors* are eligible to participate in this study. This includes physicians, residents, physician assistants, and nurses.
Participants will be asked seven questions about genetic counselors' function in facilitating care for patients and scope of practice in Pennsylvania. This short survey is estimated to take between five and seven minutes.

Optional 30 minute interviews will also be used to further understand this topic. If you wish to participate in an interview, you will be prompted to a secondary survey after completion of the initial survey.

Your participation in this study is voluntary and you can withdraw from this study at any time by closing the browser tab that this study is open in. If you choose not to participate in this study, there will be no effect on your relationship with the University of Pittsburgh, or the University of Pittsburgh Medical Center (UPMC). There are no direct benefits to you for completing this study. The only potential risk anticipated from participating in this survey is the infrequent risk of a breach of confidentiality. 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.

This study is being conducted by Lucy Galea (lug22@pitt.edu), who is a University of Pittsburgh genetic counseling student. This study has been approved by the Institutional Review Board at the University of Pittsburgh.

If you would like to participate in this study, please follow this link.
Kind regards,

Appendix C ‘InfoNet’ Intranet, ‘Physicians Flash’ and ‘Extra’ Recruitment

Do you provide direct patient care? Click HERE to participate in a brief survey about healthcare providers’ perceptions about genetic counselors!

Appendix D Main Survey Content

Thank you for your interest in this research study.

The purpose of this research study is to investigate the perspectives and understanding non-genetics providers have about genetic counselors.

We will be asking seven questions about non-genetics provider's understanding of genetic counselors' function in facilitating care for patients, and their understanding of genetic counselors' scope of practice in Pennsylvania. This will be facilitated through the following survey as well as optional interviews to further understand this topic.

It is anticipated that this survey will take between 5 and 7 minutes.

If you wish to participate in a thirty minute optional interview, you will be prompted to complete a secondary survey to consent to enter your details and book a scheduled interview time. This secondary survey will keep your original survey answers anonymous and confidential.

Your participation in this survey is voluntary and you can withdraw from this study at any time by closing the browser tab that this study is open in. If you choose not to participate in this study, there will be no effect on your relationship with the University of Pittsburgh, or the University of Pittsburgh Medical Center (UPMC). There are no direct benefits to you for completing this study.

The only risk anticipated from participating in this survey is the infrequent risk of a breach of confidentiality. This survey will collect no participant identifiers as a means to minimize the risk of a breach of confidentiality. 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.

This study is being conducted by Lucy Galea, who can be contacted at lug22@pitt.edu.
If you would like to participate in this study, please click the arrow below to proceed.

Q1 Have you ever worked or interacted with a genetic counselor?

○ Yes

○ No

○ Unsure

Q2 How have you interacted with a genetic counselor? (Choose all that apply)

☐ Regularly work with them one-on-one

☐ Work with them on a multidisciplinary team

☐ Have been in a meeting with them

☐ Had annual education/workshop on genetic counseling

☐ Have referred to a genetic counselor

☐ Have never interacted with a genetic counselor

☐ Other: ____

Q3 On average, how often do you refer patients for genetic counseling?

○ Daily

○ Weekly

○ Monthly

○ Once every six months

○ Once a year

○ Never

○ Other: ____

Q3 part 2 If you are rarely or never referring to genetic counselors, what reasons or barriers are stopping you from referring?

☐ Can provide genetic counseling myself

☐ Unsure if insurance would cover genetic testing for my patients

☐ Unsure if my patient population would be suitable for genetic testing

☐ Information would be too complicated for my patients

☐ Unsure if it would be beneficial for my patients

☐ Did not know that genetic counselors were available

☐ Not easy to find genetic counselors for referral on the EHR (eg Epic, Cerner)

☐ Other: ____

Q4 Which of the following most often prompts you to refer a patient for genetic counseling? (select all that apply)

☐ Patient's family medical history

☐ Patient request

☐ Patient's personal medical history

☐ Pharmacogenomic testing/Testing to inform correct medication or treatment

☐ Other: ____

Q4a If you were to refer to a genetic counselor, which of the following would prompt you to do so? (select all that apply)

☐ Patient's family medical history

☐ Patient request

☐ Patient's personal medical history

☐ Pharmacogenomic testing/Testing to inform correct medication or treatment

☐ Other: ____

Q5 Complete the sentence: Genetic counselors facilitate care for my patients by... (select all that apply)

- ☐ Providing psychosocial care
- ☐ Providing accurate family history assessment to determine genetic risk factors
- ☐ Applying their expert knowledge of genetics and genomics
- ☐ Providing patients with the information to make the best decision for themselves
- ☐ Helping coordinate multidisciplinary care
- ☐ Explaining the risk of a genetic condition and recurrence risk for other family members
- ☐ Explaining management options after an abnormal result (e.g. positive genetic test result, high risk prenatal screening result, variant of uncertain significance on genetic testing)
- ☐ Explaining genetic test results to patients and their families
- ☐ Other: ____

Q5a Complete the sentence: Genetic counselors could facilitate care for my patients by... (select all that apply)

- ☐ Providing psychosocial care

☐ Providing accurate family history assessment to determine genetic risk factors

☐ Applying their expert knowledge of genetics and genomics

☐ Providing patients with the information to make the best decision for themselves

☐ Helping coordinate multidisciplinary care

☐ Explaining the risk of a genetic condition and recurrence risk for other family members

☐ Explaining management options after an abnormal result (e.g. positive genetic test result, high risk prenatal screening result, variant of uncertain significance on genetic testing)

☐ Explaining genetic test results to patients and their families

☐ Other: ____

Q6 What level of education do practicing genetic counselors need?

○Bachelors

○Masters

○PhD

○MD

Q7 Which of the following roles are included in the scope of practice for genetic counselors in Pennsylvania?
a. Explain genetics, genetic testing, and genetic risk to patients

○Yes

○No

○Unsure

Q8 *b. Obtain and evaluate personal and family medical histories to determine genetic risk for genetic conditions.*

○Yes

○No

○Unsure

Q9 *c. Conduct physical examination, diagnosis and treatment of patients.*

○Yes

○No

○Unsure

Q10 *d. Discuss the features, natural history, diagnosis means and management of genetic conditions.*

○Yes

○No

○Unsure

Q11 *e. Identify and coordinate appropriate genetic testing including prior authorization.*

○Yes

○No

○Unsure

Q12 *f. Order genetic testing for patients considering their personal and family history as the ordering provider.*

○Yes

○No

○Unsure

Q13 *g. Integrate genetic testing results and other diagnostic studies with personal and family history to assess risk factors for genetic conditions and communicate these risk facts as well as explain the clinical implications of genetic test results.*

○Yes

○No

○Unsure

Q14 *h. Evaluate a patient or family's response to the risk of a genetic condition or the risk of recurrence and provide patient-centered psychosocial counseling*

○ Yes

○ No

○ Unsure

Q15 Thank you for your answers. Please answer the following demographics: Qualification (or degree):

☐ MD

☐ DO

☐ PA-C

☐ CRNP

☐ PhD

☐ Other:

Q16 Main Specialty:

○ Oncology

○ Obstetrics/Gynecology

○ Pediatrics

○ Internal Medicine

○ Family Medicine

○ Other: ____

Q17 How long have you been practicing? (please include your residency if an MD/DO)

○ 1-2 years

○ 3-5 years

○ 6-10 years

○ 11-20 years

○ 21+ years

Q18 How long have you been practicing in Pennsylvania? (please include your residency if done in Pennsylvania)

○ 1-2 years

○ 3-5 years

○ 6-10 years

○ 11-20 years

○ 21+ years

Q19 What is your primary institutional setting?

☐ Private practice

☐ University medical center

☐ Private hospital/medical facility

☐ VA System

☐ Other: ____

Q20 For this research, interviews are being conducted in addition to this survey. Would you be interested in participating in a 30 minute interview on your perspectives of genetic counselors?

○ Yes

○ No

Appendix E Interview Interest Survey

Thank you for your further interest in this research study.

The purpose of this research study is to investigate healthcare providers' awareness and perception of genetic counselors.

The interview will be asking participants approximately fifteen questions about non-genetics provider's understanding of genetic counselors' training, their function in providing care for patients and the scope of practice for genetic counselors in Pennsylvania.

This interview is expected to take approximately thirty minutes. Your interview will be recorded with your permission, and you can revoke recording rights at any time. Recordings and transcription of this interview will be stored secured and without identifiers to minimize the risk of a breach of confidentiality.

Your participation in this interview is voluntary and you can withdraw from this study at any time by cancelling your interview appointment via Calendly, by reaching out to Lucy Galea or by terminating the interview at any time. If you choose not to participate in this study, there will be no effect on your relationship with the University of Pittsburgh, or the University of Pittsburgh Medical Center (UPMC).

There are no direct benefits to you for completing this study. The potential risks anticipated from participating in this survey include the risk of a breach of confidentiality. Every effort has and will be made to maintain your confidentiality before, during and, after interviews have taken place. 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.

This study is being conducted by Lucy Galea, who can be contacted at lug22@pitt.edu.

If you would like to participate in the interview section of the study, please click the arrow below to proceed to registering your interest.

Q1 Thank you for considering participating in an interview for this research!

Interviews can take place in person, over the phone or on a video call. Interviews will be recorded with your permission. Available dates and time can be seen in the Calendly link below. If you have any further questions about this part of the research study or cannot find a time and date that suits you, please reach out to Lucy Galea at lug22@pitt.edu.

Q2 Name

____

Q3 Email Address

____

Q4 Please use this link to book your research interview.

Thank you for again for being a part of this study.

Appendix F Interview Questions

I will be recording this interview, do I have your permission to do so?

Tell me about yourself/your profession.

Demographic Questions (ask directly if not given above):

1. What is your profession?
2. What is your clinical specialty?
3. How long have you been practicing (including residency, if applicable)?
4. How long have you been practicing in the state of Pennsylvania?
5. What institutional setting do you work in? (e.g., academic medical center, private practice, VA)

Interview

1. Can you share with me the nature of the interactions you've had with genetic counselors?
2. How often do you have these kinds of interactions?
3. What do you see genetic counselors doing on a daily basis?
    a. Or could be 'what do you think genetic counselors do on a daily basis' depending on their answer to the first question.
4. What is your understanding of the training that is required to become a practicing genetic counselor?
    a. Probe further if the answer warrants it.
5. From your understanding, what are the requirements for genetic counselors to obtain and then maintain licensure in Pennsylvania?
6. What is your understanding of genetic counselors' scope of practice in Pennsylvania?
    a. Follow up question: Do you know where to find information on genetic counselors' scope of practice?
7. Which of the following genetic counselors' scope of practice do you find beneficial to your practice? Feel free to elaborate on each answer as you feel necessary.

    Examples to give interviewee:
    a. Accurate evaluation of family history and determination of whether genetic testing would be warranted
    b. Interpretation and integration of test results in relation to a patient's personal and family history
        i. Follow up question: How comfortable do you feel doing genetic testing result interpretation yourself?
    c. Risk counseling in relation to a patient's personal and family history
    d. Psychosocial counseling and resource identification
8. What is your understanding of genetic counselors' ability to be the ordering provider on a genetic test requisition?

   a. Follow up: Genetic counselors are not able to be the ordering provider for genetic tests in Pennsylvania; what do you think about this nuance of genetic counselors' scope of practice in Pennsylvania?

9. Can you share with me some of the reasons why you refer your patients to a genetic counselor?
   a. Probe further if the answer warrants it.
10. How do you see a genetic counselor's role in a patient's multidisciplinary team?
11. What do you think a genetic counselor is able to bring to your patients' care?
12. What have you found surprising in your work with genetic counselors?
13. Have you found anything unexpected in your interactions with genetic counselors?
14. Any other comments about genetics counselors that you wish to add to this interview?

Ending question: Can you share with me what motivated you to participate in this interview?

Appendix G Code Book

Appendix Table 1

Code	Definition	Examples
Positive regard for genetic counselors	Positive comments about or interactions with genetic counselors	‘It’s always been positive, which is good’ (P1) ‘I definitely see that there’s benefit to you guys [in more than a reproductive cancer way]... I appreciate you guys. You’re wonderful’ (P1) ‘There’s never been a time when a genetic counselor wronged me’ (P1) ‘Definitely empowerment, knowledge, a sense of control, a sense of calmness’ (P2) ‘I really do respect their knowledge and am blown away most of the time by their presentations’ (P2) ‘They’re kind of empowering’ (P2) ‘They really have a lot of knowledge, I mean, it seems like it’s as strong as at a doctor level’ (P2)
Confidence in genetic counselors	Statements of confidence toward genetic counselors’ abilities	‘I would definitely refer to them in that case to, you know, determine if testing would be necessary’ (P1) ‘I think we should really rely on the provider with the level of expertise and training to order appropriate testing’ (P1)
Interaction with genetic counselor	Any kind of interaction that the participant mentioned having with a genetic counselor	‘My interactions with genetic counselors is always on the back end... like a little summary when someone has seen them. Usually it’s a nice little note about what they talked about, what they’re planning as far as testing’ (P1) ‘Scheduling patients, rescheduling patients, like doing that side of the clinical as well sometimes’ (P2) ‘I would say I interact with the genetics department in some way close to a daily basis. But often that’s just a matter of umm scheduling. You know, trying to accommodate and schedule umm patients. More from a management side and less from uh, individual cases? You know, getting deep into the cases, not so much.’ (P2) ‘See them advocating for patients to get testing like sometimes’ (P2) ‘Scheduling patients, rescheduling of patients, like doing that side of the clinical as well sometimes’ (P3)

Understanding of genetic counseling process	Understanding of what genetic counseling entails and the process that occurs with genetic counseling	‘...they’re letting the couple know what all the options are, talking them through, answering their questions.... I hear them present in their presentations... here’s what their pedigree looks and based on all these other things, this is what our recommendation like to get blood work or whatever’ (P2)
		‘You have to have a physician signing that [genetic testing] order and the same thing with consent for procedures. They can draw up the consent form, explain to the patient what the test is about. But in the end, the doctor has to take responsibility and sign off’ (P2)
Importance of genetic counseling	Statements surround the importance of genetic counseling and the impact of genetic counseling on patients	‘...I had a patient more recently who knew she had BRCA, and I believe she got incidentally diagnosed with like DCIS, but like, so she’s already had, you know, genetics follow through. So I think it made her experience with pursuing kind of the oncology side of things a little bit better because she had kind of been preparing for it’ (P1)
		‘Our patients don’t understand what testing they’re having done. If it’s invasive, you know, like CVS or amniocentesis, or non invasive with the first trimester screen. They need to understand the test before we do their exam. So I do think it’s important for them to have counseling, and it’s important afterward too. So they understand that, you know, ultrasound is not... doesn’t diagnose anything and they need to understand the difference’ (P2)
		‘[on putting together all the different pieces including family history, genetic testing, and ultrasound] I don’t think that’s something that their order physician is capable of doing. It’s not their specialty. You know that they depend on it too. So we need that person in between to put it all together’ (P2)
Awareness of genetic counseling appointment happenings/ recommendations	Awareness of what happens in a genetic counseling appointment and the types of recommendations that genetic counselors make for patients	‘Breast MRI every six months and a mammogram at an earlier age’ (P1)
		‘I try and have my patient’s like, do a little homework on the front end before they see you guys to say ‘hey you know, talk to people in your family if you want mom on the zoom call or whatever, like, that’s fine’ (P1)
		‘They’re either counseling prior to, you know, ultrasound or testing or additional testing or they’re counseling after the ultrasound results have been given... give them additional resources... do some pre-authorizations and they talk about different options with them’ (P2)

		'She had to draw a pedigree. She had to draw it on a poster board. It was massive there were so many family members that were afflicted with the same disorder. It was remarkable' (P2)
Ordering of genetic testing	Statements around the ordering of genetic testing and who should be ordering genetic testing	'...my final answer will be that the genetic counselor orders the test' (P1) 'It sounds dumb to me, to be honest... I think we should really rely on the provider with the level of expertise and training to order appropriate testing' (in response to being asked how the participant felt about GCs not being able to order testing in PA) (P1) 'The only piece that I'm really familiar with, with scope of practice, is that they, from my understanding they cannot write an order for a patient. I mean they can create the order, but an ordering physician has to sign it' (P2) 'I think they should be able to order testing, genetic testing' (P2)
Genetic counseling scope of practice	Understanding and mention of the genetic counseling scope of practice that the participant talked about in their own words	'Recommending screening, giving people probably risks and benefits of screening, and then probably getting the results back. Interpreting those results to say, hey, this is what we need to do next' (P1) 'They're either counseling prior to, you know, ultrasound or testing or additional testing or they're counseling after the ultrasound results have been given... give them additional resources... do some pre-authorizations and they talk about different options with them' (P2) 'I think mostly they educate' (P2) 'They constantly have to stay on top and they also comment about different articles they read most recently' (P2)
Genetic counseling psychosocial	Mention of psychosocial counseling that genetic counselors are involved with	'There's also on the end of reassurance too...Yes this cancer is in your family. And yes it made an impact and it doesn't increase your risk of any of that kind of cancer'(P1) 'They're also doing a little bit of social like psychology with them too... they'll say AB presented with J, her husband, who they've been together for however long' (P2)
Utilization of genetic information	Integration and utilization of genetic information after the patient has received genetic test results	'...We didn't have all this genetic stuff a long time ago. But we have it now, So, we should utilize it for people' (P1)
Genetic counseling training	Understanding of the training required of genetic counselors to be a practicing genetic counseling	'So I think it's a master's, right?' (P1) 'I think it's a Master's degree, but I just really don't know' (P2)
Multidisciplinary improvement		'I feel like we're really siloed and I feel like we could really learn from each other....

	Parts of the genetic counseling and other teams' interactions that could be improved.	Understanding going both ways would be helpful' (P2) 'I that we are located in the exact same department and everybody feels, everybody things that we are the same department but really we're not like there's, you can tell that there's separation' (P3)
Awareness of referral indication	Participant awareness of any reason for referral	'They say 'oh yeah my aunt had ovarian cancer when she was like 30'. And I'm like, that's a red flag, you need to see genetics' (P1) 'I find out it was cervical cancer, which is less, you, it's HPV. It's not really genetics' (P1) 'Actually if it's any kind of defect we have them go through genetic counseling' (P2)
Referral indication	A referral indication that the participant said that they make referrals to genetic counselors for	'I'll take a family history and they'll be slightly higher risk and I'll say 'hey, it would be good for you to meet with the genetic counselor' (P1) 'Cancer like breast, colon and ovarian high risk genetic cancers' (P1) '...if we have routine outpatients that come in and we find an anomaly' (P2)
How often participant refers to genetic counselor	How often referral to genetic counselor is made	'At least once or twice a week on average. Sometimes it's more or less...' (P1)
Participant profession/role	Profession and role that the participant takes within their clinic	'Women's health nurse practitioner by training. Currently I work in gynecology and I work with just general Gyne stuff like preventative care, pap smears, all that good stuff' (P1)
	A description about the profession of the participant and the role that they play within the healthcare system. This could also include their degree or training they undertook for the position.	'I am a, by training a sonographer... I've been here for a little over 25 years... I moved into leadership...probably about ten years ago maybe. ...Now I'm the director of ultrasound department.' (P2) 'I'm the manager of the OB Ultrasound department. ...used to be a radiology tech for 20 years of my life and in the middle I went back to school setting for ultrasound... started working at Magee, I think 15 years now I've been there and then got in the leadership role probably 10 years ago' (P3)
Participant/Provider genetics training	General genetics training that the participant has received either in their graduate training or on the job.	'...mostly on the job... I think my program covered it a little bit more than like maybe like a family (medicine) training, just because all my training was was in OBGYN...' (P1) 'I have a good relationship with my old mentor too, so I asked a lot of questions' (P1)
Uncertainty	Uncertainty regarding the topic that the participant was talking about	'I'm guessing, yeah I don't, I don't really know. So I'm guessing they probably pass boards and then they probably have to reup it like every two or three years like we do I'm guessing' (P1)

		‘From listening to them give their presentations, I feel like they almost have to have a master’s degree, but I don’t know for sure’ (P2)

Bibliography

ACGC. (2022). ACGC Program Directory. https://www.gceducation.org/wp-content/uploads/2022/11/ACGC-Program-Directory-22November2022.pdf

Ademuyiwa, F. O., Salyer, P., Tao, Y., Luo, J., Hensing, W. L., Afolalu, A., Peterson, L. L., Weilbaecher, K., Housten, A. J., Baumann, A. A., Desai, M., Jones, S., Linnenbringer, E., Plichta, J., & Bierut, L. (2021). Genetic Counseling and Testing in African American Patients With Breast Cancer: A Nationwide Survey of US Breast Oncologists. Journal of Clinical Oncology, 39(36), 4020–4028. https://doi.org/10.1200/JCO.21.01426

Baldwin, L.-M., Trivers, K. F., Andrilla, C. H. A., Matthews, B., Miller, J. W., Lishner, D. M., & Goff, B. A. (2014). Accuracy of Ovarian and Colon Cancer Risk Assessments by U.S. Physicians. Journal of General Internal Medicine, 29(5), 741–749. https://doi.org/10.1007/s11606-014-2768-2

Braun, V., & Clarke, V. (2006). Using thematic analysis in psychology. Qualitative Research in Psychology, 3, 77–101.

Campbell, C., A. (2022, May 20). Improving Access to Genetic Counselors through the “Access to Genetic Counselor Services Act” [Interview]. https://www.ashg.org/publications-news/ashg-news/improving-access-to-genetic-counselors/

Catapano, F., El Hachmi, M., Ketterer-Heng, N., Renieri, A., Mari, F., Morris, M., & Cordier, C. (2022). The role of the Genetic Counsellor in the multidisciplinary team: The perception of geneticists in Europe. European Journal of Human Genetics, 30(12), Article 12. https://doi.org/10.1038/s41431-022-01189-5

Choi, J. J., Fikre, T., Fischman, A., Buck, A. K., & Ko, N. Y. (2022). The Role of Race and Insurance Status in Access to Genetic Counseling and Testing Among High-Risk Breast Cancer Patients. The Oncologist, 27(10), 832–838. https://doi.org/10.1093/oncolo/oyac132

Clyman, J. C., Nazir, F., Tarolli, S., Black, E., Lombardi, R. Q., & Higgins, J. J. (2007). The impact of a genetics education program on physicians’ knowledge and genetic counseling referral patterns. Medical Teacher, 29(6), 143-e150. https://doi.org/10.1080/01421590701477373

Medical Practice Act of 1985—Regulation of Genetic Counselors, 332, Pennsylvania General Assembly, 2011–125 (2011). https://www.legis.state.pa.us/cfdocs/legis/li/uconsCheck.cfm?yr=2011&sessInd=0&act=125

Cordier, C., Taris, N., Moldovan, R., Sobol, H., & Voelckel, M.-A. (2016). Genetic professionals' views on genetic counsellors: A French survey. Journal of Community Genetics, 7(1), 51–55. https://doi.org/10.1007/s12687-015-0250-4

Czekalski, M. A., Huziak, R. C., Durst, A. L., Taylor, S., & Mai, P. L. (2022). Mainstreaming Genetic Testing for Epithelial Ovarian Cancer by Oncology Providers: A Survey of Current Practice. JCO Precision Oncology, 6, e2100409. https://doi.org/10.1200/PO.21.00409

Desrosiers, L. R., Quinn, E., Cramer, S., & Dobek, W. (2019). Integrating genetic counseling and testing in the pediatric oncology setting: Parental attitudes and influencing factors. Pediatric Blood & Cancer, 66(10), e27907. https://doi.org/10.1002/pbc.27907

Diamonstein, C., Stevens, B., Hashmi, S. S., Refuerzo, J., Sullivan, C., & Hoskovec, J. (2018). Physicians' Awareness and Utilization of Genetic Services in Texas. Journal of Genetic Counseling, 27(4), 968–977. https://doi-org.pitt.idm.oclc.org/10.1007/s10897-017-0199-z

East, K. M., Cochran, M. E., Kelley, W. V., Greve, V., Finnila, C. R., Coleman, T., Jennings, M., Alexander, L., Rahn, E. J., Danila, M. I., Barsh, G., Korf, B., & Cooper, G. (2022). Education and Training of Non-Genetics Providers on the Return of Genome Sequencing Results in a NICU Setting. Journal of Personalized Medicine, 12(3), Article 3. https://doi.org/10.3390/jpm12030405

Evenson, S. A., Hoyme, H. E., Haugen-Rogers, J. E., Larson, E. A., & Puumala, S. E. (2016). Patient and Physician Perceptions of Genetic Testing in Primary Care. South Dakota Medicine: The Journal of the South Dakota State Medical Association, 69(11), 487–493.

GARD. (n.d.). About—GARD. About - Genetic and Rare Diseases Information Center. Retrieved January 12, 2023, from https://rarediseases.info.nih.gov/about

Hajek, C., Hutchinson, A. M., Galbraith, L. N., Green, R. C., Murray, M. F., Petry, N., Preys, C. L., Zawatsky, C. L. B., Zoltick, E. S., Christensen, K. D., Baye, J., Bell, M., Deberg, K., Forred, B., Free, C., Hajek, C., Van Heukelom, J., Hopp, A., Hutchinson, A., … Jamal, L. (2022). Improved provider preparedness through an 8-part genetics and genomic education program. Genetics in Medicine, 24(1), 214–224. https://doi.org/10.1016/j.gim.2021.08.008

Harding, B., Webber, C., Ruhland, L., Dalgarno, N., Armour, C., Birtwhistle, R., Brown, G., Carroll, J., Flavin, M., Phillips, S., & MacKenzie, J. (2019a). Primary care providers' lived experiences of genetics in practice. Journal of Community Genetics, 10(1), 85–93. https://doi.org/10.1007/s12687-018-0364-6

Harding, B., Webber, C., Ruhland, L., Dalgarno, N., Armour, C., Birtwhistle, R., Brown, G., Carroll, J., Flavin, M., Phillips, S., & MacKenzie, J. (2019b). Bridging the gap in genetics: A progressive model for primary to specialist care. BMC Medical Education, 19(1), 195. https://doi.org/10.1186/s12909-019-1622-y

Hauser, D., Obeng, A. O., Fei, K., Ramos, M. A., & Horowitz, C. R. (2018). Views Of Primary Care Providers On Testing Patients For Genetic Risks For Common Chronic Diseases. Health Affairs, 37(5), 793–800. https://doi.org/10.1377/hlthaff.2017.1548

H.R.2144—Access to Genetic Counselor Services Act of 2021, 117th Congress, 2021–2022 (2021) (testimony of Brian Higgins). http://www.congress.gov/

Hudson, P., Zajo, K., Gerhardt, C. A., Stanek, J., & Varga, E. (2019). Defining the role of a genetic counselor within pediatric hematology and oncology comprehensive care teams: Perspectives of the provider team and patients. Journal of Genetic Counseling, 28, 1139–1147.

Hyatt, C., Russo, J., & McDougall, C. (2019). Genetic counseling perspective of engagement with urology and primary care. Cancer Journal of Urology, 26(5 Supp), 52–53.

Liang, M. I., Wong, D. H., Walsh, C. S., Farias-Eisner, R., & Cohen, J. G. (2018). Cancer Genetic Counseling and Testing: Perspectives of Epithelial Ovarian Cancer Patients and Gynecologic Oncology Healthcare Providers. Journal of Genetic Counseling, 27(1), 177–186. https://doi.org/10.1007/s10897-017-0135-2

Linfield, D. T., Rothberg, M. B., Pfoh, E. R., Noss, R., Cassard, L., Powers, J. C., Lipold, L., & Martinez, K. A. (2022). Primary care physician referral practices regarding BRCA1/2 genetic counseling in a major health system. Breast Cancer Research and Treatment, 195(2), 153–160. https://doi.org/10.1007/s10549-022-06523-5

Manrriquez, E., Chapman, J. S., Mak, J., Blanco, A. M., & Chen, L. (2018). Disparities in genetics assessment for women with ovarian cancer: Can we do better? Gynecologic Oncology, 149(1), 84–88. https://doi.org/10.1016/j.ygyno.2017.10.034

Marks, J., H., & Richter, M., L. (1976). The genetic associate: A new health professional. American Journal of Public Health, 66(4), 388–390. https://doi.org/10.2105/AJPH.66.4.388

Massart, M., Berenbrok, L. A., Munro, C., Berman, N. R., & Empey, P. E. (2022). A Multidisciplinary Precision Medicine Service in Primary Care. Annals of Family Medicine, 20(1), 88. https://doi.org/10.1370/afm.2764

Mikat-Stevens, N. A., Larson, I. A., & Tarini, B. A. (2015). Primary-care providers' perceived barriers to integration of genetics services: A systematic review of the literature. Genetics in Medicine: Official Journal of the American College of Medical Genetics, 17(3), 169–176. https://doi.org/10.1038/gim.2014.101

Muller, C., Lee, S. M., Barge, W., Siddique, S. M., Berera, S., Wideroff, G., Tondon, R., Chang, J., Peterson, M., Stoll, J., Katona, B. W., Sussman, D. A., Melson, J., & Kupfer, S. S. (2018). Low Referral Rate for Genetic Testing in Racially and Ethnically Diverse Patients Despite Universal Colorectal Cancer Screening. Clinical Gastroenterology and Hepatology : The Official Clinical Practice Journal of the American Gastroenterological Association, 16(12), 1911-1918.e2. https://doi.org/10.1016/j.cgh.2018.08.038

NHGRI. (2018, April 13). Rare Genetic Diseases. Genome.Gov. https://www.genome.gov/dna-day/15-ways/rare-genetic-diseases

NSGC. (n.d.). State Licensure for Genetic Counselors Model Legislative Provisions. Retrieved June 28, 2022, from https://www.nsgc.org/Policy-Research-and-Publications/State-Licensure-for-Genetic-Counselors/Model-Legislative-Provisions

NSGC. (2021a). Genetic Counselor Workforce. https://www.nsgc.org/Policy-Research-and-Publications/Genetic-Counselor-Workforce

NSGC. (2021b). States that explicitly allow a genetic counselor to order tests. https://www.nsgc.org/Portals/0/ASD%20Cmte%20Resources/Multi-State%20Licensure/States%20that%20explicitly%20allow%20a%20genetic%20counselors%20to%20order%20tests.pdf?ver=qk5JpNM-99mYmmawkH784w%3D%3D#:~:text=Page%201-,States%20that%20explicitly%20allow%20a%20genetic%20counselors%20to%20order,as%20of%20August%202021)%3A&text=Florida%20%E2%80%A2%20Georgia%20%E2%80%A2%20Maryland%20%E2%80%A2%20Montana%20%E2%80%A2%20Indiana%20%E2%80%A2,with%20a%20physician%20collaborative%20agreement.

NSGC. (2022a). 2022 Professional Status Survey: Executive Summary (p. 25).

NSGC. (2022b). 2022 Professional Status Survey: Work Environment (p. 39).

NSGC. (2022c, July). State Licensure Map. https://datawrapper.dwcdn.net/xNc1w/1/

Olsabeck, T., Fritzler, J., Wahl, R., & Anderson, B. (2019, September). Racial Differences in the Utilization of Genetic Counseling Services among Patients. Bureau of Epidemiology and Population Health, Michigan Department of Health and Human Services, Lansing, Michigan.

Parker, J. C., Rangu, S., Grand, K. L., Bhoj, E. J., Castelo-Soccio, L., & Sheppard, S. E. (2021). Genetic skin disorders: The value of a multidisciplinary clinic. American Journal of Medical Genetics, 185(4), 1159–1167. https://doi-org.pitt.idm.oclc.org/10.1002/ajmg.a.62095

Pasca, C., Carroll, R., Cragun, R. T., & Cragun, D. L. (2021). Knowledge and perceptions of the genetic counseling profession among a national cross-sectional sample of U.S. adults. Journal of Genetic Counseling, 31(1), 206–217. https://doi.org/doi: 10.1002/jgc4.1473

Pet, D. B., Holm, I. A., Williams, J. L., Myers, M. F., Novak, L. L., Brothers, K. B., Wiesner, G. L., & Clayton, E. W. (2019). Physicians' perspectives on receiving unsolicited genomic results. Genetics in Medicine: Official Journal of the American College of Medical Genetics, 21(2), 311–318. https://doi.org/10.1038/s41436-018-0047-z

Peterson, J. M., Pepin, A., Thomas, R., Biagi, T., Stark, E., Sparks, A. D., Johnson, K., & Kaltman, R. (2020). Racial disparities in breast cancer hereditary risk assessment referrals. Journal of Genetic Counseling, 29(4), 587–593. https://doi.org/10.1002/jgc4.1250

Prochniak, C. F., Martin, L. J., Miller, E. M., & Knapke, S. C. (2012). Barriers to and Motivations for Physician Referral of Patients to Cancer Genetics Clinics. Journal of Genetic Counseling, 21(2), 305–325. https://doi.org/10.1007/s10897-011-9401-x

Quinn, C. B. (2021, February 25). House Co-Sponsorship Memoranda. House Co-Sponsorship Memoranda. https://www.legis.state.pa.us/cfdocs/Legis/CSM/showMemoPublic.cfm?chamber=H&SPick=20210&cosponId=34820

Resta, R. G., Biesecker, B. B., Bennett, R. L., Blum, S., Hahn, S. E., Strecker, M. N., & Williams, J. L. (2006). A New Definition of Genetic Counseling: National Society of Genetic Counselors' Task Force Report. Journal of Genetic Counseling, 15(2), 77–83. https://doi.org/DOI: 10.1007/s10897-005-9014-3

Riesgraf, R. J., Veach, P. M., Macfarlane, I. M., & Leroy, B. S. (2015). Perceptions and Attitudes About Genetic Counseling Among Residents of a Midwestern Rural Area. Journal of Genetic Counseling, 24(4), 565–579. https://doi.org/10.1007/s10897-014-9777-5

Seibel, E., Gunn, G., Ali, N., Jordan, E., & Kenneson, A. (2022). Primary Care Providers' Use of Genetic Services in the Southeast United States: Barriers, Facilitators, and Strategies. Journal of Primary Care & Community Health, 13, 21501319221134750. https://doi.org/10.1177/21501319221134752

Selkirk, C. G., Weissman, S. M., Anderson, A., & Hulick, P. J. (2013). Physicians' preparedness for integration of genomic and pharmacogenetic testing into practice within a major healthcare system. Genetic Testing and Molecular Biomarkers, 17(3), 219–225. https://doi.org/10.1089/gtmb.2012.0165

Shah, S., Aune, G. J., Mette, L., & Poullard, N. (2017). Successful integration of genetic counseling in a comprehensive childhood cancer survivorship clinic. Journal of Clinical Oncology, 35(5_suppl), 230–230. https://doi.org/10.1200/JCO.2017.35.5_suppl.230

Soller, H. (2022). The Acquisition of Clinical Genetics Knowledge in Medical Students. University of Pittsburgh.

StataCorp. (2021). Stata Statistical Software: Release 17 (Version 17). StataCorp LLC.

Tri, N. (2022). Improving Provider Use of NCCN HBOC Genetic Counseling Referral Guidelines Through Provider Education and Awareness [Masters, University of Pittsburgh]. http://d-scholarship.pitt.edu/42877/

Truong, T. K., Kenneson, A., Rosen, A. R., & Singh, R. H. (2021). Genetic Referral Patterns and Responses to Clinical Scenarios: A Survey of Primary Care Providers and Clinical Geneticists. Journal of Primary Care & Community Health, 12, 21501327211046736. https://doi.org/10.1177/21501327211046734

University of South Carolina. (n.d.). Transnational Alliance for Genetic Counseling—School of Medicine Columbia. Retrieved April 12, 2023, from https://sc.edu/study/colleges_schools/medicine/centers_and_institutes_new/transnational_alliance_for_genetic_counseling/index.php

Venier, R. E., Grubs, R. E., Kessler, E., Cooper, K. L., Bailey, K. M., & Meade, J. (2022). Evaluation of barriers to referral for cancer predisposition syndromes in pediatric oncology patients in the United States. Journal of Genetic Counseling. https://doi.org/10.1002/jgc4.1559

Vito, A., Kennel, V., & Rezich, B. (2022, November). Completing the team: A study of non-genetics providers' perceptions of genetic counselors in multidisciplinary clinics. National Society of Genetic Counselors 41st Annual Conference, Nashville. https://epostersonline.com/nsgc2022/poster/asd8?view=true