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Primary Care Providers' Comfort With Utilization of Genetics in Practice

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PRIMARY CARE PROVIDERS' COMFORT WITH UTILIZATION OF GENETICS IN PRACTICE

by

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Dedication

"The future belongs to those who believe in the beauty of their dreams."

Eleanor Roosevelt

For the people along the way who inspired me to believe in the power of an interdisciplinary care team. The whole is *always* greater than the sum of its parts.

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Abstract

Primary care providers (PCPs) are often the first opportunity for individuals at risk for a genetic condition to be identified and they must care for patients with known genetic conditions. However, PCPs lag behind other providers in incorporating genetics into their practice. This study aimed to understand which genetics related concepts/topics PCPs (1) find relevant to practice, (2) are currently comfortable utilizing in practice, and (3) desire further education on. A mixed methods survey was sent to internists, family medicine providers, OBGYNs, pediatricians and geriatrics providers in South Carolina via email to assess this information. This included physicians, nurse practitioners, and physician assistants providing care in these fields. A total of 71 complete responses were analyzed.

The survey found that the majority of providers felt 8/13 items analyzed were relevant to their clinical practice. Furthermore, a majority of providers did not feel comfortable utilizing 17/24 items (expanded from the 13 items used when assessing relevancy) in their clinical practice. For the five items that a majority of respondents did not find relevant for practice, they also indicated that they were not comfortable utilizing these items in practice. This suggests a correlation between perceived relevancy and provider comfort, though the exact relationship is unclear. A majority of providers reported their prior genetics education was inadequate for what is needed in clinic on 10/14 items questioned. PCPs were less comfortable reaching out to genetics health professionals than other specialty providers and the majority of providers were unaware

of 10/13 genetics-based resources available to them. Overall, the study concluded that there are multiple opportunities for genetics health professionals to aid in furthering the education of PCPs, and specific topics per specialty and provider type were identified. Genetics health professionals will need to aid these providers in remedying the education gap, as well as continue to find ways to be more accessible to PCPs.

Table of Contents

List of Tables

List of Figures

Chapter 1. Background

1.1 Introduction

Since its inception over half a century ago, clinical genetics has infiltrated nearly every arena of medicine. There has been recognition that other providers outside of genetics health professionals have a stake in engaging with genetic services to best benefit patients. In studies dating back twenty years, primary care providers (PCPs) described the integral role genetics has in medicine in the appropriate prevention, surveillance, and management of various conditions (Emery et al., 1999). By being the first point of contact for many patients, PCPs are often the first opportunity for appropriate assessments and referrals to occur. PCPs serve as a gatekeeper for genetic services, identifying those most appropriate to be referred for further assessment. Additionally, PCPs contribute to patient support and coordination of care for surveillance and management (Carroll et al., 2003; Emery et al., 1999). Since these studies, a multitude of other research projects have further supported that PCPs see the utility and importance of genetics for their patients, despite the fact that the PCP may not be currently using these skills regularly (Ahmed et al., 2016; Carroll et al., 2019; Carroll et al., 2016; Evenson et al., 2016; Houwink et al., 2011).

Due to the recognition and value of genetic education for healthcare providers, various studies and guidelines have been published to help direct the education of providers. Burke et al. (2009) attempted to identify the core needs of a genetic curriculum for PCPs. The study was conducted in the United Kingdom and produced three main

categories: identifying patients with or at risk of a genetic condition, clinical management of genetic conditions, and communicating genetic information (Burke et al., 2009). Houwink et. al. (2011) furthered the conversation by utilizing three focus groups to assess the perceived role of genetics in primary care. Four themes emerged: genetics knowledge, family history, ethical dilemmas and psychosocial effects in relation to genetics, and insight into the organization and role of clinical genetics services (Houwink et al., 2011). This information has helped shape the structure of many curriculum guidelines put forth since. In 2014, the working group of the Inter-Society Coordinating Committee for Physician Education in Genomics developed the most recent recommendations for medical school and residency program curriculum in regard to genetics, including five entrustable professional activities (EPAs) for which developing physicians should aim for mastery. These include (1) Family History: elicit, document, and act on relevant family history pertinent to the patient's clinical status; (2) Genomic Testing: use genomic testing to guide patient management; (3) Treatment Based on Genomic Results: use genomic information to make treatment decisions; (4) Somatic Genomics: use genomic information to guide the diagnosis and management of cancer and other disorders involving somatic genetic changes; and (5) Microbial Genomic Information: use genomic tests that identify microbial contributors to human health and disease, as well as genomic tests that guide therapeutics in infectious diseases. It is recognized in the report that these may need to be modified based on specific specialty of medicine (Korf et al., 2014). Shortly afterward, similar guidelines were developed for physician assistant (PA) education (Goldgar et al., 2016).

1.2 Genetics and Primary Care Providers (PCPs)

Various studies have been conducted since the publication of these guidelines in an attempt to assess the current knowledge base of genetic concepts in non-genetics health professionals. This has provided insight into whether or not current educational practices have been effective in educating various populations, as well as understanding the implications of education on the utilization of genetic testing and family history risk assessments.

A recently published study of PCPs in Ontario, Canada found that few providers could appropriately identify useful sources of genetic information or information regarding genetic testing (22% and 21% respectively). Despite the struggles identifying quality information regarding genetics and genetic testing, the large majority of providers reported being involved in various aspects of genomic medicine, including taking a family history, identifying individuals who should be offered a referral, identifying individuals with genetic conditions, and providing support to those who have a genetic test result (82.8%-93.8%). The majority of participants reported interest in further education on genetics and genetics-based resources to help increase their confidence in utilizing genetic knowledge in their patient care practices (Carroll et al., 2019). In another recent study looking at the appropriate interpretation of variants of uncertain significance (VUSs) by providers, researchers found that only 14.6% of physicians surveyed were able to answer all three case examples correctly and about half of providers (46.4%) incorrectly defined a VUS. Additionally, about half of providers reported feeling uncomfortable or somewhat uncomfortable discussing genetics and VUS results (Macklin et al., 2019). Incorrect interpretation of these common VUS results can lead to

inappropriate management and care for the patient, and increased discomfort in providers may result in avoidance of testing altogether.

Pediatricians in Utah were assessed based on their perception of the genetic evaluation of children with autism spectrum disorder (ASD). About half of those surveyed were able to correctly answer questions regarding diagnostic yield, recurrence risk, and clinical guidelines for ASD. Despite current guidelines recommending that all children with ASD receive a genetics evaluation, about a quarter of pediatricians reported never initiating a conversation about genetic testing for ASD. Various barriers to referral and testing were reported, including not knowing which children with ASD to refer and lack of confidence in ordering testing and interpreting results. The participants self-reported a lack of knowledge and confidence in this common genetics referral for pediatricians, further suggesting that providers lack the necessary information and self-assurance in their ability to utilize genetics in clinic (Rutz et al., 2019). Avoidance of these necessary conversations or incorrect interpretation of common test results in clinic can lead to inadequate or inappropriate care for these patients. Thus, it is important for these providers to feel equipped to address genomic medicine in practice.

A study conducted in 2015 reviewed OBGYN and family medicine physicians' knowledge with *BRCA1* and *BRCA2* testing. The study found that the average correct responses to knowledge questions was 73%, and about 50% of providers reported being somewhat confident in providing related information. Respondents selected genetic specialists and oncologists as the most qualified to provide cancer genetic services, suggesting that these PCPs see the duty of genetic testing as a responsibility of specialty providers over PCPs (Dekanek et al., 2020). A 2019 study assessing PCP knowledge,

attitudes, and experience with direct-to-consumer testing also assessed the respondents' perceived knowledge of key components in genetics and genomics. This study found that the large majority (90%) of providers felt they had a moderate to expert level of understanding as it pertained to basic genetic principles, yet 61% reported they had no or minimal knowledge of when and how to integrate genomic medicine into practice. Furthermore, it was found that some factors deemed important by the researchers, such as an understanding of genome-wide association studies, was not clearly understood by the large majority of respondents (>90% reported little to no knowledge) (Haga et al., 2019).

A review of internal medicine providers in South Dakota found that while 88% of respondents understand the purpose of genetic testing, only 25% felt confident in responding to questions about the impact of genetic testing on disease susceptibility (Evenson et al., 2016). Another report looking at internists' test utilization found that 65% of internists counseled patients on genetic issues, and 44% had ordered genetic testing, but the majority felt they had either very poor or somewhat poor knowledge regarding genetics (73.7%) and guidelines for genetic testing (87.1%). The study also found that about half (53.4%) of providers knew of a genetic counselor or geneticist to refer a patient to if they felt unequipped to handle the situation themselves (Klitzman et al., 2013).

More recently, a study was conducted looking into comfort of OBGYNs utilizing genetic skills required for practice. At the time that Briggs et. al (2018) conducted a study which found that 48% of OBYGNs felt comfortable discussing positive carrier screening test results, The American College of Obstetricians and Gynecologists' recommendations were that all patients be offered cystic fibrosis carrier screening for which they would

need to be appropriately counseled. Furthermore, the study found that half of those surveyed did not feel comfortable discussing positive results, and only a quarter reported utilizing a genetic counselor to discuss positive results (Briggs et al., 2018). This suggests a level of discomfort with genetic information in addition to utilizing genetics health professionals, which does not appear to have significantly changed over the most recent decade. It further suggests a disconnect between the knowledge these providers need for clinic and the information they have from previous education and available resources, including genetics health professionals.

Review of the literature suggests that PCPs utilize genetic testing and/or engage in genetics discussion the least compared to other medical providers. Previously mentioned studies reported either a lack of comfort in genetics knowledge or suggested that this decreased comfort level is a motivating factor for the difference in uptake of genetics between PCPs and other medical specialties (Briggs et al., 2018; Carroll et al., 2019; Evenson et al., 2016; Klitzman et al., 2013; Macklin et al., 2019; Maradiegue et al., 2013; Rutz et al., 2019). A qualitative study comparing PCPs to cardiologists found that over half of the cardiologists interviewed discussed genetics information with patients "almost always" or "often" and reported feeling "prepared" or "very prepared" to disclose results of genetic testing. In contrast, only 9% of PCPs interviewed reported discussing genetic information with patients "almost always" or "often", and only 18% of the PCPs interviewed felt "prepared" or "very prepared" to disclose results. Yet, when looking at ability to answer genetics questions accurately, there was no statistically significant difference between the two groups (Christensen et al., 2016). Another survey of Wisconsin physicians concluded that PCPs lagged behind other providers in various

components that would result in less incorporation, such as familiarity and experience with genetic testing, as well as perceived adequate education on the topic (McCauley et al., 2017). Yet another report from the United Kingdom suggested that the perception of being "wrong" in front of the patient or not having an answer to a question posed may further contribute to the avoidance of genetics in practice, resulting from a discomfort with their own knowledge-base and ability to adequately address genetics in clinic (Mathers et al., 2010). These studies suggest that comfort with knowledge surrounding genetics may truly be the driving factor for the lack of utilization of genetics-based skills and knowledge in a primary care setting.

1.3 Addressing the Education Gap

Some attempts to remedy these reported education gaps include continuing education, decision support models, and the incorporation of genetics rotations in education programs. Ideally, remedying these gaps in education would result in increased comfort and therefore increased utilization of genetics in practice. The continuing education interventions reported in the literature between January, 2005 and January, 2018 found that educational interventions often increased confidence and knowledge short-term, but long-term studies suggested that this information was not always retained unless the increases in knowledge and confidence were due to a prolonged educational strategy. It was additionally found that all three of the different educational approaches identified (immersive and experiential learning, interdisciplinary and interprofessional education, and electronic- and web-based approaches) could be effective strategies for education and produce long-term increases in confidence and knowledge (Paneque et al., 2016; Rubanovich et al., 2018). A controlled assessment of PAs who received a web-

based educational model prior to seeing a standardized patient found that these PAs were able to ask more relevant medical questions and identify more family members of the patient with a history of cancer than their counterparts without spending additional time in the session (Roter et al., 2012). The conclusion from these literature review analyses and patient simulation study is that additional, appropriate education is capable of increasing both confidence and knowledge in providers, but it requires the dedication of providers to learn more and willingness to learn over time.

Looking at the incorporation of genetics rotations into educational programs, a report on the effectiveness and utility of a clinical genetics rotation, from the perspective of nurse practitioner (NP) students in that rotation, found that students believed the rotation enhanced their genetic thinking skills, their ability to collect a three-generation pedigree, and their ability to assess genetic risk factors in a way that could aid them in a genetic diagnosis. They felt equipped to navigate genetic resources and felt that their clinical practice would be enhanced significantly due to the rotation experience (Sloand et al., 2018). With adequate training, the issues associated with less-than-optimal knowledge and confidence can be resolved. However, many programs and areas do not have the ability to provide all students with genetics rotations, and it does not address the needs of the providers who are no longer in educational programs. That being said, it is important to identify the core needs of PCPs in clinic regarding genetics to target continuing education towards these topics, and have genetic health professionals available as a resource to support their local PCPs in their areas of need.

Part of the gap in education and lack of provider comfort may be due to the continuing genetics education of the faculty teaching these concepts. A look into nurse

practitioner faculty integration of genetics concepts found that, while faculty comfort in teaching genetics had improved in a five-year window with targeted educational programs, 30% of study participants still did not feel comfortable educating on basic genetic concepts and a larger proportion for more advanced topics such as complex modes of inheritance and pedigrees. Majority (65%) reported not feeling comfortable using Online Mendelian Inheritance in Man (OMIM) to look up genetic information (Maradiegue et al., 2013). This indicates that genetics health professionals may need to further aid in the education of developing providers via guest lecturing, consulting with faculty on genetics lectures, continuing to provide education and support for the programs in their area, and continue to be a resource as the educational gap continues to close. Regardless of the reason why the educational gap exists, there is significant data to support that there continues to be a lag between the necessary genetics education for clinical utility and the current knowledge. Based on these findings, it is important to assess if our educational practices are targeted appropriately to the needs of PCPs based on what they find relevant in daily practice, and also to determine if there are adequate continuing education opportunities for PCPs.

As an attempt to remedy the education gap seen in PCPs, various decision support models were created for testing and referral as a possible way to remedy the gap in education. This would additionally help relieve the burden of further education for providers. However, Zazove et al. (2015) found that screening questionnaires and prompts alone could not fill the gap. They looked at the responses of providers to automated, tailored prompts based on the electronic medical records of a total of 695 visits that were deemed moderate or high-risk for heart disease, stroke, diabetes, breast,

colorectal, or ovarian cancer. Physicians reviewed the family history in 53.5% of cases, discussed the family history in 22.9% of cases, and ordered testing/referral in 0.7% of the cases. In 22.3% of cases, the prompt was not addressed at all (Zazove et al., 2015). Another report testing the impact of a virtual family history questionnaire with decision support found that it was helpful in identifying patients that could benefit from extra screening and management or should be referred to genetics, but was not independently sufficient. In that study, half of the cases referred to a genetic counselor required a change in the family health history information due to misinterpretation of the question, and some cases were missed due to the lack of consideration of second-degree relatives by the clinician (Buchanan et al., 2015). It has also been reported that without this knowledge-base, providers may not utilize the decision-support tools because they would feel unable to adequately explain to a patient why they are being referred or managed differently (Ahmed et al., 2016). This information may explain why some of the providers chose not to address the prompt or did not address the family history with the patient in the Zazove et al. (2015) study. These studies agree on the necessity for PCPs to understand the genetic concepts behind the aids in order to appropriately identify patients who would benefit from testing, referral, or altered management.

The studies conducted thus far have established the need for further education of PCPs to help aid them in their role as a key resource for the identification and referral of individuals with personal or family histories suggestive of genetic conditions. However, a clear assessment looking at a broad range of genetic healthcare components in terms of provider comfort, utilization, and desire for further education does not appear to have been previously done. Understanding this information would provide educators, creators

of continuing education opportunities, and genetic health professionals ways in which they could continue to provide support to PCPs, as well as understand where to focus educational opportunities moving forward. Due to the need for this information, the study conducted aimed to understand what genetic topics and concepts PCPs felt comfortable with utilizing in clinic, and where further education may be needed. Furthermore, an attempt to understand what providers felt was most relevant to clinic was made.

CHAPTER 2

PRIMARY CARE PROVIDERS' COMFORT WITH UTILIZATION OF GENETICS IN PRACTICE[12345]

[1] Kupneski, T.M., Fairey, J.M., Hopla, D.L., Knight, E.M., and W.G. Patterson. To be submitted to *Journal of Genetic Counseling*.

[2] Kupneski, T.M., Fairey, J.M., Hopla, D.L., Knight, E.M., and W.G. Patterson. To be submitted to *Annals of Internal Medicine*.

[3] Kupneski, T.M., Fairey, J.M., Hopla, D.L., Knight, E.M., and W.G. Patterson. To be submitted to *Annals of Family Medicine*.

[4] Kupneski, T.M., Fairey, J.M., Hopla, D.L., Knight, E.M., and W.G. Patterson. To be submitted to *Pediatrics*.

[5] Kupneski, T.M., Fairey, J.M., Hopla, D.L., Knight, E.M., and W.G. Patterson. To be submitted to *Obstetrics and Gynecology*.

2.1 Abstract

Primary care providers (PCPs) are often the first opportunity for individuals at risk for a genetic condition to be identified and must care for patients with known genetic conditions. However, PCPs lag behind other providers in incorporating genetics into their practice. This study aimed to understand which genetics related concepts/topics PCPs (1) find relevant to practice, (2) are currently comfortable utilizing in practice, and (3) desire further education on. A mixed methods survey was sent to internists, family medicine providers, OBGYNs, pediatricians and geriatrics providers in South Carolina via email to assess this information. This included physicians, nurse practitioners, and physician assistants providing care in these fields. A total of 71 complete responses were analyzed.

The survey found that the majority of providers felt 8/13 items analyzed were relevant to their clinical practice. Furthermore, majority of providers did not feel comfortable utilizing 17/24 items (expanded from the 13 items used when assessing relevancy) in their clinical practice. For the five items that a majority of respondents did not find relevant for practice, a majority of respondents also indicated that they were not comfortable utilizing these items in practice. This suggests some type of correlation between perceived relevancy and provider comfort, though the exact relationship is unclear. The majority of providers reported their prior education has been inadequate for what is needed in clinic on 10/14 items questioned. PCPs were less comfortable reaching out to genetics health professionals than other specialty providers, and the majority of providers were unaware of 10/13 genetics-based resources available to them. Overall, the study concluded that there are multiple opportunities for genetics health professionals to aid in furthering the education of PCPs, and specific topics per specialty and provider

type were identified. Genetics health professionals will need to aid these providers in remedying the education gap, as well as continue to find ways to be more accessible to PCPs.

2.2 Introduction

Since its inception over half a century ago, clinical genetics has infiltrated nearly every arena of medicine as it is recognized that providers outside of genetics health professionals have a stake in engaging with genetic services to best benefit patients. By being the first point of contact for many patients, primary care providers (PCPs) are often the first opportunity for appropriate assessments and referrals to occur, and thus serve as a gatekeeper for genetic services. Additionally, PCPs will contribute to patient support and coordination of care for surveillance and management for patients with genetic conditions (Carroll et al., 2003; Emery et al., 1999). A multitude of other research projects have further supported that PCPs see the utility and importance of genetics for their patients, despite the fact that the PCP may not currently be using these skills regularly (Ahmed et al., 2016; Carroll et al., 2019; Carroll et al., 2016; Evenson et al., 2016; Houwink et al., 2011).

Due to the recognition and value of genetic education for healthcare providers, various studies and guidelines have been published to help direct the education of providers. Burke et al. (2009) attempted to identify the core needs of a genetic curriculum for PCPs. The study, conducted in the United Kingdom, produced three main categories: identifying patients with or at risk of a genetic condition, clinical management of genetic conditions, and communicating genetic information (Burke et al., 2009). Houwink et al. (2011) furthered the conversation by utilizing three focus groups to assess the perceived

role of genetics in primary care. Four themes emerged: genetics knowledge, family history, ethical dilemmas and psychosocial effects in relation to genetics, and insight into the organization and role of clinical genetics services (Houwink et al., 2011). In 2014, the working group of the Inter-Society Coordinating Committee for Physician Education in Genomics developed the most recent recommendations for medical school and residency program curriculum in regard to genetics, including five entrustable professional activities (EPAs) for which developing physicians should aim for mastery. These include (1) Family History; (2) Genomic Testing; (3) Treatment Based on Genomic Results; (4) Somatic Genomics; and (5) Microbial Genomic Information. It is recognized in the report that these may need to be modified based on specific specialty of medicine (Korf et al., 2014). Shortly afterward, similar guidelines were developed for physician assistant (PA) education (Goldgar et al., 2016).

Since the publication of these guidelines, there has been an attempt to assess the current comfort and knowledge base of genetic concepts in non-genetics health professionals. A recently published study of PCPs in Ontario, Canada found that few providers could appropriately identify useful sources of genetic information or information regarding genetic testing (22% and 21% respectively) despite the majority being involved in various aspects of genomic medicine (82.8%-93.8%). The majority of participants reported interest in further education to help increase their confidence (Carroll et al., 2019). A study on the interpretation of variants of uncertain significance (VUSs) by providers found that 14.6% of physicians surveyed were able to answer all three case examples correctly, about half of providers (46.4%) incorrectly defined a VUS, and half of providers reported feeling uncomfortable or somewhat uncomfortable

discussing genetics and VUS results (Macklin et al., 2019). Approximately half of surveyed pediatricians in Utah were able to correctly answer questions regarding diagnostic yield, recurrence risk, and clinical guidelines for autism spectrum disorder. The participants self-reported a lack of knowledge and confidence in referral recommendations and test utilization for this common pediatric genetics referral, further suggesting that providers lack the necessary information and self-assurance in their ability to utilize genetics in clinic (Rutz et al., 2019).

A review of internal medicine providers in South Dakota found that while 88% of respondents understand the purpose of genetic testing, only 25% felt confident in responding to questions about the impact of genetic testing on disease susceptibility (Evenson et al., 2016). Another report looking at internists' test utilization found that majority felt they had poor knowledge regarding genetics (73.7%) and guidelines for genetic testing (87.1%) and 46.6% of providers did not know of a genetic counselor or geneticist to whom they could refer (Klitzman et al., 2013). At the time that Briggs et al. conducted a study which found only 48% of OBGYNs felt comfortable discussing positive carrier screening test results, the American College of Obstetricians and Gynecologists' recommendations were that all patients be offered cystic fibrosis carrier screening for which they would need to be appropriately counseled (Briggs et al., 2018). These studies suggest that there may be a disconnect between the knowledge these providers need for clinic and the information they have from previous education, available resources, and genetics health professionals (often referred to as an "education gap"). This disconnect appears to be impacting the comfort and ability of these providers to adequately provide genomic medicine for their patients. Avoidance of these necessary

conversations or incorrect interpretation of common test results in clinic can lead to inadequate or inappropriate care for these patients. Thus, it is important for these providers to feel equipped to address genomic medicine in practice.

Education gaps have been identified for nearly all non-genetics health professionals, not just PCPs. However, review of the literature suggests that PCPs utilize genetic testing and/or engage in genetics discussion the least. Previously mentioned studies suggest that this decreased comfort level is a motivating factor for the difference in uptake of genetics between PCPs and other medical specialties (Briggs et al., 2018; Carroll et al., 2019; Evenson et al., 2016; Klitzman et al., 2013; Macklin et al., 2019; Maradiegue et al., 2013; Rutz et al., 2019). Christensen et al. (2016) compared cardiologists' and PCPs' frequency of genetics conversations and provider comfort when disclosing genetic test results. While over half of the cardiologists reported discussing genetics with patients regularly and feeling confident when disclosing results, less than a fifth of PCPs reported having these conversations or feeling comfortable with results disclosure. Yet, when assessing ability to answer genetics questions accurately, there was no difference between the two groups. Furthermore, another study concluded that PCPs had less familiarity with genetic testing and perceived that they had inadequate education on genetics, leading these physicians to incorporate genetics into clinical practice less frequently than other providers. These studies suggest that comfort with knowledge surrounding genetics may truly be the driving factor for the lack of utilization of genetics in a primary care setting (McCauley et al, 2017).

Decision support models have been proposed as a method for remedying the PCP's education gap. Zazove et al. (2015) and Buchanan et al. (2015) found that, while

decision support models may be helpful, they are not independently sufficient and further education would still be required. A report from the United Kingdom suggested that the perception of being "wrong" may further contribute to the avoidance of genetics in practice, resulting from a discomfort with their own knowledge-base, which would not be corrected solely with decision support models (Mathers et al., 2010). These studies agree on the necessity for PCPs to understand the genetic concepts behind the aids in order to appropriately identify patients who would benefit from testing, referral, or altered management.

Despite no significant difference in genetics knowledge, a clear difference in comfort level exists between PCPs and other healthcare providers. This suggests that knowledge may be the limiting factor in uptake of genomic medicine by PCPs. Therefore, by providing further education to these providers and thereby increasing comfort levels, genetic practice by PCPs could also increase. The exact skills and topics for which providers are not comfortable has not been clearly reported and thus was the target of this study. This research study was designed to identify opportunities where referral to genetics health professionals could aid in the comfort and utilization needs of PCPs. Additionally, this research aimed to identify target areas for genetics education in the future pertaining to primary care. Lastly, the information gathered from this research could generate information about utilization and comfort needs of genetics by PCPs.

This study aimed to assess what genetics skills and knowledge PCPs find helpful to have in a clinical setting, which they currently feel comfortable with, and how they perceive their level of education surrounding these topics. It was predicted that PCPs would have clear opinions about which subset of the listed skills and knowledge related

to genetics are important for their individual clinical practice. Additionally, it was suspected that the specific skills and knowledge deemed necessary for primary care clinics may differ from those PCPs are comfortable with practicing. It was hypothesized that PCPs would indicate that the level of education about genetics thus far is not adequate for what is needed in a clinical practice setting.

2.3 Materials and Methods

An invitation to participate in this survey was distributed to physicians, nurse practitioners (NPs), and PAs throughout the state of South Carolina via email. Individuals were reached through affiliation with professional organizations within the state or the medical care networks the providers are associated with such as Prisma Health, McLeod Regional Medical Center, Self Regional Medical Center, and Federally Qualified Health Centers. Furthermore, the invitation may have been shared to others by those who were initially contacted, potentially causing the survey to reach others outside of this original sample population. The selection process for participation included those who self-identify into the target population of physician, NP, or PA practicing in a field related to primary care (family medicine, internal medicine, obstetrics and gynecology/women's health, pediatrics, or geriatrics).

This study was conducted via an online questionnaire designed and stored on Qualtrics XM and distributed as previously described. Electing to take the questionnaire served as participant consent. The questionnaire was composed of multiple choice, multi-select, slider scale, and open-ended items (vignettes) designed to address the research questions of this study. Additionally, demographic information was collected. The data was analyzed using SPSS (Statistical Package for the Social Sciences) software to

calculate descriptive statistics, as well as run paired t-tests and chi-square test of independence when appropriate. Qualitative data was analyzed for themes using a grounded theory approach. There were no preset themes for our study, and apparent themes were coded based on participant responses. Themes were analyzed and responses were coded by one member of the research team, then two other individuals analyzed the data and classified responses into themes. Any discrepancies were discussed until an agreed upon conclusion could be reached.

A total of 129 individuals responded to the questionnaire. Of those 129, 6 responses were not included in analysis due to the respondent being a provider other than a physician, NP, or PA. Additionally, 31 responses were excluded due to the respondent practicing in a specialty outside of family medicine, internal medicine, obstetrics and gynecology/women's health (OBGYN), pediatrics or geriatrics. Lastly, 21 responses were excluded due to being incomplete. In total, 71 complete responses were used for data analysis. Demographic information pertaining to the sample population can be found in Table 2.1.

2.4 Results

2.4.1 Objective 1: What genetic skills and knowledge do PCPs find necessary?

The initial portion of the questionnaire focused on PCP perspectives of various genetic skills, topics, and testing options that PCPs may find relevant for use in clinic. The respondents were able to select which items they felt were important for practice, and the results are described in Table 2.2 and Table 2.3. Multiple chi-square tests of independence were run on the data to identify any statistically significant differences

Table 2.1 Sample Demographics

		Percentage	Count
Provider Type	Physician	48%	34
	Nurse Practitioner	18%	13
	Physician Assistant	34%	24
Specialty	Family Medicine	24%	17
	Internal Medicine	24%	17
	OBGYN	20%	14
	Pediatrics	23%	16
	Geriatrics	10%	7
Gender	Male	21%	
	Female	63%	
	Non-binary/ Unknown	7%	
Race/Ethnicity	Caucasian	77%	
	African American	24%	
	Asian	4%	
	Hispanic/Latino	3%	
	Other	1%	
	Unknown	7%	
Years in Practice	0-4	18%	
	5-9	11%	
	10-14	18%	
	15-19	14%	
	20-24	11%	
	25-29	6%	
	30-34	8%	
	35-39	1%	
	40-44	3%	
	45+	1%	
Practice Setting	Urban	35%	
	Suburban	41%	
	Rural	17%	
	Unknown	7%	

Table 2.2 Provider attitude towards relevance of various genetics topics and skills for clinical practice by provider type

	Percent of Providers Indicated as Relevant			
Item	**Total**	**Physician**	**Nurse Practitioner**	**Physician Assistant**
Structure, function, and replication of DNA	19.7%	20.6%	30.8%	12.5%
Inheritance patterns	73.2%	85.3%	53.8%	66.7%
Karyotype/microarray findings	36.6%	52.9%	30.8%	16.7%
Genetic principles	33.8%	52.9%	7.7%	20.8%
Family history taking and interpretation	95.8%	94.1%	100.0%	95.8%
Pedigree construction	23.9%	44.1%	7.7%	4.2%
Types of genetic testing	64.8%	73.5%	69.2%	50.0%
Genetic test results	71.8%	91.2%	46.2%	58.3%
Ethical, legal, and social implications of testing on patients and family members	63.4%	70.6%	53.8%	58.3%
Ethical, legal and social implications of testing on children/minors and adults with incapacity	40.8%	50.0%	38.5%	29.2%
Cost of genetic testing and insurance coverage	85.9%	94.1%	69.2%	83.3%
Ability to identify/locate resources related to referrals and management guidelines, and patient support	78.9%	85.3%	69.2%	75.0%
Ability to refer and interact with local or regional geneticists and/or genetic counselors	71.8%	91.2%	38.5%	62.5%

Table 2.3 Provider attitude towards relevance of various genetics topics and skills for clinical practice by provider specialty

	Percent of Providers Indicated as Relevant					
Item	**Total**	**Family Medicine**	**Internal Medicine**	**OBGYN**	**Pediatrics**	**Geriatrics**
Structure, function, and replication of DNA	19.7%	29.4%	17.6%	21.4%	12.5%	14.3%
Inheritance patterns	73.2%	58.8%	70.6%	85.7%	93.8%	42.9%
Karyotype/microarray findings	36.6%	23.5%	5.9%	71.4%	68.8%	0.0%
Genetic principles	33.8%	29.4%	29.4%	35.7%	50.0%	14.3%
Family history taking and interpretation	95.8%	94.1%	94.1%	100.0%	93.8%	100.0%
Pedigree construction	23.9%	23.5%	17.6%	21.4%	43.8%	0.0%
Types of genetic testing	64.8%	52.9%	47.1%	92.9%	87.5%	28.6%
Genetic test results	71.8%	70.6%	70.6%	78.6%	81.3%	42.9%
Ethical, legal, and social implications of testing on patients and family members	63.4%	76.5%	47.1%	64.3%	68.8%	57.1%
Ethical, legal and social implications of testing on children/minors and adults with incapacity	40.8%	47.1%	23.5%	28.6%	62.5%	42.9%
Cost of genetic testing and insurance coverage	85.9%	88.2%	88.2%	100.0%	81.3%	57.1%
Ability to identify/locate resources related to referrals and management guidelines, and patient support	78.9%	64.7%	88.2%	71.4%	93.8%	71.4%
Ability to refer and interact with local or regional geneticists and/or genetic counselors	71.8%	52.9%	76.5%	85.7%	87.5%	42.9%

based on provider type when looking at each item mentioned in Table 2.2 and Table 2.3 and to identify any statistically significant differences based on provider specialty. The results of these tests can be found in Table 2.4. In general, physicians were more likely to find these items necessary for clinic while NPs were least likely. OBGYN and pediatricians were the specialties most likely to find the items in question relevant for practice.

Respondents had the opportunity to describe any other genetics topics or concepts that were deemed relevant to the respondent but not previously mentioned. A total of 19 individuals chose to respond to this question. Themes identified were cancer-related information (3/19), continued provider education and/or useful provider resources on genetics-based topics (6/19) and distinct counseling skills (3/19). The distinct counseling skills included best practices for discussing test results, counseling on genetic testing limitations, and advocating against unwarranted or unproven genetic testing.

2.4.2 Objective 2: Are PCPs comfortable with genetic skills and knowledge?

To address comfort level with genetic skills and knowledge, respondents were asked to identify items they felt comfortable with utilizing in clinic. These were broken into four thematic categories: (1) items related to genetic principles, inheritance and family history, (2) items related to genetic testing and test results, (3) ethical, legal and social implications of genetic testing, and (4) genetic resources and referrals. Results from these items analyzed by provider type and provider specialty can be seen in Figures 2.1-2.8.

Chi-square tests of independence were run on this data to determine if there were any statistically significant differences between provider types or provider specialties for

Table 2.4 Chi-squared analysis of provider attitude towards relevance of genetic components by provider type and by specialty

	X^2 results, item versus provider type		**X^2 results, item versus provider specialty**	
Item	**X^2 (df, N) =**	**p-value**	**X^2 (df, N) =**	**p-value**
Structure, function, and replication of DNA	-	Not significant	-	Not significant
Inheritance patterns	-	Not significant	X^2 (4, N=71)=9.706	p=.046
Karyotype/microarray findings	X^2 (2, N=71)=8.211	p=.016	X^2 (4, N=71)=26.645	p=.000
Genetic principles	X^2 (2, N=71)=11.330	p=.003	-	Not significant
Family history taking and interpretation	-	Not significant	-	Not significant
Pedigree construction	X^2 (2, N=71)=14.639	p=.001	-	Not significant
Types of genetic testing	-	Not significant	X^2 (4, N=71)=15.866	p=.003
Genetic test results	X^2 (2, N=71)=12.686	p=.002	-	Not significant
Ethical, legal, and social implications of testing on patients and family members	-	Not significant	-	Not significant
Ethical, legal and social implications of testing on children/minors and adults with incapacity	-	Not significant	-	Not significant
Cost of genetic testing and insurance coverage	-	Not significant	-	Not significant
Ability to identify/locate resources related to referrals and management guidelines, and patient support	-	Not significant	-	Not significant
Ability to refer and interact with local or regional geneticists and/or genetic counselors	X^2 (2, N=71)=14.475	p=.001	-	Not significant

Note: significance was determined to be p<.05. Some information on items deemed not significant has been omitted and replaced with a "-" for ease of reading. Full statistical information can be found in Table C.3.

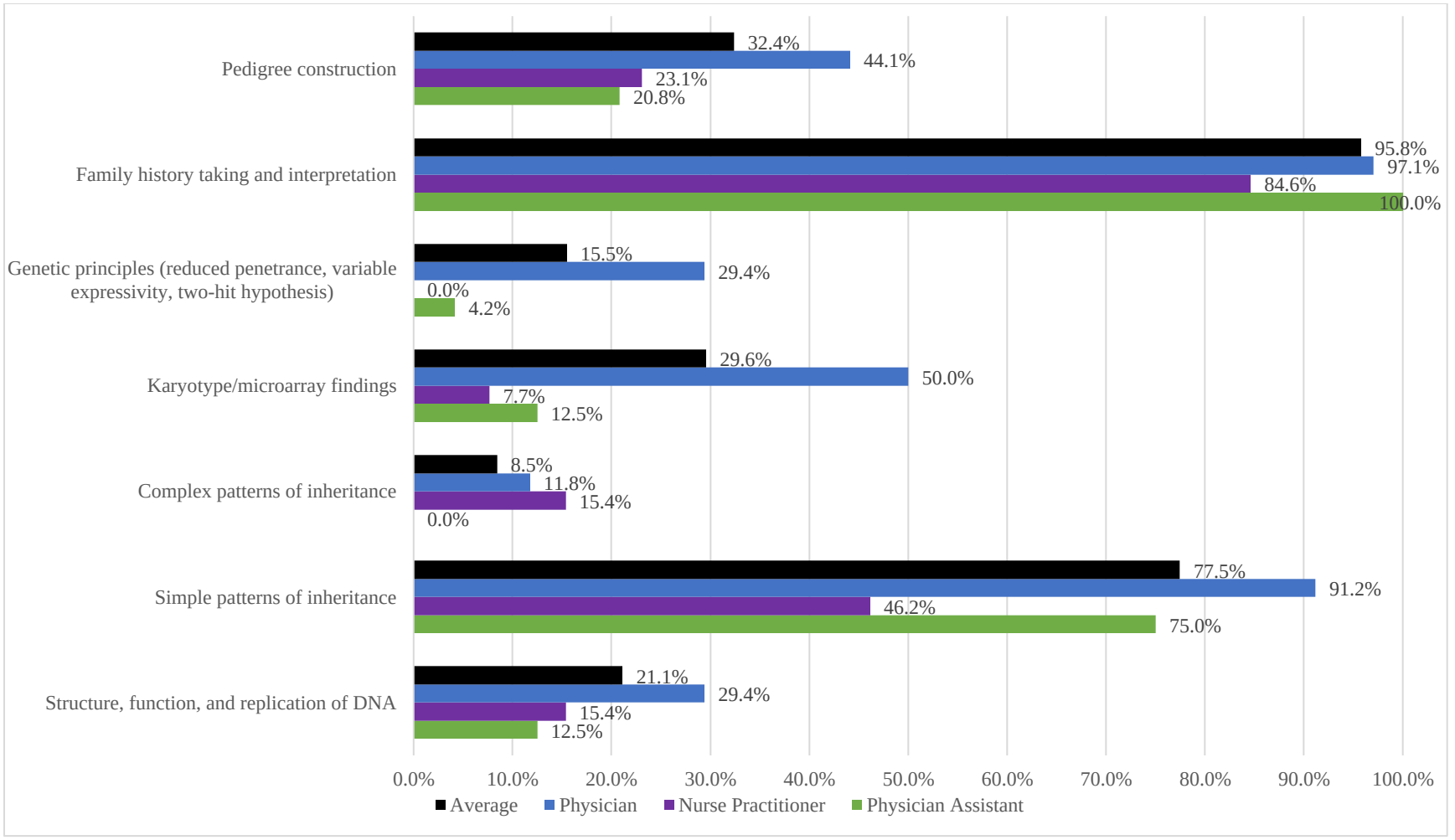

Figure 2.1 Comfort with genetic based concepts related to genetic principles, inheritance and family history by provider type

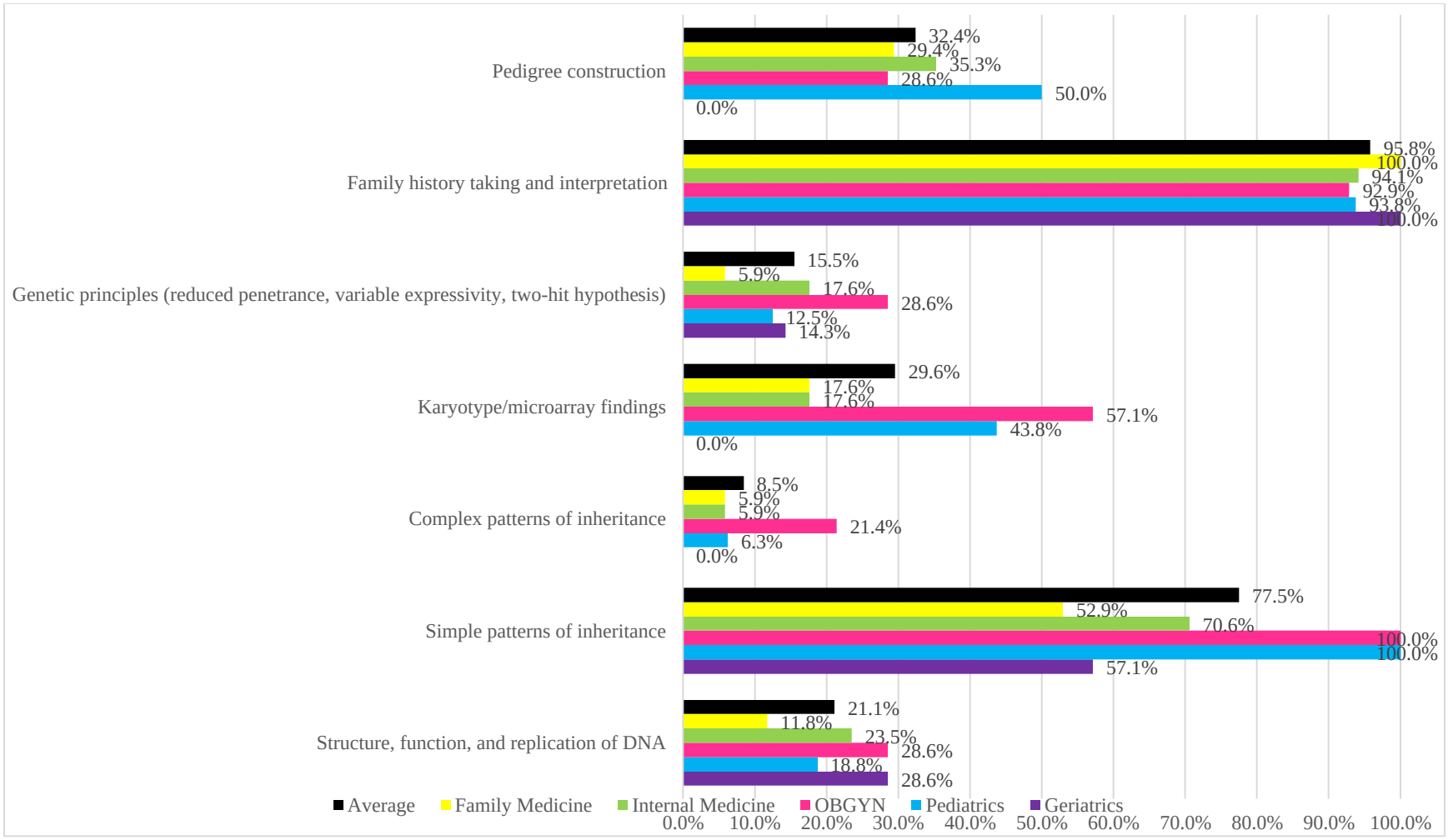

Figure 2.2 Comfort with genetic based concepts related to genetic principles, inheritance and family history by provider specialty

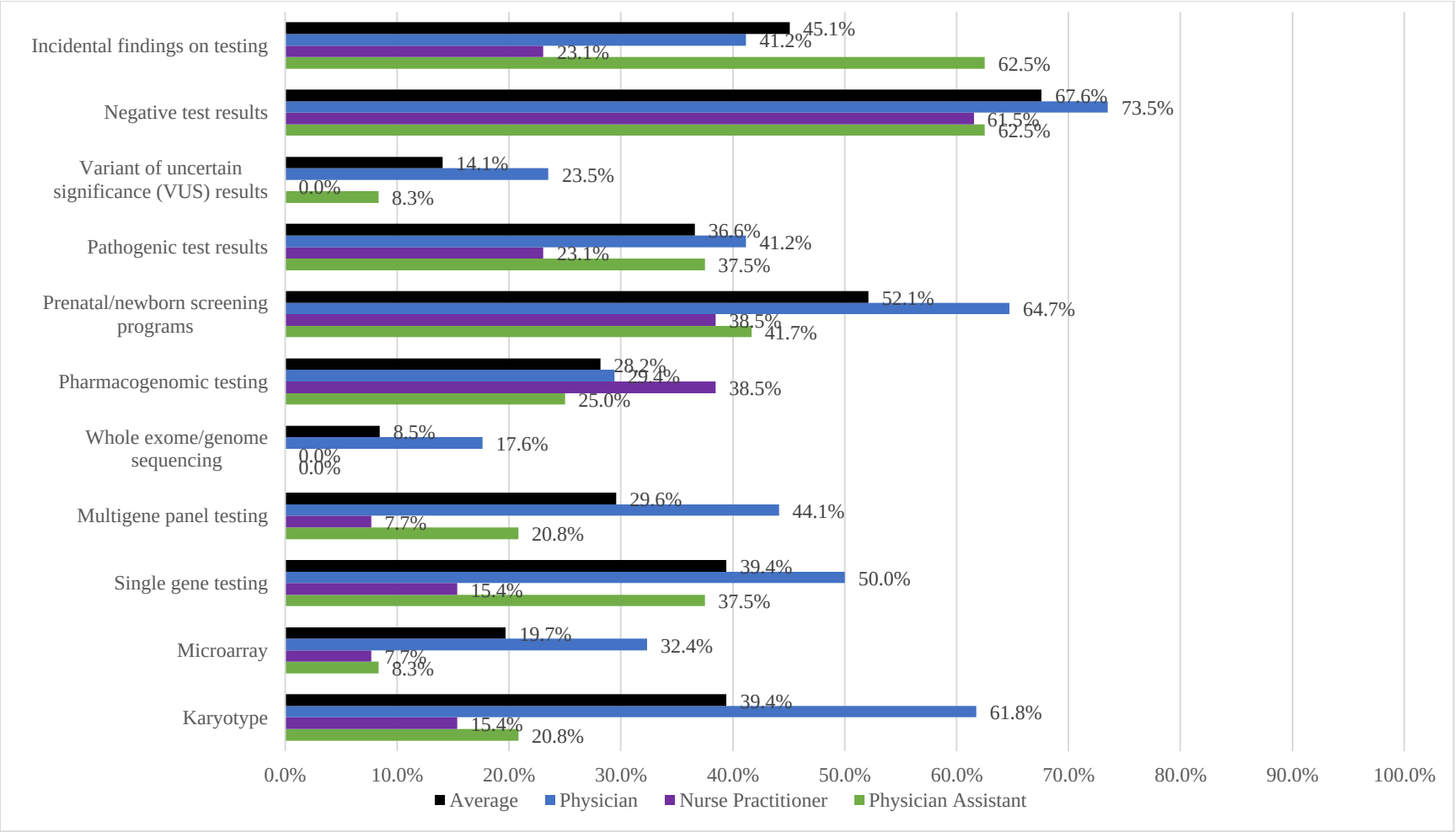

Figure 2.3 Comfort with genetic based concepts related to genetic testing and test results by provider type

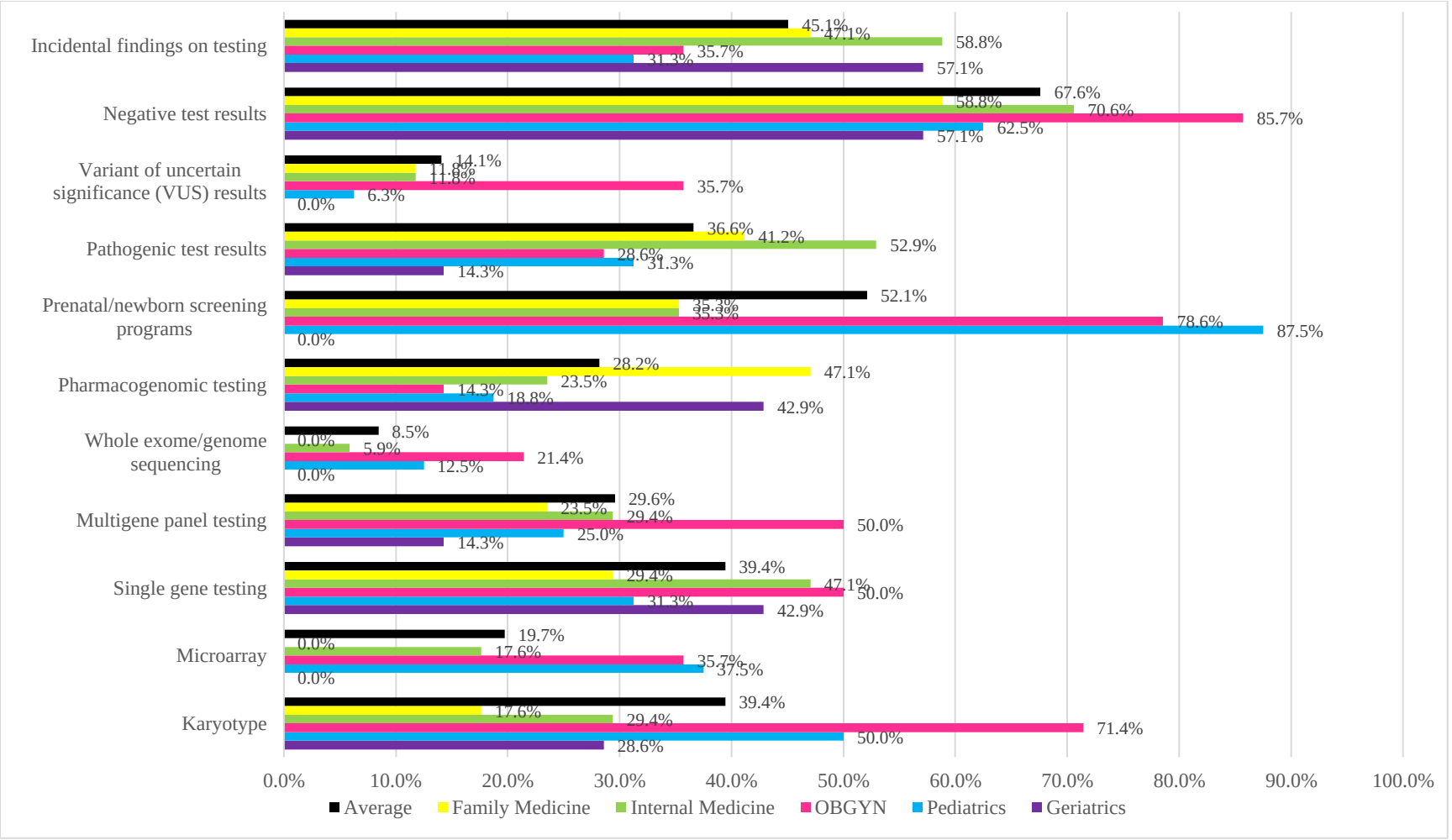

Figure 2.4 Comfort with genetic based concepts related to genetic testing and test results by provider specialty

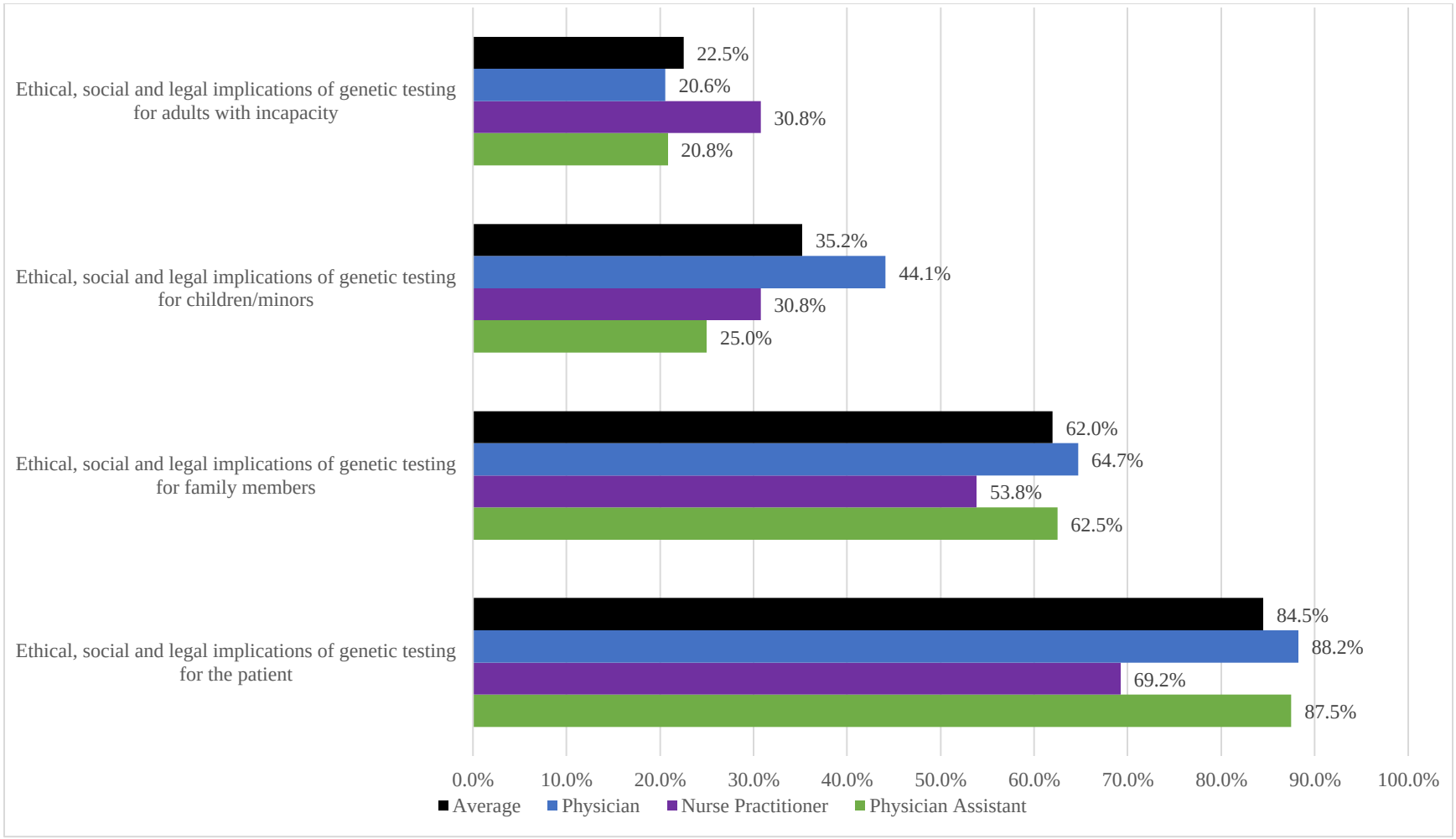

Figure 2.5 Comfort with genetic based concepts related to ethical, legal, and social implications of genetic testing by provider type

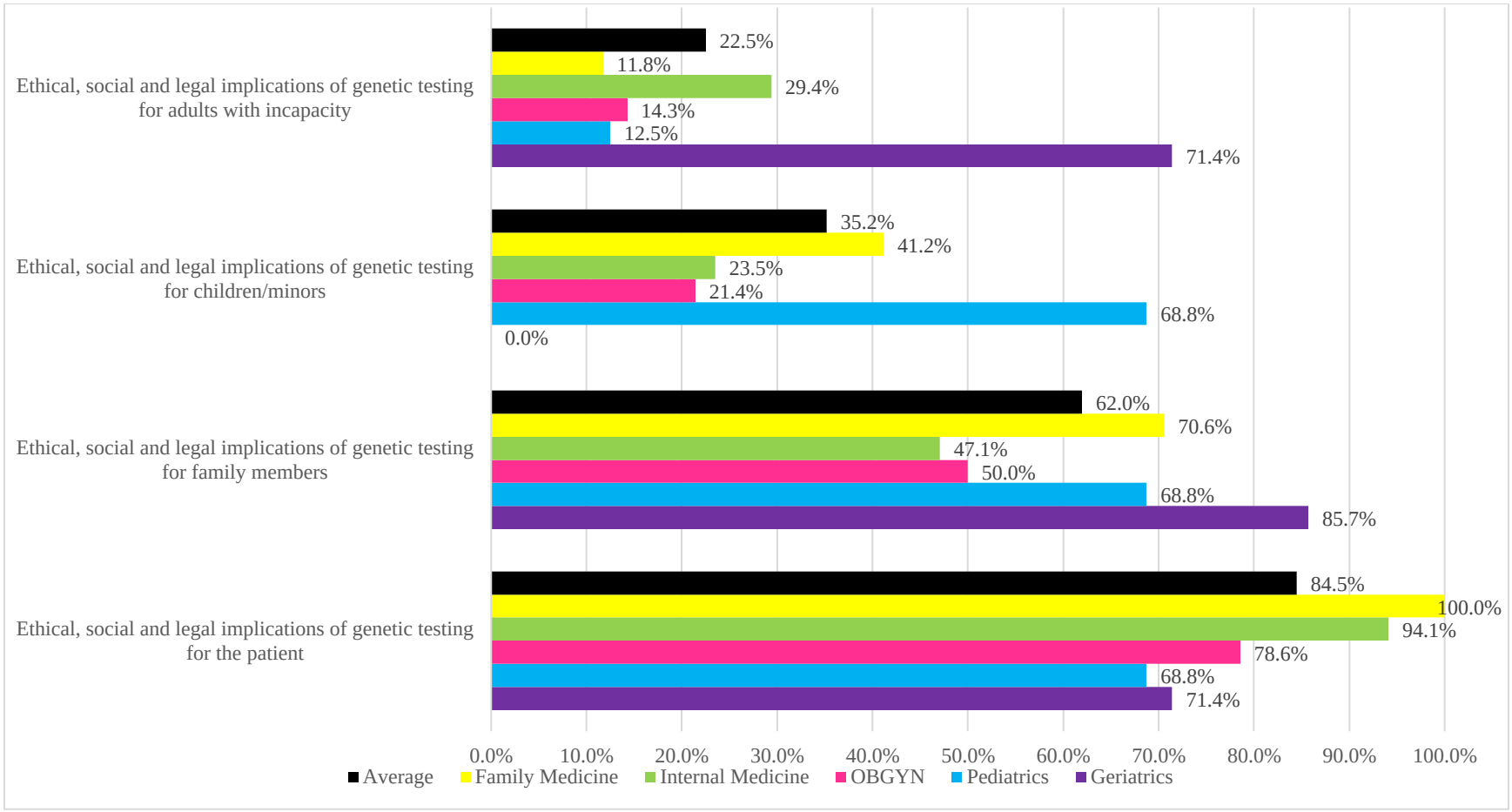

Figure 2.6 Comfort with genetic based concepts related to ethical, legal, and social implications of genetic testing by provider specialty

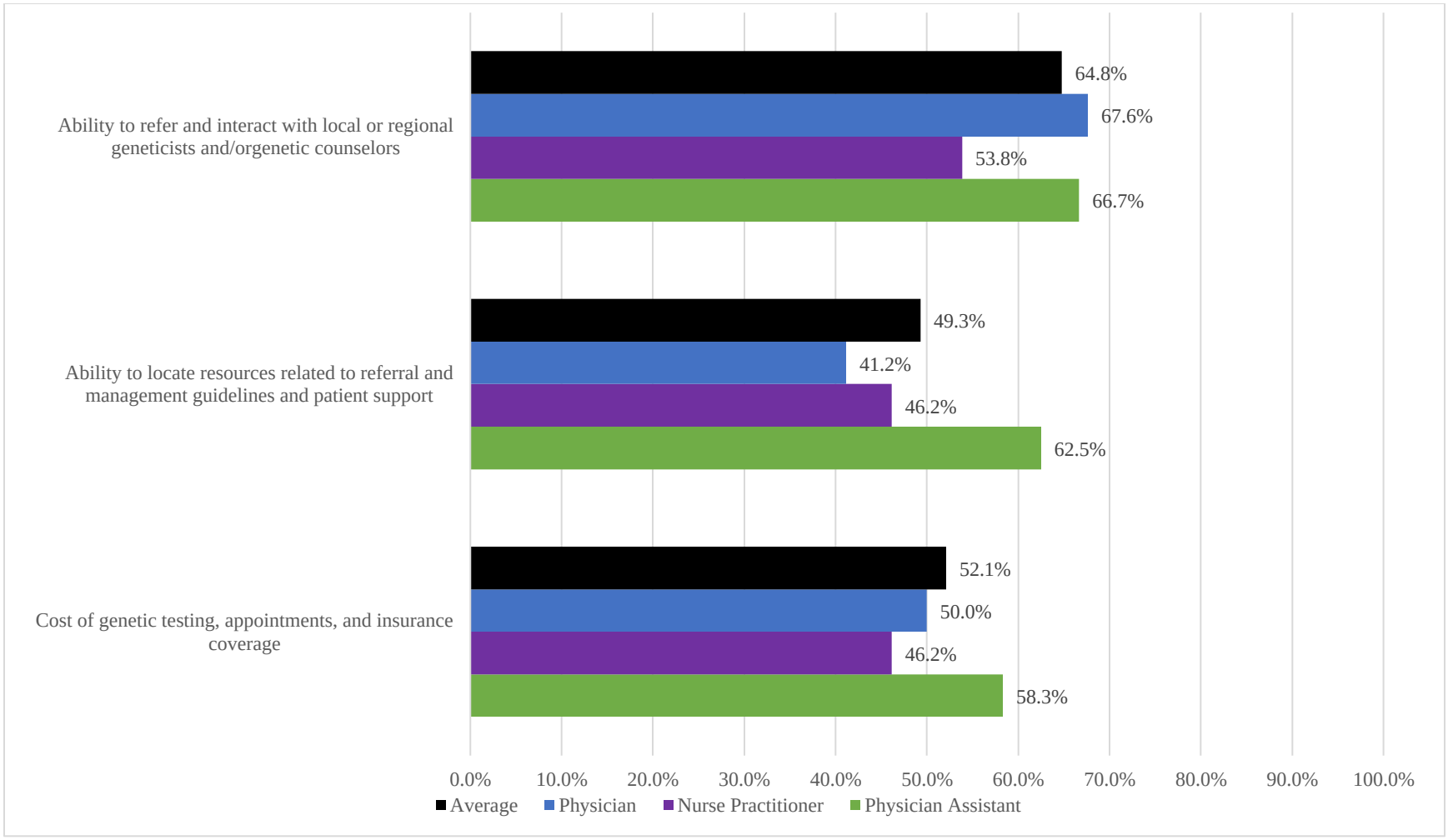

Figure 2.7 Comfort with genetic based concepts related to genetic resources and referrals by provider type

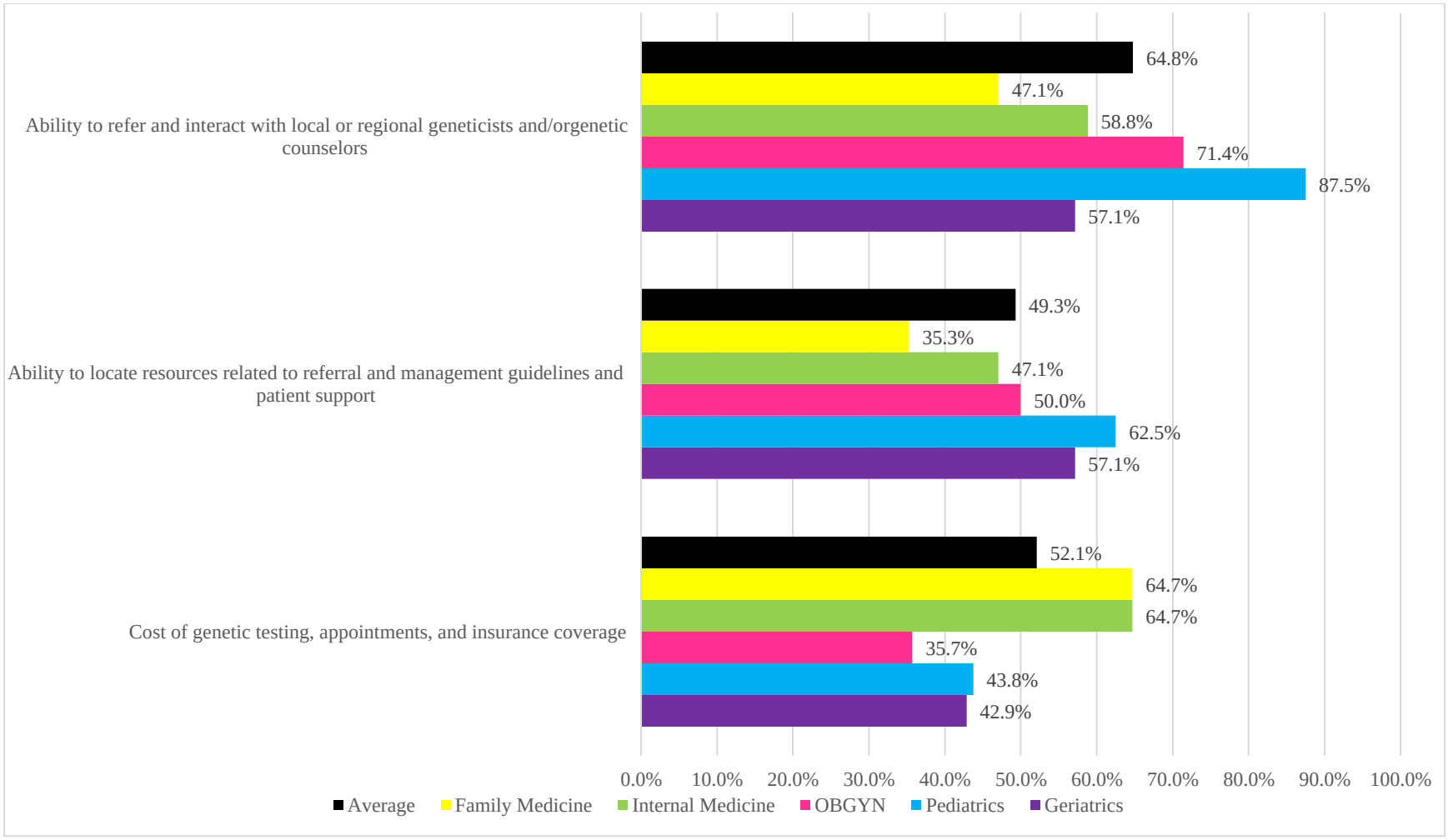

Figure 2.8 Comfort with genetic based concepts related to genetic resources and referrals by provider specialty

each item depicted in Figure 2.1-Figure 2.8. The results of this analysis can be found in Table 2.5. In general, physicians were typically more comfortable than NPs and PAs with these items, and NPs were the least comfortable. Furthermore, providers in OBGYN or pediatrics were typically more comfortable with each of these items than providers in internal medicine, family medicine, and geriatrics. In the case of ethical, legal, and social implications of genetic testing for children/minors, pediatricians were most likely to be comfortable, and in the case of ethical, legal, and social implications of genetic testing for adults with incapacity, internal medicine and geriatric providers were the most likely to be comfortable.

For each item a provider identified as being comfortable with, the respondent was then asked to rank how comfortable they felt with that item, (1) neither comfortable nor uncomfortable, (2) slightly comfortable, (3) moderately comfortable, or (4) extremely comfortable, which was quantified (as noted) and averaged for analysis. When looking at the average for all respondents, 84% (21/25) of items scored a 3.0 or higher. This indicates that on average, for the providers who felt comfortable with an item, they had at least a moderate level of comfort. The four items that scored below 3.0 were: structure, function, and DNA replication (2.93); complex patterns of inheritance (2.83); multigene panel testing (2.90); and cost of genetic testing, appointments, and insurance coverage (2.86). When looking at averages by provider type, none of the items scored below a 3.0 for physicians. Above half (54.5%, 12/22) of items were scored below 3.0 for NPs, and 22.7% (5/22) were scored below 2.5. Three items could not be considered for NPs due to no respondents indicating comfort with those items. Furthermore, PAs also averaged below 3.0 for over half of items (65.2%, 15/23) and below 2.5 for 13.0% (3/23) items.

Table 2.5 Chi-squared analysis of provider attitude towards comfort with genetic components by provider type and by specialty

	X^2 results, item versus provider type		**X^2 results, item versus provider specialty**	
Item	**X^2 (df, N) =**	**p-value**	**X^2 (df, N) =**	**p-value**
Structure, function, and replication of DNA	-	Not significant	-	Not significant
Simple patterns of inheritance	X^2 (2, N=71)=11.046	p=.004	X^2 (4, N=71)=16.700	p=.002
Complex patterns of inheritance	-	Not significant	-	Not significant
Karyotype/microarray findings (as it relates to inheritance)	X^2 (2, N=71)=13.723	p=.001	X^2 (4, N=71)=11.913	p=.018
Genetic principles (reduced penetrance, variable expressivity, two-hit hypothesis)	X^2 (2, N=71)=9.766	p=.008	-	Not significant
Family history taking and interpretation	-	Not significant	-	Not significant
Pedigree construction	-	Not significant	-	Not significant
Karyotype (as it relates to genetic testing and test results)	X^2 (2, N=71)=13.723	p=.000	X^2 (4, N=71)=12.379	p=.015
Microarray (as it relates to genetic testing and test results)	X^2 (2, N=71)=6.581, p=.001	p=.037	X^2 (4, N=71)=11.399	p=.022
Single gene testing	-	Not significant	-	Not significant
Multigene panel testing	X^2 (2, N=71)=7.321	p=.026	-	Not significant
Whole exome/genome sequencing	X^2 (2, N=71)=7.132	p=.028	-	Not significant
Pharmacogenomic testing	-	Not significant	-	Not significant
Prenatal/newborn screening programs	-	Not significant	X^2 (4, N=71)=23.428	p=.000
Pathogenic test results	-	Not significant	-	Not significant
Variant of uncertain significance (VUS) results	-	Not significant	-	Not significant
Negative test results	-	Not significant	-	Not significant
Incidental findings on testing	-	Not significant	-	Not significant

Ethical, social and legal implications of genetic testing for the patient	-	Not significant	-	Not significant
Ethical, social and legal implications of genetic testing for family members	-	Not significant	-	Not significant
Ethical, social and legal implications of genetic testing for children/minors	-	Not significant	X^2 (4, N=71)=14.141	p=.007
Ethical, social and legal implications of genetic testing for adults with incapacity	-	Not significant	X^2 (4, N=71)=12.645	p=.013
Costs of genetic testing, genetics appointments, and insurance coverage	-	Not significant	-	Not significant
Ability to identify/locate resources related to referral and management guidelines and support for genetic conditions	-	Not significant	-	Not significant
Ability to refer and interact with local or regional geneticists and/or genetic counselors	-	Not significant	-	Not significant

Note: significance was determined to be p<.05. Some information on items deemed not significant has been omitted and replaced with a "-" for ease of reading. Full statistical information can be found in Table C.4.

Two items could not be considered for PAs due to no respondents indicating comfort with those items. When looking at averages by provider specialty, none of the items scored below a 3.0 for OBGYNs. Three items (12%, 3/25) were below 3.0 for pediatricians and one was below 2.5 (4%, 1/25). Four items (23.5%, 4/17) were below 3.0 for geriatric providers, and one (5.9%, 1/17) was below 2.5. Eight items could not be considered for geriatrics due to no respondents indicating comfort with those items. Family medicine providers averaged below 3.0 for 47.8% (11/23) of items and below 2.5 for 8.7% (2/23) of items. Two items could not be considered for family medicine due to no respondents indicating comfort with those items. Internal medicine averaged below 3.0 for 56% (14/25) of items and below 2.5 for 12% (3/25) of items. Tables 2.6 and 2.7 show which items were below 3.0 for each provider type and provider specialty respectively, with their exact averages for items given if it was below 3.0.

To evaluate comfort with various genetics-based resources, providers were asked to identify any of the resources listed that they were aware of, and if they felt comfortable utilizing these resources for those that indicated awareness of the resource. The total percentage of providers who felt comfortable, aware, or unfamiliar with each of the thirteen listed resources is depicted in Figure 2.9. When asked in an open-ended question which resources providers rely on most when preparing to care for a patient with a known or suspicious for a genetic condition, 43 providers responded, and four themes emerged. The four themes identified were: UpToDate (24/43), genetic counselors/geneticists (13/43), coworkers/attendings (3/43), and Medscape (3/43). Providers were asked to rate their interest in learning about genetics-based resources available from 0-100 using a sliding scale. The average level of interest across providers was 65.09 (*SD*=22.73).

Table 2.6 Average level of comfort with genetic based concepts by provider type

	Average Level of Comfort			
Item	**Total**	**Physician**	**Nurse Practitioner**	**Physician Assistant**
Structure, function, and replication of DNA	2.93	-	2.00	-
Simple patterns of inheritance	-	+	2.83	2.94
Complex patterns of inheritance	2.83	-	2.00	n/a
Karyotype/microarray findings	-	-	2.00	2.33
Genetic principles (reduced penetrance, variable expressivity, two-hit hypothesis)	-	-	n/a	2.00
Family history taking and interpretation	-	+	2.82	-
Pedigree construction	-	-	-	2.80
Karyotype	-	+	2.50	2.60
Microarray	-	-	-	2.00
Single gene testing	-	-	2.50	2.89
Multigene panel testing	2.90	-	2.00	2.60
Whole exome/genome sequencing	+	+	n/a	n/a
Pharmacogenomic testing	-	-	-	-
Prenatal/newborn screening programs	-	+	-	2.70
Pathogenic test results	-	-	-	-
Variant of uncertain significance (VUS) results	-	-	n/a	2.50
Negative test results	-	+	-	-
Incidental findings on testing	-	-	-	2.93
Ethical, social and legal implications of genetic testing for the patient	-	-	2.67	2.90
Ethical, social and legal implications of genetic testing for family members	-	-	2.57	-

Ethical, social and legal implications of genetic testing for children/minors	-	+	-	2.67
Ethical, social and legal implications of genetic testing for adults with incapacity	-	-	-	-
Cost of genetic testing, appointments, and insurance coverage	2.86	-	2.17	2.86
Ability to locate resources related to referral and management guidelines and patient support	-	-	2.67	2.93
Ability to refer and interact with local or regional geneticists and/or genetic counselors	-	-	-	-

Note: “n/a” indicates that no providers within said provider type indicated comfort with that particular item. “–” indicates that the item averaged a 3.0-3.4 for said provider type. “+” indicates that the item averaged a 3.5 or above for said provider type.

Table 2.7 Average level of comfort with genetic based concepts by provider specialty

	Average Level of Comfort					
Item	**Total**	**Family Medicine**	**Internal Medicine**	**OBGYN**	**Pediatrics**	**Geriatrics**
Structure, function, and replication of DNA	2.93	2.50	-	-	2.67	-
Simple patterns of inheritance	-	-	-	-	-	2.75
Complex patterns of inheritance	2.83	2.00	2.00	+	2.00	n/a
Karyotype/microarray findings	-	2.33	2.67	-	-	n/a
Genetic principles (reduced penetrance, variable expressivity, two-hit hypothesis)	-	-	2.67	+	-	2.00
Family history taking and interpretation	-	-	-	-	+	-
Pedigree construction	-	2.80	-	-	-	n/a
Karyotype	-	2.67	2.80	+	-	-
Microarray	-	n/a	2.33	-	-	n/a
Single gene testing	-	-	2.75	-	-	-
Multigene panel testing	2.90	-	2.80	-	2.50	-
Whole exome/genome sequencing	+	n/a	-	+	-	n/a
Pharmacogenomic testing	-	2.75	-	+	-	+
Prenatal/newborn screening programs	-	-	2.83	-	+	n/a
Pathogenic test results	-	-	-	+	-	+
Variant of uncertain significance (VUS) results	-	2.50	+	-	-	n/a
Negative test results	-	-	2.92	+	+	-
Incidental findings on testing	-	-	2.80	+	-	-

Ethical, social and legal implications of genetic testing for the patient	-	2.76	2.88	+	-	-
Ethical, social and legal implications of genetic testing for family members	-	2.83	-	+	-	2.83
Ethical, social and legal implications of genetic testing for children/minors	-	2.86	-	+	-	n/a
Ethical, social and legal implications of genetic testing for adults with incapacity	-	+	-	+	+	-
Cost of genetic testing, appointments, and insurance coverage	2.86	2.82	2.36	+	-	-
Ability to locate resources related to referral and management guidelines and patient support	-	-	2.50	-	-	-
Ability to refer and interact with local or regional geneticists and/or genetic counselors	-	-	2.50	+	+	2.75

Note: “n/a” indicates that no providers within said specialty indicated comfort with that particular item. “–” indicates that the item averaged a 3.0-3.4 for said specialty. “+” indicates that the item averaged a 3.5 or above for said specialty.

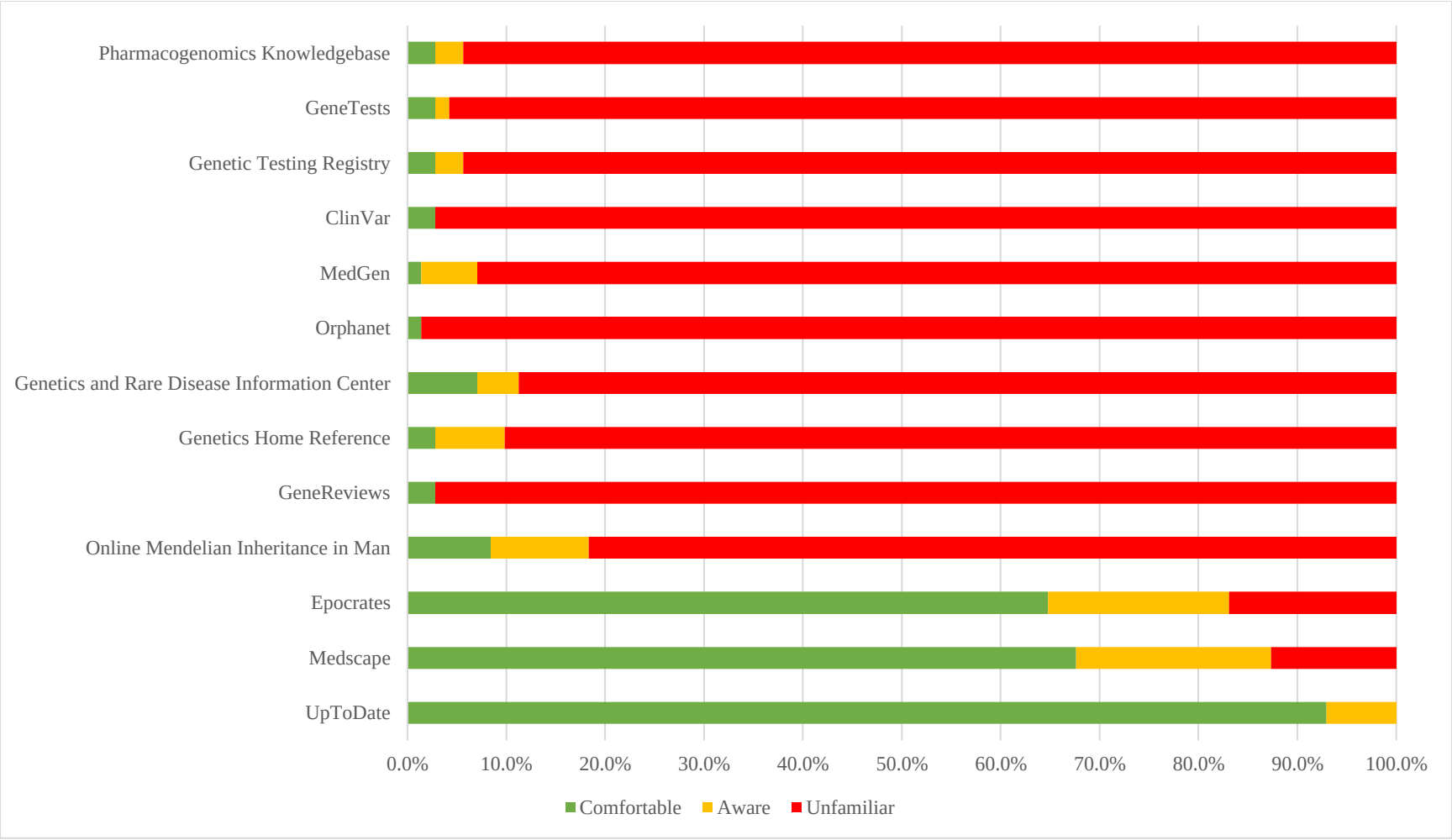

Figure 2.9 Provider familiarity and comfort with various genetic based resources

Respondents were asked to report their level of comfort reaching out to a genetics health professional to answer a question regarding referrals, test results, or any other patient-specific topic related to their specialty, as well as their level of comfort doing so with a non-genetics health specialist (such as neurology, cardiology, endocrinology). This data was analyzed utilizing a paired t-test and found that overall, providers were more comfortable reaching out to non-genetics health professionals (M=72.51, SD=24.66) than genetics health professionals (M=66.72, SD=25.80), t(70)=2.175, p=.033. When asked about experience utilizing genetic counselors as a resource/member of the interdisciplinary care team, 41 providers described their general experiences. Approximately half (20/41) of providers that responded reported little to no experience with utilizing genetic counselors, 39% (16/41) reported a generally positive experience, 9.8% (4/41) reported that their interaction was limited to referral and test reports/ summary letters, and 9.8% (4/41) reported either negative or mixed experiences. Confidence in a genetic counselor's ability to explain various genetics concepts to a provider was assessed by asking which items respondents would feel comfortable trusting a genetic counselor to explain to a provider. Overall, providers felt comfortable utilizing a genetic counselor's knowledge 64.8-85.9% of the time.

2.4.3 Objective 3: How do PCPs perceive their level of education compared to what is necessary for clinic?

Respondents were asked to rate their perceived level of education in comparison to what they consider the appropriate amount necessary for clinic for various genetics-based concepts. The results of this data can be seen in Figures 2.10 and 2.11. For 71.4% (10/14) of items, over half of respondents indicated slightly too little or far too little

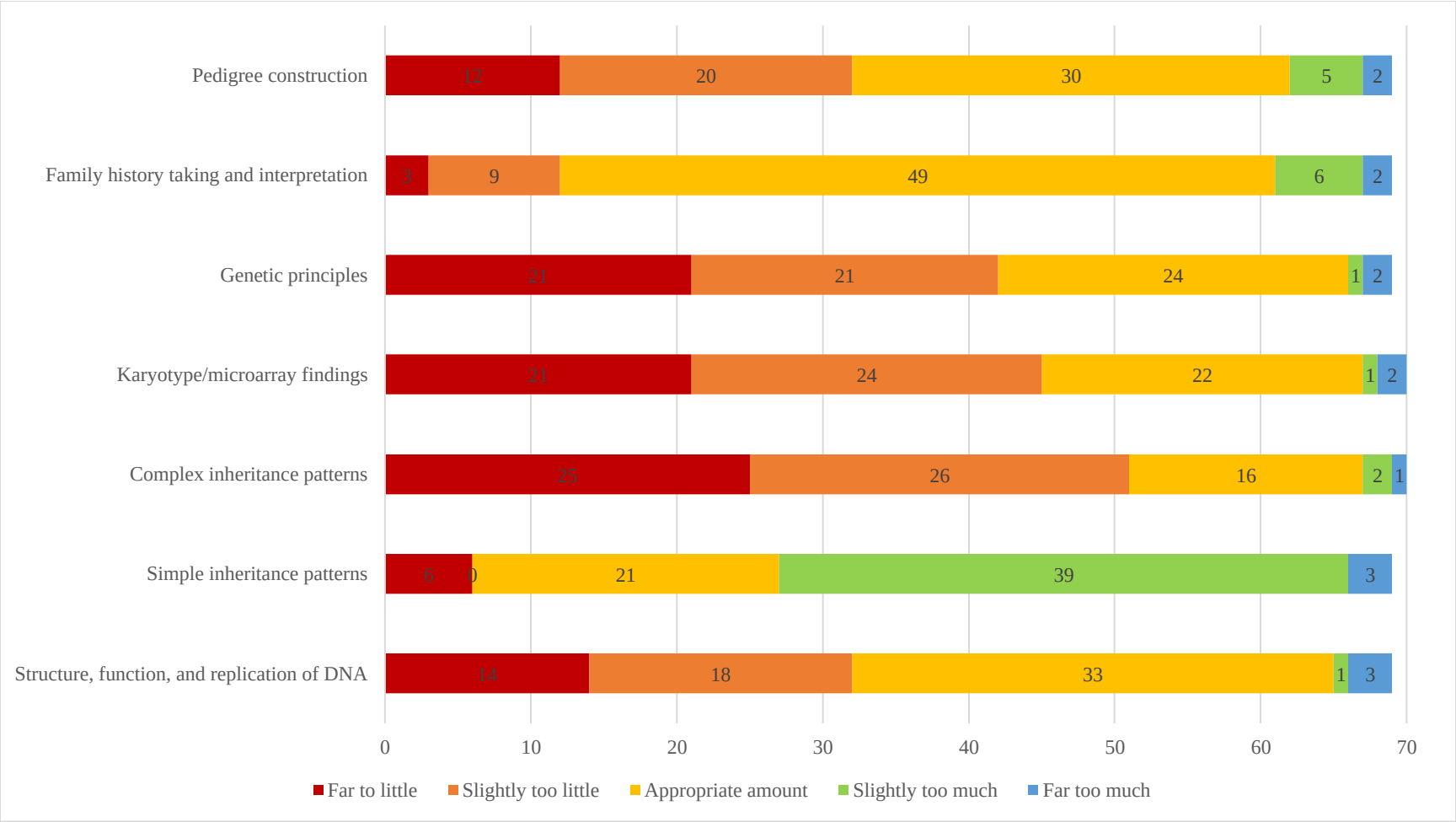

Figure 2.10 Perceived provider education based on necessity for clinic for concepts related to inheritance and family history

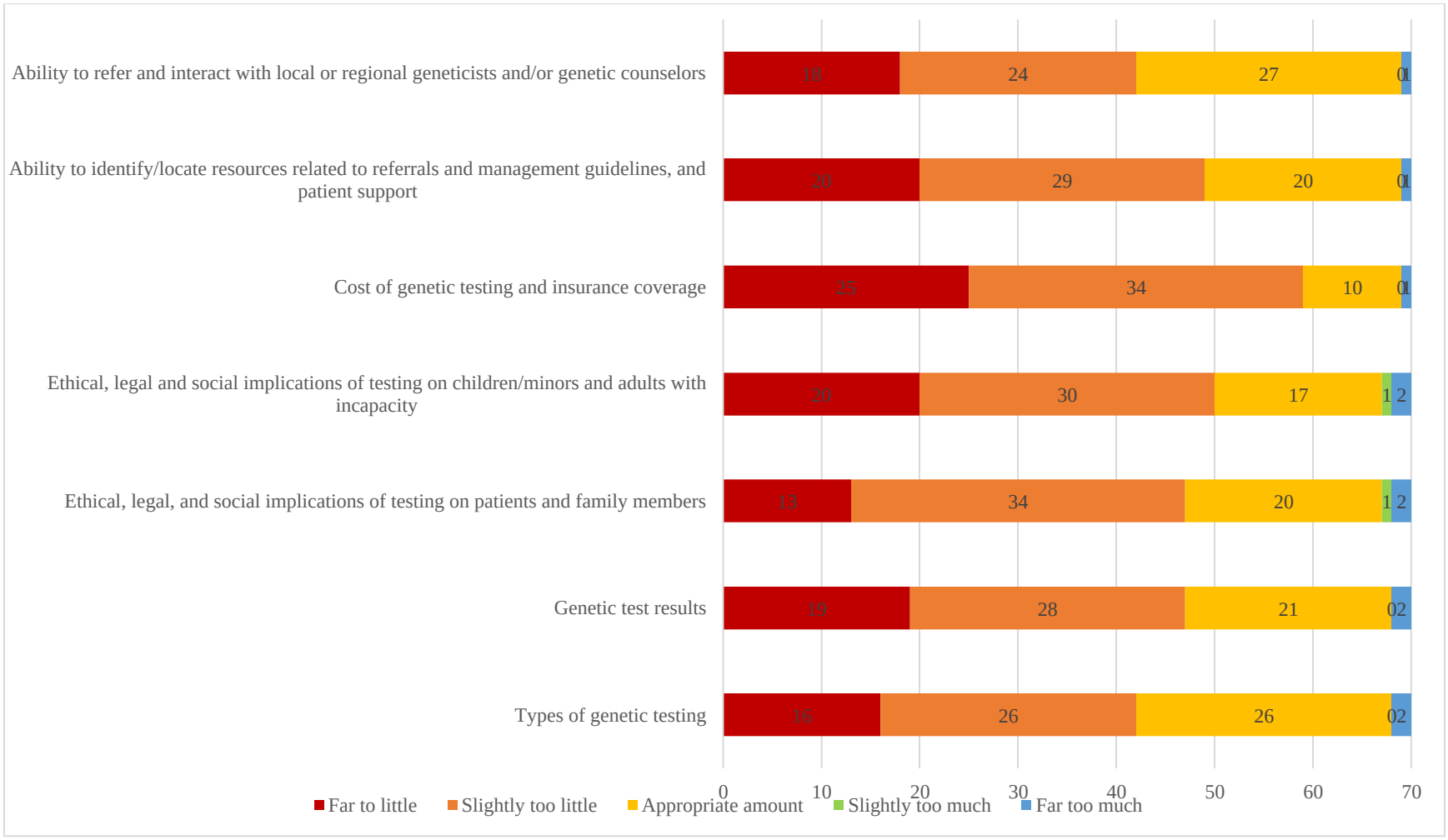

Figure 2.11 Perceived provider education based on necessity for clinic for concepts related to genetic testing, resources, and referrals

education compared to what is necessary for clinic. When asked if there were any other questions the providers would like to make regarding their genetics education up to this point in their career, 12 respondents provided additional comments. Through analysis of those additional comments, one main theme emerged. This theme was a desire for long-term/longitudinal continued education related to genetics, particularly by providers who have been done with formal education for many years (7/12). Another note made by two respondents to this question was that they had an abundance of experience with genetics either outside the clinic serving as an educator or due to mentorship by individuals with experience in clinical genetics.

2.4.4 Objective 4: How do providers prepare for patients with genetic conditions?

At the end of the questionnaire, participants were given the option of responding to three additional questions regarding how they would prepare to see a patient based on knowing some of their personal and family medical history. A total of 49.3% (35/71) of respondents opted in to participate and completed responses. Respondents were shown cases based on their clinical specialty and asked to describe what they would do for preparation, and what their plan for the appointment may be including any discussion topics, follow-up questions, referrals considered, and resources they may utilize. Themes were determined across all specialties and cases. Those practicing in internal medicine, family medicine, OBGYN, and pediatrics were given the opportunity to respond to three cases, and those practicing in geriatrics were given the opportunity to respond to two cases. Ten total themes were identified: (1) posing follow-up questions related to personal history, (2) follow-up lab work, imaging, or physical exam, (3) referral to a specialist, (4) referral to a genetic counselor/geneticist, (5) utilization of resources for

additional information, (6) patient education, (7) follow-up questions/detailed family history, (8) consideration/suggestion of a specific diagnosis, (9) assessment of patient concern, and (10) discussion of genetic testing options.

In 45.5% (45/99) of cases, providers indicated they would utilize follow-up questions to elicit more information from the patient regarding their personal medical history. Providers indicated they would utilize follow-up lab work, imaging, or physical exam for patient care in 46.5% (46/99) of cases. Referral to a specialist was considered or deemed appropriate in 31.3% (31/99) of cases. Referral to a genetic counselor or geneticist was suggested in 22.2% (22/99) of cases. Providers indicated they would utilize other resources for more information in 25.3% (25/99) of cases. Discussion of specific topics and patient education were mentioned as a likely portion of the appointment in 20.2% (20/99) of cases. Providers indicated they would ask more questions related to family history in 22.2% (22/99) of cases. A specific diagnosis was mentioned in the response of the provider in 10.1% (10/99) of cases. Patient concern was utilized by the provider as a guiding factor in the appointment in 6.1% (6/99) of cases. Lastly, providers would feel comfortable discussing genetic testing with the patient in 13.1% (13/99) of cases. Further breakdown of these themes by provider specialty can be seen in Table 2.8 to further understand utilization of these skills by provider specialty.

The cases presented to respondents were designed with their particular specialty in mind, and thus, it is important to recognize some differences may be due to the particular cases presented to them. Each case presented is described below, with breakdown of how providers from each specialty responded across all cases presented, as well as per case in order to give a clear picture of responses.

Table 2.8 Appointment plan of providers for patients with findings suggestive of genetic conditions by specialty

Item	Total	Family Medicine	Internal Medicine	OBGYN	Pediatrics	Geriatrics
Pose follow-up questions related to personal medical history	45.5% (45/99)	40.0% (6/15)	39.4% (13/33)	33.3% (7/21)	76.2% (16/21)	33.3% (3/9)
Follow-up lab work, imaging, or physical exam	46.5% (46/99)	33.3% (5/15)	63.6% (21/33)	33.3% (7/21)	57.1% (12/21)	11.1% (1/9)
Referral to specialist	31.3% (31/99)	40.0% (6/15)	33.3% (11/33)	0.0% (0/21)	57.1% (12/21)	22.2% (2/9)
Referral to genetic counselor/geneticist	22.2% (22/99)	20.0% (3/15)	36.4% (12/33)	14.3% (3/21)	14.3% (3/21)	11.1% (1/9)
Reference resources	25.3% (25/99)	13.3% (2/15)	21.2% (7/33)	4.8% (1/21)	38.1% (8/21)	77.8% (7/9)
Patient education	20.2% (20/99)	46.7% (7/15)	21.2% (7/33)	23.8% (5/21)	4.8% (1/21)	0.0% (0/9)
Follow-up questions related to family history	22.2% (22/99)	13.3% (2/15)	18.2% (6/33)	33.3% (7/21)	28.6% (6/21)	11.1% (1/9)
Consideration of a specific diagnosis	10.1% (10/99)	6.7% (1/15)	9.1% (3/33)	28.6% (6/21)	0.0% (0/21)	0.0% (0/9)
Utilization of patient concern as guiding factor	6.1% (6/99)	0.0% (0/15)	9.1% (3/33)	9.5% (2/21)	4.8% (1/21)	0.0% (0/9)
Discussion of genetic testing	13.1% (13/99)	13.3% (2/15)	3.0% (1/33)	42.9% (9/21)	0.0% (0/21)	11.1% (1/9)

Family Medicine. The following descriptions are the cases presented to providers who selected "family medicine" as their primary specialty (now referred to as "FM-Case 1", "FM-Case 2", and "FM-Case 3").

FM-Case 1. "You are seeing a 15 y.o. girl for her annual physical to clear her for sports. In the past, she has tested to be mildly anemic and has reported occasionally having some lightheadedness when standing up. She feels it is normal, as her mother has a history of fainting spells. You follow up on this and she reports that she has been doing well and has been taking the daily vitamins with iron that you recommended. Overall, she seems to be healthy and well-adjusted. Her intake form noted that her paternal grandfather had a heart-attack at age 50 but was reported to be overweight. Her paternal grandmother has diabetes. Her maternal grandmother had two heart attacks in her late 60s and early 70s, but was a chain smoker for most of her life. Her maternal aunt had a severe seizure while in college with an unknown cause. She has one cousin with ADHD and one cousin with a heart condition that she doesn't know much else about. She reports having no concerns for the appointment."

FM-Case 2. "You are seeing a 94 y.o. male patient for a regularly scheduled appointment. You notice that since the last time you have seen him, he has been diagnosed with his second colon cancer and has scheduled a colectomy. Other than the recent cancer diagnosis and becoming slightly overweight, his intake information does not suggest any new personal medical concerns. His family history indicates that his mother and grandfather both had colon cancer, and his aunt and two cousins both had uterine cancers. Furthermore, he has a son that was recently diagnosed with prostate cancer."

FM-Case 3. "You are seeing a current 24 y.o. male who has been dealing with rapidly progressing, bilateral hearing loss. He is presenting to you today with concerns that he is developing the condition his mother was recently diagnosed with, Maternally Inherited Diabetes and Deafness (MIDD). From previous visits, you know he is newly married and was planning on starting a family."

Each case was then followed with a prompt to discuss how the respondent would prepare to see the patient, and what their plan for the appointment would be including topics of discussion, follow-up questions, or referrals they would consider. Table 2.9 demonstrates the breakdown of responses by themes per case for family medicine providers.

Internal Medicine. The following descriptions are the cases presented to providers who selected "internal medicine" as their primary specialty (now referred to as "IM-Case 1", "IM-Case 2", and "IM-Case 3").

IM-Case 1. "You are seeing a 37 y.o. female patient for her annual physical visit. You are reviewing her intake form and see that she has no concerns for the appointment. At her last appointment she reported some back pain that was manageable with ibuprofen. Her family history section shows that her mother had hypertension before passing away at 59 y.o., her uncle passed due to a brain aneurysm, and her 33 y.o. cousins are on dialysis for renal failure."

IM-Case 2. "You are seeing a 22 y.o. African American female for concern of recurrent constipation and mild abdominal pain. In review of her chart, you see she recently had a cone biopsy to remove a small cervical cancer, and you noted some unusual dark spots on the inside of her mouth. She self-reported that her mother had

Table 2.9 Appointment plan of family medicine providers for patients with findings suggestive of a genetic condition by case

Item	Total	FM-Case 1	FM-Case 2	FM-Case 3
Pose follow-up questions related to personal medical history	40.0% (6/15)	60% (3/5)	20% (1/5)	40% (2/5)
Follow-up lab work, imaging, or physical exam	33.3% (5/15)	60% (3/5)	20% (1/5)	20% (1/5)
Referral to specialist	40.0% (6/15)	40% (2/5)	20% (1/5)	60% (3/5)
Referral to genetic counselor/geneticist	20.0% (3/15)	0% (0/5)	20% (1/5)	40% (2/5)
Reference resources	13.3% (2/15)	20% (1/5)	0% (0/5)	20% (1/5)
Patient education	46.7% (7/15)	20% (1/5)	80% (4/5)	40% (2/5)
Follow-up questions related to family history	13.3% (2/15)	40% (2/5)	0% (0/5)	0% (0/5)
Consideration of a specific diagnosis	6.7% (1/15)	0% (0/5)	0% (0/5)	20% (1/5)
Utilization of patient concern as guiding factor	0.0% (0/15)	0% (0/5)	0% (0/5)	0% (0/5)
Discussion of genetic testing	13.3% (2/15)	0% (0/5)	40% (2/5)	0% (0/5)

breast cancer at age 38, and her older brother had part of his intestines removed in his early teens, but she did not know why. Her grandfather died of colon cancer and mother's sister died in her early 40's from some kind of abdominal cancer."

IM-Case 3. "You are seeing a current 24 y.o. male who has been dealing with rapidly progressing, bilateral hearing loss. He is presenting to you today with concerns that he is developing the condition his mother was recently diagnosed with, Maternally Inherited Diabetes and Deafness (MIDD). From previous visits, you know he is newly married and was planning on starting a family."

Each case was then followed with a prompt to discuss how the respondent would prepare to see the patient and what their plan for the appointment would be including topics of discussion, follow-up questions, or referrals they would consider. Table 2.10 demonstrates the breakdown of responses by themes per case for family medicine providers.

Obstetrics and Gynecology/Women's Health. The following descriptions are the cases presented to providers who selected "obstetrics and gynecology/women's health" as their primary specialty (now referred to as "OBGYN-Case 1", "OBGYN-Case 2", and "OBGYN-Case 3").

OBGYN-Case 1. "You are seeing a 24 y.o. African American female in her first pregnancy. Her EDD is making her 10w5d. She nervous because her older sister has had multiple miscarriages, as did her mother. The remainder of her family history is limited."

OBGYN-Case 2. "You are seeing a 36 y.o. patient with irregular periods. She and her husband have been trying to have a second child. They already have a son with autism. They are concerned about their ability to conceive."

Table 2.10 Appointment plan of internal medicine providers for patient with findings suggestive of a genetic condition by case

Item	Total	IM-Case 1	IM-Case 2	IM-Case 3
Pose follow-up questions related to personal medical history	39.4% (13/33)	54.5% (6/11)	36.3% (4/11)	27.2% (3/11)
Follow-up lab work, imaging, or physical exam	63.6% (21/33)	63.6% (7/11)	72.7% (8/11)	54.5% (6/11)
Referral to specialist	33.3% (11/33)	18.1% (2/11)	45.4% (5/11)	36.3% (4/11)
Referral to genetic counselor/geneticist	36.4% (12/33)	18.1% (2/11)	45.4% (5/11)	45.4% (5/11)
Reference resources	21.2% (7/33)	18.1% (2/11)	9.1% (1/11)	36.3% (4/11)
Patient education	21.2% (7/33)	18.1% (2/11)	27.2% (3/11)	40% (2/11)
Follow-up questions related to family history	18.2% (6/33)	45.4% (5/11)	0% (0/11)	9.1% (1/11)
Consideration of a specific diagnosis	9.1% (3/33)	9.1% (1/11)	18.1% (2/11)	0% (0/11)
Utilization of patient concern as guiding factor	9.1% (3/33)	18.1% (2/11)	0% (0/11)	9.1% (1/11)
Discussion of genetic testing	3.0% (1/33)	0% (0/11)	9.1% (1/11)	0% (0/11)

OBGYN-Case 3. "You are seeing a 47 y.o. female for her annual visit. Her intake form shows that she is perimenopausal. She is having moderate hot flashes and some sleep irregularities. Her family history indicates that her grandmother had a history of DVT in her 90s. Her mother passed from a heart attack in her 70s, and her sister had a stroke at age 45. Her other two sisters are unable to take oral contraceptives due to heavy clotting during menstruation. Additionally, her younger brother was diagnosed with pancreatic cancer in his mid-40s, and her father died of metastatic prostate cancer. All of her siblings and the patient were reported to have melanomas, but she reported that they were 'outside kids' and sunbathers. She sees dermatology regularly to monitor her moles. She reports having no concerns to be addressed during the session."

Each case was then followed with a prompt to discuss how the respondent would prepare to see the patient and what their plan for the appointment would be including topics of discussion, follow-up questions, or referrals they would consider. Table 2.11 demonstrates the breakdown of responses by themes per case for family medicine providers.

Pediatrics. The following descriptions are the cases presented to providers who selected "pediatrics" as their primary specialty (now referred to as "Peds-Case 1", "Peds-Case 2", and "Peds-Case 3").

Peds-Case 1. "You are seeing a 3 y.o. male patient for follow up. You also see his older sister in your practice, who is 5 y.o.. Mom has expressed concerns to the nurse prior to you seeing them that her son isn't meeting his developmental milestones as quickly as his older sister did. You observe the child's speech is mildly delayed and appears disinterested with other people in the room. You also see in his chart that he has had

Table 2.11 Appointment play of OBGYNs for patients with findings suggestive of a genetic condition by case

Item	Total	OBGYN-Case 1	OBGYN-Case 2	OBGYN-Case 3
Pose follow-up questions related to personal medical history	33.3% (7/21)	28.6% (2/7)	57.1% (4/7)	14.3% (1/7)
Follow-up lab work, imaging, or physical exam	33.3% (7/21)	28.6% (2/7)	28.6% (2/7)	42.9% (3/7)
Referral to specialist	0.0% (0/21)	0% (0/7)	0% (0/7)	0% (0/7)
Referral to genetic counselor/geneticist	14.3% (3/21)	0% (0/7)	14.3% (1/7)	28.6% (2/7)
Reference resources	4.8% (1/21)	14.3% (1/7)	0% (0/7)	36.3% (0/7)
Patient education	23.8% (5/21)	28.6% (2/7)	14.3% (1/7)	28.6% (2/7)
Follow-up questions related to family history	33.3% (7/21)	57.1% (4/7)	28.6% (2/7)	14.3% (1/7)
Consideration of a specific diagnosis	28.6% (6/21)	14.3% (1/7)	28.6% (2/7)	42.9% (3/7)
Utilization of patient concern as guiding factor	9.5% (2/21)	0% (0/7)	14.3% (1/7)	14.3% (1/7)
Discussion of genetic testing	42.9% (9/21)	42.9% (3/7)	42.9% (3/7)	42.9% (3/7)

frequent colds and infections. You notice his growth, while within the normal range, is progressing slowly and he is on the small side for his age. Other notes in his chart include that he has asthma, and had some feeding difficulties as an infant."

Peds-Case 2. "You are seeing a 9 y.o. boy for the first time. His family has recently relocated to South Carolina from central Puerto Rico. His father has brought a copy of his chart from their previous pediatrician. His chart notes that he has a confirmed diagnosis of Hermansky-Pudlak syndrome."

Peds-Case 3. "You are seeing a 15 y.o. girl for her annual physical to clear her for sports. In the past, she has tested to be mildly anemic and has reported occasionally having some lightheadedness when standing up. She feels it is normal, as her mother has a history of fainting spells. You follow up on this and she reports that she has been doing well and has been taking the daily vitamins with iron that you recommended. Overall, she seems to be healthy and well-adjusted. Her intake form noted that her paternal grandfather had a heart-attack at age 50 but was reported to be overweight. Her paternal grandmother has diabetes. Her maternal grandmother had two heart attacks in her late 60s and early 70s, but was a chain smoker for most of her life. Her maternal aunt had a severe seizure while in college with an unknown cause. She has one cousin with ADHD and one cousin with a heart condition that she doesn't know much else about. She reports having no concerns for the appointment."

Each case was then followed with a prompt to discuss how the respondent would prepare to see the patient and what their plan for the appointment would be including topics of discussion, follow-up questions, or referrals they would consider. Table 2.12

Table 2.12 Appointment plan of pediatricians for patients with findings suggestive of a genetic condition by case

Item	Total	Peds-Case 1	Peds-Case 2	Peds-Case 3
Pose follow-up questions related to personal medical history	76.2% (16/21)	100% (7/7)	42.9% (3/7)	85.7% (6/7)
Follow-up lab work, imaging, or physical exam	57.1% (12/21)	71.4% (5/7)	0% (0/7)	100% (7/7)
Referral to specialist	57.1% (12/21)	100% (7/7)	57.1% (4/7)	14.3% (1/7)
Referral to genetic counselor/geneticist	14.3% (3/21)	14.3% (1/7)	28.6% (2/7)	0% (0/7)
Reference resources	38.1% (8/21)	14.3% (1/7)	100% (7/7)	0% (0/7)
Patient education	4.8% (1/21)	0% (0/7)	0% (0/7)	14.3% (1/7)
Follow-up questions related to family history	28.6% (6/21)	42.9% (3/7)	0% (0/7)	42.9% (3/7)
Consideration of a specific diagnosis	0.0% (0/21)	0% (0/7)	0% (0/7)	0% (0/7)
Utilization of patient concern as guiding factor	4.8% (1/21)	14.3% (1/7)	0% (0/7)	0% (0/7)
Discussion of genetic testing	0.0% (0/21)	0% (0/7)	0% (0/7)	0% (0/7)

demonstrates the breakdown of responses by themes per case for family medicine providers.

Geriatrics. The following descriptions are the cases presented to providers who selected "geriatrics" as their primary specialty (now referred to as "Ger-Case 1", and "Ger-Case 2").

Ger-Case 1. "You are seeing an 84 y.o. female patient in follow up for high blood pressure medication. She expresses no other concerns to you for the appointment. You note in her chart that she has some cutaneous lesions she sees dermatology for regularly and she had her uterus removed in her 30s due to painful fibroids. During casual conversation, the nurse notes that the patient mentioned her son was recently diagnosed with Reed's syndrome after his renal cell cancer diagnosis."

Ger-Case 2. "You are seeing a 76 y.o. male patient for a new patient appointment as they are transitioning into your care. Their chart indicates that the have arthritis, mild urinary leakage, and a clinical diagnosis of Type I Osteogenesis Imperfecta."

Each case was followed with a prompt to discuss how the respondent would prepare to see the patient and what their plan for the appointment would be including topics of discussion, follow up questions, or referrals. Table 2.13 demonstrates the breakdown of responses by themes per case for family medicine providers.

2.5 Discussion

The primary focus of this study was to identify which genetics skills and topics PCPs are not comfortable with and utilize this information to inform ways in which genetic counselors could provide support to these providers in the future. Additionally, identification of these topics can inform future continuing education directions and

Table 2.13 Appointment plan of geriatrics providers for patients with findings suggestive of a genetic condition by case

Item	Total	Ger-Case 1	Ger-Case 2
Pose follow-up questions related to personal medical history	33.3% (3/9)	40% (2/5)	25% (1/4)
Follow-up lab work, imaging, or physical exam	11.1% (1/9)	20% (1/5)	0% (0/4)
Referral to specialist	22.2% (2/9)	20% (1/5)	25% (1/4)
Referral to genetic counselor/geneticist	11.1% (1/9)	20% (1/5)	0% (0/4)
Reference resources	77.8% (7/9)	80% (4/5)	75% (3/4)
Patient education	0.0% (0/9)	0% (0/5)	0% (0/4)
Follow-up questions related to family history	11.1% (1/9)	20% (1/5)	0% (0/4)
Consideration of a specific diagnosis	0.0% (0/9)	0% (0/5)	0% (0/4)
Utilization of patient concern as guiding factor	0.0% (0/9)	0% (0/5)	0% (0/4)
Discussion of genetic testing	11.1% (1/9)	20% (1/5)	0% (0/4)

provide insight into where current education models may be lacking. Over half of providers indicated that 8/13 items listed were relevant to their clinical practice. Furthermore, for the five items that were deemed not relevant by majority of respondents, majority of respondents also reported not being comfortable with these items. This suggests there may be some correlation between level of comfort with an item and perceived relevance to practice. Therefore, when educating about these items it may be beneficial to also educate on the utility of these components of genetic healthcare in practice. These five items were: (1) structure, function, and DNA replication, (2) karyotype/microarray findings (as it relates to inheritance), (3) genetic principles, (4) pedigree construction, and (5) ethical, social, and legal implications of genetic testing for children/minors and adults with incapacity. Additionally, less than half of providers felt comfortable utilizing the majority (17/25) of items assessed in a clinical setting. This suggests that genetic counselors have an opportunity to support these providers across many aspects of genomic healthcare related to genetic principles, inheritance, family history, genetic testing, genetic test results, ethical, legal, and social implications of genetic testing, identification of referral and management guidelines, and cost of genetic testing and insurance coverage. It is further important to recognize provider type and specialty in assessing need, as it is clear that distinct groups have varying needs. This is to be expected as different providers have different educational and clinical exposures that would alter the comfort levels of these providers. For example, it is not surprising OBGYNs and pediatricians were significantly more comfortable with prenatal/newborn screening programs than other specialties, as they interact with these programs far more regularly. This aligns with the concept that flexibility is needed within the five

entrustable professional activities as described by Korf et al. (2014), and different groups may require more education outside of these core EPAs.

Furthermore, those attempting to remedy the education gaps seen may pay particular attention to items found to have a significant difference between groups, such as karyotype/microarray (as it relates to inheritance), and utilize the educational practices of the groups with increased comfort as a model to inform their own modules, or utilize the groups themselves as a resource to aid in educating the groups with lower levels of comfort. Genetic counselors may have the opportunity to serve these PCPs by aiding in conversations with patients directly or serving as a resource for PCPs by answering questions or concerns they may have. This may present as a clinical genetic counselor working in offices with PCPs, a local hotline for PCPs to ask clarifying questions or receive brief education themselves, or PCPs accessing lab genetic counselors with questions related to result interpretation or next steps for their patient, among many other possibilities.

Additionally, majority of these providers felt comfortable trusting the knowledge and training of genetic counselors for the items in which majority of providers themselves did not feel comfortable utilizing in clinic, suggesting that genetic counselors could fill the educational gaps reported both in this study and previous literature. However, providers need to feel comfortable and connected to genetic counselors in their area for this strategy to work successfully, and some education would likely still be needed in helping identify patients who should be initially considered for referral. Another possibility is the opportunity for genetic counselors to expand into the primary care setting where they can be useful in identifying patients who may benefit from

genetic counseling and provide better access to genetic counseling services and genetic counselor knowledge for connected providers.

The majority of providers who chose to comment on their current experiences with genetics health professionals reported that they had minimal to no experience, but when they did, the experience was typically positive. This is encouraging to recognize that genetic counselors can be a beneficial resource to these PCPs when utilized but disheartening to learn genetics health professionals are likely still being underutilized. The difference reported in comfort reaching out to genetics health professionals compared to other specialty providers found in this study or lack of awareness of available genetics health professionals described in prior literature (Klitzman et al., 2013) may be an explanation for the majority of providers still having minimal interaction with genetics health professionals. Carroll et al. (2016) even had providers in their study request having a direct contact or "buddy" in the genetics field to alleviate some of these feelings, suggesting that genetic counselors finding innovative ways to support their PCPs may be the preferred resolution to the education gap currently seen.

The identification of trust from these providers of the genetic counselor knowledgebase also provides a unique opportunity for genetic counselors to provide education to their PCPs. Of the 14 items assessed for perceived education, over half of providers found their education was less than needed for clinic for 10 of these items. This confirms what has been reported previously that providers often feel underprepared to provide genetic-based healthcare. Furthermore, prior research such as the Maradiegue et al. (2013) study looking at NP faculty integration of genetic concepts found that, while faculty comfort in teaching genetics had improved in a five-year window with targeted

educational programs, 30% of study participants still did not feel comfortable educating on basic genetic concepts and a larger proportion for more advanced topics such as complex modes of inheritance and pedigrees. Genetics health professionals may need to further aid in the education of developing providers via guest lecturing, consulting with faculty on genetics lectures, and continuing to provide education and support for the programs in their area, potentially in a larger capacity than has been done previously. Many genetic counselors recognize educating providers is a responsibility of theirs, but how many hours are truly spent on this job duty that could potentially help increase necessary and appropriate referrals to genetic counseling services?

Continuing to identify best practices for continuing education related to genetics is another important component to possibly close the education and comfort gap for these PCPs. Literature exists analyzing continuing education interventions as they relate to genetics education for healthcare providers. A controlled assessment of PAs who received a web-based educational model prior to seeing a standardized patient found that these PAs were able to ask more relevant medical questions and identify more family members of the patient with a history of cancer than their counterparts without spending additional time in the session (Roter et al., 2012). Furthermore, a review of interventions reported in the literature between January 2005 and January 2018 found that all educational approaches identified (immersive and experiential learning, interdisciplinary and interprofessional education, and electronic- and web-based approaches) could be effective strategies for education and produce long-term increases in confidence and knowledge. It also found educational interventions often increased confidence and knowledge in the short-term, but long-term studies suggested that this information was

often not retained unless the increases in knowledge and confidence were due to a prolonged educational strategy (Paneque et al., 2016; Rubanovich et al., 2018).

The conclusion from the literature review analyses and patient simulation study is that additional, appropriate education is capable of increasing both confidence and knowledge in providers, but it requires the dedication of providers to learn more and willingness to learn over time. The findings related to long-term education strategies align with the request made by some respondents for long-term/longitudinal continuing education opportunities and validate that PCPs would be willing to participate in these long-term educational opportunities should they be provided. Making these opportunities available to PCPs will require dedication from genetics health professionals to offer these opportunities over extended periods of time to implement true change in the field. Incorporating genetics rotations into educational programs has also been suggested as a remedy, and research has found this can be a beneficial strategy (Sloand et al., 2018). The difficulty with incorporating genetics rotations is that not all programs have adequate resources and accessibility to create such a rotation, and it does not address the needs of providers who are no longer in an educational program. However, providing opportunities for providers in a genetic counselor's network to sit in on genetic counseling appointments may help them embrace genetics in their own practice and aid in providers feeling connected to the genetics health professionals around them. It may also provide opportunities to educate providers firsthand on distinct counseling skills (such as counseling on test limitations or against unwarranted testing) as desired by some respondents in this study from individuals who are trained and do so regularly.

It is unlikely that one single approach will fulfill the needs of genetics education for PCPs. However, having an understanding of which topics providers feel they need more education to adequately provide genomic healthcare to their patients will be valuable in all forms of education for PCPs. Understanding how PCPs approach continuing education when working in small or private practices may be the next step in helping implement a broad-scale increase in PCP utilization of genetic practices, as connecting PCPs to continuing education opportunities is often a challenge when they are not directly connected to a large hospital or academic setting.

Another interesting finding of this study was the lack of PCP awareness of resources available to them. The resources listed on the study are commonly used resources within the genetic health professional population due to their accurate, up-to-date data regarding genetics information. Previous literature has described a desire by providers for written, accessible resources to reference and increased awareness of existing resources may be extremely valuable knowledge for providers. This study was conducted prior to the integration of Genetics Home Reference into Medscape, and it will be interesting to see how this integration may or may not improve awareness of this resource to providers. Approximately a quarter of providers who responded to the vignettes indicated they would utilize resources to help prepare for the case, and having awareness of resources based in genetics may help them both prepare for the case, but also continue to develop a deeper understanding of genetic conditions and implications the diagnosis may have on care. Furthermore, all cases presented would have been appropriate patients to refer to see a genetics health professional or for the provider to reach out to a genetic counselor for more information, if needed. However, this was only

mentioned by the provider in 22.2% of cases, whereas referral to other specialty providers was suggested in the 31.3% of cases. This further suggests a barrier between PCPs and referral to genetics that could be improved.

One of the limitations of this study include the fact that the survey was restricted to PCPs in South Carolina. Thus, the results may not be representative of other states or regions of the world. Furthermore, some of the healthcare systems utilized to recruit members are well connected to genetic counseling and genetics services, and the results may be biased due to their increased awareness of these providers within their healthcare system. However, the survey did not request respondents to indicate which health system they were connected with, and therefore the survey may be representative of a state with large academic institutions that are well connected to genetic services as well as smaller, rural institutions that are often less connected. Furthermore, a larger sample population may have found more statistically significant differences between analyzed groups. About half of respondents started practicing within the past 14 years, with nearly a fifth of respondents having started practicing in the past 4 years, and thus it does not seem the study was significantly biased by an unusually large portion of late-career respondents.

Lastly, it was interesting to note that majority of genetic based items analyzed for level of comfort found a fairly high level of comfort for providers. It is possible that this data may be slightly skewed by individuals who felt a low level of comfort, but did not indicate they were comfortable with the item because they were unaware the question would be followed up with the ability to select a magnitude of comfort. Thus, individuals who have a low level of comfort have been placed in the "not comfortable" category,

unfairly increasing this group, and the values of magnitude of comfort may be higher because the low comfort respondents were not prompted to respond to this question.

Chapter 3. Conclusions

As the field of genetics continues to grow, PCPs will undoubtedly continue to expand their own role by incorporating genetics into their practice. By understanding what providers currently need to help integrate genetics in practice, a step is taken forward in improving the overall care of patients. With a deeper understanding of what providers find relevant and where they feel they need further education, we can hopefully improve comfort levels and remedy the lag in integration of genetics in primary care. Genetics health professionals will need to continue to be a resource to these providers in their continuing education, as well as in clinic. Improving interactions between PCPs and genetics health professionals should be a continuing goal as it is important for both patient and provider understanding, as well as the appropriate connection of providers to resources and referrals.

This study has prompted a multitude of questions yet to be clearly answered. Future directions resulting from the research may include: developing genetics curricula for physicians, NPs, and PAs in training; developing continuing education modules for providers; further assessing the correlation between comfort levels and perceived relevance to practice for various genetic-based topics; understanding the role and potential utility of genetic counselors in primary care; assessing the differences between the relationships of PCPs and genetics health professionals versus PCPs and other specialty providers; and understanding the best approach to connecting PCPs to continuing education opportunities related to genetics.

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Appendix A. Survey

1. Introduction

Q0 Thank you for considering to participate in the study of providers' comfort with utilization of genetics in practice. This questionnaire will contain a series of multiple choice, multi-select, slider scale, and open-ended questions attempting to understand the current status of physician, nurse practitioner, and physician assistant comfort with various genetics concepts and skills. Your participation is completely voluntary and you may choose to skip questions if you prefer not to answer.

The last page of this survey will contain a separate link where you may enter a raffle for the chance to win one of three $25 Amazon gift cards. There is also an option to volunteer to complete three additional questions at the end of the survey.

If you are willing to participate in this study, please click the "next" button below. If not, please exit the browser.

2. Inclusion/Exclusion Criteria

Q1 Please select which of the following categories applies to you:

- Physician (1)
- Nurse Practitioner (2)
- Physician Assistant (3)
- Other (nurse, office staff, etc.) (4)

Skip To: End of Survey if Q1= Other (nurse, office staff, etc.)

Q2 Which specialty do you identify with most?

- Family medicine (1)
- Internal medicine (2)
- Obstetrics and Gynecology/Women's Health (3)
- Pediatrics (4)
- Geriatrics (5)
- Other (6)

Skip To: End of Survey if Q2= Other

3. Relevance

Q3 Please indicate which of the following genetics-based concepts related to inheritance and family history you believe are relevant for your practice in clinic (select all that apply):

- ☐ Structure, function, and replication of DNA
- ☐ Inheritance patterns (dominant, recessive, x-linked)
- ☐ Karyotype/microarray findings
- ☐ Genetic principles (reduced penetrance, variable expressivity, two-hit hypothesis)
- ☐ Family history taking and interpretation
- ☐ Pedigree construction

Q4 Please indicate which of the following aspects of genetic testing you believe are relevant for your practice in clinic (select all that apply):

- ☐ Types of genetic testing (such as" chromosome analysis, microarray, newborn screening, pharmacogenomics, single gene testing)
- ☐ Genetic test results (pathogenic, benign, variant of uncertain significance, incidental)
- ☐ Ethical, legal, and social implications of genetic testing on patients and their families, including those who are asymptomatic
- ☐ Ethical, legal, and social implications of genetic testing on minors and adults with incapacity

Q5 Please indicate which of the following aspects related to genetics resources and referrals you believe are relevant for your practice in clinic (select all that apply):

- ☐ Cost of genetic testing and insurance coverage
- ☐ Ability to identify/locate resources related to referral and management guidelines and support for patients with genetic conditions
- ☐ Ability to refer and interact with local or regional geneticists and/or genetic counselors

Q6 What other genetic topics/concepts not listed previously do you find important for practice?

[Open-ended]

4. Comfort

Q7 Please indicate which of the following genetics-based concepts related to inheritance and family history you would feel comfortable discussing and utilizing in clinic (select all that apply):

- ☐ Structure, function, and replication of DNA (1)
- ☐ Simple patterns of inheritance (autosomal recessive, autosomal dominant, sex—linked) (2)
- ☐ Complex patterns of inheritance (repeat expansion, epigenetics, mitochondrial) (3)
- ☐ Karyotype/microarray findings (4)
- ☐ Genetic principles (reduced penetrance, variable expressivity, two-hit hypothesis) (5)
- ☐ Family history taking and interpretation (6)
- ☐ Pedigree construction (7)

Skip To: Q9 if Q7= 0

Q8 Please indicate your level of comfort in discussing and utilizing the following genetics-based concepts related to family history and inheritance in clinic:

	Extremely Comfortable	Moderately Comfortable	Slightly Comfortable	Neither Comfortable or Uncomfortable
Display if Q7 = 1				
Structure, function, and DNA replication	O	O	O	O
Display if Q7 = 2				
Simple patterns of inheritance (autosomal recessive, autosomal dominant, sex-linked)	O	O	O	O
Display if Q7 = 3				
Complex patterns of inheritance (repeat expansion, epigenetics, mitochondrial)	O	O	O	O
Display if Q7 = 4				
Karyotype/microarray findings	O	O	O	O
Display if Q7 = 5				
Genetic principles (reduced penetrance, variable expressivity, two-hit hypothesis)	O	O	O	O
Display if Q7 = 6				
Family history taking and interpretation	O	O	O	O
Display if Q7 = 7				
Pedigree construction	O	O	O	O

Q9 Please indicate which of the following types of genetic testing and test results you would feel comfortable discussing and utilizing in clinic (select all that apply):

- ☐ Karyotype (1)
- ☐ Microarray (2)
- ☐ Single gene testing (3)
- ☐ Multigene panel testing (4)
- ☐ Whole exome/whole genome sequencing (5)
- ☐ Pharmacogenomic testing (6)
- ☐ Prenatal/newborn screening programs (7)
- ☐ Pathogenic test results (8)
- ☐ Variant of uncertain significance (VUS) test results (9)
- ☐ Negative test results (10)
- ☐ Incidental findings on testing (11)

Skip To: Q11 if Q9= 0

Q10 Please indicate your level of comfort in discussing and utilizing the following types of genetic testing and test results in clinic:

	Extremely Comfortable	Moderately Comfortable	Slightly Comfortable	Neither Comfortable or Uncomfortable
Display if Q9 = 1 Karyotype	O	O	O	O
Display if Q9 = 2 Microarray	O	O	O	O
Display if Q9 = 3 Single gene testing	O	O	O	O
Display if Q9 = 4 Multigene panel testing	O	O	O	O
Display if Q9 = 5 Whole exome/whole genome sequencing	O	O	O	O
Display if Q9 = 6 Pharmacogenomic testing	O	O	O	O
Display if Q9 = 7 Prenatal/newborn screening programs	O	O	O	O
Display if Q9 = 8 Pathogenic test results	O	O	O	O
Display if Q9 = 9 Variant of uncertain significance (VUS) test results	O	O	O	O
Display if Q9 = 10 Negative test results	O	O	O	O
Display if Q9 = 11 Incidental findings on testing	O	O	O	O

Q11 Please indicate if you would feel comfortable discussing the ethical, legal, and social implication of genetic testing for each of the following groups (select all that apply):

- ☐ The patient (1)
- ☐ The patient's family members (including those who are asymptomatic) (2)
- ☐ Children/minors (3)
- ☐ Adults with incapacity (4)

Skip To: Q13 if Q11= 0

Q12 Please indicate how comfortable you are discussing the ethical, legal, and social implications of genetic testing for each of the following groups:

	Extremely Comfortable	Moderately Comfortable	Slightly Comfortable	Neither Comfortable or Uncomfortable
Display if Q11 = 1 The patient	O	O	O	O
Display if Q11 = 2 The patient's family members (including those who are asymptomatic)	O	O	O	O
Display if Q11 = 3 Children/minors	O	O	O	O
Display if Q11 = 4 Adults with incapacity	O	O	O	O

Q13 Please indicate which of the following aspects related to genetics resources and referrals you would feel comfortable discussing and utilizing in clinic (select all that apply):

- ☐ Cost of genetic testing, genetics appointments, and insurance coverage (1)
- ☐ Ability to identify/locate resources related to referral and management guidelines and support for patients with genetic conditions (2)
- ☐ Ability to refer and interact with local or regional geneticists and/or genetic counselors (3)

Skip To: Q15 if Q13= 0

Q14 Please indicate your level of comfort in discussing and utilizing the following aspects related to genetics resources and referrals in clinic:

	Extremely Comfortable	Moderately Comfortable	Slightly Comfortable	Neither Comfortable or Uncomfortable
Display if Q13 = 1 Cost of genetic testing, genetics appointments, and insurance coverage	O	O	O	O
Display if Q13 = 2 Ability to identify/locate resources related to referral and management guidelines, and support for patients with genetic conditions	O	O	O	O
Display if Q13 = 3 Ability to refer and interact with local or regional geneticists and/or genetic counselors	O	O	O	O

5. Education

Q15 Please indicate your attitude towards the amount of education you have received on each of the following genetics concepts related to inheritance and family history up to this point in your career:

	Far too much	Slightly too much	Appropriate amount	Slightly too little	Far too little
Structure, function, and replication of DNA	O	O	O	O	O
Simple patterns of inheritance (autosomal recessive, autosomal dominant, sex-linked)	O	O	O	O	O
Complex patterns of inheritance (repeat expansion, epigenetics, mitochondrial)	O	O	O	O	O
Karyotype/microarray findings	O	O	O	O	O
Genetic principles (reduced penetrance, variable expressivity, two-hit hypothesis)	O	O	O	O	O
Family history taking and interpretation	O	O	O	O	O
Pedigree construction	O	O	O	O	O

Q 16 Please indicate your attitude towards the amount of education you have received on each of the following aspects of genetic testing up to this point in your career:

	Far too much	Slightly too much	Appropriate amount	Slightly too little	Far too little
Types of genetic testing (such as: chromosome analysis, microarray, newborn screening, pharmacogenomics, single gene testing)	O	O	O	O	O
Genetic test results (pathogenic, benign, VUS, incidental)	O	O	O	O	O
Ethical, legal, and social implications of genetic testing on patients and their families, including those who are asymptomatic	O	O	O	O	O
Ethical, legal, and social implications of genetic testing on minors and adults with incapacity	O	O	O	O	O

Q17 Please indicate your attitude towards the amount of education you have received on each of the following aspects related to genetics resources and referrals up to this point in your career:

	Far too much	Slightly too much	Appropriate amount	Slightly too little	Far too little
Cost of genetic testing, genetics appointments, and insurance coverage	O	O	O	O	O
Ability to identify/locate resources related to referral and management guidelines and support for patients with genetic conditions	O	O	O	O	O
Ability to refer and interact with local or regional geneticists and/or genetic counselors	O	O	O	O	O

Q18 Are there any other comments you would like to make about your genetics education up to this point in your career (including continuing education experiences):

[Open-ended]

6. Resources

Q19 Please indicate which of the following web-based resources you are aware of (select all that apply):

- ☐ UpToDate (1)
- ☐ Medscape (2)
- ☐ Epocrates (3)
- ☐ Online Mendelian Inheritance in Man (OMIM) (4)
- ☐ GeneReviews (5)
- ☐ Genetics Home Reference (6)
- ☐ Genetics and Rare Disease Information Center (GARD) (7)
- ☐ Orphanet (8)
- ☐ MedGen (9)
- ☐ ClinVar (10)
- ☐ Genetic Testing Registry (11)
- ☐ GeneTests (12)
- ☐ Pharmacogenomics Knowledgebase (PharmGkb) (13)

Skip To: Q21 if Q19= 0

Q20 Please indicate which of the following web-based resources you are comfortable with using (select all that apply):

- ☐ UpToDate
  Display if Q19=1
- ☐ Medscape
  Display if Q19=2
- ☐ Epocrates
  Display if Q19=3
- ☐ Online Mendelian Inheritance in Man (OMIM)
  Display if Q19=4
- ☐ GeneReviews
  Display if Q19=5
- ☐ Genetics Home Reference
  Display if Q19=6
- ☐ Genetics and Rare Disease Information Center (GARD)
  Display if Q19=7
- ☐ Orphanet
  Display if Q19=8
- ☐ MedGen
  Display if Q19=9
- ☐ ClinVar
  Display if Q19=10
- ☐ Genetic Testing Registry
  Display if Q19=11
- ☐ GeneTests
  Display if Q19=12
- ☐ Pharmacogenomics Knowledgebase (PharmGkb)
  Display if Q19=13

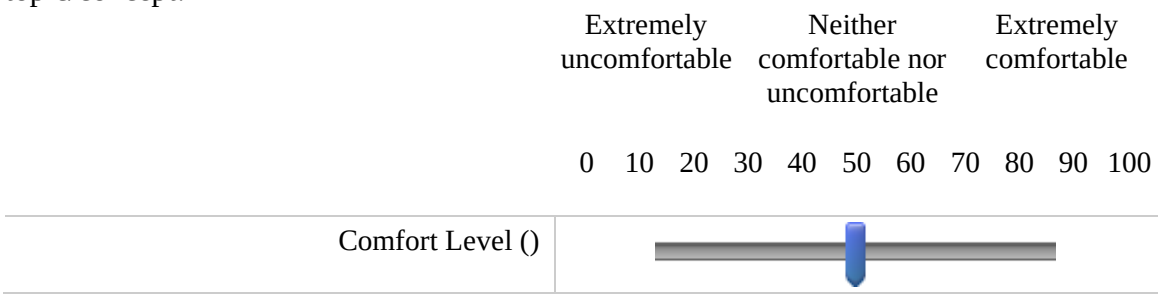
Q21 Please indicate your desire to learn more about genetics-based resources available to you:

	No interest	Little interest	Somewhat interested	Mildly interested	Extremely interested
	0 10	20 30	40 50	60 70	80 90 100
Interest ()					

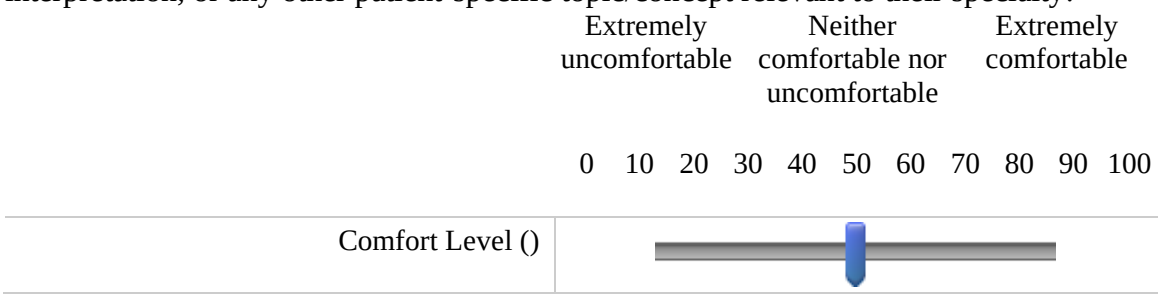
Q22 What resources do you currently rely on when preparing to care for a patient suspicious for or with a known genetic condition?

[Open-ended]

Q23 Please indicate your comfort reaching out to a genetics health professional to answer a question regarding referrals, test result interpretation, or any other patient-specific topic/concept:



Q24 Please indicate your comfort reaching out to a specialized health professional (such as a neurologist or endocrinologist) to answer a question regarding referrals, test result interpretation, or any other patient-specific topic/concept relevant to their specialty:



Q25 Please indicate which of the following genetics topics/concepts you would feel comfortable trusting the knowledge of a genetic counselor to explain to you (select all that apply):

- ☐ Structure, function, and replication of DNA
- ☐ Inheritance patterns (dominant, recessive, X-linked)
- ☐ Karyotype/microarray findings
- ☐ Genetic principles (reduced penetrance, variable expressivity, two-hit hypothesis)
- ☐ Family history taking and interpretation
- ☐ Pedigree construction
- ☐ Types of genetic testing (such as: chromosome analysis, microarray, newborn screening, pharmacogenomics, single gene testing)
- ☐ Genetic test results (pathogenic, benign, variant of uncertain significance, incidental)
- ☐ Ethical, legal, and social implications of genetic testing on patients and their families, including those who are asymptomatic
- ☐ Ethical, legal, and social implication of genetic testing on minors and adults with incapacity
- ☐ Cost of genetic testing and insurance coverage
- ☐ Ability to identify/locate resources related to referral and management guidelines and support for patients with genetic conditions
- ☐ Ability to refer and interact with local or regional geneticists and/or genetic counselors

Q26 What has your experience been utilizing genetic counselors as a resource/member of your interdisciplinary care team?

[Open-ended]

7. Vignettes

Q27 Thank you for your responses so far! You now have the option of completing three additional questions prior to answering demographic information. These three questions will contain three short vignettes of cases you may see in clinic and request you share your thoughts on how you would approach seeing the patient. This information will be valuable insight in understanding the approach of practitioners to address patients who are diagnoses with or are suspicious for a genetic condition.

If you do choose to answer these three questions, you will have the opportunity to enter a second raffle for a fourth $25 Amazon gift care exclusive to those who complete these questions. You will still be eligible to win one of the three $25 gift cards from completing the previous questions.

Are you interested in answering these three additional questions?

- Yes (1)
- No (2)

Skip To: Q43 if Q27= 2

Display this question: If Q2= Family medicine

Q28 You are seeing a 15 y.o. girl for her annual physical to clear her for sports. In the past, she has tested to be mildly anemic and has reported occasionally having some lightheadedness when standing up. She feels it is normal, as her mother has a history of fainting spells. You follow up on this and she reports that she has been doing well and has been taking the daily vitamins with iron that you recommended. Overall she seems to be healthy and well-adjusted. Her intake form noted that her paternal grandfather had a heart-attack at age 50 but was reported to be overweight. Her paternal grandmother has diabetes. Her maternal grandmother had two heart attacks in her late 60s and early 70s, but was a chain smoker for most of her life. Her maternal aunt had a severe seizure while in college with an unknown cause. She has one cousin with ADHD and one cousin with a heart condition that she doesn't know much else about. She reports having no concerns for the appointment.

How would you prepare before walking into the room with this patient, and what would be your plan for the appointment? (topics you may discuss, follow-up questions you may ask, referrals you would consider making, resources you may reference, etc.)

[Open-ended]

Display this question: If Q2= Family medicine

Q29 You are seeing a 94 y.o. male patient for a regularly scheduled appointment. You notice that since the last time you have seen him, he has been diagnosed with his second colon cancer and has scheduled a colectomy. Other than the recent cancer diagnosis and becoming slightly overweight, his intake information does not suggest any new personal medical concerns. His family history indicates that his mother and grandfather both had colon cancer, and his aunt and two cousins both had uterine cancers. Furthermore, he has a son that was recently diagnosed with prostate cancer.

How would you prepare before walking into the room with this patient, and what would be your plan for the appointment? (topics you may discuss, follow-up questions you may ask, referrals you would consider making, resources you may reference, etc.)

[Open-ended]

Display this question: If Q2= Family medicine

Q30 You are seeing a current 24 y.o. male who has been dealing with rapidly progressing, bilateral hearing loss. He is presenting to you today with concerns that he is developing the condition his mother was recently diagnosed with, Maternally Inherited Diabetes and Deafness (MIDD). From previous visits, you know he is newly married and was planning on starting a family.

How would you prepare before walking into the room with this patient, and what would be your plan for the appointment? (topics you may discuss, follow-up questions you may ask, referrals you would consider making, resources you may reference, etc.)

[Open-ended]

Display this question: If Q2= Internal medicine

Q31 You are seeing a 37 y.o. female patient for her annual physical visit. You are reviewing her intake form and see that she has no concerns for the appointment. At her last appointment she reported some back pain that was manageable with ibuprofen. Her family history section shows that her mother had hypertension before passing away at 59 y.o., her uncle passed due to a brain aneurysm, and her 33 y.o. cousins are on dialysis for renal failure.

How would you prepare before walking into the room with this patient, and what would be your plan for the appointment? (topics you may discuss, follow-up questions you may ask, referrals you would consider making, resources you may reference, etc.)

[Open-ended]

Display this question: If Q2= Internal medicine

Q32 You are seeing a 22 y.o. African American female for concern of recurrent constipation and mild abdominal pain. In review of her chart, you see she recently had a cone biopsy to remove a small cervical cancer, and you noted some unusual dark spots on the inside of her mouth. She self-reported that her mother had breast cancer at age 38, and her older brother had part of his intestines removed in his early teens, but she did not know why. Her grandfather died of colon cancer and mother's sister died in her early 40's from some kind of abdominal cancer.

How would you prepare before walking into the room with this patient, and what would be your plan for the appointment? (topics you may discuss, follow-up questions you may ask, referrals you would consider making, resources you may reference, etc.)

[Open-ended]

Display this question: If Q2= Internal medicine

Q33 You are seeing a current 24 y.o. male who has been dealing with rapidly progressing, bilateral hearing loss. He is presenting to you today with concerns that he is developing the condition his mother was recently diagnosed with, Maternally Inherited Diabetes and Deafness (MIFF). From previous visits, you know he is newly married and was planning on starting a family.

How would you prepare before walking into the room with this patient, and what would be your plan for the appointment? (topics you may discuss, follow-up questions you may ask, referrals you would consider making, resources you may reference, etc.)

[Open-ended]

Display this question: If Q2= Obstetrics and Gynecology/Women's Health

Q34 You are seeing a 24 y.o. African American female in her first pregnancy. Her EDD is making her 10w5d. She nervous because her older sister has had multiple miscarriages, as did her mother. The remainder of her family history is limited.

How would you prepare before walking into the room with this patient, and what would be your plan for the appointment? (topics you may discuss, follow-up questions you may ask, referrals you would consider making, resources you may reference, etc.)

[Open-ended]

Display this question: If Q2= Obstetrics and Gynecology/Women's Health

Q35 You are seeing a 36 y.o. patient with irregular periods. She and her husband have been trying to have a second child. They already have a son with autism. They are concerned about their ability to conceive.

How would you prepare before walking into the room with this patient, and what would be your plan for the appointment? (topics you may discuss, follow-up questions you may ask, referrals you would consider making, resources you may reference, etc.)

[Open-ended]

Display this question: If Q2= Obstetrics and Gynecology/Women's Health

Q36 You are seeing a 47 y.o. female for her annual visit. Her intake form shows that she is perimenopausal. She is having moderate hot flashes and some sleep irregularities. Her family history indicates that her grandmother had a history of DVT in her 90s. Her mother passed from a heart attack in her 70s, and her sister had a stroke at age 45. Her other two sisters are unable to take oral contraceptives due to heavy clotting during menstruation. Additionally, her younger brother was diagnosed with pancreatic cancer in his mid 40s, and her father died of metastatic prostate cancer. All of her siblings and the patient were reported to have melanomas, but she reported that they were "outside kids" and sunbathers. She sees dermatology regularly to monitor her moles. She reports having no concerns to be addressed during the session.

How would you prepare before walking into the room with this patient, and what would be your plan for the appointment? (topics you may discuss, follow-up questions you may ask, referrals you would consider making, resources you may reference, etc.)

[Open-ended]

Display this question: If Q2= Pediatrics

Q37 You are seeing a 3 y.o. male patient for follow up. You also see his older sister in your practice, who is 5 y.o.. Mom has expressed concerns to the nurse prior to you seeing them that her son isn't meeting his developmental milestones as quickly as his older sister did. You observe the child's speech is mildly delayed and appears disinterested with other people in the room. You also see in his chart that he has had frequent colds and infections. You notice his growth, while within the normal range, is progressing slowly and he is on the small side for his age. Other notes in his chart include that he has asthma, and had some feeding difficulties as an infant.

How would you prepare before walking into the room with this patient, and what would be your plan for the appointment? (topics you may discuss, follow-up questions you may ask, referrals you would consider making, resources you may reference, etc.)

[Open-ended]

Display this question: If Q2= Pediatrics

Q38 You are seeing a 9 y.o. boy for the first time. His family has recently relocated to South Carolina from central Puerto Rico. His father has brought a copy of his chart from their previous pediatrician. His chart notes that he has a confirmed diagnosis of Hermansky-Pudlak syndrome.

How would you prepare before walking into the room with this patient, and what would be your plan for the appointment? (topics you may discuss, follow-up questions you may ask, referrals you would consider making, resources you may reference, etc.)

[Open-ended]

Display this question: If Q2= Pediatrics

Q39 You are seeing a 15 y.o. girl for her annual physical to clear her for sports. In the past, she has tested to be mildly anemic and has reported occasionally having some lightheadedness when standing up. She feels it is normal, as her mother has a history of fainting spells. You follow up on this and she reports that she has been doing well and has been taking the daily vitamins with iron that you recommended. Overall she seems to be healthy and well-adjusted. Her intake form noted that her paternal grandfather had a heart-attack at age 50 but was reported to be overweight. Her paternal grandmother has diabetes. Her maternal grandmother had two heart attacks in her late 60s and early 70s, but was a chain smoker for most of her life. Her maternal aunt had a severe seizure while in college with an unknown cause. She has one cousin with ADHD and one cousin with a heart condition that she doesn't know much else about. She reports having no concerns for the appointment.

How would you prepare before walking into the room with this patient, and what would be your plan for the appointment? (topics you may discuss, follow-up questions you may ask, referrals you would consider making, resources you may reference, etc.)

[Open-ended]

Display this question: If Q2= Geriatrics

Q41 You are seeing a 84 y.o. female patient in follow up for high blood pressure medication. She expresses no other concerns to you for the appointment. You note in her chart that she has some cutaneous lesions she sees dermatology for regularly and she had her uterus removed in her 30s due to painful fibroids. During casual conversation, the nurse notes that the patient mentioned her son was recently diagnosed with Reed's syndrome after his renal cell cancer diagnosis.

How would you prepare before walking into the room with this patient, and what would be your plan for the appointment? (topics you may discuss, follow-up questions you may ask, referrals you would consider making, resources you may reference, etc.)

[Open-ended]

Display this question: If Q2= Geriatrics

Q42 You are seeing a 76 y.o. male patient for a new patient appointment as they are transitioning into your care. Their chart indicates that the have arthritis, mild urinary leakage, and a clinical diagnosis of Type I Osteogenesis Imperfecta.

How would you prepare before walking into the room with this patient, and what would be your plan for the appointment? (topics you may discuss, follow-up questions you may ask, referrals you would consider making, resources you may reference, etc.)

[Open-ended]

8. Demographics

Q43 What gender do you identify as?

- Male
- Female
- Non-binary

Q44 What best describes your ethnicity?

- ☐ Caucasian
- ☐ African-American
- ☐ Latino or Hispanic
- ☐ Asian
- ☐ Native American
- ☐ Native Hawaiian or Pacific Islander
- ☐ Other: _____
- ☐ Unknown

Q45 What is the highest degree or level of education you have completed?

- Some high school
- High school/GED
- Some college
- Associates Degree
- Bachelors Degree
- Masters Degree
- Doctoral Degree

Q46 How many years have you been in practice?

Option Range: 1 year or less, 1 year, 2 years, … 49 years, 50+ years

Q47 In what type of setting do you practice?

- Urban
- Suburban
- Rural

Appendix B. Recruitment Material

1. Direct Email

Subject Line: Research Survey--Amazon Gift Card Raffle for Completion!

Body Content:

Hello [Name],

You are being invited to participate in a student-led graduate research study through the University of South Carolina genetic counseling program. This study will help complete the principle investigator's degree requirements.

We are conducting a survey to understand provider comfort with genetics in their current daily practice. The survey will assess your attitudes towards which genetics-based skills and knowledge are relevant in your practice, how comfortable you feel utilizing genetics concepts with a patient, and how you perceive your level of genetics education up to this point in your medical career.

We are interested in the responses of physicians, nurse practitioners, and physician assistants practicing in the following areas:

- Family Medicine
- Internal Medicine
- Obstetrics and Gynecology/Women's Health
- Pediatrics
- Geriatrics

At the completion of the survey, you will have the opportunity to enter into a raffle for one of three $25 Amazon gift cards. Respondents may elect to complete an additional three questions at the end of the survey, with the opportunity to enter an exclusive raffle for completion of these additional questions. The survey is voluntary and anonymous. You may choose to skip questions or exit the survey at any time, and no identifiable information will be collected.

If you are interested in participating in the survey, please click the link below! The survey should take you approximately 10-15 minutes to complete.

[Survey Link]

Please feel free to share this link with other providers in your network.

If you have questions regarding the survey, please contact the principal investigator: Taylor Kupneski at [email] or Jessica Fairey, MS, CGC at [email].

2. Invitation Blurb

Primary Care Providers' Comfort with Utilization of Genetics in Practice
We are interested in understanding the attitudes of physicians, NPs, and PAs practicing in family medicine, internal medicine, OB/GYN/Women's Health, pediatrics, and geriatrics regarding the relevance of various genetics skills and knowledge in clinical practice. In addition, we want to understand your current comfort utilizing these concepts, and your perceived level of education on each of these topics. The survey takes approximately 10-15 minutes with the chance to win one of three $25 Amazon gift cards for completing the survey. Respondents may elect to complete an additional three questions at the end of the survey, with the opportunity to enter an exclusive raffle for completion of these additional questions. You may choose to skip questions or exit the survey at any time, and no identifiable information will be collected.

Complete the survey at the following link:

[survey link]

For any questions regarding the study, please contact Taylor Kupneski at [email] or Jessica Fairey, MS, CGC at [email].

Appendix C. Supplemental Tables

Table C.1 Average level of comfort with genetic based concepts by provider type

	Average Level of Comfort			
Item	**Total**	**Physician**	**Nurse Practitioner**	**Physician Assistant**
Structure, function, and replication of DNA	2.93	3.00	2.00	3.33
Simple patterns of inheritance	3.31	3.61	2.83	2.94
Complex patterns of inheritance	2.83	3.25	2.00	n/a
Karyotype/microarray findings	3.00	3.18	2.00	2.33
Genetic principles (reduced penetrance, variable expressivity, two-hit hypothesis)	3.00	3.10	n/a	2.00
Family history taking and interpretation	3.35	3.52	2.82	3.38
Pedigree construction	3.04	3.13	3.00	2.80
Karyotype	3.29	3.52	2.50	2.60
Microarray	3.00	3.18	3.00	2.00
Single gene testing	3.14	3.35	2.50	2.89
Multigene panel testing	2.90	3.07	2.00	2.60
Whole exome/genome sequencing	3.50	3.50	n/a	n/a
Pharmacogenomic testing	3.15	3.33	3.00	3.00
Prenatal/newborn screening programs	3.27	3.55	3.20	2.70
Pathogenic test results	3.23	3.29	3.33	3.11
Variant of uncertain significance (VUS) results	3.20	3.38	n/a	2.50
Negative test results	3.33	3.52	3.25	3.07

Incidental findings on testing	3.09	3.29	3.00	2.93
Ethical, social and legal implications of genetic testing for the patient	3.07	3.30	2.67	2.90
Ethical, social and legal implications of genetic testing for family members	3.09	3.32	2.57	3.00

Table C.2 Average level of comfort with genetic based concepts by provider specialty

	Average Level of Comfort					
Item	**Total**	**Family Medicine**	**Internal Medicine**	**OBGYN**	**Pediatrics**	**Geriatrics**
Structure, function, and replication of DNA	2.93	2.50	3.00	3.25	2.67	3.00
Simple patterns of inheritance	3.31	3.22	3.25	3.43	3.44	2.75
Complex patterns of inheritance	2.83	2.00	2.00	3.67	2.00	n/a
Karyotype/microarray findings	3.00	2.33	2.67	3.25	3.14	n/a
Genetic principles (reduced penetrance, variable expressivity, two-hit hypothesis)	3.00	3.00	2.67	3.50	3.00	2.00
Family history taking and interpretation	3.35	3.12	3.38	3.38	3.53	3.43
Pedigree construction	3.04	2.80	3.00	3.25	3.13	n/a
Karyotype	3.29	2.67	2.80	3.80	3.25	3.00
Microarray	3.00	n/a	2.33	3.20	3.17	n/a
Single gene testing	3.14	3.40	2.75	3.43	3.20	3.00
Multigene panel testing	2.90	3.00	2.80	3.14	2.50	3.00
Whole exome/genome sequencing	3.50	n/a	3.00	4.00	3.00	n/a
Pharmacogenomic testing	3.15	2.75	3.00	4.00	3.33	3.67
Prenatal/newborn screening programs	3.27	3.17	2.83	3.27	3.50	n/a
Pathogenic test results	3.23	3.00	3.11	4.00	3.00	4.00
Variant of uncertain significance (VUS) results	3.20	2.50	3.50	3.40	3.00	n/a
Negative test results	3.33	3.20	2.92	3.67	3.60	3.25
Incidental findings on testing	3.09	3.00	2.80	3.60	3.40	3.00

Ethical, social and legal implications of genetic testing for the patient	3.07	2.76	2.88	3.64	3.27	3.00
Ethical, social and legal implications of genetic testing for family members	3.09	2.83	3.25	3.57	3.09	2.83
Ethical, social and legal implications of genetic testing for children/minors	3.24	2.86	3.00	4.00	3.36	n/a
Ethical, social and legal implications of genetic testing for adults with incapacity	3.31	3.50	3.00	4.00	3.50	3.20
Cost of genetic testing, appointments, and insurance coverage	2.86	2.82	2.36	3.60	3.00	3.33
Ability to locate resources related to referral and management guidelines and patient support	3.00	3.17	2.50	3.29	3.10	3.00
Ability to refer and interact with local or regional geneticists and/or genetic counselors	3.17	3.25	2.50	3.50	3.50	2.75

Table C.3 Chi-squared analysis of provider attitude towards relevance of genetic components by provider type and by specialty

	X^2 results, item versus provider type		**X^2 results, item versus provider specialty**	
Item	**X^2 (df, N) =**	**p-value**	**X^2 (df, N) =**	**p-value**
Structure, function, and replication of DNA	X^2 (2, N=71)=1.809	p=.405	X^2 (4, N=71)=1.738	p=.784
Inheritance patterns	X^2 (2, N=71)=5.545	p=.063	X^2 (4, N=71)=9.706	p=.046
Karyotype/microarray findings	X^2 (2, N=71)=8.211	p=.016	X^2 (4, N=71)=26.645	p=.000
Genetic principles	X^2 (2, N=71)=11.330	p=.003	X^2 (4, N=71)=3.383	p=.496
Family history taking and interpretation	X^2 (2, N=71)=0.804	p=.669	X^2 (4, N=71)=1.319	p=.858
Pedigree construction	X^2 (2, N=71)=14.639	p=.001	X^2 (4, N=71)=6.071	p=.194
Types of genetic testing	X^2 (2, N=71)=3.552	p=.169	X^2 (4, N=71)=15.866	p=.003
Genetic test results	X^2 (2, N=71)=12.686	p=.002	X^2 (4, N=71)=3.946	p=.413
Ethical, legal, and social implications of testing on patients and family members	X^2 (2, N=71)=1.534	p=.464	X^2 (4, N=71)=3.527	p=.474
Ethical, legal and social implications of testing on children/minors and adults with incapacity	X^2 (2, N=71)=2.65	p=.277	X^2 (4, N=71)=6.371	p=.173
Cost of genetic testing and insurance coverage	X^2 (2, N=71)=5.013	p=.082	X^2 (4, N=71)=7.523	p=.111
Ability to identify/locate resources related to referrals and management guidelines, and patient support	X^2 (2, N=71)=1.783	p=.410	X^2 (4, N=71)=5.765	p=.217
Ability to refer and interact with local or regional geneticists and/or genetic counselors	X^2 (2, N=71)=14.475	p=.001	X^2 (4, N=71)=9.358	p=.053

Table C.4 Chi-squared analysis of provider attitude towards comfort of genetic components by provider type and by specialty

	X^2 results, item versus provider type		**X^2 results, item versus provider specialty**	
Item	**X^2 (df, N) =**	**p-value**	**X^2 (df, N) =**	**p-value**
Structure, function, and replication of DNA	X^2 (2, N=71)=2.730	p=.255	X^2 (4, N=71)=1.706	p=.790
Simple patterns of inheritance	X^2 (2, N=71)=11.046	p=.004	X^2 (4, N=71)=16.700	p=.002
Complex patterns of inheritance	X^2 (2, N=71)=3.506	p=.173	X^2 (4, N=71)=4.084	p=.395
Karyotype/microarray findings (as it relates to inheritance)	X^2 (2, N=71)=13.723	p=.001	X^2 (4, N=71)=11.913	p=.018
Genetic principles (reduced penetrance, variable expressivity, two-hit hypothesis)	X^2 (2, N=71)=9.766	p=.008	X^2 (4, N=71)=3.206	p=.524
Family history taking and interpretation	X^2 (2, N=71)=5.198	p=.074	X^2 (4, N=71)=1.631	p=.803
Pedigree construction	X^2 (2, N=71)=4.114	p=.128	X^2 (4, N=71)=5.846	p=.211
Karyotype (as it relates to genetic testing and test results)	X^2 (2, N=71)=15.604	p=.000	X^2 (4, N=71)=12.379	p=.015
Microarray (as it relates to genetic testing and test results)	X^2 (2, N=71)=6.581	p=.037	X^2 (4, N=71)=11.399	p=.022
Single gene testing	X^2 (2, N=71)=4.775	p=.092	X^2 (4, N=71)=2.266	p=.687
Multigene panel testing	X^2 (2, N=71)=7.321	p=.026	X^2 (4, N=71)=4.049	p=.399
Whole exome/genome sequencing	X^2 (2, N=71)=7.132	p=.028	X^2 (4, N=71)=5.747	p=.219
Pharmacogenomic testing	X^2 (2, N=71)=0.734	p=.693	X^2 (4, N=71)=5.559	p=.235
Prenatal/newborn screening programs	X^2 (2, N=71)=4.181	p=.124	X^2 (4, N=71)=23.428	p=.000
Pathogenic test results	X^2 (2, N=71)=1.339	p=.512	X^2 (4, N=71)=4.197	p=.380
Variant of uncertain significance (VUS) results	X^2 (2, N=71)=5.294	p=.071	X^2 (4, N=71)=7.523	p=.111
Negative test results	X^2 (2, N=71)=1.049	p=.592	X^2 (4, N=71)=3.304	p=.508
Incidental findings on testing	X^2 (2, N=71)=5.693	p=.058	X^2 (4, N=71)=3.468	p=.483

Ethical, social and legal implications of genetic testing for the patient	X^2 (2, N=71)=2.842	p=.241	X^2 (4, N=71)=8.641	p=.071
Ethical, social and legal implications of genetic testing for family members	X^2 (2, N=71)=0.475	p=.789	X^2 (4, N=71)=4.978	p=.290
Ethical, social and legal implications of genetic testing for children/minors	X^2 (2, N=71)=2.392	p=.302	X^2 (4, N=71)=14.141	p=.007
Ethical, social and legal implications of genetic testing for adults with incapacity	X^2 (2, N=71)=0.619	p=.734	X^2 (4, N=71)=12.645	p=.013
Costs of genetic testing, genetics appointments, and insurance coverage	X^2 (2, N=71)=0.618	p=.734	X^2 (4, N=71)=4.358	p=.360
Ability to identify/locate resources related to referral and management guidelines and support for genetic conditions	X^2 (2, N=71)=2.622	p=.270	X^2 (4, N=71)=2.659	p=.616
Ability to refer and interact with local or regional geneticists and/or genetic counselors	X^2 (2, N=71)=0.841	p=.657	X^2 (4, N=71)=6.675	p=.154

Appendix D. Supplemental Figures

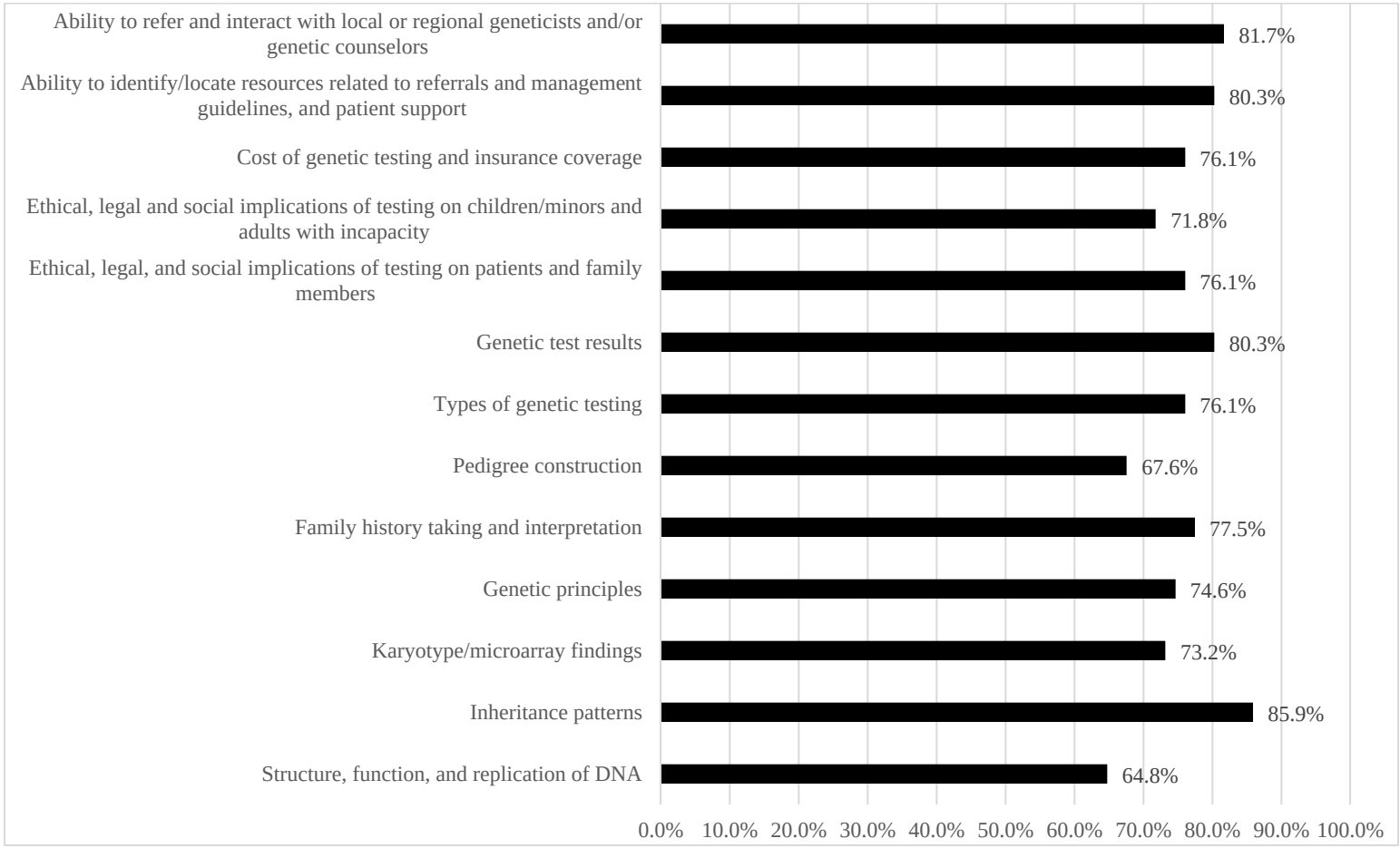


Figure D.1 Provider comfort with genetic counselor ability to explain genetic based concepts