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Assessing OBGYN Residents' Knowledge, Attitudes, and Current Practices for Carrier Screening

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Abstract

Obstetricians and gynecologists (OBGYNs) order carrier screening on a regular basis, so it is important to understand their knowledge, attitudes, and current practices regarding this screening. There are two primary professional organizations that have established practice guidelines for carrier screening, the American College of Obstetrics and Gynecology (ACOG) and American College of Medical Genetics (ACMG). With the growth of pan-ethnic carrier screening, these guidelines have become remarkably different. This study aimed to assess resident OBGYN's utilization of pan-ethnic carrier screening, discern any discrepancies between knowledge, attitudes, and current practices, identify possible practice resources that may be beneficial, and assess the need for standardized guidelines. A survey was shared with OBGYN residents across the nation that included topics related to knowledge, attitudes, and current practices about carrier screening. The average knowledge score for our participants (n=23) was 90%. A notable difference existed between the number of participants who believed pre-test counseling was their responsibility (65.2%) and those that felt confident in providing pre-test counseling (43.5%). Additionally, the majority of participants were considering a patient's ethnicity when selecting a panel (60.9%), and the most popular panel size included four or less conditions.

Findings from this study provided insight into which domains and concepts of carrier screening OBGYN providers may find additional practice resources beneficial. We identified the most significant discrepancies for test selection, points of counseling,

and results disclosure. There is significant variability in current practices among OBGYN residents which suggests the need for standardization of guidelines. We recommend that practice resources include recommendations from both ACOG and ACMG and promote pan-ethnic carrier screening. Based on residents' opinions, these resources should be quick and direct summaries of practice guidelines that outline important topics of discussion. With improved provider knowledge and comfortability with carrier screening, this may improve equitable access of reproductive knowledge for patients.

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Chapter 1: Literature Review

1.1 What is Carrier Screening?

Carrier screening is a genetic test used to identify carriers and determine the reproductive risk for autosomal recessive and X-linked conditions. The conditions screened for with this type of testing generally have infantile or early-onset of symptoms and would have impacts on reproductive clinical management. To be a carrier means an individual has one variant allele and one functional allele in a gene. Carriers are typically healthy and have little to no symptoms, with exceptions for some X-linked female carriers. When both members of a couple are identified as carriers for the same autosomal recessive condition, there is a 25% chance for a pregnancy to be affected. For X-linked conditions, female carriers have a 25% chance to have an affected male pregnancy and a 25% chance for a pregnancy to be a female carrier (Committee Opinion No. 691, 2017).

1.2 History of Carrier Screening

Carrier screening first began in 1970. Throughout its early history, it was only targeted at certain ethnic populations with high carrier frequencies. The first condition that carrier screening was conducted for was Tay-Sachs disease (Kaback, 2000). Tay-Sachs disease is a progressive neurodegenerative disorder. The classic form is characterized by progressive weakness, loss of motor skills, and decreased visual attentiveness within the first few months of life. Seizures and a plateau in development can also be seen in the first year of life. The majority of affected individuals generally pass away at an average age of two or three years old (Ramani & Sankaran, 2023; Toro et

al., 2020). This initial carrier screening targeted Ashkenazi Jewish populations, as their carrier frequency was much higher for this condition compared to the general population. This method reduced the incidence of Tay-Sachs disease in the Jewish population by 90% (Kaback, 2000).

The second condition to be routinely screened for was Beta Thalassemia in the late 1970s (Cao et al., 1997). Beta Thalassemia is a condition that affects the production of hemoglobin by reducing the production of the beta globin chain. Characteristic features of Beta Thalassemia Major are severe anemia and hepatosplenomegaly, which can result in failure to thrive, recurrent infections, and jaundice. Symptom onset typically begins within the first two years of life and is primarily treated with blood transfusions (Langer, 2023; Needs et al., 2023). Education and screening for Beta Thalassemia was primarily targeted towards Mediterranean populations (Cao et al., 1997).

The next condition to have routine carrier screening was cystic fibrosis (CF). CF is a multisystem disease that primarily affects the respiratory tract, pancreas, intestines, liver, and sweat glands. Some characteristic features include progressive obstructive pulmonary disease, pancreatic insufficiency, pancreatitis, and bilateral absence of the vas deferens. Transmembrane conductance regulator modulator targeted therapy is a proven treatment to help reduce symptom manifestation (Savant et al., 2023). CF carrier screening became available after the identification of the *CFTR* gene in 1989 (Riordan et al., 1989). This became one of the first conditions recommended to be screened in all pregnant women or couples planning to become pregnant (Ioannou et al., 2014).

1.3 The Evolution of Carrier Screening

In the more than 50 years since its inception, carrier screening has undergone many advancements in technology and changes to practice but continues to provide couples with information about their likelihood of having a child with an autosomal recessive or X-linked condition.

Pan-ethnic carrier screening is the use of carrier screening without consideration of the ethnicity or ancestry of patients. It was first offered in 2009 (Goldberg et al., 2023). With advancements in technology of DNA analysis, specifically in next generation sequencing (NGS), the feasibility of expanding carrier screening beyond targeted populations increased. This was due to decreased costs and the increased scale of DNA analysis that NGS provided. Due to the success that the Tay-Sachs screening programs had in the Jewish community, it was desired to scale a similar program to the general population; it was also thought that universal carrier screening could be beneficial because couples could receive testing while planning a pregnancy and was not dependent on family history (Srinivasan et al., 2010). One of the first pan-ethnic carrier screening panels offered was a saliva-based assay that screened for over 100 Mendelian diseases across the population with high specificity and sensitivity (Srinivasan et al., 2010).

Due to the increasing diversity and multiethnicity of the population, ethnicity-based screening becomes increasingly inaccurate. Studies have shown that offering pan-ethnic carrier screening to all patients is clinically superior to using ethnicity-based screening paradigms. The use of pan-ethnic carrier screening has been shown to significantly increase the identification of at-risk carrier couples. In a study of more than 380,000 individuals, it was found that using NGS pan-ethnic carrier screening greatly

improved the detection of carriers (Westemeyer et al., 2020). It was found that carrier rates across ethnicities were higher than expected; for example, with conditions that are commonly associated with Ashkenazi Jewish ancestry, over 80% of carriers in this study did not report any Ashkenazi Jewish ancestry (Westemeyer et al., 2020). Additionally, ethnicity-based carrier screening relies on accurate knowledge of ancestry for proper test selection and risk assessment. Self-reported ethnicity can be an imperfect indicator of genetic ancestry. A study found that there were significant discordant results between genetic ancestry and self-reported ethnicity for about 10% of patients (Kaseniit et al., 2020). This study also determined that ethnicity-based guidelines would only identify about a quarter of all carriers in the cohort compared to pan-ethnic screening, although this varies by ethnicity. This data highlights the need for pan-ethnic screening in order to adequately identify carriers in a population, as ethnicity-based screening is missing many individuals (Kaseniit et al., 2020).

1.4 Current Guidelines for Carrier Screening

Even though pan-ethnic carrier screening has been available for more than a decade, it is still not routinely used. In a 2017 study, there were only three conditions being screened for on all panels: CF, maple syrup urine disease 1b, and Niemann-Pick disease (Chokoshvili et al., 2018). Panels can range in size from less than 50 conditions to over 1,500 conditions (Chokoshvili et al., 2018). While some of this variability is due to the different offerings by genetic testing laboratories, it can also be attributed to the difference across professional guidelines and their lack of consistency.

The two main organizations offering guidelines for carrier screening are the American College of Obstetrics and Gynecology (ACOG) and American College of

Medical Genetics (ACMG). The guidelines set forth by both organizations are markedly different.

ACOG currently recommends offering carrier screening to all couples regardless of their race or ethnicity for two conditions: CF and spinal muscular atrophy (SMA). They include five other conditions in their guidelines to be screened based on race or ethnicity, including alpha thalassemia, beta hemoglobinopathies, Tay-Sachs disease, Canavan disease, and familial dysautonomia (Committee Opinion No. 691, 2017). Conversely, since 2021, ACMG has recommended offering carrier screening to all couples for 112 different conditions regardless of ethnicity or race (Gregg et al., 2021). Prior to this, ACMG recommended carrier screening for all couples for SMA and CF regardless of ethnicity and an additional eight conditions for individuals of Ashkenazi Jewish descent (Gross et al., 2008; Prior, 2008; Watson et al., 2004).

There are also differing criteria between these two organizations regarding the conditions that should be included on a pan-ethnic carrier screening panel. ACOG has seven criteria that need to be met for a condition to be included on a pan-ethnic carrier screening panel. The criteria include: 1. Have a carrier frequency of 1 in 100 or greater, 2. Have a well-defined phenotype, 3. Have a detrimental effect on the quality of life, 4. Cause physical or cognitive impairment, 5. Require medical intervention, 6. Have an early age of onset, and 7. Prenatal diagnosis possible (Committee Opinion No. 690, 2017). ACMG recommendations for panel design criteria have some overlap but include broader criteria. Their cutoff for carrier frequency is 1 in 200 or greater for autosomal recessive and greater than 1 in 40,000 for X-linked conditions. They also include that a predictable genotype-phenotype correlation exists, with at least a moderate gene-disease

association. They also address severity, citing conditions with phenotypes that could impact decision-making, especially those categorized as moderate, severe, or profound. Like ACOG, they indicate that there should be prenatal diagnosis and reproductive options available, as well as established analytical validity of screening methods (Gregg et al., 2021).

With these differences in guidelines and a lack of specificity regarding some criteria, it can be difficult for providers to decide what should be included. Clear guidelines and panel criteria are needed to ensure consistency and quality throughout all carrier screening offerings (Johansen Taber et al., 2022). The use of consistent carrier screening guidelines and panels is important for reducing racial inequality and disparate health outcomes by providing equal opportunity for reproductive knowledge (Johansen Taber et al., 2022).

1.5 Carrier Screening: A Complement to Newborn Screening

Carrier screening does not replace the need for newborn screening (NBS), but their roles are complementary. Carrier screening can be a critical tool in early detection and intervention for affected children. Knowledge of at-risk couples can help expedite the testing process post-birth. Diagnostic testing can be done from cord blood rather than waiting for NBS results, allowing for a quicker turn-around-time to diagnosis and a faster initiation of treatments that may be available for the condition (Rose & Wick, 2018). Furthermore, for these treatable conditions, an earlier diagnosis, including a prenatal diagnosis, could improve prognosis (Henneman et al., 2016). Additionally, the knowledge of an affected child prenatally could help educate and prepare the parents and pediatricians for any special medical care the child will require after birth and may

influence management of the delivery to optimize outcomes (Committee Opinion No. 690, 2017).

1.6 Opinions and Attitudes of Physicians about Carrier Screening

Early opinions of physicians regarding carrier screening were generally hesitant. In a study surveying physicians on their perspectives of CF carrier screening, providers were generally concerned about having enough time to educate patients and respond to their questions, in addition to their liability if patients do not undergo screening (Rowley et al., 1993). They were not typically concerned about the sensitivity or specificity of the testing or possible discrimination due to a positive result. They also believed it would be more difficult to screen non-pregnant patients as they do not routinely return for follow-up, receive mailings, or have phlebotomy (Rowley et al., 1993).

There has been limited research on the attitudes and knowledge of providers on comprehensive carrier screening, with the most recent being conducted in 2015 to 2016 prior to the shift in ACMG guidelines to pan-ethnic carrier screening (Briggs et al., 2018). Briggs et al. (2018) surveyed general OBGYNs, maternal fetal medicine (MFM) and reproductive endocrinologist (REI) specialists, and OBGYN residents and fellows on their utilization and attitudes towards carrier screening. They found that most providers have continued to prefer ethnic-based carrier screening from 2010 to 2016, and only 1 in 4 providers offered pan-ethnic carrier screening. Only about half of providers were comfortable offering pan-ethnic carrier screening. They also found that providers were significantly more confident in discussing negative results than positive results. Overall, they concluded that pan-ethnic carrier screening is still far from being the standard of care due to the lack of provider comfort and competence (Briggs et al., 2018).

Two other major studies were conducted between 2010 and 2012 regarding provider attitudes and utilization of pan-ethnic carrier screening. Ready et al. (2011) surveyed women's healthcare providers to understand their perceptions of pan-ethnic carrier screening. They found an overall positive attitude towards pan-ethnic carrier screening but there were misconceptions regarding the confidentiality of results, the risk of passing on a mutation, and the risks of having an affected child (Ready et al., 2011). Roughly 40% of providers were concerned that these results would not remain confidential and over one-third had concerns for insurance increasing rates based on results. Additionally, nearly half of participants had misperceptions about autosomal recessive inheritance; only 56% of participants correctly identified the risk of two carrier parents having an affected child as 25% (Ready et al., 2011). Similar results were found by Benn et al. (2013); they surveyed ACOG fellows to understand their practices and opinions on pan-ethnic carrier screening. They found that 15% of providers were routinely offering pan-ethnic carrier screening, and a little over 50% were offering it only at patient request, putting the burden of knowledge on the patient. Additionally, they highlighted that the majority of providers thought that the most optimal time for carrier screening was preconceptionally, but it continues to be completed routinely during early pregnancy. Overall, they determined an overall positive opinion of carrier screening, and while its utilization has increased, it is still not routinely used in practice by the majority of providers (Benn et al., 2013).

A study assessing the attitudes and comfort levels of OBGYN residents regarding genetics in general found that almost half of residents do not feel that their genetics education in residency is sufficient (Kathrens-Gallardo et al., 2021). While comfort was

increased through training, gaps still existed in their comfort with offering genetic tests and interpreting results. Attending physicians who were knowledgeable about genetics and reinforced this information in clinic and working with genetic counselors were key learning tools for participants (Kathrens-Gallardo et al., 2021). Overall, residents felt that pre- and post-test counseling were within their scope of practice, but that genetics education needs to be reinforced to increase their comfortability.

1.7 Perspectives of Genetic Counselors on Carrier Screening

Prenatal genetic counselors have supported the use of pan-ethnic, comprehensive carrier screening since its advent. A survey of genetic counselors conducted in 2012 found that 80% of genetic counselors would offer a larger, pan-ethnic panel if the costs were the same, with most thinking this would be the future of routine clinical care (Lazarin et al., 2016). There was some debate among genetic counselors on what conditions should be included on carrier screening, but they overall agreed that in 2012, ACMG and ACOG recommendations were too limited (Lazarin et al., 2016). One of the most significant concerns that genetic counselors had with carrier screening was the practicality of implementation; about half of reproductive genetic counselors surveyed reported concern regarding the amount of time spent on discussing results or follow-ups from these larger panels. They noted that pre-test counseling should focus on simplicity rather than providing all the information. The majority of participants preferred that all positive results have formal genetic counseling but understood that prenatal genetic counselors are a limited resource (Lazarin et al., 2016).

The National Society of Genetic Counselors has recently published updated practice guidelines for carrier screening (Sagaser et al., 2023). They recommend pan-

ethnic carrier screening be offered to everyone considering pregnancy, currently pregnant, or able to contribute to a pregnancy, as it can identify the reproductive risk for a greater number of pregnancies (Sagaser et al., 2023). They discuss that only offering this comprehensive carrier screening to those who specifically request it attributes to systemic inequalities and limits information available to individuals with fewer medical resources or lower health literacy. Informed consent is an essential part of the process for comprehensive carrier screening and should be made accessible to all. Five elements are highlighted as what should be included in the process of obtaining informed consent. These elements include the benefits and limitations of knowledge of one's carrier status (i.e., potential reproductive impacts, anxiety, not all genetic conditions are screened for, etc.), discussion of inheritance patterns (autosomal recessive and x-linked), the normalization of carrier status and that often this has little impact on one's health, the limitations of GINA, and the potential for incidental findings that may impact one's own health (Sagaser et al., 2023). Methods for obtaining informed consent can include counseling, videos, fact sheets, or other techniques that would meet the needs of the population. The process for deciding to pursue comprehensive carrier screening should be a shared decision-making process involving the values and preferences of the couple. Barriers to comprehensive carrier screening should be identified and addressed to ensure equitable care (Sagaser et al., 2023).

1.8 Barriers to Carrier Screening

Many barriers exist to the implementation of comprehensive carrier screening, especially in the preconception setting, which is considered the optimal time for this screening. These barriers can include insurance coverage, cost, lack of knowledge, and

lack of consistent guidelines. In general, insurance does not always cover carrier screening for one or both partners; this is in part due to varying Medicaid coverage by state (Hull et al., 2023). Insurance companies do not always recognize the medical necessity for this type of genetic screening and its potential impacts on reproductive decisions. Ensuring insurance coverage for both members of the couple can delay the time to results (Sagaser et al., 2023). Among clinicians there appears to be an uneven distribution of knowledge and comfort regarding comprehensive carrier screening, which can in part be attributed to the nonspecific and conflicting guidelines from professional organizations (Hull et al., 2023). There are reports of a lack of understanding of the importance and value of preconception carrier screening (Hull et al., 2023).

Additional barriers to the implementation of comprehensive carrier screening include the lack of patient follow-up and low uptake of partner screening following a positive result. In a 2023 study evaluating the implementation of carrier screening in an urban population, only 70.6% of patients with an abnormal screening received post-test genetic counseling (Strauss et al., 2023). Of the individuals who received a positive result, less than half (48%) went on to pursue partner screening (Strauss et al., 2023). Barriers to completion included difficulty in contacting patients and setting up a follow up visit to educate and counsel regarding the significance of testing and obtaining the partner's sample. The time from submitting the patient's sample to receiving the partner's result was approximately 46 days which can have a significant impact on decision making regarding an ongoing pregnancy (Strauss et al., 2023). This delay between partner testing also highlights the importance of preconception carrier screening. Additionally, patients of a higher gestational age were less likely to pursue partner testing

(Strauss et al., 2023). Other reasons for the lack of partner screening include insufficient insurance coverage and the inability to afford the additional testing (Carlotti et al., 2021). Patients reported their partners may have been more likely to pursue screening if they had been screened at the same time and if they had had more information about the condition they screened positive for (Carlotti et al., 2020).

In addition to these logistical barriers, providers are often learning information regarding genetics and carrier screening as the need arises in practice. In a study surveying the rotation schedules of American Council for Graduate Medical Education (ACGME)-accredited OBGYN residency programs, only 14.3% had opportunities for medical genetics and genomics rotations. For those that were available, they were significantly shorter than other non-core rotations, on average lasting only two days compared to about two weeks (Putra et al., 2019). Additionally, there is currently no formal genetics curriculum available for residency programs especially in cases where there are no specialists available or comfortable teaching the content (Dotters-Katz et al., 2019). With the growing use of genetics in OBGYN practice, providers are often having to learn this information through their independent practice, which leads to wide discrepancies in the care that patients receive.

Chapter 2: Assessing OBGYN Residents' Knowledge, Attitudes, and Current Practices for Carrier Screening[1]

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2.1 Abstract

Obstetricians and gynecologists (OBGYNs) order carrier screening on a regular basis, so it is important to understand their knowledge, attitudes, and current practices regarding this screening. There are two primary professional organizations that have established practice guidelines for carrier screening, the American College of Obstetrics and Gynecology (ACOG) and American College of Medical Genetics (ACMG). With the growth of pan-ethnic carrier screening, these guidelines have become remarkably different. This study aimed to assess resident OBGYN's utilization of pan-ethnic carrier screening, discern any discrepancies between knowledge, attitudes, and current practices, identify possible practice resources that may be beneficial, and assess the need for standardized guidelines. A survey was shared with OBGYN residents across the nation that included topics related to knowledge, attitudes, and current practices about carrier screening. The average knowledge score for our participants (n=23) was 90%. A notable difference existed between the number of participants who believed pre-test counseling was their responsibility (65.2%) and those that felt confident in providing pre-test counseling (43.5%). Additionally, the majority of participants were considering a patient's ethnicity when selecting a panel (60.9%), and the most popular panel size included four or less conditions.

Findings from this study provided insight into which domains and concepts of carrier screening OBGYN providers may find additional practice resources beneficial. We identified the most significant discrepancies for test selection, points of counseling, and results disclosure. There is significant variability in current practices among OBGYN residents which suggests the need for standardization of guidelines. We recommend that

practice resources include recommendations from both ACOG and ACMG and promote pan-ethnic carrier screening. Based on residents' opinions, these resources should be quick and direct summaries of practice guidelines that outline important topics of discussion. With improved provider knowledge and comfortability with carrier screening, this may improve equitable access of reproductive knowledge for patients.

2.2 Introduction

Carrier screening is a genetic test used to identify carriers and determine the reproductive risk for autosomal recessive and X-linked conditions. To be a carrier of a genetic condition means an individual has one variant allele and one functional allele in a gene. Carriers are typically healthy and have little to no symptoms, with exceptions for some X-linked female carriers. When both members of a couple are identified as carriers for the same autosomal recessive condition, there is a 25% chance for a pregnancy to be affected. For X-linked conditions, female carriers have a 25% chance to have an affected male pregnancy and a 25% chance for a pregnancy to be a female carrier (Committee Opinion No. 691, 2017). At its inception, carrier screening was designed to target certain ethnic populations with known high carrier frequencies (Kaback, 2000). Due to the increasing diversity and multiethnicity of the nation's population, ethnicity-based carrier screening has become increasingly inaccurate (Westemeyer et al., 2020). Pan-ethnic carrier screening, the use of carrier screening without consideration of the ethnicity or ancestry of patients, has become a clinically superior option compared to using ethnicity-based screening paradigms (Goldberg et al., 2023; Westemeyer et al., 2020). Its utilization has been shown to significantly increase the identification of at-risk carrier couples (Westemeyer et al., 2020).

There is significant variability in the size and content of carrier screening panels, which can in part be attributed to the lack of consistency across professional guidelines. For example, the guidelines put forth by the American College of Obstetrics and Gynecology (ACOG) and American College of Medical Genetics (ACMG), the two main organizations with guidelines for carrier screening, are markedly different.

ACOG currently recommends offering carrier screening to all couples regardless of their race or ethnicity for two conditions: CF and spinal muscular atrophy (SMA). They include five other conditions in their guidelines to be screened based on race or ethnicity, including alpha thalassemia, beta hemoglobinopathies, Tay-Sachs disease, Canavan disease, and familial dysautonomia (Committee Opinion No. 691, 2017). Conversely, ACMG has recommended carrier screening to all couples for 112 different conditions regardless of ethnicity or race since 2021 (Gregg et al., 2021).

With these differences in guidelines across professional organizations, it can be difficult for providers to select the most appropriate test for carrier screening. Clear guidelines and panel criteria are needed to ensure consistency and quality throughout all carrier screening offerings which is important for reducing racial inequality and disparate health outcomes (Johansen Taber et al., 2022).

There has been limited research on the attitudes and knowledge of providers on comprehensive carrier screening, with the most recent being conducted in 2015 to 2016 prior to the shift in ACMG guidelines to pan-ethnic carrier screening (Briggs et al., 2018). Briggs et al. (2018) surveyed general OBGYNs, maternal fetal medicine (MFM) and reproductive endocrinologist (REI) specialists, and OBGYN residents and fellows on their utilization and attitudes towards carrier screening. They found that most providers

have continued to prefer ethnic based carrier screening from 2010 to 2016, and only 1 in 4 providers offered pan-ethnic carrier screening. Only about half of providers were comfortable offering pan-ethnic carrier screening, and providers were significantly more confident in discussing negative results than positive results. Overall, they concluded that pan-ethnic carrier screening is still far from being the standard of care due to the lack of provider comfort and competence (Briggs et al., 2018).

Two other major studies were conducted between 2010 and 2012 regarding provider attitudes and utilization of pan-ethnic carrier screening. Ready et al. (2011) surveyed women's healthcare providers to understand their perceptions of pan-ethnic carrier screening. They found an overall positive attitude towards pan-ethnic carrier screening but there were misconceptions regarding the confidentiality of results, the risk of passing on a mutation, and the risks of having an affected child (Ready et al., 2011). Nearly half of participants had misperceptions about autosomal recessive inheritance; only 56% of participants correctly identified the risk of two carrier parents having an affected child as 25% (Ready et al., 2011). Similar results were found by Benn et al. (2013); they surveyed ACOG fellows to understand their practices and opinions on pan-ethnic carrier screening. They found that 15% of providers were routinely offering pan-ethnic carrier screening, and a little over 50% were offering it only at patient request, putting the burden of knowledge on the patient. Additionally, they highlighted that the majority of providers thought the most optimal time for carrier screening was preconceptionally, but it continues to be completed routinely during early pregnancy. Overall, they determined a positive opinion of carrier screening, and while its utilization

has increased, it is still not routinely used in practice by the majority of providers (Benn et al., 2013).

A study assessing the attitudes and comfort levels of OBGYN residents regarding genetics in general found that almost half of residents do not feel that their genetics education in residency is sufficient (Kathrens-Gallardo et al., 2021). While comfort was increased through training, gaps still existed in their comfort with offering genetic tests and interpreting results. Attending physicians who were knowledgeable about genetics and reinforced this information in clinic and working with genetic counselors were key learning tools for participants (Kathrens-Gallardo et al., 2021). Overall, residents felt that pre- and post-test counseling were within their scope of practice, but that genetics education needs to be reinforced to increase their comfortability.

This study assessed current OBGYN residents' knowledge, attitudes, and current practices regarding genetic carrier screening; as recent graduates from a medical institution and entry-level OBGYN providers, residents offer a unique insight into their genetics education and its implementation into practice. This study aimed to determine any discrepancies between knowledge and current practice for carrier screening and to identify resources which may offer guidance for incorporating carrier screening into practice. Additionally, this study aimed to identify which practice guidelines are currently used by resident OBGYNs to offer carrier screening and to assess the need for standardization of these guidelines. It was predicted that this study would identify a discrepancy between the knowledge and current practices of carrier screening in OBGYN residents and identify resources that residents would find helpful for guidance about carrier screening.

2.3 Method

This study was approved by the Institutional Review Board at the University of South Carolina in August 2023 (Pro00129845).

2.3.1 Participants

Participants of this study included obstetrician and gynecological residents. To be included, participants must be currently enrolled in residency in the field of OBGYN. Physicians who have completed their residency and students still in medical school were excluded from the study. Additionally, residents in other specialties were excluded. This specialty was targeted as the majority of carrier screening is ordered by OBGYN physicians. These participants were obtained by sending an electronic survey invitation (Appendix B) to residency program directors who then dispersed it to their residents.

2.3.2 Research Methods

An original electronic survey based on past literature and background research was developed by the study team (Appendix B). An incentive was included with the purpose of increasing participation. This incentive was presented as an optional raffle to win one of 10 $20 gift cards. A participation invitation letter with a link to the survey was shared with residency programs via email by a University of South Carolina OBGYN resident. The survey was open to participants from October 2023 through January 2024. It was sent out three times to maximize participation. Participants were invited to take the survey and their choice to participate served as their consent. The survey consisted of 41 items and assessed the current practices of OBGYN residents when offering carrier screening. The survey consisted of four sections including inclusion criteria/demographics, knowledge of carrier screening and genetics, attitudes about

carrier screening, and current practices at their institution. The survey consisted of Likert scale, multiple choice, true or false, and open-ended items. The goal of the survey was to identify the level of knowledge of residents in regard to carrier screening, their attitudes, and their current practices for offering it and disclosing results. No identifying information was collected in the survey, so the participants remained anonymous. Identifying information collected for the optional raffle was separated from survey responses, so the surveys remained anonymous.

2.3.3 Statistical Analysis

In order to address our research goals, we used descriptive statistical analysis. Since the survey consisted primarily of categorical information, percentages and frequencies were calculated. Excel software was used to conduct quantitative analysis. For all open-ended questions, the responses were organized and coded by theme, which were then calculated for their frequency.

2.4 Results

2.4.1 Demographics

There were 30 responses to the survey. Seven of them were incomplete, so a total of 23 responses were analyzed for the study. The majority of participants were female (78.3%) and white (60.9%). Participants' ages ranged from 26 to 40 years old. The majority of participants practice primarily in an urban setting (78.3%), and 65.2% of respondents were in their first two years of residency. Respondents (N=23) practiced in Virginia (n=4), Maryland (n=2), Illinois (n=1), Pennsylvania (n=2), Oregon (n=3), Connecticut (n=5), Rhode Island (n=2), New Jersey (n=3), and Colorado (n=1). The following ethnicities were represented in the patient populations of participants:

American Indian or Alaska Native (n=7), Asian (n=17), Black or African American (n=20), Native Hawaiian or Other Pacific Islander (n=7), White (n=22), and Hispanic (n=21). A summary of the demographics of the study population can be seen in Table 2.1.

Table 2.1 Demographics.

	Frequencies (%)
Gender (n=23)	
Male	2 (8.7%)
Female	18 (78.3%)
Other	3 (13.0%)
Age (n=21)	
25-29	10 (43.5%)
30-34	10 (43.5%)
35-39	0 (0%)
≥40	1 (4.3%)
Race/Ethnicity (n=21)	
Asian	3 (13.0%)
Black or African American	2 (8.7%)
White	14 (60.9%)
Hispanic	2 (8.7%)
PGY Residency Year (n=23)	
1	7 (30.4%)
2	8 (34.8%)
3	5 (21.7%)
4	2 (8.7%)
>4	1 (4.3%)
Which best describes the location of your primary practice? Select all that apply (n=23)	
Urban	18 (78.3%)
Suburban	5 (21.7%)
Rural	1 (4.3%)

2.4.2 Knowledge

The average knowledge score was 90% with a range from 60% to 100%. A summary of the knowledge scores of respondents is displayed in Table 2.2.

Table 2.2 Knowledge about carrier screening.

Question (correct answer)	Correct (%)	Incorrect (%)
If both individuals in a couple test negative for a specific genetic condition, it is still possible for their child to have that condition. (True)	20 (87.0%)	3 (13%)
An individual can be a carrier for a genetic condition even when there is no history of the condition in the family. (True)	22 (95.7%)	1 (4.3%)
Individuals in certain ethnic groups have an increased risk of being carriers of certain genetic conditions. (True)	23 (100%)	0 (0%)
All variants in a gene (i.e. pathogenic, benign, and variants of uncertain significance) are reported by carrier screening. (False)	17 (73.9%)	6 (26.1%)
If both individuals in a couple are carriers of a mutation in a gene that causes an autosomal recessive condition, the probability that their child will have the condition is __. (25%)	22 (95.7%)	1 (4.3%)

2.4.3 Attitudes

The majority of respondents either agreed or strongly agreed (68.8%) with a statement that if costs were the same, they would prefer to order a larger panel. The majority of participants (78.2%) stated they were confident in disclosing negative results, while 39.1% reported feeling confident disclosing positive results. Table 2.3 summarizes the attitudes of respondents about carrier screening including what should be included on a panel, when to order, pre-test counseling, and results disclosure.

Figure 2.1 Respondents' attitudes about carrier screening.

2.4.4 Current Practices

Table 2.3 summarizes the circumstances, timing, and offerings that participants utilize in their current practice.

Table 2.3 Current practices for offering carrier screening.

	Frequencies (%)
As of 2023, under what circumstances does your practice offer genetic carrier screening? Select all that apply.	
Offers to patients considered high risk (based on personal or family medical history) prior to conception	6 (26.1%)
Offers to patients considered high risk (based on personal or family medical history) during pregnancy	5 (21.7%)
Offers to all patients of reproductive age prior to conception	6 (26.1%)
Offers to all patients during pregnancy	20 (87.0%)
At what point in time does your practice offer carrier screening? Select all that apply.	
Only on patient request	3 (13.0%)
During annual well-woman exams	1 (4.3%)
During appointments scheduled to specifically address fertility	12 (52.2%)
During follow-up care for miscarriages	12 (52.2%)
During appointments for ongoing pregnancy	23 (100%)
Which genetic carrier screening option does your practice prefer to offer patients? Select all that apply.	
ACOG recommendation of cystic fibrosis, spinal muscular atrophy, and hemoglobinopathies	18 (78.3%)
Specific carrier testing based on personal or family history only	10 (43.5%)
A pan-ethnic genetic carrier screening approach, which screens for hundreds of genetic mutations including common recessive childhood illnesses, regardless of ethnicity	11 (47.8%)
Other	1 (4.3%)

Panel Selection. There was a range of three to 200 for the number of conditions respondents typically screened for. Most commonly, participants screened for four or less conditions (39.1%) and 21.7% of participants were unsure of how many they screened for. The distribution of responses can be seen in Figure 2.2. Additionally, more than half (60.9%) of participants stated that they consider a patient's ethnicity when selecting a panel.

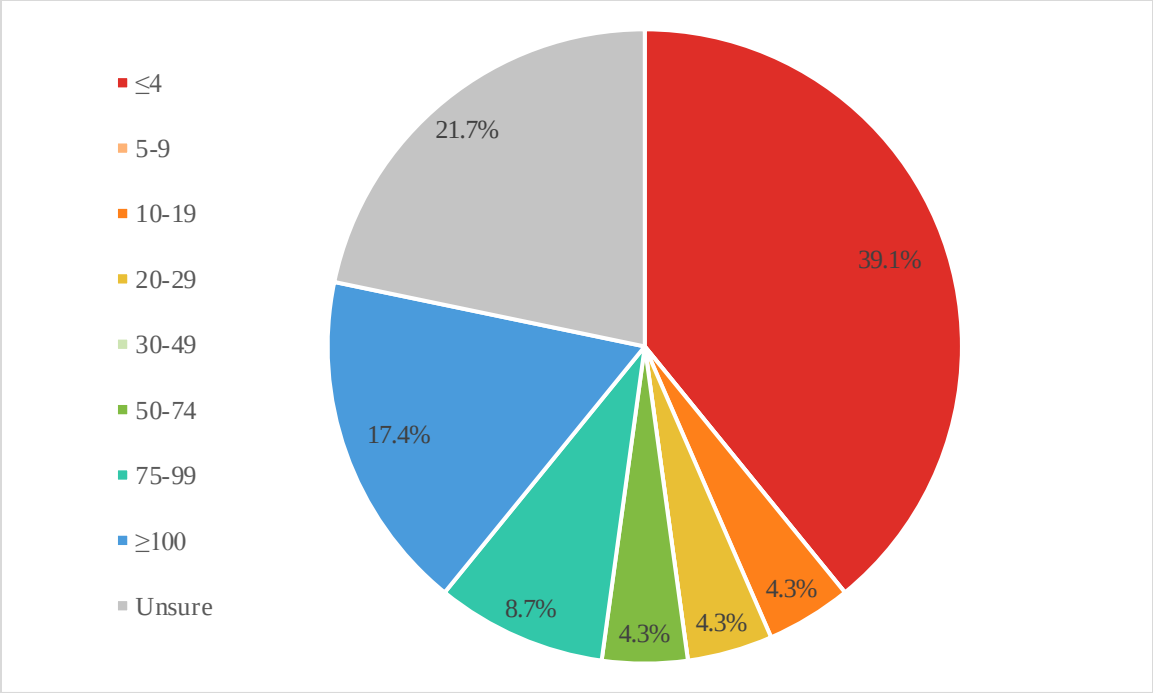

Figure 2.2 Number of conditions participants screened for on a typical carrier screening panel.

Pre- and Post-Test Counseling. The majority of participants do not discuss residual risk when disclosing negative results (65.2%). Data for concepts discussed in pre-test counseling and results follow up practices are summarized in Table 2.4

Table 2.4 Pre- and post-test counseling current practices.

	Frequencies (%)
Which concepts of genetic carrier screening do you include in pre-test counseling? Select all that apply.	
Frequency of carrier detection	4 (17.4%)
Clinical significance of conditions being tested	16 (69.6%)
Autosomal recessive inheritance	9 (39.1%)
Prenatal diagnosis options	9 (39.1%)
Limitations of carrier screening (i.e. does not test for all conditions, only reports pathogenic or likely pathogenic variants, only applicable for current reproductive partner, cannot identify de novo variants)	17 (73.9%)
None of the above	1 (4.3%)

How do you notify patients about their positive genetic carrier testing results? Select all that apply.	
Phone call from obstetrician	13 (56.5%)
Phone call from nurse	2 (8.7%)
Letter/Email/MyChart	6 (26.1%)
Phone call from genetic counselor	10 (43.5%)
Office appointment	17 (73.9%)
Other (I don't know)	1 (4.3%)
How do you notify patients about their negative genetic carrier testing results? Select all that apply.	
Phone call from obstetrician	9 (39.1%)
Phone call from nurse	1 (4.3%)
Letter/Email/MyChart	15 (65.2%)
Phone call from genetic counselor	3 (13.0%)
Office appointment	10 (43.5%)
Other (I don't know)	1 (4.3%)
When disclosing positive results, what is your follow-up? Select all that apply.	
Schedule partner carrier screening	13 (56.5%)
Refer to genetic counseling	18 (78.3%)
Other (I don't know)	1 (4.3%)

Professional Organizations. All participants primarily follow ACOG for clinical practice guidelines related to carrier screening, with one participant selecting that they also follow ACMG guidelines. The majority of respondents (78.3%) never consult ACMG practice guidelines. The frequency at which respondents consult ACOG and ACMG carrier screening practice guidelines is displayed in Figure 2.3. Participants' preferences for receiving updates on practice guidelines is summarized in Figure 2.4.

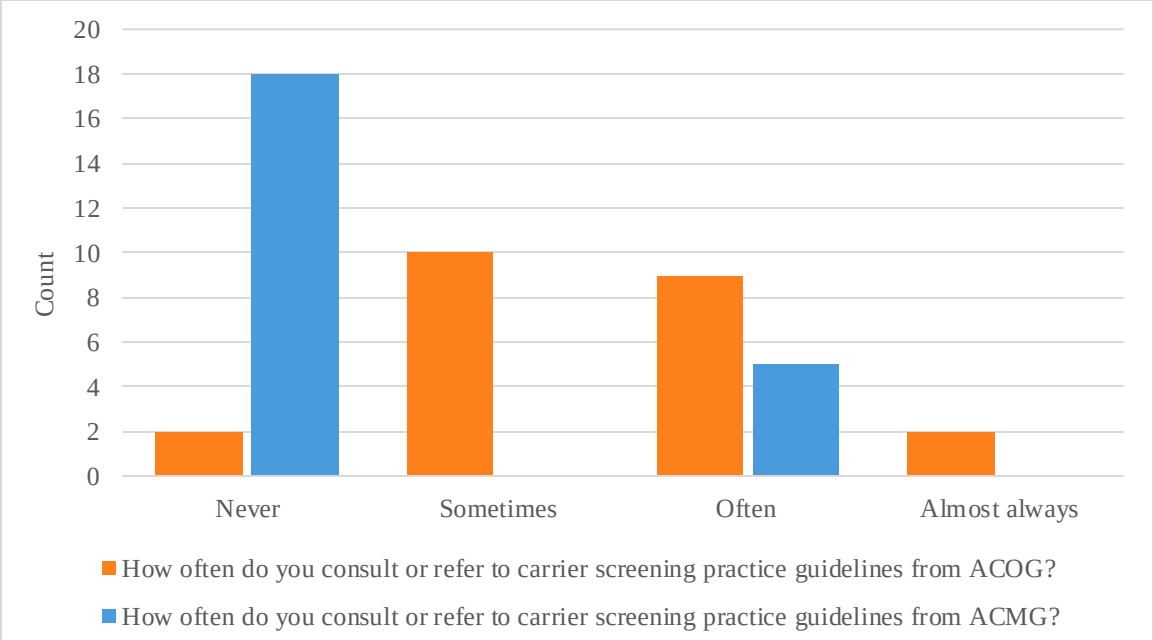

Figure 2.3 Frequency of which participants refer to ACOG and ACMG practice guidelines.

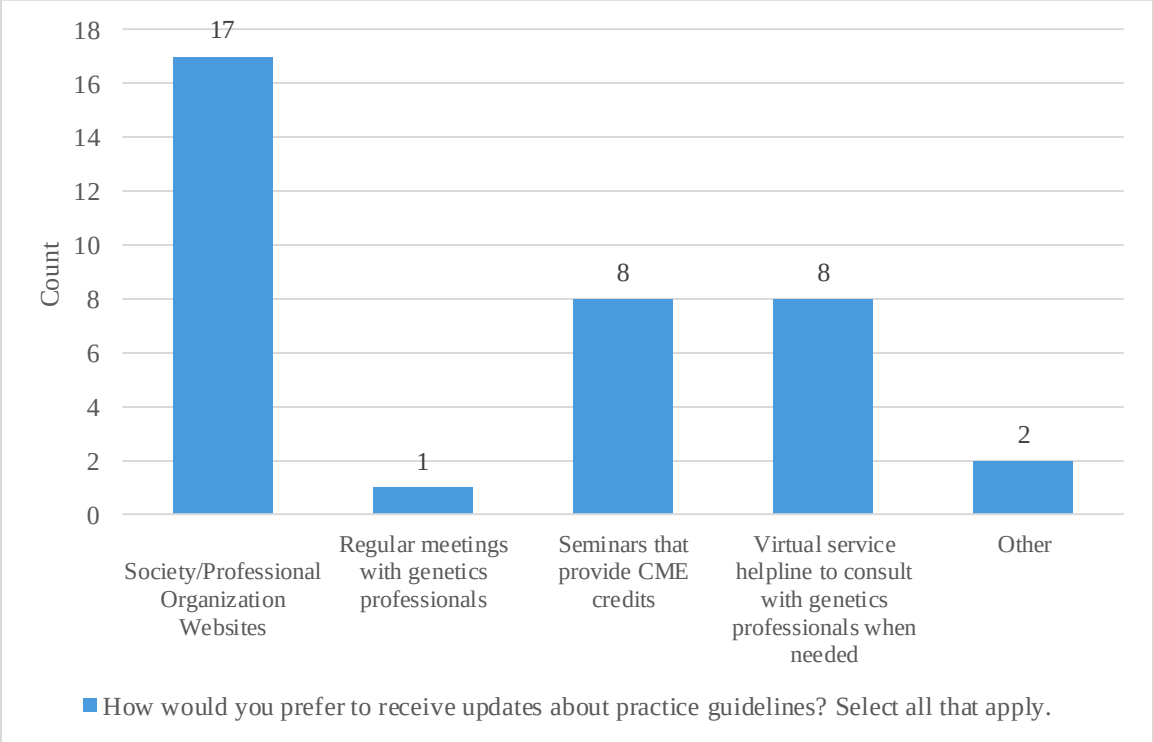

Figure 2.4 Preference for receiving updates about practice guidelines.

Resources. Six respondents stated they would reach out to an MFM if they had questions about genetic testing, and nine responded that they would reach out to a genetic counselor. All but two respondents (91.3%) stated they have access to a genetic counselor or genetics team within or near their practice. When asked what resources they would like as guidance for offering and delivering results for carrier screening, responses included "a flow diagram of how to choose which screening for which patients from ACOG," "a comprehensive guide of current recommendations for screening and counseling patients about their testing results," and "a short and direct slide set or guide."

2.5 Discussion

This study aimed to assess resident OBGYN's knowledge, attitudes, and current practices regarding genetic carrier screening. As entry level OBGYN providers, their perspective offers a unique insight into genetics education and how this knowledge is incorporated into practice with carrier screening. While previous literature has focused on overall OBGYN physicians' knowledge, attitudes, and current practices, this is the first study to have focused solely on this unique perspective of residents (Benn et al., 2013; Briggs et al., 2018; Ready et al., 2011). Our findings provide valuable insights into these domains of knowledge, attitudes, and current practices and highlight the need for standardized practice guidelines and resources to improve the incorporation of pan-ethnic carrier screening to ensure equitable access to carrier screening.

As expected, all OBGYN residents follow ACOG clinical practice guidelines for carrier screening. The majority stated they never consult ACMG practice guidelines. Given the variability between the guidelines and the specialist-specific nature of the organizations, standardization of practice guidelines between organizations would allow

for more comprehensive health care. Given their markedly different guidelines, collaboration between and support for either organization's recommendations would lead to more consistent practices. Standardization of health care practices is important for reducing racial inequality and disparate health outcomes by providing equal opportunity for reproductive knowledge (Johansen Taber et al., 2022).

2.5.1 Pre-test Counseling

Given that informed consent is an essential part of the comprehensive carrier screening process, pre-test counseling should be routine. Roughly two-thirds of providers agreed or strongly agreed with a statement that they felt it was their responsibility to provide pre-test counseling, while less than half of participants agreed that they feel confident in offering pre-test counseling for carrier screening. This highlights that there is room for improvement in increasing providers' confidence and comfortability with discussing topics related to pre-test counseling and informed consent. Further highlighting this need, 96% of participants correctly identified the risk for an affected pregnancy when both partners are carriers, but despite this knowledge, it does not appear to be translated into clinical practice routinely; less than half of respondents stated they discuss this inheritance or risk with patients. The discussion of inheritance is considered a necessary element of achieving informed consent for carrier screening, so providing this information to patients should be standard (Sagaser et al., 2023). Additionally, ACMG practice resource highlights information that all providers should be comfortable discussing, which includes topics like the limitations of carrier screening, that carrier screening is optional, and what it means to be a carrier of an X-linked and/or autosomal recessive condition (Gregg et al., 2021). A practice resource can be developed that

includes recommendations from both ACOG and ACMG and clearly outlines these key discussion points for pre-test counseling. A brief reference guide may improve provider confidence in pre-test counseling for carrier screening. Providers may also benefit from continuing education about carrier screening to improve their comfortability with the content.

2.5.2 Results Disclosure

Regarding the disclosure of carrier screening results, similar to the findings of Briggs et al. (2018), this study identified that residents are more comfortable disclosing negative carrier screening results than positive results. Over three-quarters of respondents agreed that they felt comfortable disclosing negative results, while less than half of respondents agreed they felt comfortable disclosing positive results. This finding highlights a need to increase providers' confidence in discussing positive results, which standardized practice guidelines may promote. Additionally, some recommended points of discussion are not being routinely implemented in post-test counseling. ACMG practice guidelines recommend the discussion of residual risk with patients as a negative carrier screen result does not eliminate the possibility of an affected child but rather reduces the risk (Gregg et al., 2021). More than half of participants stated they do not discuss residual risk with a patient, while the majority of participants also correctly answered that it is still possible for a child to be affected with a genetic condition even if their parents' carrier screening is negative for that condition. This underscores that this discrepancy with current practices is not due to a lack of knowledge about residual risk. Standardized guidelines outlining the key recommended discussion points for results disclosures may improve provider's confidence in disclosing results, especially for

positive results. Practice resources that include topics like condition education and variable expressivity, partner testing, residual risk, and diagnostic testing and reproductive decision making may increase comfortability with these discussions and results disclosures (Gregg et al., 2021). In addition to the benefit of standardized guidelines, access to a multidisciplinary team may also improve provider confidence, as the majority of residents would speak to a genetic counselor or MFM attending physician if they had questions regarding genetics.

2.5.3 Addressing Misconceptions

Similar to preceding studies, this study identified that most providers are not utilizing pan-ethnic carrier screening (Benn et al., 2013; Briggs et al., 2018). The majority of providers still consider patient's ethnicity when ordering carrier screening and almost half of respondents stated they are ordering panels of four conditions or less for carrier screening. This is consistent with ACOG's current guidelines for carrier screening recommending screening based on a patient's ethnicity (Committee Opinion No. 691, 2017). Interestingly, even some providers who stated they refer to ACMG guidelines for carrier screening, which recommends screening all patients for 112 conditions, still only screen for four or less conditions. One possible reason for this discrepancy is a misconception about the cost of screening. The majority of providers agreed that if costs were the same, they would prefer to order a panel consisting of more conditions rather than a smaller panel. Knowledge of cost and insurance coverage for various screening options is important for providers ordering carrier screening, as many tests have similar costs regardless of the number of genes analyzed. If practice guidelines or continuing education highlighted detailed information about cost, providers may elect

to screen for more conditions. Pan-ethnic carrier screening has been shown to significantly increase the identification of at-risk carrier couples, leading to improved knowledge about a couple's reproductive risk (Westemeyer et al., 2020).

While overall knowledge scores were high, more than a quarter of participants incorrectly selected that all variants in a gene, including variants of uncertain significance (VUS) and benign variants, are reported by carrier screening. As ACMG recommends, the majority of genetic testing laboratories will only routinely report pathogenic and likely pathogenic variants; VUSs are only recommended to be reported when one partner is found have a pathogenic or likely pathogenic variant in the same gene (Gregg et al., 2021). The partner's carrier status would need to be reported to the genetic testing laboratory to ensure any VUSs in the same gene are reported. If providers are unaware of this, potential risks due to a suspicious VUS could remain hidden. These findings highlight a need for further education or resources that clarify for providers exactly what is reported with carrier screening.

2.5.4 Development of Practice Resources

Participants suggested ways in which they would prefer to receive updates regarding practice guidelines. It is valuable to know that the majority of residents prefer to receive updates directly from professional organizations. It is possible that methods like regular meetings with a genetics professional may be too time intensive, making it a less convenient option for providers; the less frequent and as-needed resources seemed to be more popular among residents. These findings highlight additional possible routes that could be explored for distributing practice resources. Residents expressed a desire for resources for carrier screening; these included a flow diagram or short slide set outlining

the recommendations. Given that the majority of residents preferred updates and resources directly from the professional organizations, it is proposed that practice guidelines and resources are developed in collaboration with and including consideration of the recommendations from both governing organizations. OBGYN residents expressed a desire for a simplified resource that could be utilized to select the best panel for each patient. We propose the development of a resource that includes recommendations for test selection, points of counseling, and results disclosure given these were areas that we noticed the most discrepancy between knowledge and current practice. Future research may include development of these practice resources and assessment of their utility in practice.

2.5.5 Limitations and Future Directions

There were limitations to this study that do not allow for wide generalization of our findings. The primary limitation was the small sample size. There are more than 5,000 active OBGYN residents in the United States, and this study only received a total of 23 complete responses, which is a response rate of less than 0.5% (Association of American of Medical Colleges, 2017). This study only represents a small portion of this population. Additionally, the majority of participants practiced primarily in an urban region, with only one participant practicing in a rural region. Respondents were also primarily from the northeastern region of the United States; there were no participants from the southeast region and there was limited data from other regions. With this lack of geographic diversity, these findings cannot be extrapolated to all OBGYN residents; it is possible that residents in regions not represented could have different knowledge, attitudes, and current practices. Ideally, all regions would be represented, allowing for a

comparison between the knowledge, attitudes, and current practices of providers in different regions. Future research may consider identifying if there are differences in these domains for carrier screening between urban and rural regions or different geographical locations of the United States. Another potential limitation could be recall bias which is inherent in all survey studies. Also, senior level residents could have provided more insight into knowledge at the end of residency. Our study was not powered to study those differences. Additionally, another possible limitation was question structure. For multiple questions, preference was asked but participants were able to select all that apply, so preference was not able to be accurately assessed.

While this study was not designed specifically to develop resources for the implementation of pan-ethnic carrier screening, the valuable insights into the knowledge, attitudes, and current practices of OBGYN residents it provided can further guide the development and establishment of practice resources. Based on the resources proposed in this study, future research can include the development of and validation of such resources. Additionally, the need for the standardization of practice guidelines was highlighted by this study; future research can determine best practices for collaboration between organizations and measure its overall effectiveness. This study provided valuable information that can be utilized in future research to help improve access to pan-ethnic carrier screening and promote the reproductive knowledge of patients.

Chapter 3: Conclusion

With the growth of pan-ethnic carrier screening, practice guidelines for carrier screening from professional organizations such as the American College of Obstetrics and Gynecology (ACOG) and the American College of Medical Genetics (ACMG) have become remarkably different. This study assessed resident obstetrician and gynecologists' (OBGYN) utilization of pan-ethnic carrier screening, identified discrepancies between knowledge, attitudes, and current practices, identified possible practice resources that may be beneficial, and assessed the need for standardized guidelines. Findings from this study provided insight into what domains OBGYN providers may benefit from additional practice resources. The most significant discrepancies between these domains existed in test selection, points of counseling, and results disclosure, highlighting the need for resources outlining recommended practice. Significant variability existed in the current practice of residents, with most ordering carrier screening panels assessing four conditions while others select upward of 200 conditions. Given this and that the majority of participants are considering a patient's ethnicity when ordering carrier screening, pan-ethnic carrier screening is not routinely utilized. The significant variability in the current practices of surveyed OBGYN residents suggests the need for the standardization of guidelines between ACOG and ACMG. Practice resources that are produced should include recommendations from both governing bodies to ensure this standardization and promote pan-ethnic carrier screening as it has been determined to be the clinically superior screening paradigm. Based on the

opinions of residents, new resources should be quick and direct summaries of practice guidelines that outline important topics of discussion. Future research can further guide the development and establishment of these practice resources and aid in the access to pan-ethnic carrier screening. With an increase in provider knowledge and comfortability with carrier screening, patients' access to reproductive knowledge may be improved.

References

Association of American of Medical Colleges. (2017, August 27) *Table B3: Number of active residents, by type of medical school, GME specialty, and sex.* https://www.aamc.org/data-reports/students-residents/data/table-b3-number-active-residents-type-medical-school-gme-specialty-and-sex

Benn, P., Chapman, A. R., Erickson, K., Defrancesco, M. S., Wilkins-Haug, L., Egan, J. F., & Schulkin, J. (2014). Obstetricians and gynecologists' practice and opinions of expanded carrier testing and noninvasive prenatal testing. *Prenatal Diagnosis, 34*(2), 145–152. https://doi.org/10.1002/pd.4272

Briggs, A., Nouri, P. K., Galloway, M., O'Leary, K., Pereira, N., & Lindheim, S. R. (2018). Expanded carrier screening: A current survey of physician utilization and attitudes. *Journal of Assisted Reproduction and Genetics*, *35*(9), 1631–1640. https://doi.org/10.1007/s10815-018-1272-8

Cao, A., Saba, L., Galanello, R., & Rosatelli, M. C. (1997). Molecular diagnosis and carrier screening for beta thalassemia. *JAMA, 278*(15), 1273–1277.

Chokoshvili, D., Vears, D., & Borry, P. (2018). Expanded carrier screening for monogenic disorders: Where are we now? *Prenatal Diagnosis, 38*(1), 59–66. https://doi.org/10.1002/pd.5109

Committee Opinion No. 690: Carrier Screening in the Age of Genomic Medicine. (2017). *Obstetrics and Gynecology*, *129*(3), e35–e40. https://doi.org/10.1097/AOG.0000000000001951

Committee Opinion No. 691: Carrier Screening for Genetic Conditions. (2017). O*bstetrics & Gynecology*, *129*(3), e41–e55. https://doi.org/10.1097/aog.0000000000001952

Dotters-Katz, S. K., Vora, N., & Kuller, J. (2019). Genetics for the Women's Health Trainee: A Five-Module Curriculum. *MedEdPORTAL*, *15*, 10797. https://doi.org/10.15766/mep_2374-8265.10797

Goldberg, J. D., Pierson, S., & Johansen Taber, K. (2023). Expanded carrier screening: What conditions should we screen for? *Prenatal Diagnosis*, *43*(4), 496–505. https://doi.org/10.1002/pd.6306

Gregg, A. R., Aarabi, M., Klugman, S., Leach, N. T., Bashford, M. T., Goldwaser, T., Chen, E., Sparks, T. N., Reddi, H. V., Rajkovic, A., & Dungan, J. S. (2021). Screening for autosomal recessive and X-linked conditions during pregnancy and preconception: A practice resource of the American College of Medical Genetics and Genomics (ACMG). *Genetics in Medicine*, *23*(10), 1793–1806. https://doi.org/10.1038/s41436-021-01203-z

Gross, S. J., Pletcher, B. A., Monaghan, K. G., & Professional Practice and Guidelines Committee (2008). Carrier screening in individuals of Ashkenazi Jewish descent. *Genetics in Medicine*, *10*(1), 54–56. https://doi.org/10.1097/GIM.0b013e31815f247c

Henneman, L., Borry, P., Chokoshvili, D., Cornel, C., van El, C., Forzano, F., Hall, A., Howard, H., Janssens, S., Kayserili, H., Lakeman, P., Lucassen, A., Metcalfe, S., Vidmar, L., de Wert, G., Dondorp, W., & Peterlin, B. (2016). Responsible implementation of expanded carrier screening. *European Journal of Human*

Genetics, *24*(6), e1–e12. https://doi.org/10.1038/ejhg.2015.271

Hull, L. E., Flannery, K., Kaimal, A. J., Sepucha, K., Rehm, H. L., & Haas, J. S. (2023). Multilevel barriers and facilitators to widespread use of preconception carrier screening in the United States. *Genetics in Medicine*, *25*(12), 100946–100946. https://doi.org/10.1016/j.gim.2023.100946

Ioannou, L., McClaren, B. J., Massie, J., Lewis, S., Metcalfe, S. A., Forrest, L., & Delatycki, M. B. (2014). Population-based carrier screening for cystic fibrosis: A systematic review of 23 years of research. *Genetics in Medicine*, *16*(3), 207–216. https://doi.org/10.1038/gim.2013.125

Johansen Taber, K., Ben-Shachar, R., Torres, R., Arjunan, A., Muzzey, D., Kaseniit, K. E., Goldberg, J., & Brown, H. (2022). A guidelines-consistent carrier screening panel that supports equity across diverse populations. *Genetics in Medicine*, *24*(1), 201–213. https://doi.org/10.1016/j.gim.2021.09.009

Kaback, M. M. (2000). Population-based genetic screening for reproductive counseling: The Tay-Sachs disease model. *European Journal of Pediatrics*, 159 Suppl 3, S192–S195. https://doi.org/10.1007/pl00014401

Kaseniit, K. E., Haque, I. S., Goldberg, J. D., Shulman, L. P., & Muzzey, D. (2020). Genetic ancestry analysis on >93,000 individuals undergoing expanded carrier screening reveals limitations of ethnicity-based medical guidelines. *Genetics in Medicine*, *22*(10), 1694–1702. https://doi.org/10.1038/s41436-020-0869-3

Kathrens-Gallardo, A., Propst, L., Linn, E., Pothast, R., Wicklund, C., & Arjunan, A. (2021). OB/GYN residents' training, attitudes, and comfort level regarding genetics. *Journal of Assisted Reproduction and Genetics*, *38*(11), 2871–2880.

https://doi.org/10.1007/s10815-021-02310-1

Langer, A. L. (2023, July 20). *Beta-Thalassemia.* Nih.gov; University of Washington, Seattle. https://www.ncbi.nlm.nih.gov/books/NBK1426/

Lazarin, G. A., Detweiler, S., Nazareth, S. B., & Ashkinadze, E. (2016). Genetic counselors' perspectives and practices regarding expanded carrier screening after initial clinical availability. *Journal of Genetic Counseling*, *25*(2), 395–404. https://doi.org/10.1007/s10897-015-9881-1

Needs, T., Gonzalez-Mosquera, L. F., & Lynch, D. T. (2023, May). *Beta Thalassemia*. Nih.gov; StatPearls Publishing. https://www.ncbi.nlm.nih.gov/books/NBK531481/

Prior, T. W., & Professional Practice and Guidelines Committee (2008). Carrier screening for spinal muscular atrophy. *Genetics in Medicine*, *10*(11), 840–842. https://doi.org/10.1097/GIM.0b013e318188d069

Putra, M., Idler, J., Dai, J., Blitzer, M. G., Feldman, G., & Sokol, R. J. (2019). Medical genetics and genomics training in obstetrics and gynecology residencies: Are we ready for the future? *The Journal of Maternal-Fetal & Neonatal Medicine*, *32*(7), 1184–1190. https://doi.org/10.1080/14767058.2017.1402879

Ramani, P. K., & Sankaran, B. P. (2023, January 25). Tay-Sachs Disease. Nih.gov; StatPearls Publishing. https://www.ncbi.nlm.nih.gov/books/NBK564432/

Ready, K., Haque, I. S., Srinivasan, B. S., & Marshall, J. R. (2012). Knowledge and attitudes regarding expanded genetic carrier screening among women's healthcare providers. *Fertility and Sterility*, *97*(2), 407–413. https://doi.org/10.1016/j.fertnstert.2011.11.007

Riordan, J. R., Rommens, J. M., Kerem, B., Alon, N., Rozmahel, R., Grzelczak, Z., Zielenski, J., Lok, S., Plavsic, N., & Chou, J. L. (1989). Identification of the cystic fibrosis gene: Cloning and characterization of complementary DNA. *Science* (New York, N.Y.), 245(4922), 1066–1073. https://doi.org/10.1126/science.2475911

Rose, N. C., & Wick, M. (2018). Carrier screening for single gene disorders. *Seminars in Fetal & Neonatal Medicine, 23*(2), 78–84. https://doi.org/10.1016/j.siny.2017.06.001

Rowley, P. T., Loader, S., Levenkron, J. C., & Phelps, C. E. (1993). Cystic fibrosis carrier screening: Knowledge and attitudes of prenatal care providers. *American Journal of Preventive Medicine, 9*(5), 261–266.

Sagaser, K. G., Malinowski, J., Westerfield, L., Proffitt, J., Hicks, M. A., Toler, T. L., Blakemore, K. J., Stevens, B. K., & Oakes, L. M. (2023). Expanded carrier screening for reproductive risk assessment: An evidence-based practice guideline from the National Society of Genetic Counselors. *Journal of Genetic Counseling, 32*(3), 540–557. https://doi.org/10.1002/jgc4.1676

Savant, A., Lyman, B., Bojanowski, C., & Jariya Upadia. (2023, March 9). *Cystic Fibrosis.* Nih.gov; University of Washington, Seattle. https://www.ncbi.nlm.nih.gov/books/NBK1250/

Srinivasan, B. S., Evans, E. A., Flannick, J., Patterson, A. S., Chang, C. C., Pham, T., Young, S., Kaushal, A., Lee, J., Jacobson, J. L., & Patrizio, P. (2010). A universal carrier test for the long tail of Mendelian disease. *Reproductive BioMedicine Online, 21*(4), 537–551. https://doi.org/10.1016/j.rbmo.2010.05.012

Strauss, T. S., Schneider, E., Boniferro, E., Brockhoff, E., Johnson, A., Stoffels, G., Feldman, K., Grubman, O., Cole, D., Hussain, F., Ashmead, G., Al-Ibraheemi, Z., & Brustman, L. (2023). Barriers to completion of expanded carrier screening in an inner city population. *Genetics in Medicine, 25*(7), 100858. https://doi.org/10.1016/j.gim.2023.100858

Toro, C., Shirvan, L., & Tifft, C. (2020, October). *HEXA Disorders.* Nih.gov; University of Washington, Seattle. https://www.ncbi.nlm.nih.gov/books/NBK1218/

Watson, M. S., Cutting, G. R., Desnick, R. J., Driscoll, D. A., Klinger, K., Mennuti, M., Palomaki, G. E., Popovich, B. W., Pratt, V. M., Rohlfs, E. M., Strom, C. M., Richards, C. S., Witt, D. R., & Grody, W. W. (2004). Cystic fibrosis population carrier screening: 2004 revision of American College of Medical Genetics mutation panel. *Genetics in Medicine, 6*(5), 387–391. https://doi.org/10.1097/01.gim.0000139506.11694.7c

Westemeyer, M., Saucier, J., Wallace, J., Prins, S. A., Shetty, A., Malhotra, M., Demko, Z. P., Eng, C. M., Weckstein, L., Boostanfar, R., Rabinowitz, M., Benn, P., Keen-Kim, D., & Billings, P. (2020). Clinical experience with carrier screening in a general population: Support for a comprehensive pan-ethnic approach. *Genetics in Medicine, 22*(8), 1320–1328. https://doi.org/10.1038/s41436-020-0807-4

Appendix A: Participant Invitation Letter Email

Dear OBGYN Residency Program Coordinators & Program Directors,

I am writing to let you know about an opportunity for your residents to participate in a voluntary research survey study called "Assessing OB GYN residents' knowledge, attitudes, and current practices for carrier screening" (IRB Study number: Pro00129845). This study is being conducted by Aubrey Surian, (Principal Investigator), Karlee Gibbon MD-R, Jessica Fairey MS, CGC (Thesis Advisor), and Sumit Saraf MD through the University of South Carolina School of Medicine USC Genetic Counseling Program.

Please forward the email below to your residents to facilitate participation

Dear OBGYN residents,

I am Dr. Karlee Gibbon, a third year OBGYN resident at the Prisma Health/University of South Carolina Columbia OB/GYN Residency Program. I am reaching out on behalf of Aubrey Surian, a genetic counseling graduate student at the University of South Carolina School of Medicine. We are conducting a voluntary research survey to assess OB GYN residents' knowledge, attitudes, and current practices for carrier screening.

The goal of this study is to understand the knowledge, attitudes, and current practices of OB/GYN residents regarding genetic carrier screening. We aim to assess the utility of current practice guidelines and identify if there are barriers to implementing these guidelines. Participation and completion of this survey is entirely voluntary and should take no longer than 10 minutes. If you begin the survey and choose to continue, please answer each question to the best of your ability. You may choose to withdraw from the study at any time by exiting the survey. No personal identifying information will be used, and anonymity will be maintained. There is no anticipated risk to participants.

At the end of the survey, there will be an option to enter a raffle. There will be a link for participants to enter their email address. Ten participants will be randomly selected to win a $20 gift card, which will be sent directly to their email address. Immediately after the winners have been chosen, the email addresses will be deleted from the survey software. Your email address will not be linked to your survey responses in any way.

Thank you for taking this research study into consideration. If you are comfortable doing so, please share this invitation letter and survey with your colleagues and/or residency program. Please reach out to myself, Aubrey Surian, or our faculty advisor, Jessica Fairey, if you have any questions concerning this research. For more specific questions or concerns about participating in research and your rights please communicate with the Office of Research Compliance at the University of South Carolina.

Contact information for each party is below.

Link to survey: https://redcap.healthsciencessc.org/surveys/?s=TPDLLPDRXPAMC4D9
It is estimated to take approximately 5-10 minutes to complete.

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Appendix B: Participant Questionnaire

Assessing OB GYN residents' knowledge, attitudes, and current practices for carrier screening

Dear Potential Participant,

I am a genetic counseling graduate student at the University of South Carolina School of Medicine. This letter is an invitation to participate in a research study that involves completing an online survey. The intent is for this survey to reach obstetric and gynecology residents.

The goal of this study is to understand the knowledge, attitudes, and current practices of OB/GYN residents regarding genetic carrier screening. We aim to assess the utility of current practice guidelines and identify if there are barriers to implementing these guidelines.

Participation and completion of this survey is entirely voluntary and should take no longer than 10 minutes. If you begin the survey and choose to continue, please answer each question to the best of your ability. You may choose to withdraw from the study at any time by exiting the survey. No personal identifying information will be used, and anonymity will be maintained. There is no anticipated risk to participants.

At the end of the survey, there will be an option to enter a raffle. There will be a link where participants can enter their email address. Ten participants will be randomly selected to win a $20 gift card, which will be sent directly to their email address. Immediately after the winners have been chosen, the email addresses will be deleted from the survey software. Your email address will not be linked to your survey responses in any way.

Thank you for taking this research study into consideration. If you are comfortable doing so, please share this invitation letter and survey with your colleagues and/or residency program.

Please reach out to myself or my faculty advisor, Jessia Fairey, if you have any questions concerning this research. For more specific questions or concerns about participating in research and your rights please communicate with the Office of Research Compliance at the University of South Carolina. Contact information for each party is below.

It is estimated to take approximately 5-10 minutes to complete.

Aubrey Surian, B.S.

Genetic Counseling Candidate

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Section 1	
If both individuals in a couple test negative for a specific genetic condition, it is still possible for their child to have that condition.	◯ True ◯ False
An individual can be a carrier for a genetic condition even when there is no history of the condition in the family.	◯ True ◯ False
Individuals in certain ethnic groups have an increased risk of being carriers of certain genetic conditions.	◯ True ◯ False
All variants in a gene (i.e. pathogenic, benign, and variants of uncertain significance) are reported by carrier screening.	◯ True ◯ False
If both individuals in a couple are carriers of a mutation in a gene that causes an autosomal recessive condition, the probability that their child will have the condition is	◯ 25% ◯ 33% ◯ 50% ◯ 75% ◯ 100%

Section 2

Definitions of terms:

Carrier screening is a genetic test that looks at a variety of conditions that can be passed on in an autosomal recessive or X-linked manner. Parents are screened to determine the risk for their children to inherit these conditions. The definition remains the same throughout the survey.

Pretest counseling is completed prior to carrier screening in order to help patients make informed decisions about testing. As recommended by the ACMG, this should include discussions of clinical significance, autosomal recessive inheritance, limitations of carrier screening (i.e. does not test for all conditions, only reports pathogenic or likely pathogenic variants, only applicable for the current reproductive partner, cannot identify de novo variants, does not eliminate risk), and that it is an optional screening.

	Strongly Disagree	Disagree	Neutral	Agree	Strongly Agree
If the costs were the same, I would prefer to order a panel testing a larger number of genetic conditions rather than for a smaller number.	○	○	○	○	○
Only genetic conditions that severely impact life should be screened for with carrier screening.	○	○	○	○	○
Carrier screening should only be offered for conditions in which there is treatment or management available.	○	○	○	○	○

	Strongly Disagree	Disagree	Neutral	Agree	Strongly Agree
Preconception is the best time to offer carrier screening.	○	○	○	○	○
All patients of reproductive age should be able to receive carrier screening if they desire.	○	○	○	○	○

	Strongly Disagree	Disagree	Neutral	Agree	Strongly Agree
I feel confident in offering pretest counseling for carrier screening.	○	○	○	○	○
I believe it is my responsibility to provide pretest counseling to all my patients pursuing carrier screening.	○	○	○	○	○

	Strongly Disagree	Disagree	Neutral	Agree	Strongly Agree

A post-test consultation with a genetic counselor would be helpful regardless of the carrier screening results.	○	○	○	○	○
A post-test consultation with a genetic counselor would be helpful only for positive carrier screening results.	○	○	○	○	○
I feel confident in disclosing negative results from carrier screening.	○	○	○	○	○
I feel confident in disclosing positive results from carrier screening.	○	○	○	○	○

Section 3	
As of 2023, under what circumstances does your practice offer genetic carrier screening? Select all that apply.	☐ Does not offer genetic carrier screening under any circumstances ☐ Offers to patients considered high risk (based on personal or family medical history) prior to conception ☐ Offers to patients considered high risk (based on personal or family medical history) during pregnancy ☐ Offers to all patients of reproductive age prior to conception ☐ Offers to all patients during pregnancy ☐ Only offers upon patient request
At what point in time does your practice offer carrier screening? Select all that apply.	☐ Only on patient request ☐ During annual well-woman exams ☐ During appointments scheduled to specifically address fertility ☐ During follow-up care for miscarriages ☐ During appointments for ongoing pregnancy
For a typical patient, how many conditions do you screen for with carrier screening?	_____
Which genetic carrier screening option does your practice prefer to offer patients? Select all that apply.	☐ ACOG recommendation of cystic fibrosis, spinal muscular atrophy, and hemoglobinopathies ☐ Specific carrier testing based on personal or family history only ☐ A pan-ethnic genetic carrier screening approach, which screens for hundreds of genetic mutations including common recessive childhood illnesses, regardless of ethnicity ☐ Do not offer carrier screening ☐ Other
Please specify "other"	_____
Do you consider a patient's ethnicity when selecting a panel to offer?	○ Yes ○ No
Which concepts of genetic carrier screening do you include in pretest counseling? Select all that apply.	☐ Frequency of carrier detection ☐ Clinical significance of conditions being tested ☐ Autosomal recessive inheritance ☐ Prenatal diagnosis options ☐ Limitations of carrier screening (i.e. does not test for all conditions, only reports pathogenic or likely pathogenic variants, only applicable for current reproductive partner, cannot identify de novo variants) ☐ None of the above
How do you notify patients about their positive genetic carrier testing results? Select all that apply.	☐ Phone call from obstetrician ☐ Phone call from nurse ☐ Letter/Email/MyChart ☐ Phone call from genetic counselor ☐ Office appointment ☐ Other

Please specify "other"	_____
How do you notify patients about their negative genetic carrier testing results? Select all that apply.	☐ Phone call from obstetrician ☐ Phone call from nurse ☐ Letter/Email/MyChart ☐ Phone call from genetic counselor ☐ Office appointment ☐ Other
Please specify "other"	_____
When disclosing positive results, what is your follow-up?	☐ Schedule partner carrier screening ☐ Refer to genetic counseling ☐ No follow-up ☐ Other
Please specify "other"	_____
When disclosing negative results, do you discuss residual risk? Residual risk definition: the risk for a child to be affected with a genetic condition after a negative genetic test.	◯ Yes ◯ No
Which professional organization do you primarily follow for clinical practice guidelines related to carrier screening?	☐ ACMG ☐ ACOG ☐ Other
Please specify "other"	_____

	Never	Sometimes	Often	Almost always
How often do you consult or refer to carrier screening practice guidelines from ACOG?	◯	◯	◯	◯
How often do you consult or refer to carrier screening practice guidelines from ACMG?	◯	◯	◯	◯

How would you prefer to receive updates about practice guidelines? Select all that apply.	☐ Society/Professional Organization Websites ☐ Regular meetings with genetics professionals ☐ Seminars that provide CME credits ☐ Virtual service helpline to consult with genetics professionals when needed ☐ Other
Please specify "other"	_____
Who would you reach out to if you have questions about genetic testing?	_____

Do you have access to a genetic counselor or genetics team within or near your practice?	◯ Yes ◯ No
What resources would you like as guidance for offering and delivering results for carrier screening?	_____
Do you have any other comments?	_____

Demographics	
Age	_____
Gender	◯ Male ◯ Female ◯ Non-binary ◯ Transgender male ◯ Transgender female ◯ Prefer not to answer ◯ Other
Ethnicity/race (Select all that apply)	☐ American Indian or Alaska Native ☐ Asian ☐ Black or African American ☐ Native Hawaiian or Other Pacific Islander ☐ White ☐ Hispanic ☐ Other ☐ Prefer not to say
What is your PGY residency year?	_____

Which state or territory of the United States do you primarily practice in?	◯ Alabama ◯ Alaska ◯ Arizona ◯ Arkansas ◯ California ◯ Colorado ◯ Connecticut ◯ Delaware ◯ Florida ◯ Georgia ◯ Hawaii ◯ Idaho ◯ Illinois ◯ Indiana ◯ Iowa ◯ Kansas ◯ Kentucky ◯ Louisiana ◯ Maine ◯ Maryland ◯ Massachusetts ◯ Michigan ◯ Minnesota ◯ Mississippi ◯ Missouri ◯ Montana ◯ Nebraska ◯ Nevada ◯ New Hampshire ◯ New Jersey ◯ New Mexico ◯ New York ◯ North Carolina ◯ North Dakota ◯ Ohio ◯ Oklahoma ◯ Oregon ◯ Pennsylvania ◯ Puerto Rico ◯ Rhode Island ◯ South Carolina ◯ South Dakota ◯ Tennessee ◯ Texas ◯ Utah ◯ Vermont ◯ Virginia ◯ Washington ◯ West Virginia ◯ Wisconsin ◯ Wyoming
Which best describes the location of your primary practice?	☐ Urban ☐ Suburban ☐ Rural
What ethnicities are represented in your patient population? Select all that apply.	☐ American Indian or Alaska Native ☐ Asian ☐ Black or African American ☐ Native Hawaiian or Other Pacific Islander ☐ White ☐ Hispanic ☐ Other
Please specify "other"	_____

Appendix C: Raffle Information

Raffle

Thank you for your participation in this survey. If you would like to be entered into a raffle to win one of 10 $20 gift cards, please select yes below. You will be taken to a separate survey where you can enter your email. Your email will be separate from your survey responses, so these will remain anonymous.

Would you like to be entered into the raffle?

◯ Yes
◯ No

Email:

_____