Spring 2022

Exploring Genetic Counselors' Experiences, Language, and Discussion of Consanguinity in Clinical Practice: A Multinational Perspective

Romy Isabel Fawaz

EXPLORING GENETIC COUNSELORS' EXPERIENCES, LANGUAGE, AND DISCUSSION OF CONSANGUINITY IN CLINICAL PRACTICE: A MULTINATIONAL PERSPECTIVE

By

Romy Isabel Fawaz

Bachelor of Science
University of Oklahoma, 2018

Submitted in Partial Fulfillment of the Requirements

For the Degree of Master of Science in

Genetic Counseling

School of Medicine

University of South Carolina

2022

Accepted by:

Janice Edwards, Director of Thesis

Rawan Awwad, Reader

Robin Bennett, Reader

Tracey L. Weldon, Interim Dean and Vice Provost and Dean of the Graduate School

ACKNOWLEDGMENTS

I would like to thank my thesis committee, Janice Edwards, Rawan Awwad, and Robin Bennett. I greatly appreciate your commitment to the project and shared excitement over the last year. Thank you all for your thorough review and suggestions. You all provided guidance and incredible insight into this idea through your own personal and professional experiences throughout the entire thesis process. I cannot thank you all enough for your unwavering confidence, encouragement, trust, and belief in the success of this project. Thank you, Amy Wardyn, for sharing your expertise and taking the time to read and review every aspect of this study.

I would like to thank my family and friends for their support throughout my graduate school career. To my family, I appreciate all the kind words, encouragement, and patience at every point throughout my graduate training. You provide such a comfort of love and support as I pursue my goals and aspirations. I am eternally grateful to all of my friends who have listened, offered advice, read this project, provided words of encouragement and great morale throughout this process. To Grace Hollingsworth and Rachel Nicely, I am forever thankful to have experienced all the laughs, memories, tears, and challenges we have faced together these last two years during our graduate school experience. To my entire class, thank you for all the collaboration, teamwork, and love as we all have grown and developed into spectacular genetic counselors

ABSTRACT

Consanguinity, defined as the degree of relationship between closely related individuals, is a widespread historical practice that is not specific to any one religion, population, or region of the world. Genetic counselors regularly ask whether a reproductive couple is consanguineous as part of the review of family history and risk assessment. Couples who are in consanguineous relationship may be subject to negative attitudes and stigma, potentially due to cultural differences and norms at a population level and these attitudes may interplay with questions, answers, and discussions around consanguinity. We hypothesized that genetic counselors may experience some level of discomfort discussing and responding to patients when inquiring about consanguinity due to cultural stigma and misconceptions of genetic risks associated with consanguinity. We aimed to gain an understanding of strategies genetic counselors used to discuss consanguinity. Over 140 genetic counselors from 15 various specialties, with years of experience ranging from 2 to 35 years, and from 16 countries responded to a survey exploring experiences with consanguinity to understand strategies used in clinical practice. Overall, the study found that most genetic counselors feel comfortable counseling on consanguinity and there is great variation in how genetic counselors ask about consanguinity, how genetic counselors initially react when consanguinity is disclosed, and methods used to address patient discomfort.

Counselors who report more experience with consanguineous couples were more easily able to educate patients and respond properly to their reactions than those with lesser experience. Additionally, 10% of respondents disclosed history of consanguinity within their own families and shared experiences with attitudes of colleagues and others, some of which included apparently biased interactions. Given the multiple perspectives of genetic counselors represented and their experience in discussing consanguinity, we can infer from this study that the way a genetic counselor intentionally asks and responds to this question has impact. The genetic counselor can create the opportunity to ask without prejudice and is uniquely positioned to educate patients and others about accurate risk assessment among consanguineous couples. This study also supports the importance that healthcare providers, particularly genetic counselors, continue to reflect on personal thoughts, feelings, and potential biases related to consanguinity as we serve others with acceptance.

.

TABLE OF CONTENTS

LIST OF TABLES

CHAPTER 1: BACKGROUND

1.1 Introduction to Consanguinity

Consanguinity directly translates from the Latin derivative meaning "of the same blood." In medical genetics, consanguinity is defined as any union between biological relatives descending from one or more common ancestors and, more specifically, between second cousins or closer in relation (Bittles, 2012). Consanguinity is often interchanged with the word *inbreeding* when describing population genetics. It often can be misinterpreted for the term incest which represents sexual unions between first-degree consanguineous relatives, such as father-daughter or brother-sister relations (Bittles, 2012).

All forms of consanguineous relationships share alleles inherited from common ancestors. The degree of consanguinity is calculated using the coefficient of inbreeding (*F*). The coefficient of inbreeding determines the probability of a couple's offspring inheriting two identical alleles, one from each parent (Bittles, 2001). The closer the biological relationship, the higher the coefficient of inbreeding and the higher likelihood of homozygosity in offspring (Hamamy, 2012). Consanguineous relationships are commonly discovered through the utilization of a pedigree in a clinical setting. The coefficient of inbreeding will vary by degree of first cousin, first cousin once removed, , pedigree may also consist complex consanguinity loops due to successive generations of cousin unions, leading to a higher coefficient of inbreeding.

1.2 Cultural Implications of Consanguinity

1.2.1 Historical Beliefs and Acceptance of Consanguinity

The practice of marriage and childbearing between closely related individuals is a widespread historical practice that is not specific to any one religion, population, or region of the world. It is therefore difficult to compare the rates of consanguinity in different populations because many reports claim third-degree cousins or more distantly related relationships as consanguinity. However, it is known that consanguineous marriages are more frequently customary and preferred in some populations and communities in some parts of the world such as Africa, the Middle East, and West, Central, and South Asia (Bittles & Black, 2010). The highest rates of consanguinity have been shown in communities that follow more traditional lifestyles of marriage and familial values (Bittles, 2008; Hamamy, 2012). Consanguineous marriages across populations are thought to have social and cultural advantages including, but not limited to, stable marital relationships, improved female autonomy, better compatibility with in-laws, less domestic violence, and lower divorce rates. Consanguineous marriages are noted to preserve culture and overall improve familial relationships for future generations (Bhinder et al., 2019). However, there are various contradicting beliefs between the acceptance of consanguineous unions to aversions of such relationships throughout the world as shown through religious beliefs, cultural acceptance, and civil laws.

1.2.2 Stigmatizations Around Consanguinity

While there is historical evidence of long-standing consanguinity between families throughout history and throughout the world, blood-related relationships are often treated with suspicion, embarrassment, and disdain (Bittles, 2012). Consanguineous

relationships are viewed to cause physical harm and severe intellectual disability. These stigmatizations may have stemmed from historical laws prohibiting specific relationships and deeply rooted misconceptions about the medical consequences of consanguineous unions within the medical community. In the United States, consanguinity has been associated with rural communities of low income, socioeconomic status, and specific United States populations, particularly in the southeast region (Bittles, 2012). For example, Appalachian mining towns in the 1930's made national news when images of rundown shacks, barefoot children, and rural mountain communities were broadcasted, and reporters claimed that "incest" was the explanation for their social conditions (Harkins, 2001).

Additional stigmatizations around consanguineous marriages may have stemmed from the civil and legal regulations that are in place throughout the world. Rules and regulations surrounding incestuous relationships (i.e. full siblings or direct descendants) have been in place since the late 16th century, dating back to the era of royalty in Great Britain (Bittles, 2012). These laws are known as 'prohibited degree of kinship' and stated what degree of relation was acceptable under civil law and which relationships were prohibited. The laws in the United States are the most strictly prohibiting, with each state defining incestuous relationships with different degrees of relations. Many states consider first cousin marriage as incest and in 2018, first cousin marriage was considered a criminal offense in nine states with a total of 30 states prohibiting first cousin marriages.

1.3 Consanguinity in Medical Genetics and Genetic Counseling

Individuals in consanguineous relationships are often misinformed on the risks of disease for future children (Cupp et al., 2020; Modell & Darr, 2002; Shaw & Hurst,

2009). This underscores the importance of disseminating correct information to patients and healthcare providers to understand the biological and social implications of consanguineous relationships (Alnaqeb et al., 2018).

1.3.1 Risk Assessment

Documenting consanguinity is an important step in the risk assessment portion of a genetic counseling session. The likelihood of a couple sharing common deleterious alleles is typically assessed. Disorders with an autosomal recessive inheritance pattern, in which both parents are heterozygous for a genetic mutation with a 25% chance that each offspring would be affected with a specific condition, are more commonly seen when the parents are related (Hamamy et al., 2011). The effect of consanguinity is particularly marked in rare recessive disorders because it is unlikely that a carrier finds a partner who is also a carrier for the same rare disorder unless related (Modell & Darr, 2002). Numerous studies have shown that the risk for having an affected child will depend on several factors including population risk, degree of consanguineous relationship, and previous family history (Bennett et al., 2002; Hamamy et al., 2011; Oniya et al., 2019). This reinforces the concept that medical conditions do not occur due to the practices of consanguineous relationships, but because of recessive genes and the likelihood that two individuals would both be carriers for the same condition.

In the United States, it has been shown that overall, there is a 1.7-2.8% average increased risk for any congenital disorder in children of parents who are first cousins over the general population's 2-3% risk (Bennett et al. 2002; Hamamy et al, 2011). Recommendations for prenatal screening for consanguineous couples are almost identical to screening for the general population. Currently, it is recommended that expectant

couples are offered at least genetic carrier screening for cystic fibrosis and spinal muscular atrophy and based on ancestry, screening for hemoglobinopathies and Jewish genetic disease carrier status (Deignan et al, 2020). Several professional organizations recommend that consanguineous couples be offered an expanded genetic carrier screen, which is a more comprehensive test to include 100-500 recessive genetic disorders (Gregg et al., 2021). The American College of Medical Genetics (ACMG) has recently recommended that expanded carrier screening should not be restricted based on the ethnicity of patients and could potentially be offered to everyone while considering the most comprehensive level of testing for those with known consanguinity (Gregg et al., 2021).

1.3.2 Revealing Consanguinity through Genetic Testing

Chromosomal microarray analysis (CMA) is utilized in both prenatal and pediatric genetic specialties for a variety of indications including multiple congenital anomalies, intellectual disability, developmental delay, autism spectrum disorder, or other specific medical conditions (Shao, 2021). Long strands of continuous homozygosity shown on CMA are termed "regions of homozygosity" (ROH) or sometimes called "absence of heterozygosity". Multiple large homozygous regions involving multiple chromosomes indicate that the patient is the offspring of related parents (Sund, 2014). The theoretical degree of homozygosity shown on test results ranges based on the degree of relationship. Fifth-degree relatives' ROH are equivalent to about 1.5%, whereas 25% homozygosity would be representative of a first-degree parent-child relationship (Rehder et al., 2013; Sund et al., 2014). The ACMG issued guidelines focused on reporting ROH and consanguinity following CMA result reporting. Laboratories are responsible for

determining when a segment of homozygosity is considered large enough to represent consanguinity (Rehder et al., 2013).

Clinical providers and genetic counselors are then responsible for interpreting and disclosing the results to the patient from the CMA laboratory report. Comfortability surveys of clinicians who disclose microarray ROH results stated they are very comfortable receiving and counseling about these results (Grote et al., 2014). Clinicians' experiences with reporting these results showed that some families acknowledged the relationship, some continued to deny parental relationship even after explanation of results, and others offered a cultural explanation. The discomfort of receiving a report indicating extensive ROH by clinicians was reported to stem from the fear of a nonconsensual relationship or concern for abuse or incest in patients (Grote et al., 2014; Sund et al., 2014).

A focused revision on the genetic counseling and genetic testing screening of consanguineous couples and their offspring provides a practice resource incorporating the newest screening and testing options available. In additional to CMA, whole exome sequencing and whole genome sequencing for patients with a suspected genetic disorder yields results showing homozygosity. These genetic tests offer an improved diagnostic yield and identifying more pathogenic and likely pathogenic variants in autosomal recessive variants (Bennett et al. 2021). These recommendations state that pre-test counseling should always include the possibility of detecting biological relationship, especially when a close biological relationship was not previously known.

1.3.3 Genetic Counseling Scope of Practice with Consanguinity

While the focus of consanguinity in genetic counseling is most often related to genetic evaluation in a prenatal and pediatric setting, the question of relation is asked in almost every genetic counseling specialty when documenting family history. In 1994 and 1996, the World Health Organization Regional Office of the Eastern Mediterranean determined that genetic counselors were best equipped to educate consanguineous couples on the risk of recessive disease transmission through families (Alwan and Modell, 1997). Genetic counseling services for consanguineous couples have been shown to reduce anxiety, address prior misperceived risks of consanguineous relationships, provide an accurate risk assessment based on the degree of relationship and family history, and empower couples in their decision-making process (Thain et al., 2019).

The question of consanguinity first presents itself in a genetic counseling session during the family history portion while drawing the pedigree. The most recent recommendations for counseling consanguineous couples suggest that genetic counselors should obtain a three- to four-generation pedigree, followed by risk assessment and appropriate testing based on family history and ethnicity, with the same approach as for non-consanguineous couples (Bennett et al., 2020). Resources for asking about consanguinity are limited to suggestions in training modules or textbooks that pose the general question as "Are you and your partner, or parents or grandparents related as cousins?" (Bennett, 2010).

Recommendations for inquiring about consanguinity share common themes of trying to reassure patients and state that the purpose of taking a family history and asking about consanguinity is to understand familial relationships, understand the inheritance

pattern of a condition, potentially confirm a diagnosis, and calculate the risk of genetic disease (Ahmen, 2013; Bennett, 2010). However, parents who have a child with a genetic condition and a personal history of consanguinity may be defensive in discussing their medical history and often provide examples of other individuals in their family who share a similar consanguineous relationship whose children have no genetic condition and are healthy. The defensiveness may come from feeling targeted or blamed for their child's medical condition (Ahmed, 2013). Other studies have reported that patients may fear seeking genetic advice for the same fear of being blamed. It is encouraged that all genetic health professionals avoid using language that may be stigmatizing (Shaw and Hurst, 2008).

1.4 Microaggressions When Discussing Consanguinity

Opinions on consanguineous marriage often differ in the genetic and scientific community, where some highlight the importance of social and cultural implications, while others are more concerned over the potential health risks to offspring (Bhinder et al., 2019; Hamamy et al., 2011; Teeuw et al., 2011). Due to the mixed prevalence of consanguinity across different regions and populations, the most practical means of confirming consanguinity is to ask every patient (Oniya et al., 2019). However, the stigma surrounding these relationships may make patients feel uncomfortable when asked, "are you and your partner's families related?" The stigma surrounding couples who are consanguineous in the medical community could perpetuate the stereotypes that are present in current culture. The language that genetic counselors use around consanguinity has the potential to be considered microaggressions.

The term microaggression refers to the 'commonplace behavioral indignities whether intentional or unintentional communicating hostile, derogatory or negative attitudes toward marginalized groups' (Pierce, 1974). Consanguineous couples would be subject to microaggressions under this definition. Individuals are not immune to social and cultural prejudices and many healthcare providers have exaggerated the idea of genetic disadvantages in consanguineous relationships, especially when a couple who is consanguineous has a child with a medical condition, whether it is an inherited disorder or not (Alnaqeb et al., 2018).

The National Society of Genetic Counselors (NSGC) provides a clear statement regarding justice, equality, diversity, and inclusion (JEDI) within their governing body, with the intention to "empower our members to advocate for themselves, each other, and the diverse people we strive to serve" (NSGC, 2022). The characteristics that fall under this mission statement include but are not limited to underrepresented cultures, languages, religion, health history, and spiritual beliefs. The duties of a genetic counselor are to then promote justice, equity, diversity, and inclusion to all patients and to colleagues in the field.

1.5 Rationale

While most patient-facing genetic counselors inquire about consanguinity, there is no research to determine how genetic counselors elicit this information. Most individuals have no known consanguinity in their family and may therefore not view the wording as harmful. However, it is impossible to assess who is consanguineous without asking. It would be inappropriate and offensive to only ask individuals from geographical areas suspected to have a higher prevalence of consanguinity. Therefore, there must be careful

consideration in how it is discussed and ensure that genetic counselors are using culturally appropriate and non-offensive language with all patients.

1.6 Purpose

Genetic counseling includes genetic risk assessment. Genetic counselors aim to educate individuals about the causes of genetic disease with correct information and to inform them of additional resources available through testing, treatment, and prevention. (Alwan and Modell, 1997; Thain et al., 2019). Genetic counseling standard practice includes genetic counselors asking about consanguinity as they obtain a pedigree during a genetic counseling encounter. However, negative attitudes and stigma, potentially due to cultural differences and norms at a population level, may interplay with questions, answers, and discussions around consanguinity. Consanguineous couples may deny their relationship during a medical intake because they fear discrimination, ostracization, and even legal prosecution in some areas (Bennett et al., 2002).

There are varied practices among genetic counselors in asking patients questions regarding consanguinity. Although few resources provide recommendations for addressing consanguinity in a genetic counseling session, it is unclear the usage and utility of these resources in practice (Bishop, 2008; Modell & Darr, 2002; Teeuw, 2012). Assessing communication around consanguinity in clinical practice is essential to informing genetic counselor education and guidelines for practice. To our knowledge, no study to date represents a transnational perspective of genetic counselor experiences focused on consanguinity. Through lived and shared experiences, we believe that the phrasing and questioning regarding consanguinity in a genetic counseling session may influence attitudes, stereotypes, and misconceptions surrounding consanguineous

relationships. This study explored genetic counselors' perspectives on discussing consanguinity with patients. By surveying genetic counselors from diverse educational backgrounds, cultures, and countries, we aimed to gain a multinational understanding of varied strategies used to discuss consanguinity and potentially formulate recommendations for addressing the questions tailored to all patients, regardless of biological family relations. We primarily aimed to determine the following objectives:

1. Assess the phrasing and context in which consanguinity is discussed;
2. Determine genetic counselors' responses when addressing consanguinity in a clinical session;
3. Assess if specific phrases used in common discussion are considered microaggressions or perpetuate stereotypes; and
4. Explore opportunities to better understand and educate on consanguinity

We hypothesized that there is some level of discomfort discussing or responding to patients when inquiring about consanguinity due to the cultural stigma and misconceptions of genetic risks around consanguineous unions. We predicted that genetic counselors who have more experience working with consanguineous couples may offer a more robust understanding of counseling guidelines and provide better discussion and management strategies when compared to those genetic counselors with less experience.

CHAPTER 2: EXPLORING GENETIC COUNSELORS' EXPERIENCES, LANGUAGE, AND DISCUSSION OF CONSANGUINITY IN CLINICAL PRACTICE: A MULTINATIONAL PERSPECTIVE[1]

[1] Fawaz, R.I., Edwards, J.G., Awwad, R., & Bennett, R. To be submitted to *Journal of Genetic Counseling*

2.1 Abstract

Consanguinity, defined as the degree of relationship between closely related individuals, is a widespread historical practice that is not specific to any one religion, population, or region of the world. Genetic counselors regularly ask whether a reproductive couple is consanguineous as part of the review of family history and risk assessment. Couples who are in consanguineous relationship may be subject to negative attitudes and stigma, potentially due to cultural differences and norms at a population level and these attitudes may interplay with questions, answers, and discussions around consanguinity. We hypothesized that genetic counselors may experience some level of discomfort discussing and responding to patients when inquiring about consanguinity due to cultural stigma and misconceptions of genetic risks associated with consanguinity. We aimed to gain an understanding of strategies genetic counselors used to discuss consanguinity. Over 140 genetic counselors from 15 various specialties, with years of experience ranging from 2 to 35 years, and from 16 countries responded to a survey exploring experiences with consanguinity to understand strategies used in clinical practice. Overall, the study found that most genetic counselors feel comfortable counseling on consanguinity and there is great variation in how genetic counselors ask about consanguinity, how genetic counselors initially react when consanguinity is disclosed, and methods used to address patient discomfort. Counselors who report more experience with consanguineous couples were more easily able to educate patients and respond properly to their reactions than those with lesser experience. Additionally, 10% of respondents disclosed history of consanguinity within their own families and shared experiences with attitudes of colleagues and others, some of which included apparently

biased interactions. Given the multiple perspectives of genetic counselors represented and their experience in discussing consanguinity, we can infer from this study the way that a genetic counselor intentionally asks and responds to this question has impact. The genetic counselor can create the opportunity to ask without prejudice and is uniquely positioned to educate patients and others about accurate risk assessment among consanguineous couples. This study also supports the importance that healthcare providers, particularly genetic counselors, continue to reflect on personal thoughts, feelings, and potential biases related to consanguinity as we serve others with acceptance.

2.2 Introduction

In medical genetics, consanguinity is defined as any union between biological relatives descending from one or more common ancestors and, more specifically, between second cousins or closer in relation (Bittles, 2012). All forms of consanguineous relationships share alleles inherited from common ancestors. The degree of consanguinity is calculated using the coefficient of inbreeding (*F*). The coefficient of inbreeding determines the probability of a couple's offspring inheriting two identical alleles, one from each parent (Bittles, 2001). The closer the biological relationship, the higher the coefficient of inbreeding and the higher likelihood of homozygosity in offspring (Hamamy, 2012).

The practice of marriage and childbearing between closely related individuals is a widespread historical practice that is not specific to any one religion, population, or region of the world. It is known that consanguineous marriages are more frequently customary and preferred in some populations and communities in some parts of the world such as Africa, the Middle East, and West, Central, and South Asia (Bittles & Black,

2010). The highest rates of consanguinity have been shown in communities that follow more traditional lifestyles of marriage and familial values (Bittles, 2008; Hamamy, 2012). Consanguineous unions have been prevalent in many communities including rural and low socioeconomic class, middle-class families with large holdings or significant personal wealth, and upper-class society including royal families throughout history and around the world. Consanguineous marriages across populations are thought to have social and cultural advantages including, but not limited to, stable marital relationships, improved female autonomy, better compatibility with in-laws, less domestic violence, and lower divorce rates (Bhinder et al., 2019). Consanguineous marriages are noted to preserve culture and overall improve familial relationships for future generations However, there are various contradicting beliefs between the acceptance of consanguineous unions to aversions of such relationships throughout the world as shown through religious beliefs, cultural acceptance, and civil laws (Bittles, 2008).

While there is historical evidence of long-standing consanguinity between families throughout history and throughout the world, blood-related relationships are often treated with suspicion, embarrassment, and disdain (Bittles, 2012). Consanguineous relationships are viewed to cause physical harm and severe intellectual disability. These stigmatizations may have stemmed from historical laws prohibiting specific relationships and deeply rooted misconceptions about the medical consequences of consanguineous unions within the medical community. Specific regions of the United States are particularly vulnerable to ridicule regarding incest and consanguinity that are linked to the state's poverty level and rural communities. In the United States, consanguinity has

been associated with rural communities of low income, socioeconomic status, and specific United States populations, particularly in the southeast region (Bittles, 2012).

Additional stigmatizations around consanguineous marriages may have stemmed from the civil and legal regulations that are in place throughout the world. Rules and regulations surrounding incestuous relationships (i.e. full siblings or direct descendants) have been in place since the late 16th century, dating back to the era of royalty in Great Britain (Bittles, 2012). These laws are known as 'prohibited degree of kinship' and stated what degree of relation was acceptable under civil law and which relationships were prohibited. The laws in the United States are the most strictly prohibiting, with each state defining incestuous relationships with different degrees of relations. Many states consider first cousin marriage as incest and in 2018, first cousin marriage was considered a criminal offense in nine states with a total of 30 states prohibiting first cousin marriages.

Individuals in consanguineous relationships are often misinformed on the risks of disease for future children (Cupp et al., 2020; Modell & Darr, 2002; Shaw & Hurst, 2009). This underscores the importance of disseminating correct information to patients and healthcare providers to understand the biological and social implications of consanguineous relationships (Alnaqeb et al., 2018).

Documenting consanguinity is an important step in the risk assessment portion of a genetic counseling session. The effect of consanguinity is particularly significant in rare recessive disorders because it is unlikely that a carrier finds a partner who is also a carrier for the same rare disorder unless related (Modell & Darr, 2002). Numerous studies have shown that the risk for having an affected child will depend on several factors including population risk, degree of consanguineous relationship, and previous family history

(Bennett et al., 2002; Hamamy et al., 2011; Oniya et al., 2019). This reinforces the concept that medical conditions do not occur due to the practice of consanguineous relationships, but because of recessive genes and the likelihood that two individuals would both be carriers for the same condition. In the United States, it has been shown that overall, there is a 1.7-2.8% average increased risk for any congenital disorder in children of parents who are first cousins over the general population's 2-3% risk (Bennett et al. 2002; Hamamy et al., 2011;). Recommendations for prenatal screening for consanguineous couples are almost identical to screening for the general population.

While the focus of consanguinity in genetic counseling is most often related to genetic evaluation in a prenatal and pediatric setting, the question of relation is asked in almost every genetic counseling specialty when documenting family history. In 1994 and 1996, the World Health Organization Regional Office of the Eastern Mediterranean determined that genetic counselors were best equipped to educate consanguineous couples on the risk of recessive disease transmission through families (Alwan and Modell, 1997). Genetic counseling services for consanguineous couples have been shown to reduce anxiety, address prior misperceived risks of consanguineous relationships, provide an accurate risk assessment based on the degree of relationship and family history, and empower couples in their decision-making process (Thain et al., 2019).

The question of consanguinity first presents itself in a genetic counseling session during the family history portion while drawing the pedigree. The most recent recommendations for counseling consanguineous couples suggest that genetic counselors should obtain a three- to four-generation pedigree, followed by risk assessment and appropriate testing based on family history and ethnicity, with the same approach as for

non-consanguineous couples (Bennett et al., 2020). Education on how to elicit consanguinity are limited to suggestions in training modules or textbooks that pose the general question as "Are you and your partner, or parents or grandparents related as cousins?" (Bennett, 2010).

Recommendations for inquiring about consanguinity share common themes of trying to reassure patients and state that the purpose of taking a family history and asking about consanguinity is to understand familial relationships, understand the inheritance pattern of a condition, potentially confirm a diagnosis, and calculate the risk of genetic disease (Ahmen, 2013; Bennett, 2010). However, parents who have a child with a genetic condition and a personal history of consanguinity may be defensive in discussing their medical history and often provide examples of other individuals in their family who share a similar consanguineous relationship whose children have no genetic condition and are healthy. The defensiveness may come from feeling targeted or blamed for their child's medical condition (Ahmed, 2013). Other studies have reported that patients may fear seeking genetic advice for the same fear of being blamed. It is encouraged that all genetic health professionals avoid using language that may be stigmatizing (Shaw and Hurst, 2008).

Opinions on consanguineous marriage often differ in the genetic and scientific community, where some highlight the importance of social and cultural implications, while others are more concerned over the potential health risks to offspring (Bhinder et al., 2019; Hamamy et al., 2011; Teeuw et al., 2011). Due to the mixed prevalence of consanguinity across different regions and populations, the most practical means of confirming consanguinity is to ask every patient (Oniya et al., 2019). However, the

stigma surrounding these relationships may make patients feel uncomfortable when asked, “are you and your partner’s families related?” The stigma surrounding couples who are consanguineous in the medical community could perpetuate the stereotypes that are present in current culture. The language that genetic counselors use around consanguinity could be considered as microaggressions.

The term microaggression refers to the ‘commonplace behavioral indignities whether intentional or unintentional communicating hostile, derogatory or negative attitudes toward marginalized groups’ (Pierce, 1974). Consanguineous couples would be subject to microaggressions under this definition. Individuals are not immune to social and cultural prejudices and many healthcare providers have exaggerated the idea of genetic disadvantages in consanguineous relationships, especially when a couple who is consanguineous has a child with a medical condition, whether it is an inherited disorder or not (Alnaqeb et al., 2018).

The National Society of Genetic Counselors (NSGC) provides a clear statement regarding justice, equality, diversity, and inclusion (JEDI) within their governing body with the intention to “empower our members to advocate for themselves, each other, and the diverse people we strive to serve” (NSGC, 2022). The characteristics that fall under this mission statement include but are not limited to underrepresented cultures, languages, religion, health history, and spiritual beliefs. The duties of a genetic counselor are to then promote justice, equity, diversity, and inclusion to all patients and to colleagues in the field.

There are varied practices among genetic counselors in asking patients questions regarding consanguinity. Although few resources provide recommendations for

addressing consanguinity in a genetic counseling session, it is unclear the usage and utility of these resources in practice (Bishop, 2008; Modell & Darr, 2002; Teeuw, 2012). Assessing communication around consanguinity in clinical practice is essential to informing genetic counselor education and guidelines for practice. To our knowledge, no study to date represents a transnational perspective of genetic counselor experiences focused on consanguinity. Through lived and shared experiences, we believe that the phrasing and questioning regarding consanguinity in a genetic counseling session may influence attitudes, stereotypes, and misconceptions surrounding consanguineous relationships. This study explored genetic counselors' perspectives on discussing consanguinity with patients. By surveying genetic counselors from diverse educational backgrounds, cultures, and countries, we aimed to gain a multinational understanding of varied strategies used to discuss consanguinity and potentially formulate recommendations for addressing the questions tailored to all patients, regardless of biological family relations.

2.3 Materials and Methods

This study was approved by the University of South Carolina Institutional Review Board (Pro00112301).

2.3.1 Participants and Recruitment

Participants included English-speaking genetic counselors who have worked in a full or partial patient-facing setting within the last five years. Genetic counselors invited to participate in this study were required to have at least one year of clinical work experience, have previously worked with consanguineous couples, and be able to recall their experiences. Only English- speaking participants were included in this study due to

limited resources for translation and interpretation from English to other languages. Other genetic professionals such as physicians or physician assistants were excluded from this survey.

Participation in the study was voluntary; therefore, reading the initial introduction and continuing the questionnaire served as the participant's consent. Participants were recruited through four online platforms: (1) National Society of Genetic Counseling listserv; (2) Transnational Alliance for Genetic Counseling listserv; (3) Arab Association of Genetic Counselors LinkedIn social media platform; (4) Minority Genetics Professional Network Slack channel. The survey was distributed electronically, including a description of the study and a link to the online questionnaire.

2.3.2 Procedure

The questionnaire was designed and administered online through Qualtrics.com and consisted of Likert scale questions, multiple-choice questions, and open-ended questions. The questionnaire included items regarding scope of practice to assess educational background and experiences. A total of 27 items included multiple-choice, free response, and one matrix question asking participants to identify actions within a session. Skip logic was utilized for participants who answered that they have consanguineous members in their family. Participants were able to skip any question or leave the questionnaire at any time, therefore completion rates varied. The survey was used to obtain qualitative and quantitative data that assessed current practices and experiences of genetic counselors when discussing consanguinity with all patients. Demographic information was placed at the end of the questionnaire to learn participants' current country of practice, previous countries of practice, number of years in practice,

and race and ethnicity. Participants remained completely anonymous, and the questionnaire did not include items about personally identifying information.

Responses were collected from July to November 2021. Of the 171 individuals who began the questionnaire, 152 participants submitted responses that were eligible for data analysis. Eligible responses were reviewed, and 149 participants provided answers to the entire questionnaire. Four participants began the questionnaire but did not meet the criteria to continue. The final analysis included 145 responses; of these, 141 respondents completed 100% of the survey. We defined completion of the survey as more than 80% items answered.

2.3.3 Data Analysis

Quantitative data were analyzed using Microsoft Office Excel software using descriptive statistical analysis with frequencies and percentages. Quantitative analysis was performed using data transferred from Excel spreadsheets into Statistical Package for Social Sciences (SPSS). Chi square analysis was used to compare comfortability with actions that genetic counselors exhibit during sessions. A constant comparison approach was utilized to analyze qualitative data gathered from free-response questions. Comparative statistical analysis was performed to assess the differences in wording, phrasing, and order of discussion of consanguinity between countries. Derived themes were coded based on participant responses, and thematic frequency was reported.

Figures and tables were constructed using Microsoft Word and Microsoft Excel software. Quotations from open-text entries were extracted and organized into categories. The level of comfort and agreeability to several statements was assessed using a Likert scale and represented descriptive statistics (percentages, frequencies, and means).

2.4 Results

2.4.1 Demographic Information

The country in which a genetic counseling graduate degree was obtained, total countries worked in and years of experience were reported for 142 participants, and specialty and race were reported for 141 of these (Table 2.1) The majority of participants acquired their graduate degree and had only worked in the United States of America; however, representation from other countries accounted for about 20% in both variables. About 48% of individuals had been working in a clinical setting for less than 5 years, and there was a multitude of specialties represented. Race categories were taken from the NSGC Professional Status Survey (PSS) that annually provides racial demographics on currently practicing genetic counselors (90% white). The respondents represented a more diverse racial group (only 74% white) than reported by the PSS. Participants were able to select more than one race.

Table 2.1 Participant Demographic Information

Variable	Descriptor	n (%)
Graduate Degree Location (*n=142)*	USA	120 (85)
	Canada	8 (6)
	South Africa	4 (5)
	United Kingdom	3 (2)
	Norway	2 (2)
	Spain	1 (1)
	Australia	1 (1)
	India	1 (1)
	Israel	1 (1)
Countries Worked In (*n=142)*[1]	USA	118
	Canada	11
	United Kingdom	5
	Saudi Arabia	5
	South Africa	4
	Oman	3
	Israel	2

	Spain	1
	Australia	1
	India	1
	Austria	1
	Iceland	1
	Norway	1
	Mexico	1
	Germany	1
Total Years Working	1-4	68 (48)
(n=142)	5-10	48 (34)
	11-15	13 (9)
	16-20	6 (4)
	21-25	5 (4)
	>26	2 (1)
Specialty	Pediatric	62
(n=141)[1]	Prenatal	55
	Cancer	37
	Other	36
	ART/Preconception	32
	Laboratory/Industry	19
Race	White	104 (73.8)
N=141[1]	Middle Eastern/West Asian/ North African	10 (7.1)
	South Asian	6 (4.3)
	East Asian or Southeast Asian, White	4 (2.8)
	Latinx/Hispanic, White	4 (2.8)
	Other	4 (2.8)
	East Asian/Southeast Asian	3 (2.1)
	African American or Black	2 (1.4)
	Latinx/Hispanic	2 (1.4)
	African American/Black, East Asian/Southeast Asian	1 (0.8)
	African American/Black, White	1 (0.8)
	America Indian or Alaska Native	0 (0)
	Native Hawaiian or Pacific Islander	0 (0)

[1]Individuals were allowed to select more than one option

2.4.2 Assessment of phrasing and discussion in which consanguinity is discussed

Genetic counselors were asked to provide the exact wording used when asking patients about consanguinity in their family. Participant responses were placed in two general categories: asking directly or asking using qualifiers. Asking directly was shown in only 30% of genetic counselor responses, with the most common phrase being "Are

you and your partner related by blood?" The majority genetic counselors (70%) ask the question using qualifiers, such as "We ask everyone this question. As far as you know, is there any chance that you and your partner are related?" Of the total responses (N=145), there were no responses that were identical. Each category was further divided into subcategories based on direct phases or qualifying phrases (Table 2.2). Participants were able to give multiple examples of how they may ask this question. One complete response such as "Any chance that you and your partner are related by blood, such as cousins?" was categorized into three different phrase subcategories: 'related by blood', 'provide an example', and 'any chance'.

Table 2.2 Phrases used when asking about consanguinity

Category	Phrase Examples	Total Count
Direct phrases	Related by blood Provide an example Share a common ancestor	223
Qualifying phrases	Any possible way/any chance We ask everyone Assess geographical location	130

Participants were asked to describe their initial thoughts, feelings, and reactions when a patient answers 'yes' to the question of consanguinity (Table 2.3). Respondents were mainly focused on how this information would impact the genetic clinical risk assessment for the patient and what additional testing or diagnosis was clinically indicated. *"Very technical thoughts and how it will affect the counseling: how is the couple related, how to draw the pedigree, degree of consanguinity."* Some genetic counselors expressed their emotional responses as their first reaction. Many described their desire to want to be non-judgmental and accepting of their patient's relationship, while feeling proud of the rapport and trust that was built within a session. One genetic

counselor stated: *"I am always grateful when patients are comfortable sharing this information, because I know it can be difficult to share this in a medical setting where they know they can be judged for it or that it contributed to a medical issue."*

Genetic counselors' cultural responses ranged from deep awareness of cultural norms to discomfort with certain degrees of relationships. An example of cultural awareness was as follows:

> *"I have no response to this, and document it as I would any other information on the family history/pedigree. This is a common occurrence in many populations, and I treat all with the same respect and understanding. Consanguineous marriages are not common in my population group, but I have always understood it and respected choice and culture/religion. Currently working in the Middle East, I have learned a lot about the meaning and reasoning behind this practice and came to respect the practice of consanguineous marriages even more."*

While others shared why they feel discomfort with certain degrees of relation:

> *"...If it were an illegal level of consanguinity (such as brother/sister, father/daughter etc) then I would find this quite confronting because it is not a socially acceptable level of consanguinity, I would be worried about harm and would feel obligated to take some level of action but would not feel confident in knowing the steps. If it were a 'socially acceptable' level of consanguinity, then I don't have any feelings or reactions to this information."*

Table 2.3 Initial thoughts, feelings, or reactions when consanguinity is determined in couples

Category		Count	%
Clinical risk assessment response	Determine degree of relationship	55	53%
	Risk assessment	24	
	Expanded carrier screening for autosomal recessive conditions	17	
	Adjust test counseling/planning clinical management	15	
	Use information for differential diagnosis	14	
	Drawing pedigree correctly	11	
	Documented in previous records	3	
Emotional response	Neutral feeling	29	36%
	Desire to be non-judgmental, understanding, accepting	22	
	Surprise, not a common response	15	
	Explaining the reason for asking	11	
	Thankful patients shared information	7	
	Normalize	7	
	Grad school trained to be open minded and accepting	3	
Cultural response	Aware of cultural differences	13	11%
	Address cultural stigma	7	
	Discomfort with closely related individuals/ concern for abuse or non-consensual relationship	9	
	Consider ancestry	2	

2.4.3 Patient discomfort

When asked how often patients laugh when asked about consanguinity, 60% of genetic counselors reported that is an often occurrence, 39% reported that laughter happens occasionally, and 11% selected that it rarely occurs. Genetic counselors were asked to recount responses they have heard from patients after the question of consanguinity was presented. Responses to the question often included a number of common phrases heard by numerous genetic counselors, with the most common response

heard from patients being a variation of "I hope not! That would be horrible if we were." Three major categories were identified within the sample response: aversion, joking, and disbelief (Table 2.4). Aversion included comments that represented disgust about the generalized practice of consanguineous unions or insinuate that the practice is wrong. One genetic counselor shared that patient will usually say something like 'oh I sure hope not' while many patients laugh and say no or look disgusted then say no. Joking comments ranged from statements such as, ""Well we are from [insert stereotypical state] so I can see why you'd ask!" or a sarcastic "yes, we are brother and sister" comment. Disbelief describes the reactions from patients that were unsure of why the question was being asked and that the practice of consanguinity is common enough that the genetic counselors ask everyone. Genetic counselors who work in countries other than the USA also reported hearing these common phrases in clinical practice. Most responses show that there is a distinct level of discomfort from the patient perspective when discussing consanguinity that may need to be addressed by the genetic counselor.

Table 2.4 Generalized patient responses following the question of consanguinity

Category	Examples	Count
Aversion	'I hope not'	72
	'Eww [sic], no'	46
	'That would be horrible'	11
	'Isn't it illegal?"	9
Joking	Jokingly answer 'yes'	16
	'We are all related if you go back far enough'	9
	'That explains my family'	7
Disbelief	'Do people ever say yes? Does that still happen?'	49
	'We are from different regions/areas'	11
	'Do you ask everyone that?'	6

2.4.4 Genetic counselor comfortability of addressing consanguinity in clinical context

Experience working with consanguinity was measured by asking how often genetic counselors see consanguineous couples in their specific practice. Genetic counselors self-reported that they rarely see (29%), sometimes see (50%), and often see couples who are consanguineous (28%). Of those, 75% of individuals selected that they were extremely comfortable and 23% were somewhat comfortable asking the question of consanguinity, while two respondents total reported that they were somewhat uncomfortable or extremely uncomfortable asking this question.

Ten common behaviors were utilized to assess how genetic counselors respond when patients make comments and frequencies were reported (Table 2.5). Genetic counselors were able to select yes or no if they have said or done any of these behaviors.

Table 2.5 Frequencies of common genetic counselor responses (n=145)

Behavior	Yes (%)	No (%)
Address the patient about the comment directly	85	15
Apologized for asking the question of any discomfort the question brought	24	76
Asked directly why the patient is laughing	7	93
Did not acknowledge the comment and moved on	62	38
Explained the reason behind why we ask the question of consanguinity	98	2
Laughed with the patient	51	49
Made a joke about being from a small town or specific state	4	96
“I know it’s a strange question”	49	51
“We have to ask everyone”	85	15
“Yes, I know it’s a funny question”	32	68

Genetic counselor reactions to patient's comments, jokes, and discomforts were analyzed and compared based on how frequently they worked with consanguineous couples. Of these ten behaviors, three behaviors were noted to have significant differences between how often genetic counselors see consanguineous couples (Table 2.6). This table lists the frequency and row percentages within each answer listed. Genetic counselors who rarely work with consanguineous couples were more likely to not acknowledge the comment and move on, whereas the sometimes or often group may have asked additional question or made a statement to address the comment.

Table 2.6 Comparison between behaviors exhibited by genetic counselors who rarely, sometimes, or often counsel couples who are consanguineous

Behavior	Answer	Rarely	Sometimes	Often	P value
Did not acknowledge comment and moved on	Yes	33 (36.3%)	45 (49.5%)	13 (14%)	.025
	No	9 (16.6%)	31 (57.4%)	14 (25.9%)	
"I know it's a strange question"	Yes	26 (41.9%)	30 (48.3%)	16 (25.8%)	.036
	No	16 (21.9%)	46 (63%)	11 (15.1%)	
"Yes, it's a funny question"	Yes	18 (38.2%)	17 (36.2%)	12 (25.5%)	.025
	No	24 (24.5%)	59 (60.2%)	15 (15.3%)	

2.4.5 Genetic counselor opinions on the continuation of asking about consanguinity

Genetic counselors were asked to provide their opinion on whether the question of consanguinity should be routinely asked in a clinical setting and to explain their reasoning. The majority of genetic counselors (70%, n=104) firmly stated yes, the question should continue to be asked, and 30% (n=41) were undecided or said the question should not be asked routinely. Most (n=99) who responded yes agreed that it is indicated and relevant for use of clinical risk assessment for recurrence risk, autosomal

recessive disease risk, microarray interpretation, or likelihood of inheriting a familial condition. Many who discussed importance for risk assessment mentioned that it would change testing strategy based on indication and would help inform other family members who may be at risk for a certain condition. Some genetic counselors (n=6) stated that the continuation of asking the question provides additional opportunities to educate on other genetic concepts, not just risk assessment:

> *"Yes - I do think that this can inform our thoughts about inheritance. And we have the opportunity to normalize consanguinity for our families in which cousin-cousin marriages are common. It's also a great way to provide some general genetics education for those who think that consanguineous relationships are automatically "bad"."*

Some genetic counselors who stated yes, also provided a detailed explanation with case scenarios:

> *"Yes, when indicated by clinical presentation (not based on reported ancestry). In several pediatric cancer cases I have been involved in with consanguinity, the parents shared feelings of guilt based in the closeness of their relationship. This has included families where the relationship wasn't known until after the couple got together, cases where cousin marriage and other marriage between blood relatives is more common in the family's culture, and other circumstances. Knowing about the consanguinity helps me address those feelings of guilt and provide context like the fact that most children with recessive conditions have parents who are not related. While this could be brought up on the back end (e.g. once a test identifies a homozygous variant), I think that might seem more*

accusatory. In other words, rather than a question I ask everyone (value neutral), it may appear that I am only asking you this because of your child's test result (could be perceived as a judgement).”

For genetic counselors who said they were undecided, some explained this is due to the advances of technology to identify consanguinity (n=18), and therefore self-reported responses during pre-test counseling from patients may not be necessary to the risk assessment initially, but more important as a follow-up question when indicated through testing results (n=10). However, genetic counselors in both categories provided the importance of pre-testing counseling to state that consanguinity may be revealed on a genetic test result. Genetic counselors in the yes categories added that discussing consanguinity before ordering testing may lessen the stress in discussing the results if consanguinity is revealed. Other genetic counselors in the “no” category stated numerous reasons for not asking about consanguinity. Genetic counselors who work in certain specialties, specifically cancer, neurology, cardiology, and pediatrics, (n=15) indicated that since it does not directly impact their testing strategy or risk assessment counseling, they do not ask about consanguinity regularly. Few individuals (n=6) additionally mentioned that asking the question may unintentionally create a negative impact on the provider patient relationship. One genetic counselor stated,

“ *... I also think asking patients about consanguinity might make them feel judged, when it’s not my intention to judge them. A patient’s belief that they are being judged, whether true or not, can damage the provider-patient relationship and may lead the patient to withhold information.”*

2.4.6 Genetic counselors' personal experiences with consanguinity

A supplemental question asked if genetic counselors have any family or they themselves are part of a consanguineous relationship. Fourteen participants (10%) selected yes and were asked an additional three questions regarding their experiences when discussing consanguinity with patients, any wording or comments from colleagues that have felt judgmental or uncomfortable, and any additional advice for other genetic counselors in the field when discussing this topic in general. Ten genetic counselors provided various experiences. Listed below are quotes from genetic counselors regarding their personal thoughts or feelings that have come up when discussing consanguinity with other genetic colleagues or patients:

1. *I find that some people are ashamed of their consanguinity history, whether it's them and their partner, or their parents, etc. I understand why they may feel that way, and I try to empathize with them and explain why it's helpful for me as the GC to know this information.*
2. *"… This is the first time I've really thought about it in the context of these discussions. I guess I don't see it as a "big deal" unless there is an abusive situation going on. Just like there are some higher genetic risks for pregnant people who are over 35, there are some risks with consanguinity, but you don't see the same stigma for AMA as you do for this."*
3. *"Recently, with a colleague, when I was discussing our recent project about starting a genetic counseling training program and the high demand for such a program in a country with a high consanguinity rate, one GC*

advised me not to add this information in a US-based grant application as people will be turned off by this and may reduce the chances of getting the grant funding."

4. *"I definitely sense the surprise when I disclose this. More with my colleagues than with patients. Patients are often very interested in hearing about this the few times I've disclosed. I think it helps with some of the patients who have very visceral reactions when I try to explain that in part of the world it is a common practice, and it does have some associated increased risks, but it is very possible for cousins to marry and have healthy children. One time a couple blurted out "but you're so beautiful and smart" as a reaction to this disclosure and it felt like it was a genuine response because in their mind all cousin marriages result in disfigured or disabled children"*
5. *"To be honest, I don't often even think of the consanguinity in my family because it's in a third-degree relative pairing. The previous question caused me to consider it. I generally think people in nonconsanguineous relationships don't consider that consanguineous relationships can still be typical and loving."*
6. *"Over the years, I have learned that I should be more comfortable discussing this with my colleagues and found better ways to address the comments that patients might have after asking about consanguinity. I try to think about how we need to ensure that we need to be empathetic and*

learn more about our diverse patient populations to provide them with the best care."

7. *"I feel uncomfortable when other colleagues keep talking about consanguinity as the only reason for genetic disorders, and initiate media campaigns against this social practice especially in our area where there are few numbers of GCs and testing is not widely available, this creates a wave of discrimination against the only marriage system that those simple people are allowed to practice."*
8. *"One of the MFM providers looked at a pedigree I drew for some of my distant relatives who are double first cousins and have two children with severe intellectual disabilities that I was attempted to coordinate testing for internationally and stated 'yikes'."*
9. *"I get more uncomfortable when colleagues insinuate that all Ashkenazi Jewish individuals are "inbred" or consanguineous. There were comments while I was in my grad program from other female GC students about "not dating a Jewish guy" because of the genetic diseases related to being a historically genetically isolated population. It does hurt but I feel like people will perceive me as too sensitive if I say something about it."*
10. *"I had a patient who responded to my question by saying "Oh right, like [my cultural group] - they intermarry right? To keep their riches between them. No, if we were rich maybe we'd have intermarried but haha no, not us" It hurt because there was both a negative cultural connotation and a negative association about consanguinity."*

Genetic counselors with consanguineous family members shared additional advice and perspective for their colleagues. Most responses stated the importance of addressing the topic without blame or judgement, and the first step at achieving this comes from being culturally informed. One genetic counselor suggested,

> *"Visit a local community center or a mosque to see how consanguineous couples are no different than non-consanguineous ones. The cultural and social ties between consanguineous couples strengthens the ties between members of the community. In terms of the number of cases with hereditary disorders, think about all the autosomal recessive cases you have come across where there was no consanguinity (e.g., cystic fibrosis)."*

2.4.7 Recommendations for addressing consanguinity

Genetic counselors were given the opportunity to provide any additional phrases they utilize after patients make a comment or joke (n=61), provide advice for discussing consanguinity (n=69), and any other additional comments regarding this questionnaire did not specifically address (n=53).

For additional wording or phrasing to use, most responses suggest genetic counselors simply state that it is common in many cultures or communities or explain that the information can be helpful to know from a genetics perspective because it may potentially have an impact on risk assessment. Some genetic counselors reported that they do use a phrase like "we ask every patient this question" but indicate that they must be wary of the tone that it is spoken in as that could introduce some stereotypical bias to the question. One counselor stated, "*I hate saying 'we have to ask everyone the question' and I hate that I learned that in school. I haven't used the phrase in years, and I try to*

teach my student to avoid phrasing it like that." A few responses noted that if the ancestry reported were from different regions or countries, then they may respond with "*given your different backgrounds, I assumed that the answer would be no."* Other responses indicated some genetic counselors in a prenatal setting educate on the risk by saying,

> "*It is really common in the population and the risks associated with it are much lower than media taught us. I try to give statistics comparing 2-3% chance of major birth defects/genetic condition for every non-consanguineous pregnancy, versus 4-6% for first cousins, and reflect back that's as least 94% chance of a healthy, unaffected pregnancy."*

Advice for how to discuss consanguinity varied greatly between the responses. Most genetic counselors advise to ask objectively and in a straightforward manner without prefacing the question with additional qualifiers as you would ask any other question during the family history portion. Utilizing neutral language and neutral tone allows for a direct response to the jokes or comments, while educating, normalizing, and maintaining rapport with the patient. Many genetic counselors mentioned that addressing the stigmas or taboos around the question can be helpful depending on how intensely the patient reacts. Providing the explanation in context in advance may assist in reducing the number of comments or jokes made by patients. Many providers stated that it is important to be culturally aware of the norms in different parts of the world and to understand how different degrees of consanguinity relate to genetic risk assessment. A few comments referenced that it is particularly important to continually ask each patient

routinely and as consistently as possible to avoid only asking patients based on ancestry, as that perpetuates stereotypical biases.

2.5 Discussion

This study aimed to gain insight into the experience of genetic counselors working with consanguineous couples in a variety of specialties and to explore the range of comfortability discussing or responding to patients regarding consanguinity in clinical practice. While there are few resources that offer guidance on how to inquire about consanguinity in a clinical session, little to no research has been conducted to assess the experiences of genetic counselors. The main finding of the study highlighted that while most genetic counselors feel extremely comfortable with counseling on consanguinity, there is great variation in how genetic counselors phrase the question and methods in which genetic counselors respond to patient discomfort after the question is presented.

Resources for how to phrase the question to inquire about consanguinity during a family history are noted to be limited; however, there are few utilized throughout genetic counseling training programs that offer suggestions of how to ask the question (Bennett, 2010; Modell & Darr, 2002; Teeuw, 2012). This study showed there are a variety of phrases utilized by genetic counselors in practice to address consanguinity with patients. Most genetic counselors introduced the question of consanguinity with qualifying phrases, but also incorporated direct phrases in the overall question.

While utilizing qualifiers may not cause direct harm to patients, it may perpetuate a certain bias or stereotype of consanguineous couples when initially presenting the question. “We ask everyone” was a common qualifying phrase stated by genetic counselors. While the intention of this phrase is to state that this is a standard question in

family history intake, its usage at the preface of the question may imply that consanguinity is rarely expected between two individuals, but that we are obligated to ask. Adding any qualifier phrase, including but not limited to “any possible way”, “any chance”, or “no chance”, may perpetuate a similar bias. Understanding best practice for genetic counselors to ask this question may provide further utility into patient comfortability of sharing consanguineous status with providers (Thain et al., 2019). Utilizing only direct phrases when prefacing the question may reduce the risk of misunderstanding, confusion, or hurting rapport that has been built throughout the session. Given the multiple perspectives of genetic counselors and experiences with patients, we can infer from this study that the way that a genetic counselor intentionally asks this question is important, creates opportunities to ask without prejudice, and also creates opportunities to educate.

An important finding of this study was that most genetic counselors continue to find utility in asking the question of consanguinity to all patients routinely due to its clinical utility or to address any psychosocial concerns that may arise. Because the practice of consanguinity is a widespread practice in a multitude of regions, cultures, and ethnic backgrounds, genetic counselors cannot inquire about consanguinity solely based on self-reported ancestry or their own perceptions of cultures of where consanguinity is a more common practice (Bennett et al.,2020; Hamamy & Bittles, 2009),. Those who felt undecided or stated that it should not be continuously asked were referencing that the identification of consanguinity no longer impacts clinical decision making in their specialty, but only in the context of test result implications. It is necessary to acknowledge that some genetic counselors are questioning the utility of asking about

consanguinity routinely due to the advancements of genetic testing technology including whole exome sequencing and chromosomal microarray, the newest recommendations for expanded carrier screening for all prenatal couples, and broad-based panel testing regardless of whether there is known consanguinity or not for all patients (Gregg et al, 2021). Changes in attitudes about the utility of this question are not surprising given that each question asked by genetic counselors during a session has a specific purpose for clinical risk assessment.

Genetic counselors were asked to provide comments or jokes they had heard from patients after inquiring about consanguinity within a patient's family. The results showed the overwhelming amount of discomfort from a patient perspective. Patients may be making these comments in response to not fully understanding the relevance of the question during the family history. This study revealed that genetic counselors are highly aware of the common jokes and stigmatizations that are often present in a session. The discomfort that patients express directly influence how genetic counselors respond.

Behaviors exhibited by genetic counselors provided insight into how genetic counselors chose to address the comments made by patients. Almost every participant indicated that genetic counselors would explain the reasoning behind asking the question of consanguinity after a patient responds with a comment or joke. Genetic counselors are uniquely positioned to educate every family and patient about the reason we ask this question. By explaining the purpose of this question, genetic counselors have the opportunity to inform the general public about consanguinity and educate on the misconceptions and stereotypes regarding this common cultural practice. Many providers stated that they have evolved in their practice, and that they no longer use or say many of

these phrases after the first few years practicing. The word choices and tone that genetic counselor use should not show judgment in any way. Couples who are consanguineous are often aware of the cultural stigma that surrounds their relationship and could have had negative experiences with other providers because of their relationship.

Laughing with the patient was reported to occur in half of the responses. While a nervous laugh may be an involuntary reaction to the response of a patient laughing, this continues to perpetuate the known bias against consanguinity. Genetic counselors are to be allies to patients of all backgrounds, ethnicities, and cultures and the response to patient behavior is best represented by how we educate patients on accepting other types of cultural norms. Many of these behaviors are done by genetic counselors regardless of how frequently they have experienced working with couples who are consanguineous. Interestingly, the differences in experience presented in a few of the behaviors were genetic counselors who rarely see consanguinity in practice. Individuals who rarely see consanguineous couples in practice were more likely to not acknowledge the comment, whereas genetic counselors who often see consanguineous couples were responding to the patient by normalizing their reaction of the question being "funny" but then further educating on the reason.

An additional assessment of genetic counselor comfortability was determined by asking about initial responses when consanguinity was defined between a couple. Clinical risk assessment of consanguinity was the most common response by genetic counselors which further supports the continuation of asking this question and its relevance in clinical genetics. Interestingly, many initial responses focused on genetic counselors' personal emotions and cultural implications. An emotional response to the patient

disclosing this information informs that those genetic counselors were aware of the stigmas around consanguinity and were often conscious of their biases as they work to respond to patients in an accepting and non-judgmental manner. For patients who are consanguineous, this is simply a fact about their relationship, and they are typically not the ones who are exhibiting any behaviors or awkwardness to the session.

Normalizing the question was an overall theme present in this study. Many responses included normalizing the question as one of the main goals after a patient makes a comment or joke. By first acknowledging that the question may make patients uncomfortable, genetic counselors are then able to understand why the patient may laugh or make a joke and can therefore respond in a neutral tone and explain the reason for asking. Normalizing the question preemptively may also decrease the worry that asking the question will hurt rapport or perpetuate the fear that asking the question interpreted as judgmental. For couples who are consanguineous, acknowledging their relationship, normalizing, and addressing their emotions, and exploring their understanding of the genetic risks would allow accurate patient education and allows the genetic counselor to explore the patients' attitudes and feelings.

The JEDI initiative of the NSGC is implemented in every practice of genetic counseling. Respect of lifestyle and marriage choices arguably fit into the bigger objective of JEDI. The word choice and actions of genetic counselors when discussing consanguinity, whether conscious or subconscious, could be considered microaggressions and could even be considered a form of genetic discrimination. One example was demonstrated by the initial reactions of discomfort with degree of relation or concern for abuse when a couple states that they are consanguineous. This assumption and belief are

a continuation of the misconception that consanguineous couples are unnatural or forced upon individuals and do not take into consideration the circumstances of individual choices or norms within a certain culture or community.

Genetic counselors who stated to have consanguineous family or are in a consanguineous relationship themselves offer a unique perspective. The overarching theme of their responses were feelings of hurt or being victimized because of comments that were made to them regarding consanguinity in general. Most of the comments stated to this subset of genetic counselors were primarily told by other genetic counseling colleagues or healthcare providers. While this study did not assess the patient perspective, these genetic counselors and consanguineous couples being seen in clinic may feel similarly about these microaggressions. The responses call attention to how common microaggressions happen around the discussion of consanguinity in genetic counseling and in other healthcare professions. The study results reinforce the importance of raising awareness around consanguinity and increasing cultural competency and acceptance.

The experiences shared by currently practicing genetic counselors provides insight for best practices to discuss and explore consanguinity with patients. The initial question can be simply stated in a neutral tone and without the use of qualifying phrases, such as “Are you and your partner’s families related by blood, such as cousins?” or simply “Are your two families related?”. Genetic counselors could benefit from having different ways of asking the question in the event that it needs to be restated or explained. If a patient responds with a joke, comment, or laugh, it is appropriate and within the genetic counselor’s scope of practice to explain the impact of consanguinity on the genetic risk assessment or consider briefly stating that the practice is common in many

communities. A simple explanation to a patient's comment can be "This question is something that we consider in genetics that your other providers may not" or "We see families from a variety of places and cultures where these relationships may be more common." This allows genetic counselors to provide education around the practice of consanguinity, increase cultural competency, and potentially prevent discrimination while simultaneously dismissing a common misconception. However, this study revealed the difficulty of standardization of the wording of this question due to the variability of responses and nuances of each individual's counseling style. One nuance that impacts the standardization is each individual's implicit bias towards consanguinity. An important self-reflection activity would be to acknowledge those biases around consanguinity and assumptions about consanguinity due to ancestry or country of origin.

Clinical supervisors provide an environment to educate students on how to address consanguinity in clinical practice. Clinical supervisors who are working with graduate students should notice and advise on how to best ask this question, by not prefacing the question with any particular phrase and ask it as any other question in the family history. Supervisors should also provide feedback on how genetic counseling students ask this question. Some individuals stated that their graduate school program emphasized this topic during their training. Genetic counseling students are also in a unique position to provide feedback to their supervisors who may not recognize the potential impact of an ill-phrased question or response, and students are encouraged to discuss how the question is addressed.

2.6 Limitations

One limitation of this study includes the small sample size from the multinational perspective of genetic counselors who work or have studied in other countries. The majority of the responses were from genetic counselors who have worked and practiced in the United States of America. However, responses and experiences of genetic counselors working in other countries shared similar experiences and were therefore combined in the overall analysis. Additional limitations include potential confirmatory biases, as only two individuals reviewed the themes and data found from this study.

2.7 Future Directions

The original questionnaire included a question regarding genetic counselor experiences of patients initially denying consanguinity but then confirmed later in the session or during a follow-up appointment. This research is intended to be published as a perspective piece but did not fit into the original question of this particular study.

Further studies need to be conducted on the continuation of asking the question of consanguinity to every patient in every clinical specialty. While the majority stated yes, many individuals were undecided and said that the question should only be asked when relevant, but these responses did not expand on what is considered relevant and what is not. Qualitative interviews rather than free response text may provide better context and allow for further follow-up questions. It would also be informative to gauge what patient feel when asked the question of consanguinity and to further assess the thoughts of couples who are consanguineous about their experiences. This could provide greater insight into what would be best practices for genetic counselors to ensure patient understanding.

CHAPTER 3: CONCLUSION

The multiple perspectives of genetic counselors from a variety of specialties, years of experience, and countries represented indicate the importance of how we address consanguinity in a clinical setting and implications to minimize the stigmatizations and microaggressions surrounding the topic. The question of consanguinity is unique in that genetic counselors are usually the only healthcare provider to ask this question because it does have clinical implications for the standard practice of care. While it is just one question in a multitude of important questions during a family history, it is one of the few that elicits a unique response from patients. Genetic counselors are therefore in a position to not only provide genetic education, but cultural education as well. Being aware of potential microaggressions presented by patients or other genetics professionals allows us to address these comments directly by providing accurate information. By asking the question in a culturally sensitive manner and providing a non-judgmental and professional assessment, genetic counselors alleviate the stigmatization of consanguinity and promote public awareness around a common cultural practice. This study provides evidence of the importance that all healthcare providers, particularly genetic counselors, continue to reflect on their personal thoughts, feelings, and biases related to consanguinity.

REFERENCES

Ahmed, M. (2013). Communicating with clients from the Pakistani Muslim community. In J. Wiggins & A. Middleton (Eds.) *Getting the Message Across: Communicating with Diverse Populations in Clinical Genetics* (pp.91-11). Oxford University Press.

Alnaqeb, D., Hamamy, H., Youssef, A. M., & Al-Rubeaan, K. (2018). Assessment of knowledge, attitude, and practice toward consanguineous marriages among a cohort of multiethnic health care providers in Saudia Arabia. *Journal of Biosocial Science, 50*(1), 1-18.

Alwan, A. A. & Modell, B. Community Control of Genetic and Congenital Disorders. EMRO Technical Publication Series 24 (WHO Regional Office for the Eastern Mediterranean Region, Egypt, 1997)

Barbour, B., & Salameh, P. (2009). Consanguinity in Lebanon: prevalence, distributions and determinants. *Journal of Biosocial Science*, *41*(4), 505–517.

Bennett, R.L. (2010) *The Practical Guide to the Genetic Family History.* Wiley

Blackwell. Bennett, R. L., Malleda, N. R., Byers, P. H., Steiner, R. D., & Barr, K. M. (2021). Genetic counseling and screening of consanguineous couples and their offspring practice resource: Focused revision. *Journal of Genetic Counseling*, 30(5), 1354–1357. https://doi.org/10.1002/jgc4.1477

Bennett, R. L., Motulsky, A. G., Bittles, A., Hudgins, L., Uhrich, S., Doyle, D. L., Silvey,

K., Scott, C. R., Cheng, E., McGillivray, B., Steiner, R. D., & Olson, D. (2002). Genetic counseling and screening of consanguineous couples and their offspring: Recommendations of the National Society of Genetic Counselors. Journal of Genetic Counseling, 11(2), 97–119

Bhinder, M. A., Sadia, H., Mahmood, N., Qasim, M., Hussain, Z., Rashid, M. M., Zahoor, M. Y., Bhatti, R., Shehzad, W., Waryah, A. M., & Jahan, S. (2019). Consanguinity: A blessing or menace at population level?. *Annals of Human Genetics*, *83*(4), 214–219.

Bittles A. (2001). Consanguinity and its relevance to clinical genetics. *Clinical Genetics*, 60(2), 89–98.

Bittles A. H. (2008). A community genetics perspective on consanguineous marriage. *Community Genetics*, *11*(6), 324–330.

Bittles, A. H. (2012) *Consanguinity in Context.* Cambridge University Press.

Bittles, A. H. & Black, M.L. (2010) Consanguineous marriage and human evolution. *Annual Review of Anthropology.* 39,193-207.

Cupp, M. A., Adams, M., Heys, M., Lakhanpaul, M., Alexander, E. C., Milner, Y., Huq, T., Peachey, M., Shah, L., Mirza, I. S., & Manikam, L. (2020). Exploring perceptions of consanguineous unions with women from an East London community: Analysis of discussion groups. *Journal of Community Genetics*, *11*(2), 225–234.

Fathzadeh, M., Babaie Bigi, M. A., Bazrgar, M., Yavarian, M., Tabatabaee, H. R., & Akrami,S. M. (2008). Genetic counseling in southern Iran: Consanguinity and reason for referral. *Journal of Genetic Counseling*, *17*(5), 472–479.

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., & ACMG Professional Practice and Guidelines Committee (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.

Grote, L., Myers, M., Lovell, A., Saal, H., & Sund, K. L. (2014). Variable approaches to genetic counseling for microarray regions of homozygosity associated with parental relatedness. *American Journal of Medical Genetics. Part A*, *164A*(1)

Guttmacher, A. E., Porteous, M. E., & McInerney, J. D. (2007). Educating healthcare professionals about genetics and genomics. *Nature Reviews. Genetics*, *8*(2), 151–157.

Hamamy, H., & Bittles, A. H. (2009). Genetic clinics in Arab communities: Meeting individual, family, and community needs. *Public Health Genomics, 12*(1), 30–40.

Hamamy, H., Antonarakis, S. E., Cavalli-Sforza, L. L., Temtamy, S., Romeo, G., Kate, L. P.,Bennett, R. L., Shaw, A., Megarbane, A., van Duijn, C., Bathija, H., Fokstuen, S., Engel, E., Zlotogora, J., Dermitzakis, E., Bottani, A., Dahoun, S., Morris, M. A., Arsenault, S., … Bittles, A. H. (2011). Consanguineous marriages, pearls and perils: Geneva International Consanguinity Workshop Report. *Genetics in Medicine, 13*(9), 841–847.

Hoskovec, J. M., Bennett, R. L., Carey, M. E., DaVanzo, J. E., Dougherty, M., Hahn, S. E., LeRoy, B. S., O'Neal, S., Richardson, J. G., & Wicklund, C. A. (2018). Projecting the supply and demand for certified genetic counselors: A workforce study. *Journal of Genetic Counseling*, *27*(1), 16–20.

Julian-Reynier, C., Nippert, I., Calefato, J. M., Harris, H., Kristoffersson, U., Schmidtke,

J., Ten Kate, L., Anionwu, E., Benjamin, C., Challen, K., Plass, A. M., & Harris, R. (2008). Genetics in clinical practice: general practitioners' educational priorities in European countries. *Genetics in Medicine*, *10*(2), 107–113.

Modell, B., & Darr, A. (2002). Science and society: genetic counseling and customary consanguineous marriage. *Nature Reviews: Genetics*, *3*(3), 225–229.

Oniya, O., Neves, K., Ahmed, B., & Konje, J. C. (2019). A review of the reproductive consequences of consanguinity. *European Journal of Obstetrics, Gynecology, and Reproductive Biology*, *232*, 87–96.

Powis, Z., Farwell, K. D., Alamillo, C. L., & Tang, S. (2016). Diagnostic exome sequencing for patients with a family history of consanguinity: Over 38% of positive results are not autosomal recessive patterns. *Journal of Human Genetics*, 61(2), 173-175.

Rehder, C. W., David, K. L., Hirsch, B., Toriello, H. V., Wilson, C. M., & Kearney, H. M. (2013). American College of Medical Genetics and Genomics: Standards and guidelines for documenting suspected consanguinity as an incidental finding of genomic testing. *Genetics in medicine*, *15*(2), 150–152.

Shao, L., Akkari, Y., Cooley, L. D., Miller, D. T., Seifert, B. A., Wolff, D. J., Mikhail, F. M., & ACMG Laboratory Quality Assurance Committee (2021). Chromosomal microarray analysis, including constitutional and neoplastic disease applications, 2021 revision: A technical standard of the American College of Medical Genetics and Genomics (ACMG). *Genetics in Medicine: 23*(10), 1818–1829.

Shaw A, & Hurst JA. (2009). "I don't see any point in telling them": attitudes to sharing

genetic information in the family and carrier testing of relatives among British Pakistani adults referred to a genetics clinic. *Ethnicity & Health*, 14(2), 205–224.

Sund, K. L., & Rehder, C. W. (2014). Detection and reporting of homozygosity associated with consanguinity in the clinical laboratory. *Human Heredity*, *77*(1-4), 217–224.

Tadmouri, G. O., Nair, P., Obeid, T., Al Ali, M. T., Al Khaja, N., & Hamamy, H. A. (2009). Consanguinity and reproductive health among Arabs. *Reproductive Health*, *6*, 17.

Teeuw, M. E., Hagelaar, A., ten Kate, L. P., Cornel, M. C., & Henneman, L. (2012). Challenges in the care for consanguineous couples: An exploratory interview study among general practitioners and midwives. *BMC Family Practice*, *13*, 105.

Thain, E., Shuman, C., Miller, K., Sappleton, K., Myles-Reid, D., Chitayat, D., & Gibbons, C. (2019). Prenatal and preconception genetic counseling for consanguinity: Consanguineous couples' expectations, experiences, and perspectives. *Journal of Genetic Counseling*, *28*(5), 982–992.

Yavarna, T., Al-dewik, N., Al-mureikhi, M., Ali, R., Al-mesaifri, F., Mahmoud, L., Shahbeck, N., Lakhani, S., Almulla, M., Nawaz, Z., Vitazka, P., Alkuraya, F. S., & Ben-omran, T. (2015). High diagnostic yield of clinical exome sequencing in Middle Eastern patients with Mendelian disorders. Human Genetics, 134(9), 967-980.

APPENDIX A: PARTICIPANT RECRUITMENT INVITATION

Genetic counseling standard practice includes genetic counselors asking about consanguinity as they obtain a genetic pedigree during a genetic counseling encounter. However, negative attitudes and stigma potentially due to cultural differences and norms at a population level may interplay with questions, answers, and discussion around consanguinity. Through lived and shared experiences, we believe that the phrasing and questioning regarding consanguinity in a genetic counseling session may influence attitudes, stereotypes, and misconceptions surrounding consanguineous relationships.

The study will be collecting data via an online survey. The survey should take 10-15 minutes to complete. We invite genetic counselors working in any country, not limited to the United States and Canada, to participate in this survey and share experiences of discussing consanguinity in clinical practice. You are eligible to participate in this study if you are:

1. A genetic counselor who has worked in a full or partial patient-facing setting within the last five years
2. Previously worked with consanguineous couples and can recall experiences
3. At least one year of clinical work experience.

Those interested in participating can access the survey at https://uofsc.co1.qualtrics.com/jfe/form/SV_4ZrdZvJe1rSh5Ou

This study is being conducted by Romy Fawaz, a genetic counseling trainee at the Master of Genetic Counseling Program at the University of South Carolina. If you have any questions about participating in this research project, please feel free to email Romy Fawaz (Principal Investigator) at romy.fawaz@uscmed.sc.edu or Janice Edwards, MS, CGC (Thesis Advisor) at janice.edwards@uscmed.sc.edu.

APPENDIX B : PARTICIPANT QUESTIONNAIRE

Start of Block: Introduction

Q-Intro Thank you for considering to participate in the study of **Exploring Genetic Counselors' Experiences, Language, and Discussion of Consanguinity in Practice: A Multinational Perspective.** This questionnaire will contain a series of multiple-choice, multi-select, and free response questions attempting to understand how genetic counselors experiences when inquiring and discussing consanguinity with all patients as they obtain a family history. Your participation is completely voluntary and you may choose to skip questions if you prefer not to answer. All responses gathered from the survey will be kept anonymous and confidential. The results of this study might be published or presented at academic meetings; however, participants will not be identified. If you are willing to participate in this study, please click the "next" button below. If not, please exit the browser.

End of Block: Introduction

Start of Block: Inclusion Criteria

QA How often do you encounter consanguineous couples in your practice?

- ○ Never (1)
- ○ Rarely (2)
- ○ Sometimes (3)
- ○ Often (4)

Skip To: End of Survey If QA = Never

QB Have you worked in a patient-facing/clinical setting within the last 5 years?

- Yes (1)
- No (2)

Skip To: End of Survey If QB = No

End of Block: Inclusion Criteria

Start of Block: Intro Questions

Q1 When obtaining a family history, do you ask every patient about consanguinity?

- Yes (1)
- No (2)

Display This Question:
If Q1 = No

Q2 Please provide a brief explanation why you do not ask every patient?

_____

Q3 In general, which do you inquire about first: ancestry or consanguinity?

- Ancestry (1)
- Consanguinity (2)
- Depends (3)

Display This Question:
If Q3 = Depends

Q4 Please explain the circumstances that it would depend?

_____

Q5 How comfortable are you asking the question of consanguinity?

- ○ Extremely comfortable (1)
- ○ Somewhat comfortable (2)
- ○ Somewhat uncomfortable (3)
- ○ Extremely uncomfortable (4)

End of Block: Intro Questions

Start of Block: genetic counselors ask the question of consanguinity

Q. ***We are interested to know how genetic counselors ask the question and discuss consanguinity during a family history with patients.***

Q6 To the best of your ability please provide the exact wording you use to ask about consanguinity.

_____

Q7 What are your initial thoughts/feelings/reactions when a patient answers "yes" to the question of consanguinity and why?

_____

Page Break

Q8 In your personal experience, how often do patients laugh when asked about consanguinity?

- ○ Always (1)
- ○ Often (2)
- ○ Occasionally (3)
- ○ Rarely (4)

Q9 Have patients ever made jokes/comments after you asked the question of consanguinity?

- ○ Yes (1)
- ○ No (2)

Display This Question:

If Q9 = Yes

Q10 If yes, please provide some of the comments that you have heard.

_____

Page Break

Q11 Have you experienced a situation that a patient initially denied consanguinity but then confirmed consanguinity in the family later in the session or in a follow up appointment?

- ○ Yes (1) _____
- ○ No (2)

Q12 In your opinion, should genetic counselors continue asking about consanguinity? Why or why not?

_____

End of Block: genetic counselors ask the question of consanguinity

Start of Block: GC responses to consanguinity

Q13 We are interested in how genetic counselors respond to patient comments after being asked the question of consanguinity. Please select “yes” or “no” if you have said or done any of the following. Have you ever….

	Yes (1)	No (2)
Addressed the patient about the comment directly? (1)	○	○
Apologized for asking the question or any discomfort the question brought? (2)	○	○
Asked directly why the patient is laughing? (3)	○	○
Did not acknowledge the comment and moved on? (4)	○	○
Explained the reasoning behind why we ask the question of consanguinity? (5)	○	○
Laughed with the patient? (6)	○	○
Made a joke about being from a small town or specific state (7)	○	○
Said something like “I know it's a strange question”? (8)	○	○
Said “We have to ask everyone that question”? (9)	○	○
Said "Yes, I know its a funny question" (10)	○	○

Q14 Please provide any additional wording, phrases, or responses that you have used in response to patients if not represented in the question above.

_____

End of Block: GC responses to consanguinity

Start of Block: Additional Comments

Q15 Many counselors have worked in countries where consanguinity is seen at higher prevalence or have had more experience counseling consanguineous patients. If applicable, what advice would you offer to other counselors on discussing consanguinity?

_____

Q16 Please provide any additional comments or feelings that have not been addressed in this questionnaire.

_____

Page Break

Q17 Do you or anyone in your family share a consanguineous relationship?

- ○ Yes (1)
- ○ No (2)

Skip To: End of Block If Q17 = No

Display This Question:
If Q17 = Yes

Q18 ***Your personal or familial experience could help inform other genetic counselors.***

Q19 To the best of your ability, please describe any thoughts/feelings/discomforts that may have come up when discussing consanguinity with other genetic colleagues or patients.

_____

Q20 Has any particular wording or comment by a colleague or patient made you feel judged, discriminated against, or uncomfortable due to your personal relationship or familial relationships? Please explain.

_____

Q21 What advice would you give to other genetic counselors to be culturally sensitive when talking with patients about consanguinity? Please provide any additional thoughts on this that was not represented in this questionnaire.

_____

End of Block: Additional Comments

Start of Block: Demographics

Q22 From which country did you graduate with your degree of genetic counseling?

_____

Q23 What country do you work in currently?

_____

Q24 If applicable, please list all countries where you have previously worked.

_____

Q25 How many years have you been a practicing genetic counselor?

_____

Q26 What specialty of genetic counseling do you currently practice? (*Select all that apply)*

- ☐ ART/Preconception (1)
- ☐ Cancer (2)
- ☐ Laboratory/Industry (3)
- ☐ Pediatric (4)
- ☐ Prenatal (5)
- ☐ Other (6) _____

Q27 What is your race/ethnicity? *(Select all that apply)*

- ☐ African American/Black (1)
- ☐ East Asian/Southeast Asian (2)
- ☐ Latinx/Hispanic (3)
- ☐ Middle Eastern/West Asian (4)
- ☐ Native American/Alaska Native (5)
- ☐ Native Hawaiian/Pacific Islander (6)
- ☐ North African (7)
- ☐ South Asian (8)
- ☐ White (9)
- ☐ Other (10) _____

End of Block: Demographics