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Self-reported patient ethnicity in genetic counseling practice: a closer look at its current role
and room for improvement

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May 2017

Abstract

Despite the importance of patient ethnicity in clinical genetics, its usage in genetic counseling has not been characterized. This study looked at attitudes of genetic counselors (GCs) towards the role of patient self-reported ethnicity and its incorporation into their practice, specifically related to carrier screening. 475 GCs were recruited through the National Society of Genetic Counselors Listserv. Respondents answered an online survey consisting of qualitative and quantitative questions. Questions addressed how patient ethnicity is elicited and used in clinical practice. Case studies involving patients with varying ethnicities were presented for evaluation. Participants' attitudes towards the use of ethnicity in clinical practice were evaluated before and after reviewing data showing patient self-reported ethnicity is not always a good proxy for genetic ancestry. We found that 96% of respondents elicited patient ethnicity information during the family history. Terms like "comes from originally" and "ancestry" were most often used (66% and 47% respectively), possibly to better inform assessment of disease or carrier risk. In response to the case studies, many participants asked the same questions regardless of patient ethnicity. Post-data review participants did not think patient ethnicity was as good a proxy for genetic ancestry as they had prior ($p < .001$). They also thought it was less useful for clinical risk assessment ($p < .001$), but did still have some clinical utility. Overall, surveyed GCs showed an awareness of the limitations of patient reported ethnicity but still found clinical utility in obtaining the information. This may be for residual risk calculation, determination of which screening to offer when insurance coverage is not available, or risk assessment when one partner is unavailable for testing. Future research is needed to understand these reasons. GCs may need to reconsider the role of ethnicity in their practice given its limitations and increased availability of expanded carrier screening.

Keywords: ethnicity, race, ancestry, genetic counseling, carrier screening, expanded carrier screening, panethnic carrier screening, self-reported ethnicity

Introduction

The concept of ethnicity is one with which the field of clinical genetics is intimately involved. Guidelines by professional bodies such as the American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics and Genomics (ACMG) recommend ethnicity-specific carrier screening for most tested conditions (Burke, Tarini, Press, & Evans, 2011). The logic behind such recommendations is well-understood as specific genetic conditions are more prevalent in certain groups as a result of evolutionary

forces such as positive selection and founder effects. For example, sickle cell anemia-causing alleles are more commonly seen in individuals of sub-Saharan African descent because of the alleles' protective effects against malaria (Rees, Williams, & Gladwin, 2010). Another example is the high prevalence of conditions such as Tay-Sachs and hereditary breast and ovarian cancer in the Ashkenazi Jewish population due to specific founder mutations (Rubinstein, 2004; Shi et al., 2017). Additionally, a number of other genetic conditions such as cystic fibrosis (CF) and familial Mediterranean fever are known to cluster in certain populations (Cutting et al., 1992; Yepiskoposyan & Harutyunyan, 2007). These examples show the importance of using information about patient genetic ancestry as a starting point in clinical genetics practice. In the past, knowledge about patient ancestry allowed clinicians to narrow down potential patient diagnoses, to better focus their resources in the face of high testing costs, and to give better residual risk estimates.

It is thought that information about patient ancestry is difficult to capture in the absence of molecular techniques (Mersha & Abebe, 2015). Because of this, measures such as race and self-reported ethnicity are commonly used as proxies for genetic ancestry. Race is a construct based on phenotypic differences between groups such as skin colour and hair texture whereas ethnicity is predicated on shared culture including language, diet, customs, and mythology (Race, Ethnicity, and Genetics Working Group, Ethnicity, 2005). While both measures are used as indicators of genetic ancestry, it is well documented in the literature that neither metric is completely concordant with actual patient ancestry (Mersha & Abebe, 2015).

In general, broad race/ethnicity categories are used in medical research with the United States Census categories of race being the most typically used. The first concrete classification of races was outlined by Linnaeus as *Americanus rubescus* (red), *Europaeus albus* (white), *Asiaticus luridus* (yellow), and *Afer niger* (black) (Hunt & Megyesi, 2008). Since then, racial

categories have remained similar in that they are based off of supposedly endogamously mating continental groups. The census in the U.S. uses five categories to identify race/ethnicity: American Indian, Asian/Pacific Islander, Black (not Hispanic), White (not Hispanic), or Hispanic . According to Hunt and Megyesi (2008), these categories were created under political and administrative contexts and were not designed to be transposed to scientific research, much less to indicate groups of genetically similar people. Most researchers simply follow whichever broad categories their institutions have in place, if not using the U.S. Census categories, as an ability to compare work to previous studies and be compared to in future studies is incredibly valuable to a researcher (Hunt & Megyesi, 2008). However, it is unknown whether more specific categories will lead to more accurate information about a patient's background. In general, while self-identified ethnicity can give information regarding genetic ancestry (such as continental group or super-population), more specific details may not be brought to light (Banda et al., 2015; Lee, Teitelbaum, Wolff, Wetmur, & Chen, 2010; Smith et al., 2014; Yaeger et al., 2008).

Recombine, a genetic testing company, has investigated the discrepancy between what genetic counseling patients report as their ethnicity and their actual genetic ancestry, determined through the use of ancestry informative markers (AIMs) (Shraga et al., n.d.). In this study, 4,466 patients were chosen from Recombine's CarrierMap patient pool. Ethnicity was self-reported by patients on a form with twelve categories: African, Jewish, Native American, East Asian, Latin American, South Asian, European, Mediterranean, Southeast Asian, French Canadian, Middle Eastern, and Other – which includes a write-in area. The self-reported ethnicity was compared against the ethnicity recorded by the genetic counselor while taking a family history and also compared to their ancestry as determined via AIMs (Shraga et al., 2017). Aside from this research, there has been no investigation into the usage of ethnicity

and race in genetic counseling clinical practice.

The results of this study show categories such as European and East Asian have high concordance between the sources of information about patient ethnicity: self-reported, genetic counselor recorded, and algorithm-derived ancestry. The results also show some other geographic categories invite discrepancies. The confusion between Southeast Asian and South Asian is significant and the overlapping of the Mediterranean category with others causes confusion as well. Some populations, such as Latin Americans, have a high degree of admixture and range from 1% to 96% of a European component, according to the algorithm (Shraga et al., 2017).

These discrepancies are important to consider because the field of genetic counseling is one deeply connected with carrier screening. Often occurring preconceptionally or prenatally, carrier screening is testing that determines if an individual is a carrier for an inherited genetic condition. These programs have been in existence in the United States since the early 1970s and have historically been ethnicity-specific (Burke, Tarini, Press, & Evans, 2011). In recent years, recommendations by professional bodies such as the ACMG and ACOG have stated that screening for CF and spinal muscular atrophy (SMA) should occur on the basis of patient ethnicity or family history. In recent years, both organizations have made recommendations regarding pan-ethnic carrier screening for CF and SMA. However, there is some discordance between the guidelines, with ACOG recommending pan-ethnic screening for CF while ACMG still recommends carrier screening on the basis of ethnicity and family history (ACOG Committee on Genetics, 2011).

Expanded carrier screening (ECS) is “the practice of screening all individuals for dozens to hundreds of diseases, some with lower frequencies or severity grades, typically without tailoring to a person’s reported ethnicity” (Lazarin & Haque, 2016). ECS has been

commercially available since 2009 and is gaining increasing acceptance among genetic counselors (Lazarin, Detweiler, Nazareth, & Ashkinadze, 2016). Both ACOG and ACMG, in conjunction with the National Society of Genetic Counselors (NSGC), the Society for Maternal Fetal Medicine (SMFM), and Perinatal Quality Foundation (PQF), have released a joint statement to act as a guide for clinicians and laboratories (Edwards et al., 2015). While this paper clearly lays out how ECS should be used, neither ACOG nor ACMG endorse ECS here or elsewhere. Despite this, the ethnicity-blind practice of ECS is growing in popularity in clinical genetic

The inconsistencies in the practice guidelines put out by professional bodies along with the increasing prevalence of ECS in clinical genetics, suggests that how patient ethnicity information is dealt with in clinical genetics is not standardized and is in flux. Despite this, there remains a dearth of studies looking at the present-day incorporation of ethnicity in genetic counseling. While the role of reported ethnicity and race has been investigated in other domains of biomedical research, there has been no such study in the realm of genetic counseling. This study aims to fill this void and take a deeper look at what ethnicity and race mean in genetic counseling and how these concepts affect today's clinical practice.

Additionally, this study looks at how molecularly derived data about patient ethnicity affects genetic counselors' opinions and attitudes about the use of ethnicity in their clinical practice.

Methods

Participants

An anonymous and voluntary online survey was used to characterize how self-reported patient ethnicity is currently used in clinical practice and how this might change. Exemption from Sarah Lawrence College's Institutional Review Board was received on December 16, 2016. A link to the survey was distributed by email on January 17, 2017 to the National

Society of Genetic Counselors (NSGC) listserv and remained open for a month, hosted on SurveyMonkey. One reminder email was sent out on January 31, 2017. At the end of the survey, participants were given the option to enter their emails into a raffle for five \$100 gift cards to Amazon.com. Participants in this study were practicing genetic counselors who currently see patients or have seen patients in the past. Participants gave informed consent prior to starting the survey.

Study design and procedures

Both quantitative and qualitative questions were asked to ascertain participant attitudes towards the use of self-reported patient ethnicity and how this affects current clinical practice. Both personal and professional demographic information was collected from participants. Case studies were presented for genetic counselors to evaluate and determine which follow-up questions related to ethnicity they would ask in each scenario. Lastly, the attitudes of participants towards the use of self-reported ethnicity in clinical practice were evaluated before and after reviewing the data from Recombine's research. This data (Figures 1, 2 and 3) show the discrepancies between the self-reported ethnicity and genetic ancestry of genetic counseling patients.

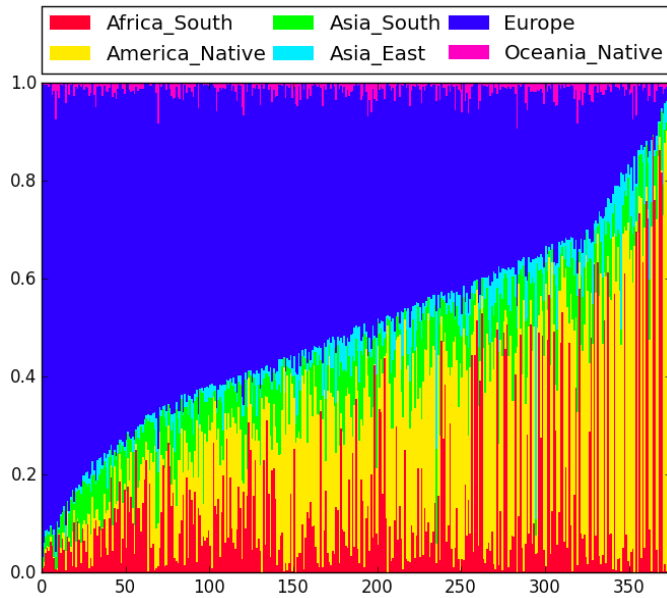


Figure 1: Patients represented here selected “Latin American” ethnicity on test requisition forms. Each patient is represented as a thin vertical line, where each color shows the proportion of ancestry predicted from each continental group. Analysis showed high levels of admixture; samples ranged from having 96% European component to only having a 0.01% European component.

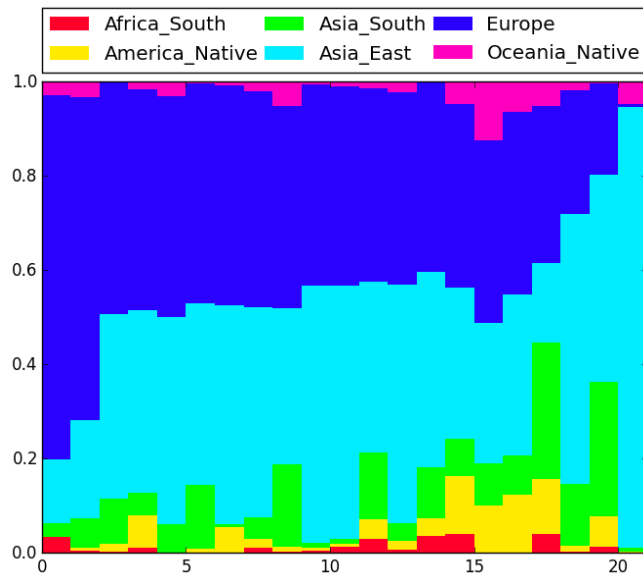


Figure 2: Patients represented here selected both “East Asian” and “European” ethnicity of test requisition forms. Each patient is represented as a thin vertical line, where each color shows the proportion of ancestry predicted from each continental group. Analysis predicted that the actual East Asian component ranged from 13% to 94%. That is, of the patients who reported having both East Asian and European ethnicity, some were predicted to be almost fully East Asian while others were predicted to have a much larger European component.

Sickle Cell Anemia				
	carrier rate < threshold	carrier rate > threshold	p-value	African Ancestry Threshold (80th percentile)
African	6.760%	15.741%	0.00605	93.439%
Latin American	1.261%	4.587%	0.00372	18.677%
Cystic Fibrosis				
	carrier rate < threshold	carrier rate > threshold	p-value	European Ancestry Threshold (80th percentile)
African	1.631%	6.482%	0.01095	19.928%
Latin American	1.606%	3.670%	0.06128	68.715%

Figure 3: Differences in carrier rates of sickle cell anemia and cystic fibrosis by proportion of African and European ancestry among self-reported Africans and Latin Americans. For each group, an ancestry threshold was chosen by computing the 80th percentile of ancestry proportion. For example the 80th percentile of African ancestry among Latin Americans is 18.68%. Thus, the carrier rate of sickle cell anemia of Latin Americans below this 80th percentile threshold is 1.261% and the carrier rate above it is 4.587%.

Statistical Analysis of Quantitative Data

It was not mandatory for participants to complete all 28 survey questions for their responses to be included in analysis. Data analysis was performed using SPSS. Chi square tests were used to determine whether the clinical and personal demographics of the participants are consistent with the demographics of the NSGC as collected through the NSGC's 2016 Professional Status Survey (PSS). A paired t-test and a Chi-squared test were performed to determine the difference between participants' opinions about ethnicity before and after viewing the data from Recombine.

Qualitative analysis

Several questions included an open-ended or write-in component. Qualitative analysis was performed by the two authors by open-coding the responses into major themes. Each author coded four randomly chosen questions with the other author reviewing their completed coding. All discrepancies were discussed and reconciled.

Results

Clinical and personal demographics

The survey received 480 total responses. Five participants were excluded from the study, two of which had never seen patients before with the remaining three self-identifying as students. The clinical and professional demographics of the participants were compared to the NSGC's 2016 PSS and were found to have varied in the participants' ethnicity, age and specialty. There was no difference in the gender/sex of the participants and the regions in which they practiced.

Participants' primary areas of practice included cancer (45.26%), prenatal (33.26%) and pediatric genetics (24.84%). 49.31% of participants currently practice in a non-prenatal specialty, and have practiced in prenatal in the past. This distribution of specialties differed from the PSS (Chi-square (20) = 435.35, $p < .001$). Other significant differences included age (Chi-square (6) = 56.27, $p < .001$) and years of practice (Chi-square (3) = 65.10, $p < .001$) with the study sample being slightly younger and having less experience. While 9.0% of the PSS sample identified as non-white, 14.6% of the study sample identified as non-white. The proportion of those identifying as Hispanic/Latino in the PSS and in this study were similar (1.8% versus 2.2%). Overall the differences between the PSS and our study sample in terms of ethnicity were significant (Chi-square (2) = 12.80, $p < .01$). There were no differences between the samples in terms of sex (Chi-square (1) = 1.20, n.s.) and regions practiced (Chi-square (5) = 3.33, n.s.).

Ethnicity in clinical practice

Participants were asked when they elicit information about patient ethnicity. The distribution of responses is listed in Table 1 (N=454).

Table 1: Methods used by participants to elicit information about patient ethnicity

Source of information	N (%)
Used intake form	13 (2.9%)
Asked patient about ethnicity	226 (49.8%)
Visually assessed patient and asked about ethnicity	26 (5.7%)
Used intake form and asked about ethnicity	115 (25.3%)
Used intake form, asked about ethnicity, and visually assessed the patient	74 (16.3%)
Used information from electronic medical record	4 (0.9%)

Participants were asked about the wording they use to discuss patient ethnicity and were instructed to write in the question or statement as they would ask it of a patient. The 448 responses were broken down into different elements with multiple elements potentially being present within one response. The frequencies of occurrence of these different elements in the participant responses are summarized in Table 2 along with examples.

Table 2: Summary of elements in participant responses to question “What wording do you use when you ask about patient ethnicity? Please write the question/statement below, as you would ask it of a patient.”.

Elements	N (%)	Examples (relevant element italicized)
Comes from originally	294 (65.63%)	“If you had to say <i>where</i> your family/ ancestors <i>come from</i> , <i>what country or countries</i> would you say?” “ <i>Where did your ancestors come from before coming to USA?</i> For example some people would say Irish, Dominican, etc....?”
Ancestry/ancestors	209 (46.65%)	“Regarding ethnicity, what country do your <i>ancestors</i> come from as far back as you know, ie: German, Ireland, Italy, etc?”
Ethnic background	108 (24.11%)	“What would you say is your <i>ethnic background</i> ?”
Ethnicity	92 (20.54%)	“How would you describe your <i>ethnicity</i> ?”
Example of ethnicity	86 (19.20%)	“Do you know your family's ethnic background, <i>like French, Spanish, English</i> ?”
Mother/father	65 (14.51%)	“Do you know your <i>mother's/father's</i> ancestry? Did your <i>mother/father</i> ever mention what countries her/his family was from before they were in the US?”

Tailoring question to participant	21 (4.69%)	"What would you say your ancestry is on your mother's side?" *visually assess patient to provide examples most relevant to them* "For example, Irish, German, ...?" After taking one side of family history, I typically ask "Do you know where your family is from before America?" <i>If they say no, and they look white, I might ask do you typically consider yourself to be caucasian or white? If they say no and they look black I might ask if they consider themselves to be african american or if they are aware of any other background in their family. If I need to get more specific I would probably ask them what race they identify with</i>
Other term	17 (3.79%)	"Where is your family from, <i>ethnically or historically?</i> " "Can you describe your <i>nationality</i> on your mother's side and your father's side of the family?"
Clarification	15 (3.35%)	"What is your ethnic background or ancestry?" <i>If that is not understood: "what is your country of origin or your family's country of origin?"</i> "What would you consider your ethnicity to be?" <i>Follow up if confused: "Some people might say they are white while others may say they are German."</i>
Race	14 (3.13%)	"What do you consider your <i>race</i> or your ethnicity to be?"

The majority of participants (92.72%, n=420) do not use pre-selected categories for indicating patient ethnicity. A number of participants used an intake form with pre-selected categories as well as asking patients about their ethnicity.

Three genetic counseling cases were presented to the participants. Each case was a preconception genetic counseling consultation with the ethnicities of the patients varying by case. Participants were asked to choose all follow-up questions that applied for each respective scenario as displayed in Tables 3, 4, and 5.

Table 3: Follow-up questions asked in a preconception genetics consultation in which both members of the couple identified their ethnicity as Vietnamese

Follow-up question	N (%)
I would not ask any follow-up questions	72 (16.00%)
I would ask them what general region of Vietnam they are from	70 (15.56%)

I would ask them the name of their hometown	18 (4.00%)
I would ask them if they had any Jewish ancestry	186 (41.33%)
I would ask the couple if they are consanguineous	404 (89.78%)

Table 4: Follow-up questions asked in a preconception genetics consultation in which one member of the couple identified as Dominican and the other member identified as Caucasian from Maine.

Follow-up question	N (%)
I would not ask them any follow-up questions	7 (1.56%)
I would ask the Caucasian individual if they know specifically which country their family originates from	405 (90.00%)
I would ask the Caucasian individual if they have any French Canadian ancestry	115 (25.56%)
I would ask the Caucasian individual if they have any Acadian ancestry	27 (6.00%)
I would ask the Dominican individual about where in the Dominican Republic their family is from.	30 (6.67%)
I would ask the Dominican individual if they have any Jewish ancestry	257 (57.11%)
I would ask the Caucasian individual if they have any Jewish ancestry	330 (73.33%)
I would ask the couple if they are consanguineous	343 (76.22%)

Table 5: Follow-up questions asked in a preconception genetics consultation in which one member of the couple identified as Portuguese with the other member saying that she was a “descendent of the Vikings”.

Follow-up question	N (%)
I would not ask them any follow-up questions	13 (2.90%)
I would ask if the Portuguese individual has Azorean ancestry	23 (5.12%)
I would ask the Portuguese individual if they have any Jewish ancestry	289 (64.37%)
I would ask the “Viking” individual if they have any Jewish ancestry	282 (62.81%)
I would ask the “Viking” individual if they know specifically which country their family originates from	402 (89.53%)

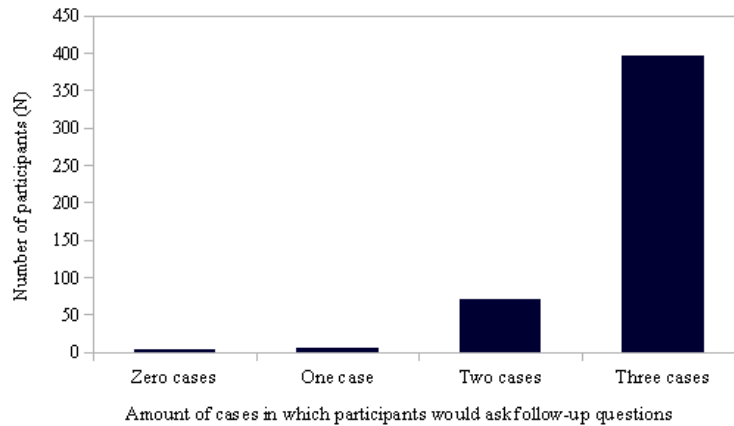
I would ask the “Viking” individual if they have any Icelandic ancestry	73 (16.26%)
I would ask the couple if they are consanguineous	340 (75.72%)

Some interesting findings emerged when comparing the follow-up questions that participants would ask for each case. Data about the number of participants who would ask at least one follow-up question in all three cases, in two cases, in one case, and in none of the cases is presented in Graph 1. Of the 70 respondents who would ask follow-up questions in all but one of the cases, the majority (90%) did not ask in the case where the couple was Vietnamese. Four (6%) did not ask in the Portuguese/Viking ancestry case and three (4%) in the Dominican/Caucasian case. All five respondents who would not ask follow-up questions in two of three cases said they would not ask in the Vietnamese case and in the Portuguese/Viking case.

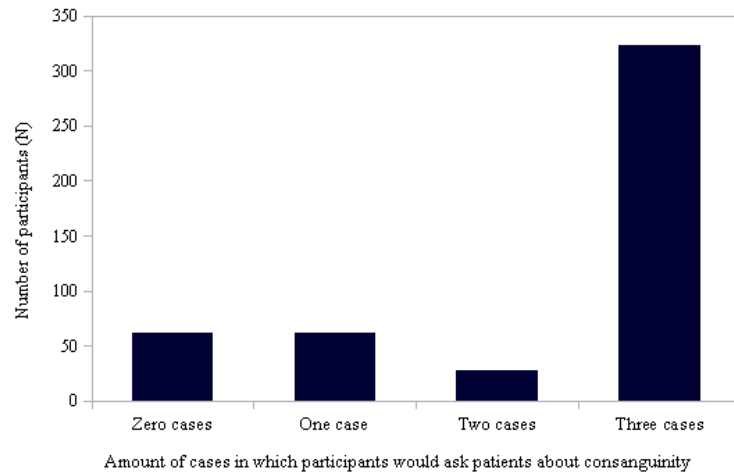
Graph 2 shows data about the number of participants who would ask about consanguinity in zero, one, two, and all three cases. Of the 62 respondents that would ask couples if they are consanguineous in only one of three cases, 61 of those responses (98%) were from the Vietnamese case. The remaining one response was in the Dominican/Caucasian case. For the 28 respondents that would ask couples if they are consanguineous in two of three cases, 12 (44%) would ask in the Vietnamese and Dominican/Caucasian case, 8 (28%) would ask in the Vietnamese and Portuguese/Viking case, and 8 (28%) would ask in the Dominican/Caucasian and Portuguese/Viking case.

Graph 3 shows data about the number of participants who would ask about Jewish ancestry in one, two, three, four, and all five patients. The Vietnamese couple in the first case presented were considered one patient in our data analysis as both members of the couple were from the same country. 127 of the participants responded that they would not ask any of the

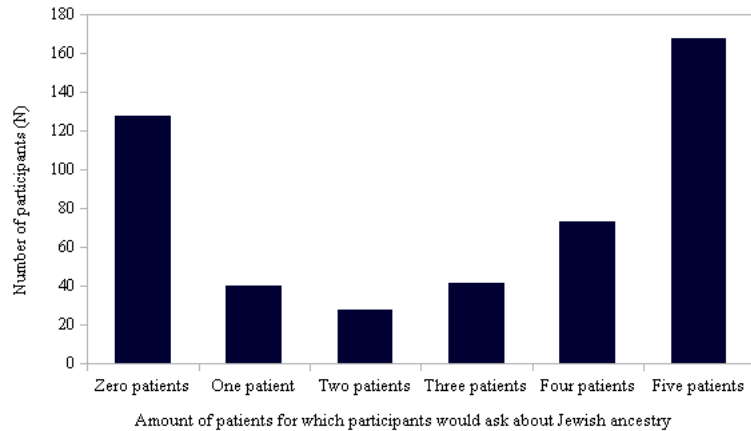
patients if they have Jewish ancestry while 167 said that they would ask it of all of the patients. However the count for those who did not ask any of the patients about Jewish ancestry (127) includes those who did not provide an answer to the question and thus may be a slightly inflated value.



Graph 1: The number of participants who asked follow-up questions in zero, one, two, and all three cases.



Graph 2: The number of participants who asked follow-up questions about consanguinity in zero, one, two, and all three cases.



Graph 3: The number of participants who asked follow-up questions about Jewish ancestry in zero, one, two, three, four, and all five patients.

Carrier screening

The respondents were asked about the situations in which they would offer carrier screening, if there was a particular professional guideline they followed when ordering carrier screening, and what degree of carrier screening they offer. Multiple responses were allowed.

The results are summarized in Tables 6 and 7.

Table 6: Situations participants would offer carrier screening to patients.

Situation	N
I offer it to all prenatal patients	177
I rarely offer carrier screening	33
I offer it based on the presence of risk factors like ethnicity or consanguinity	44
I offer it based on patient interest	12
I offer it to preconception patients	20

Table 7: The professional guidelines participants follow when ordering carrier testing.

Guideline/Recommendation	N
ACMG	34
ACOG	27

Joint NSGC/ACMG/ACOG/SMFM/PQF Expanded Screening Statement	129
SOGC	5
Center-specific	14
Combination	16
Expanded	9

The majority of participants offered an expanded panel to their patients in their clinical practice while fewer offered limited carrier screening. Many participants offered carrier screening on a case by case basis with regards to ethnicity, consanguinity, and family history, among other factors. A smaller number of participants offered all options to the patient and allowed them to choose the degree of testing that best suited them.

Opinions about ethnicity in clinical practice

There were statistically significant differences in participant attitudes and opinions towards self-reported patient ethnicity before and after reviewing Recombine’s research. There was a statistically significant difference between how useful respondents thought it was to gather patient self-reported ethnicity in risk assessment before and after looking at the data ($t(418) = 8.00, p < .001$). On a rating of 1 to 5 where 1 was “Not at all useful” and 5 was “Very useful” the mean before looking at the data was 4.08 (SD=.76), slightly above “Somewhat useful” and the mean afterwards was 3.83 (SD=.85). There was not a significant change between before and after viewing the data as to how useful participants thought it was to gather patient reported ethnicity for risk assessment if they do not know it or are unsure ($T(417) = .83, n.s.$).

There was a significant difference between the attitudes of the participants towards gathering self-reported patient ethnicity as a good proxy for genetic ancestry before and after viewing Recombine’s data (Chi-square (4) = 289.02, $p < .001$). Of those who said self-reported

ethnicity was a good proxy (N = 53) before viewing the data, 56.6% changed their answer to “Sometimes” after seeing the data. Of those who initially said it was “Sometimes” a good proxy, 90.1% (N= 291) did not change their response. The remaining 9.9% (N=32) of those who initially said it was “Sometimes” a good proxy changed their answer to “No” post- data review. Of those who initially said it was not a good proxy, 92.9% also said it was not a good proxy after seeing the data.

Of the participants who answered the question “Would you consider changing what testing you order based on this data?”, 60.44 % (N=246) indicated they would not change what testing they offer to patients post-data review, whereas 38.57% of participants (N=157) said they would offer screening more often for more conditions. No participants indicated they would screen less often for less conditions while 3 participants said they would screen for less conditions more often and 1 participant would screen less often for more conditions.

Discussion

This study is the first of its kind to examine the attitudes and opinions of genetic counselors towards the use of self-reported patient ethnicity in clinical practice. Our objective was to determine how practicing genetic counselors assess patient ethnicity in a clinical setting and determine if there is a shift in these attitudes after looking at data showing the discordance between self-reported ethnicity and genetic ancestry.

Given that family history taking is a cornerstone of genetic counseling, it is consistent that the majority of surveyed genetic counselors elicit information about patient ethnicity while taking a family history. Many use additional sources during the session to determine information about patient ethnicity; visual assessment happens naturally, and intake forms may be a part of their institutional practice. When eliciting this information about ethnicity, the majority of the study participants did not use pre-selected categories, suggesting that genetic

counselors generally ask patients about how they self-identify rather than using limited options. Ethnicity terms were used less commonly than terms eliciting information about ancestry (such as “comes from originally”) suggesting that participants are aware that patient ethnicity is not a perfect proxy for genetic ancestry and may not be the best source of direct information about disease frequencies. Few participants used the word ‘race’ when eliciting information about patient ethnicity, demonstrating genetic counselors understand race and ethnicity are distinct concepts and are not mutually exclusive (Race, Ethnicity, and Genetics Working Group, Ethnicity, 2005).

Because participants are typically using conversation as a tool to gather information about patient ethnicity/ancestry, there is a level of subjectivity that comes with gathering this information. The information given by patients in conjunction with counselors’ personal judgement may lead to them tailoring the follow-up questions they ask the patients. This was reflected in the results of the three preconception consultation cases. While many participants selected the same questions they would ask regardless of how the patient was described in the case scenario, several chose follow-up questions ostensibly based on patient self-reported ethnicity. This highlights a lack of standardization in the elicitation of patient ethnicity and shows that each genetic counselor decides which risk factors to include in their evaluation for carrier screening. For example, 78% of participants asked about consanguinity in all three cases, indicating that this is standard practice but not universally implemented. Another indication of a lack of standardization in practice is highlighted by how many participants asked all patients about Jewish ancestry but some did not ask any of the patients this question at all.

In general, the number of follow-up questions asked appears to be related to the specificity of the ethnicity information provided. For example, the most follow-up questions

were asked of the Caucasian individual from Maine likely because this information is not specific and people from Maine may originate from a variety of geographic areas. However overall, participants were more inclined to ask about general geographic regions rather than specific geographic information in their follow-up questions. Research has shown that self-identified race/ethnicity gives broad information about genetic ancestry but may lose some of the specific details (Banda et al., 2015; Lee et al., 2010; Smith et al., 2014; Yaeger et al., 2008). Participants may view the general information that they get about patient ethnicity and ancestry as sufficient for their needs.

This is corroborated by the attitudes and opinions of participants prior to the review of the Recombine data. Many surveyed genetic counselors were aware that self-reported patient ethnicity is an imperfect proxy for genetic ancestry, yet still found it somewhat clinically useful to determine ethnicity for their risk assessment. This was also the case post-data review: while there was a statistically significant decrease in how useful respondents thought it was to determine ethnicity in risk assessment, the majority of participants still found it somewhat useful. Some respondents thought that it was useful even when the patient did not know their ethnicity or was unsure. Potential reasons for why this general information still has clinical utility in the age of pan-ethnic screening may include residual risk estimates, cases in which insurance coverage is not available, and cases in which one of the partners is unavailable for testing. Patient ethnicity information may be useful in a social context by understanding a patient's background.

While most participants recognize that ethnicity is not always concordant with ancestry, the majority of respondents said that they would not change the testing that they offer based on this data. One reason why understanding this may not translate to a change in practice is that the testing offered by a genetic counselor is also influenced by outside factors

such as insurance coverage and institutional support. Given that many participants already offer ECS and use the joint NSGC/ACMG/ACOG/SMFM/PQF Expanded Screening Statement as a guideline in their clinical practice, willingness to change the testing offered may not necessarily be a good proxy for changes in attitudes regarding patient ethnicity. Participant attitudes towards ECS are consistent with a survey distributed in early 2012 shortly after ECS initially became clinically available which found that genetic counselor attitudes and practices are mainly supportive of ECS (Lazarin et al., 2016).

Overall, this study illustrates a lack of standardization in the genetic counseling profession in the elicitation of patient ethnicity information as well as the attitudes towards its utility in the field. This is highlighted greatly in the follow-up questions chosen by participants in each of the preconception cases. While most counselors asked the same types of questions for each case, there was variety in the quantity and specificity of the questions themselves. Inherent in this variety is room for error and subjectivity in collecting information. Despite their awareness of the limitations of patient self-reported ethnicity, participants still used this in their risk assessment for carrier screening. This means that subjectivity may be involved in the decision process for carrier screening - leading to fundamental differences between what different genetic counselors might offer. That said, many genetic counselors are using ECS and following the updated joint guidelines. However, the majority of surveyed counselors are using other guidelines meaning that the field as a whole may not be following the same overall practices.

Demographics

We found that while both study participants and those surveyed in the 2016 NSGC PSS were concordant in certain respects, specifically in terms of gender/sex and location of practice, there were also distinct differences that could have allowed for a discrepancy

between the findings of this study and the attitudes of genetic counselors as a whole. Our participants were significantly younger and less professionally experienced than the PSS respondents. This may mean that participants have had more experience with ECS than other more traditional methods of screening, and may generally have more open attitudes about carrier screening. Furthermore, the non-white proportion of our participants was greater than that of the PSS which may indicate a special interest of non-white genetic counselors towards the subject of race and ethnicity.

Limitations

While we believe that the participants recruited through the NSGC listserv are a good reflection of the genetic counseling community, they may not have reflected the attitudes of all practicing genetic counselors. There were differences between the way we asked about demographics and how the PSS worded their equivalent questions. We included a non-binary option in gender, added an extra bracket for age, and asked about years of practice rather than year of graduation. With regards to the ethnicities of our participants, we classified their open answers into white and non-white, which may be discrepant with how they actually identified. Lastly, we offered a preset list of questions for the cases without a write-in component. As such, there may be other questions that participants may potentially have asked in those scenarios.

Next steps and conclusion

This study was the first of its kind, looking at the usage of and attitudes towards patient self-reported ethnicity in genetic counseling. It details when and how genetic counselors are asking questions about ethnicity, how they incorporate that information into their sessions, and how they are implementing carrier screening in their practice. This study additionally looks at how reviewing data about the discrepancy between self-reported ethnicity and ancestry may

change the opinions and practices of genetic counselors. There was a statistically significant change in opinions and attitudes pre- and post-data review towards the utility of self-reported ethnicity. However, this change was not large and may highlight an existing awareness among the genetic counseling community that ethnicity and ancestry are not interchangeable. There was additionally variation in the questions that each participant would ask the same patients, indicating a lack of standardization in the profession regarding the ascertainment of patient ancestry. This lack of standardization highlights that there are differences in practice among different genetic counselors and that there is a subjective element of personal judgement in risk assessment.

This study was an initial, exploratory look at how ethnicity information is used by genetic counselors in a clinical setting. Additional studies may help to clarify why many genetic counselors think that self-reported patient ethnicity still has clinical utility and the motivation behind why they ask the questions and make the testing decisions that they do. More qualitative analysis conducted with personal interviews may also be useful for conducting more in-depth research on these topics

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