African American mothers' attitudes towards genetic testing in the InterGEN study

Michelle L. Wright^{1,2} · Kevin Newhall³ · Veronica Barcelona⁴ · Jacquelyn Y. Taylor⁵

Received: 14 February 2019 / Accepted: 2 October 2019 / Published online: 7 December 2019
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Abstract

The paucity of representation of people of color, particularly those of African ancestry, is a major issue in contemporary omics research. Metadata summarizing genome-wide association studies from 2005 to 2015 suggest that nearly 80% of participants are of European ancestry and only 2.4% are of African ancestry. Negative attitude towards genetic testing is a commonly cited belief as to why there is low representation of Americans of African ancestry participating in genetic studies. Using the attitudes towards genetic testing survey, administered as part of our parent (epi)genome-wide association study, we characterized the perceptions of genetic research among our cohort of African ancestry women ($n = 168$). Our data show generally favorable perceptions of genetic testing among our cohort. Further, we demonstrate that more favorable attitudes towards genetic testing correlated with higher levels of income, even when accounting for commonly cited negative predictors such as maternal age, education, country of origin, and religion. Overall, our data characterize generally positive perceptions of genetic testing among women of African ancestry.

Keywords (4-6) Genetic testing · African American · Attitudes · InterGEN · Women

Introduction

The push for precision medicine offers hope and opportunity to improve health on both the individual and community levels. Genetic testing related to health risk, response to medications, and disease prognosis, such as inborn errors of

metabolism and cancer, has led to improvements in identifying people at risk for disease and improving health outcomes (Taylor et al. 2017; Ashley 2015; Taylor and Barcelona de Mendoza 2018). Others have warned that despite the benefits of genetic testing and promise of precision health initiatives, the focus on the individual may minimize research and interventions to address significant social and structural determinants of health, such as racism and poverty, and will therefore not broadly improve public health (Khoury and Galea 2016). To further complicate the progression of precision medicine to help all individuals, a majority of genetic studies have included only participants of European ancestry (Popejoy and Fullerton 2016). Metadata from the European Bioinformatics Institute and the National Human Genome Research Institute analyzing the ancestry of participants in genome-wide association studies (GWAS) from 2005 to 2016 indicated that persons of European ancestry made up 78% and persons of African ancestry made up 2.4% of the over 110 million individuals that participated in over 4500 studies (European Molecular Biology Laboratory 2017).

To specifically address the lack of African Americans in omics research, the Precision Medicine Initiative by the National Institute of Health has explicit requirements for targeting enrollment of minority and historically underrepresented populations (Sankar and Parker 2017). Recently, media

✉ Michelle L. Wright
michelle.wright@utexas.edu

Kevin Newhall
ken2012@med.cornell.edu

Veronica Barcelona
veronica.barcelona@yale.edu

Jacquelyn Y. Taylor
jt139@nyu.edu

¹ School of Nursing, University of Texas, Austin, TX, USA

² Dell Medical School, Department of Women's Health, Austin, TX, USA

³ Department of Medicine, Division of Hematology and Medical Oncology, Weill Cornell Medicine, New York, NY, USA

⁴ Yale School of Nursing, Orange, CT, USA

⁵ New York University, Rory Meyers College of Nursing, New York, NY, USA

outlets have reported conflicting views by people of color regarding the Precision Medicine Initiative; some are enthusiastic about increasing representation in research data, while others are hesitant due to historic legacies of mistreatment, particularly of African Americans in biomedical research [e.g., The Tuskegee Study (Stein 2017)]. Several research groups have formally investigated reasons why minority populations are underrepresented in genetic studies (Furr 2002; Wendler et al. 2006; BeLue et al. 2006; Suther and Kiros 2009; Bates et al. 2011; Sussner et al. 2011; Taylor et al. 2013). Negative attitudes towards genetic testing, specifically among African Americans, have been associated with fear of racialization of genetic test results, general mistrust of physicians and the medical system, and lower levels of education (Furr 2002; Suther and Kiros 2009; Taylor et al. 2013; Rogers et al. 2018). However, others have found favorable attitudes towards genetic testing among African Americans and willingness to participate in genetic research studies (Wendler et al. 2006; Bates et al. 2011; Taylor et al. 2013).

As women are often the main consumers of health care for their families, it is important to better understand African American women's views on this increasingly important healthcare option for them and their children. Since both positive and negative attitudes towards genetic testing among African Americans have been reported, we aimed to evaluate factors among African American women, more specifically mothers, that may result in women having more positive or negative attitudes towards genetic testing. Using survey data from our parent study (InterGEN), an (epi)genome-wide association study (EWAS), we aimed to examine the attitudes of mothers of African ancestry towards genetic testing (Crusto et al. 2016; Taylor et al. 2016). This descriptive, cross-sectional study assessed if there were relationships among African American mothers' attitudes towards genetic testing (benefits, awareness, and outcomes) and maternal age, country of origin (US vs. non-US born), level of income, level of education, or religious affiliation.

Methods

Data used for this study are a subset from our ongoing, IRB-approved study: the Intergenerational Impact of Genetic and Psychological Factors on Blood Pressure [InterGEN, (Crusto et al. 2016; Taylor et al. 2016)]. Extensive details on the InterGEN study recruitment, data cleaning, and statistical analysis have been reported elsewhere (Crusto et al. 2016; Taylor et al. 2016). The primary aims of the parent study are to evaluate the contributions of psychological, genetic, and epigenetic factors and hypertension among African American mothers and their children. Pertinent to this secondary analysis, African American mothers of children between the ages of 3 and 5 years old were recruited from the

community to participate in a study evaluating factors that contribute to hypertension in African American mothers and their children. All women self-identified as Black or African American. Baseline demographic and survey data were collected at the initial interview (time point 1 of 4 total visits) of this ongoing longitudinal study, via audio self-assisted interviewing software.

The attitudes towards genetic testing (AGT) tool was administered at time point 2 of the InterGEN study. The AGT instrument has been validated among African American women and girls (Taylor et al. 2013). The questionnaire is comprised of ten questions (Cronbach's $\alpha = 0.66$) using a 4-point Likert scale (1 = strongly agree, 2 = somewhat agree, 3 = somewhat disagree, 4 = strongly disagree), and lower scores are associated with more positive attitudes towards genetic testing. Subscales within the instrument assess attitudes towards the benefits of testing, awareness of genetic testing results, and outcomes of having genetic tests. We computed descriptive statistics and conducted correlation analyses to determine if sociodemographic factors, previously described as barriers to enrollment in genetic studies (e.g., age, income, education, country of origin, religious), were associated with negative attitudes towards genetic testing among African American mothers. All analyses were completed in the R statistical computing environment using the psych package (R Development Core Team 2008; Revelle 2018). Significance of correlations was determined using Spearman rank correlation testing with Bonferroni correction assuming an alpha of 0.05.

Results

The subset of women from the InterGEN study who completed the AGT ($n = 168$) were on average 31.60 (5.64) years old, were born in the USA (81%, $n = 135$), made less than \$20,000 per year (55%, $n = 92$), had some college or more (63%, $n = 105$), were single/divorced/separated (73%, $n = 123$), and belonged to a religious faith community (55%, $n = 95$). Most women not born in the USA were from the Caribbean (67%, $n = 22$) or Africa (24%, $n = 8$), 1 participant was from Canada, and 3 did not list their country of origin. Most women who reported belonging to a religious community were Christian (88%, $n = 82$), and the remaining reported other (9%, $n = 10$) or Islamic (3%, $n = 2$) affiliations. Mothers in our study also generally had favorable attitudes towards genetic testing, per the low mean scores for each question (Table 1). The mothers in our study that were born in the USA were younger than foreign born mothers (mean age, 30.8. vs. 34.9), and age was positively correlated with income, education, and identifying as religious (Table 2). Higher income was also significantly correlated with more positive

Table 1 Attitudes towards genetic testing questions and scores, InterGEN ($n = 168$)

Subscale	Questions	Mean	SD
Benefits	1) You are confident that the results of your genetic tests will be kept confidential	1.30	0.64
	2) Genetic testing is relevant to you and/or your child's health	1.40	0.70
	3) Genetic testing is beneficial in early identification of disease	1.35	0.60
	4) Genetic testing is beneficial in treatment of disease	1.35	0.62
	5) Genetic testing is beneficial in preventing disease	1.49	0.73
Awareness	6) You would like to know if you and/or your child test positive for a genetic disorder	1.36	0.74
	7) If you or your child tested positive for a genetic disorder, you would seek medical care immediately to minimize you or your child's chances of developing the disease	1.14	0.53
Outcomes	8) If you or your child tested positive for a genetic disorder, you wait until experienced signs and symptoms of the disease before obtaining medical care	1.94*	1.20
	9) If you or your child tested positive for a genetic disorder, you and/or your child would be treated differently by health care providers	2.68	1.16
	10) If you test positive for a genetic disorder, it is likely that your child is at risk for testing positive for the same disease.	2.23	0.98

*This item is reverse scored

attitudes towards genetic testing, given that lower scores on the AGT tool are indicative of more positive attitudes.

Discussion

The women in our study generally had favorable attitudes towards genetic testing, consistent with what other research groups have reported (Wendler et al. 2006; Bates et al. 2011; Taylor et al. 2013). In our previous study analyzing attitudes towards genetic testing in grandmother-mother-daughter triads, being younger and less educated was associated with more negative attitudes towards genetic testing (Taylor et al. 2013). However, among our current cohort, age and level of education were not associated with less positive attitudes towards genetic testing. Surprisingly, women with higher levels of income had more favorable attitudes towards genetic testing, and the significance of this relationship persisted, even

when accounting for variables correlated with level of income that have traditionally been associated with negative attitudes towards genetic or medical research (Shavers et al. 2001; Wendler et al. 2006; Long et al. 2011).

In the USA, there are ongoing attempts to increase the inclusion of people of color and ethnic minorities in federally funded research projects. For example, research studies including human subjects are mandated to include women and minorities unless sufficient justification for exclusion is given. Some have argued that it is difficult to recruit people of color and other minority populations because of increased levels of distrust of the medical and research communities, as well as beliefs that genetic research may result in societal harm (Furr 2002; Suther and Kiros 2009). However, a separate large study including responses from more than 70,000 individuals, including more than 25,000 African Americans primarily from the USA, did not find any significant differences among individuals of diverse racial and ethnic backgrounds in

Table 2 Correlation matrix of maternal characteristics and attitudes towards genetic testing, InterGEN study ($n = 168$)

	Age	Cohabiting	US born	Income	Education	Religious	Benefits	AGT awareness	AGT outcomes	AGT total score
Age	1.00									
Cohabiting	−0.16	1.00								
US born	−0.27	0.22	1.00							
Income	0.29	−0.19	−0.18	1.00						
Education	0.21	−0.22	−0.08	0.62	1.00					
Religious	0.17	−0.17	−0.05	0.26	0.24	1.00				
AGT benefits	−0.04	−0.06	0.08	−0.20	−0.03	−0.08	1.00			
AGT awareness	−0.07	−0.13	−0.01	−0.15	0.04	0.03	0.51	1.00		
AGT outcomes	−0.07	−0.00	0.14	−0.21	−0.10	−0.11	0.11	0.07	1.00	
AGT total score	−0.06	−0.07	0.12	−0.28	−0.07	−0.11	0.79	0.57	0.62	1.00

Bolded values indicate significant Spearman correlation $\alpha < 0.05$ with Bonferroni correction

willingness to participate in healthcare research (Wendler et al. 2006). This large study by Wendler et al. (2006) evaluated willingness to participate in healthcare research, not specifically genetic research; nor did they evaluate differences by sex. Their study was seminal in that it removed the burden of inclusion from marginalized groups and places the burden squarely upon researchers to more actively engage with, and recruit, non-European ancestry research participants. Our work adds to and supports the aforementioned conclusions of Wendler and colleagues (Wendler et al. 2006), in that African American women, particularly mothers, generally have positive perceptions of genetic testing. Further, by enrolling in our parent study that is specifically focused on evaluating genetics related to health disparities, it is demonstrated that African American women are willing and ready to participate in genetic research studies.

The AGT tool does not directly measure participants' level of trust in the medical system or of research personnel, both frequently cited barriers to inclusion of African Americans in genetic studies. However, regardless of our participants' level of trust in the medical system or of researchers, they generally maintained positive attitudes towards genetic testing (i.e., low average scores for all AGT questions). For example, the mothers in our study had slightly less positive scores (Question 9, mean = 2.68) related to how they felt healthcare providers would treat them if they, or their children, had a genetic disorder. However, this question does not allow qualitative responses for interpretation of why the mother thought they may be treated differently. For instance, if the child screened positive for sickle cell disease, did the mother expect for her and her child to be treated differently because they would need monitoring and care or because they thought they may be discriminated against because of the disorder?

The main strength of this study is that we examine attitudes towards genetic testing in all African American sample of women and that we had a fairly large sample size for a dyadic focused study on genetic and environmental factors related to health. As the parent study is an (epi)genome-wide association study (EWAS), our sample population may be biased towards more favorable views of genetic testing. While this may be a limitation in that it may not be generalizable to all mothers of African ancestry, it is also a strength because mothers of African ancestry are actively engaging in genetic research. Interestingly, 94% of women approached for participation agreed to enroll in the study, which we attribute to our commitment to cultural competence and diversity of our research field staff and principal investigators. To that end, we aim to extend these analyses and results by evaluating attitudes towards genetic testing among a larger and more diverse population of people of color and to be inclusive of men and other family members who may influence health care decision-making, such as grandparents.

Conclusion

Inclusion of people of color within genetic research studies has been extremely limited to date. Over the past decade, there has been some improvement in recruiting minority populations into genetic research studies, increasing from 4% non-European ancestry participants to 19% (Popejoy and Fullerton 2016). This increase, however, is largely represented by an increase of participants of Asian ancestry, primarily from East, South, and Southeast Asia and in particular due to an increase in GWAS studies completed in Asia (Popejoy and Fullerton 2016). The continued underrepresentation of people of color in genetic studies may also contribute to diminished accuracy in identifying variants that are associated with disease. For example, many SNPs associated with health risks are present in varying amounts and effect sizes in different populations (Carlson et al. 2013; Bryc et al. 2015; Wright et al. 2016; Yudell et al. 2016). In some cases, a SNP associated with a trait can be identified among non-European and replicated within a European population because the SNP is more prevalent in the minority population, as in the case of SNP associated fatty acid metabolism and height among Greenlandic Inuit (Fumagalli et al. 2015). Conversely, SNPs identified as significant among European ancestry populations may be false positives among other ancestry groups, as is frequently observed with variants associated with cancer and cardiovascular diseases (Taylor et al. 2011; Carlson et al. 2013; Dietze et al. 2015; Wright et al. 2018). In order to improve accuracy in precision medicine and ensure equity of the provision of precision medicine, people of color and other ethnic minorities must be included in increased numbers in genetic research.

Others have published recommendations and approaches for recruiting and engaging racial and ethnic minority populations to participate in health and genetic research studies (National Academy of Sciences 2018). For example, individuals from racial and ethnic minority groups are more likely to engage in research projects where they have a relationship with the research team and feel the needs of their community are being addressed, have a lead investigator of color, and are treated with respect as an individuals and not merely as a research participant (Taylor and Barcelona de Mendoza 2018). Other groups have found that engaging with the community provides important insights and can help to guide research questions that will be valuable among groups traditionally excluded from research studies (Franck et al. 2018; National Academy of Sciences 2018). Our study further supports the works of others that African Americans in the USA do not have inherently negative attitudes towards genetic testing that are causing them to be underrepresented in genetic studies. The onus is on researchers to engage with broader communities, treat them with respect, and provide

access and opportunities for understudied population to participate in research.

Funding information This study was funded by grants R01NR013520 (JYT), K01NR017010 (VB), and K01NR017903 (MLW) from the National Institute of Nursing Research of the National Institutes of Health.

Compliance with ethical standards

Conflict of interest All authors declare that they have no conflicts of interest. Research data is included as a supplementary file with this manuscript. Further study and data inquiries can be made via the InterGEN website (<https://intergen.yale.edu/>).

Ethical approval All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

Statement of informed consent Informed consent was obtained from all individual participants included in the study.

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