Perceptions About Genetic Testing for the Susceptibility to Alcohol Dependence and Other Multifactorial Diseases

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Background: Beliefs, attitudes, and preferences about the risk and benefits of genetic testing are important determinants of willingness to undergo testing. **Aims:** The purpose of this study was to evaluate the perceived importance of genetic testing for alcohol dependence compared with other multifactorial diseases among African Americans. **Methods:** Surveys were conducted with 258 participants using the Genetic Psycho-Social Implications (GPSI) questionnaire to evaluate several areas of hypothetical genetic testing for alcohol dependence. Respondents were divided into two groups: those who perceived testing for alcohol dependence to be equally important as testing for cancer and those who did not. Using chi-square, the groups' responses were compared for nine GPSI items measuring beliefs about the severity of alcohol dependence, general benefits of genetic testing, and specific benefits of genetic testing for diabetes, hypertension, or a disease affecting a family member. **Results:** Nearly 86% of respondents believed that genetic testing for alcoholism was equally as important as testing for cancer. Those who reported parity of importance of alcohol dependence and cancer screening were more likely to believe that alcoholism is a deadly disease ($p < 0.001$) and genetic testing influences health ($p < 0.001$). **Conclusion:** African Americans reported favorable attitudes and beliefs in possible availability of susceptibility genetic testing for alcohol dependence. The perceived importance of testing for alcohol dependence was associated with beliefs about the severity of alcoholism and certain benefits of genetic testing in general.

Introduction

FAMILY, TWIN, AND ADOPTION studies have revealed a genetic contribution to a positive family history of alcohol dependence (Goodwin *et al.*, 1974; Bohman, 1978; Cadoret *et al.*, 1980; Schuckit, 1981; Gurling *et al.*, 1984). However, the preponderance of research evidence demonstrates that alcohol dependence is a multifactorial disease resulting from genes and environment (Cloninger *et al.*, 1981; Schuckit, 1981; Pickens *et al.*, 1991; McGue *et al.*, 1992; Prescott and Kendler, 1999). Emerging genetic medicine research on other multifactorial diseases such as diabetes, cardiovascular disease, and cancer has provided a greater need for health care professionals to provide genetic services (Hotlzman, 1993; Shields *et al.*, 2008).

More importantly, the patient who is knowledgeable about genetic concepts is better equipped to make informed decisions about genetic testing, medical treatment, and continuity of care. Earlier research studies have shown that most patients do not understand some of the basic genetic terminology and concepts (Emery *et al.*, 1998; Lanie *et al.*, 2004). For example, Lanie *et al.* (2004) conducted in depth one-on-one telephone

interviews to explore the understanding of basic genetic terms and belief as to the location of genes in the human body. It was found that participants had limited understanding of genetic terms and the location of genes in the human body. Additionally, some participants demonstrated frustration when trying to answer the questions during the interview.

Variation in perceptions and utilization of genetic testing has been linked to sociodemographic differences. Literature has shown that greater education is associated with higher rates of genetic counseling and testing (Lerman *et al.*, 1999b). In addition, ethnic differences in beliefs and behaviors related to genetic testing have been evaluated in recent research studies (Lerman *et al.*, 1999a; Thompson *et al.*, 2002; Peters *et al.*, 2004; Singer *et al.*, 2004). Results reveal lower awareness about genetic testing and medical research among some ethnic groups due, partially, to factors such as acculturation and language barriers (Thompson *et al.*, 2003). As a result, not all socioeconomic and population groups have benefited from the growing use of genetic counseling. Kessler *et al.* (2005) evaluated concepts relating to knowledge about family history, types of disorders, and medical genetics terminology among a cohort of African American women. This study

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revealed that attitudes about the benefits of genetic testing were endorsed at a higher rate relative to limitations and risks of genetic testing (Kessler *et al.*, 2005). However, favorable attitudes toward genetic testing do not necessarily translate to the uptake of genetic testing. According to Thompson *et al.* (2002), having a limited amount of knowledge about genetic counseling and testing may be a barrier to utilization of genetic services specifically among African Americans. While these studies are informative, historically in the United States, ethnic group comparisons about medical research have been limited due to a lack of minority participants and other factors such as poor access to health care settings where research is conducted, limited knowledge of medical research, societal distrust of medical researchers, and effects of historical events such as the US Policy Health Syphilis Study at Tuskegee (Shavers-Hornaday *et al.*, 1997; Shavers *et al.*, 2000; Corbie Smith *et al.*, 2002; Chandra, 2003).

Inclusion of ethnic minorities in research on genetic testing is vital because this area represents the frontier in the battle for health equity. While no specific genetic tests are currently available to determine whether a person is at risk for becoming alcohol dependent, genetic testing for alcohol use disorders and related diseases can serve as an example of the processes needed to maximize clinical benefits of the rapidly transforming field of genetic medicine (Burke, 2002). The ability to assure that individuals adequately understand genetic testing in the context of environmental contributions may be the linchpin of the future of genetic medicine.

Research literature has evaluated the interest and concerns of susceptibility of genetic testing in medical conditions such as cancer (Halbert, 2005; Kessler *et al.*, 2005; Edwards *et al.*, 2008; Rantala *et al.*, 2009); however, there is very limited research on interests and concerns regarding susceptibility to genetic testing about alcoholism (Quaid *et al.*, 1996; Gamm *et al.*, 2004a, 2004b). There is a stigma associated with alcohol use disorders that may not be associated with multifactorial diseases such as cancer, diabetes, or hypertension. Studies have suggested that a greater public stigma is associated with people who drink alcohol, experience problems with alcohol, or those less likely to perceive alcoholism as a disease (Beckman and Amaro, 1986; Wilsnack *et al.*, 1991; Weisner and Schmidt, 1992; Fortney *et al.*, 2004). A study by Gamm *et al.* (2004a) explored beliefs about the cause of alcoholism and risk perception among individuals with affected relatives. Data from this study suggested that participants' interest in genetic testing for susceptibility to alcoholism was moderate. In addition, Gamm *et al.* (2004b) explored interest and concerns about the cause of alcoholism and risk perception among individuals with affected relatives. Data from this second analysis suggested that participants perceived themselves being at risk for alcoholism stemming from a belief in a genetic or biological cause of alcoholism (Gamm *et al.*, 2004b). Overall the participant's belief in a genetic cause for alcohol dependence was associated with a significantly increased risk perception of genetic susceptibility for this study.

Each year millions of people are diagnosed with alcohol dependence, and of those who are diagnosed, ethnic minority populations make up a significant proportion (Hasin *et al.*, 2004). The purpose of this study was to evaluate the perception of hypothetical genetic testing for alcoholism compared with other medical conditions among an urban population of African Americans.

Materials and Methods

Study population

This cross-sectional study included a community sample of 304 African American participants (113 men and 191 women) whose ages ranged from 18 to 83 years old. To be eligible for the study, participants had to self-identify as being African American, be at least 18 years old, and not meet the Alcohol Use Identification Test (AUDIT) criteria for alcohol dependence. Of the 304 participants, 258 (99 men and 159 women) agreed to a hypothetical test for alcohol dependence. This was the precursory statement from which participants, if agree, proceed to the statement "It is equally as important to screen for alcoholism as it is to screen for certain types of cancer." The data presented in this manuscript represent the 258 participants mentioned previously. Data from 46 participants were excluded from the analysis. Of these 46 participants, 45 answered "no" or "don't know" to the precursory statement and hence did not proceed to the statement on parity of testing, while one participant did not respond to the question on parity of testing. All 46 participants were similar to the 258 participants in most demographic characteristics except education and perception of risk compared with the general population. The 258 participants were predominantly those with some college education and were more likely to believe that they were at a higher risk for alcoholism compared with the general population. This study protocol was approved by the Howard University Institutional Review Board and the project advisory committee of the Howard University Alcohol Research Center (HU-ARC).

Procedures

The community cohort was recruited by advertisement through newspapers, radio, television, flyers, and self-referrals. Individuals interested in participating were instructed to call the study's recruitment phone line to complete a voluntary and confidential telephone screener. This confidential telephone screener was used to evaluate different drinking patterns among individuals and obtain data on race and age to determine eligibility for the study. Additionally, AUDIT was used to screen for excessive drinking, alcohol use disorders, and identify individuals who were hazardous drinkers. Eligible participants were then invited for a face-to-face interview.

The study took place at the Howard University Hospital General Clinical Research Center (GCRC). Upon arrival at the GCRC facility, participants were given an overview of the study requirements and procedures and were informed of use of study-related data in accordance with the Health Insurance Portability and Accountability Act. After providing informed consent, the study measures were administered in a private setting by a research assistant trained in the study protocol. The study measures were completed during a single session ranging in length from 1 to 2 h. Each participant was compensated \$20 upon completion of the interview.

Measures

Participants were interviewed using the Self-Rating of Ethanol Use (SRE) form and a Genetic Psycho-Social Implications (GPSI) questionnaire. The SRE is a 12-item self-report

questionnaire that measures the level of response to alcohol by asking participants to report the number of standard alcohol drinks required to experience behavioral effects of intoxication, including (1) begin to feel different, feel a bit dizzy, or begin to slur your speech; (2) begin stumbling, or walking in an uncoordinated manner; and (3) pass out, or fall asleep when you did not want to (Schuckit *et al.*, 1997). The GPSI is a 90-item self-report questionnaire. The GPSI questionnaire reviewed several main areas: (1) the participant's interest in predispositional genetic testing for alcohol dependence, defined as testing that evaluates transference of genetic risk of alcoholism from parent to offspring and what they believe might be the impact of this type of testing; (2) the participant's concerns about genetic susceptibility testing for alcohol dependence; (3) the participant's beliefs about the causes of alcohol dependence and the possible role of genetic factors; (4) the participant's feelings of personal control over his/her fate with alcoholism, and whether belief in the role of genetic factors influences those feelings; (5) the participant's interests, concerns, and beliefs about research in general. Responses to each item are given on a 5-point Likert scale where 1 represents "strongly disagree" and 5 represents "strongly agree." This questionnaire was developed specifically for this study based on the results of Gamm *et al.* (2004a) and Kessler *et al.* (2005).

A pilot test of the GPSI questionnaire was conducted using a convenience sample that was representative of the study population. The GPSI questionnaire includes five subscales: (1) perceptions, (2) interests, (3) concerns, and (4) beliefs about alcohol dependence, and (5) general research as it relates to hypothetical genetic testing for alcoholism. These questions were answered on a scale of 1 through 5 with the number 1 indicating "disagree strongly" and the number 5 indicating "agree strongly." The questionnaire was administered to a test group of seven individuals within 7 days of original survey administration as a test-retest for reliability to measure internal consistency of individuals' response to the items within the subscale. Cronbach's alpha for the subscales ranged from 0.68 to 0.71. The overall internal consistency coefficient for the GPSI questionnaire was Cronbach's alpha = 0.76.

Belief about the importance of hypothetical genetic testing for alcohol dependence compared with other medical conditions was measured by the GPSI question, "It is equally as important to screen for alcoholism as it is to screen for certain types of cancer." The nine GPSI items were selected to test whether the perception of parity of testing importance was associated with interest in testing, attitudes toward genetic tests/screening, and willingness to participate in genetic testing. Additionally, the nine GPSI items represent a participant's knowledge of genetic testing. Sociodemographic data including age, gender, education, income, and having children were collected along with a detailed family history of alcohol dependence including affected first-, second-, and third-degree relatives.

Data analysis

All data were analyzed using SPSS 17.0 software (computer program, 2009) (SPSS, Inc., Chicago, IL). For the purposes of the analysis, the participants' responses were recoded into three categories: (1) responses that were either "Agree" or

"Agree Strongly" were recoded as "Agree"; (2) responses that were either "Disagree" or "Strongly Disagree" were recoded as "Disagree"; and (3) responses of "Uncertain" remained as a separate category. One-way ANOVA was used to compare continuous data (age) across parity of testing. Pearson's χ^2 was used to determine whether there were significant associations between equal importance of genetic testing to medical testing and variables of interest, attitudes, and willingness to test. Significance was established at $p < 0.05$.

Results

Evaluation of the demographic variables revealed the following: 62% were single, never married; 44.2% earned less than \$10,000 annually; and 52% had children (see Table 1). Additionally, over 70% of the participants had relatives with alcohol-related issues, of which over 50% had relatives who were alcohol dependent. However, more than 66% of the participants did not perceive themselves to be at a higher risk of alcoholism compared with the general population. Participants who agreed to the question stating "genetic testing is equally as important as cancer testing" did not differ on most demographics from participants who disagreed or were uncertain; however, they did differ in age ($p = 0.033$) and level of education ($p = 0.002$). Participants who strongly agreed to the question stating "genetic testing is equally as important as cancer testing" were older, and fewer had college degrees compared with those participants who disagreed.

Table 2 shows a few variables that were associated with the belief that testing for alcoholism was equally as important as cancer testing. Participants who held this belief were more likely to report that they would take a genetic test for alcoholism because they believed alcoholism was a deadly disease (Pearson's $\chi^2 = 35.49$, $p < 0.001$). Data revealed that participants who believed genetic tests for alcoholism were equivalent to genetic tests for cancer were more likely to also believe that genetic testing influences one's health (Pearson's $\chi^2 = 34.18$, $p < 0.001$). Perception of importance of alcoholism and cancer testing was not associated with the belief that genetic testing is preventative, should only be offered to those at risk, or should be given to children if treatment exists. Upon adjusting for age and education, both statements remained significantly different between those who believe that genetic testing for alcoholism is equally as important as genetic testing for cancer, and those who do not. Participants who would also take a genetic test because they believe alcoholism is a deadly disease were still more likely to agree that genetic testing for alcoholism was as important as genetic testing for cancer, regardless of age or level of education. Likewise, the age and education of the participant did not affect the finding that more of the participants who agree that genetic testing influences one's health also agree that alcoholism and cancer genetic testing are of equal footing. Although there was a trend toward significance among those who would participate in research for genetic risk of diabetes (Pearson's $\chi^2 = 9.39$, $p = 0.052$), there were no differences between groups for willingness to participate in research if a relative had the condition being researched or if participation gives risk for high blood pressure.

Evaluation of other variables, such as belief that genetic test is preventative or likelihood of considering research

TABLE 1. DEMOGRAPHICS (N=258)

	Equally important to screen for alcoholism as cancer				p
	Disagree (n=29)	Uncertain (n=29)	Agree (n=200)	Total (n=258)	
Age (mean±SD)	35.48±14.4	34.52±14.83	40.66±14.45	39.39±14.62	0.033 ^a
Gender (n, %)					
Male	15, 51.7%	10, 34.5%	74, 37.0%	99, 38.4%	0.282
Female	14, 48.3%	19, 65.5%	126, 63.0%	159, 61.6%	
Marital status (n, %)					
Married	4, 13.8%	3, 10.3%	28, 14.0%	35, 13.6%	0.239
Single	21, 72.4%	22, 75.9%	117, 58.5%	160, 62.0%	
Separated/divorced/widowed	4, 13.8%	4, 13.8%	55, 27.5%	63, 24.4%	
Education (n, %)					
Some high school/diploma	7, 24.1%	1, 3.4%	69, 34.5%	77, 29.8%	0.002 ^a
Some college	11, 37.9%	16, 55.2%	92, 46.0%	119, 46.1%	
BS/advanced degree	11, 37.9%	12, 41.4%	39, 19.5%	62, 24.0%	
Household income (n, %)					0.611
<\$10,000	14, 48.3%	11, 37.9%	89, 44.5%	114, 44.2%	
\$10,000–\$39,999	8, 27.6%	11, 37.9%	78, 39.0%	97, 37.6%	
\$40,000 or more	7, 24.1%	7, 24.1%	33, 16.5%	47, 18.2%	
Have children (n, %)					0.153
No	16, 55.2%	18, 62.1%	89, 44.7%	123, 47.9%	
Yes	13, 44.8%	11, 37.9%	110, 55.3%	134, 52.1%	
Ethnicity (n, %)					0.414
African American	28, 96.6%	28, 96.6%	198, 99.0%	254, 98.4%	
Other	1, 3.4%	1, 3.4%	2, 1.0%	4, 1.6%	
Drinking caused problems w/health, family job, police (n, %)					0.871
Don't know	1, 3.4%	1, 3.4%	4, 2.0%	6, 2.3%	
No	9, 31.0%	6, 20.7%	55, 27.5%	70, 27.1%	
Yes	19, 65.5%	22, 75.9%	141, 70.5%	182, 70.5%	
Any family dependent on alcohol (n, %)					0.341
Don't know	1, 3.4%	3, 10.3%	9, 4.5%	13, 5.0%	
No	16, 55.2%	14, 48.3%	83, 41.5%	113, 43.8%	
Yes	12, 41.4%	12, 41.4%	108, 54.0%	132, 51.2%	
Any family diagnosed with illicit or unprescribed drugs (n, %)					0.778
Don't know	1, 3.4%	3, 10.3%	11, 5.5%	15, 5.8%	
No	15, 51.7%	16, 55.2%	107, 53.5%	138, 53.5%	
Yes	13, 44.8%	10, 34.5%	82, 41.0%	105, 40.7%	
Would be tested (n, %)	29, 100.0%	29, 100.0%	200, 100.0%	258, 100.0%	n/a
Risk increased compared with general population (n, %)					0.239
No	18, 81.8%	19, 70.4%	110, 64.3%	147, 66.8%	
Yes	4, 18.2%	8, 29.6%	61, 35.7%	73, 33.2%	

^ap<0.05.

SD, standard deviation.

participation if relative has condition being researched, show that there are just as many participants who agree genetic tests are as important as health screenings as those who do not.

Discussion

Although some research literature suggest ethnic minority populations have attitudes that are less likely to favor genetic testing than of European descent due to barriers and challenges, our data demonstrated that over 90% of all respondents would participate in genetic testing for diabetes, hypertension, or other diseases afflicting family members and 85% were interested in genetic testing for alcoholism risk. Our data further revealed that having perceptions of parity between the perceived importance of testing for alcoholism and cancer was

associated with the beliefs about the severity of alcoholism and perception of certain benefits of genetic testing.

Barriers for genetic testing may include sociodemographic status such as age, gender, race, or education. Data for this study suggested that there was a difference in age and education for participants who agree genetic testing is equally as important as cancer testing. The participants were older and self-reported some college education. This outcome suggests that lack of formal education may pose a barrier to person's utilization of genetic testing. For this study purposes, formal education was defined as completing high school or general education development. These findings complement previous studies that have shown that greater education is associated with higher rates of genetic counseling and testing (Lerman *et al.*, 1999b). Lerman *et al.* (1999b) evaluated racial differences

TABLE 2. INTEREST AND ATTITUDES IN GENETIC TESTING ASSOCIATED WITH EQUAL IMPORTANCE OF ALCOHOL AND CANCER SCREENING

	<i>Equally important to screen for alcoholism as cancer</i>			x	p
	<i>Disagree</i>	<i>Uncertain</i>	<i>Agree</i>		
Would choose to be tested because:					
You believe alcoholism is a deadly disease	Disagree 10, 34.48%	4, 13.79%	8, 4.00%	35.49	<0.001 ^a
	Uncertain 1, 3.45%	4, 13.79%	10, 5.00%		
	Agree 18, 62.07%	21, 72.41%	182, 91.00%		
What is your attitude toward genetic testing and screening in general:					
Genetic testing is for preventive care	Disagree 2, 6.90%	—	11, 5.50%	2.12	0.714
	Uncertain 1, 3.45%	2, 6.90%	11, 5.50%		
	Agree 26, 89.66%	27, 93.10%	178, 89.00%		
With genetic testing can find out if develop disease before have symptoms	Disagree 3, 10.34%	2, 6.90%	8, 4.00%	5.99	0.200
	Uncertain 4, 13.79%	3, 10.34%	11, 5.50%		
	Agree 22, 75.86%	24, 82.76%	181, 90.50%		
Use of genetic testing can influence one's health	Disagree 9, 31.03%	2, 6.90%	11, 5.50%	34.18	<0.001 ^a
	Uncertain 1, 3.45%	8, 27.59%	15, 7.50%		
	Agree 19, 65.52%	19, 65.52%	174, 87.00%		
Genetic screening should only be offered to people at risk for a disease	Disagree 22, 75.86%	25, 86.21%	161, 80.50%	1.15	0.887
	Uncertain 2, 6.90%	1, 3.45%	13, 6.50%		
	Agree 5, 17.24%	3, 10.34%	26, 13.00%		
Genetic screening should be offered to children if treatment exists	Disagree 6, 20.69%	6, 20.69%	34, 17.00%	0.64	0.959
	Uncertain 3, 10.34%	2, 6.90%	18, 9.00%		
	Agree 20, 68.97%	21, 72.41%	148, 74.00%		
You would participate in medical research if it involves:					
A disease and/or condition you or your family member is diagnosed with	Disagree—	1, 3.45%	2, 1.00%	2.11	0.716
	Uncertain 1, 3.45%	2, 6.90%	10, 5.00%		
	Agree 28, 96.55%	26, 89.66%	188, 94.00%		
A genetic test to give your risk for diabetes	Disagree—	1, 3.45%	2, 1.00%	9.39	0.052
	Uncertain 1, 3.45%	3, 10.34%	3, 1.50%		
	Agree 28, 96.55%	25, 86.21%	195, 97.50%		
A genetic test to give your risk for high blood pressure	Disagree—	1, 3.45%	4, 2.00%	4.38	0.357
	Uncertain 1, 3.45%	2, 6.90%	3, 1.50%		
	Agree 28, 96.55%	26, 89.66%	193, 96.50%		

Results are presented as (n, %).

^ap < 0.001.

in the effects of education only versus education and counseling on testing intentions, blood sample provision, and psychological distress for the breast cancer susceptibility gene (BRCA1). Their study revealed that pretest education and counseling may motivate genetic testing. The lack of genetic testing education may have limited the ability of some individuals to comprehend the information about genetic counseling and testing.

The results of this study suggest that the GPSI is a useful research questionnaire. It is important to note some of the limitations and strengths of this study. First, we did not assess personal history of genetic testing, nor did we ascertain whether respondents' had relatives or friends who had undergone genetic testing. Actual experiences with genetic testing may influence perceptions of genetic testing. Second, this convenience sample of community participants may include some self-selection bias. Despite these limitations, this is one of the few studies that evaluate the perceived importance of genetic testing for alcohol dependence compared with other medical conditions among a community sample of African Americans. Our initial sample size included over 300 African Americans within a robust age range. Furthermore, this study's strength is that important findings about the high level of interest in ge-

netic testing and medical research among African Americans. Future directions may include (1) improving knowledge of and access to genetic testing as an important goal for achieving equity in health care; (2) efforts to tailor and market education about genetic testing to the diversity within communities; and (3) providing genetic counseling and testing for alcoholism.

In conclusion, our current understanding of the beliefs and perceptions of genetic testing for susceptibility to alcoholism is limited in literature, especially among ethnic minorities. Patients who consider genetic testing need to have an accurate understanding and know the benefits and limitations (Richards and Ponder, 1996; Lanie *et al.*, 2004). An understanding of elements associated with beliefs and perceptions of risk in genetic testing for susceptibility to alcohol use disorders is important for providing genetic counseling, education, and testing services.

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Disclosure Statement

The authors have no conflicts of interest to disclose.

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