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Public Perspectives About Pharmacogenetic Testing and Managing Ancillary Findings

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Aims: Pharmacogenetic (PGx) tests are intended to improve therapeutic outcomes through predicting a patient's likelihood to respond to or experience an adverse effect from a specific treatment. In addition, PGx testing may also generate ancillary, or incidental, disease information unrelated to the purpose for which the test was ordered. To assess public attitudes toward PGx testing, ancillary disease risk information and related clinical issues, we conducted a series of focus groups. *Results:* Forty-five individuals recruited from Durham, NC, participated in four focus groups. Overall, participants were enthusiastic about PGx testing, though expressed concerns about privacy, confidentiality, and psychological harms associated with ancillary information. Focus group participants believed that physicians had a responsibility to disclose ancillary risk information, but were concerned about managing and coping with unexpected disease risk information. *Conclusion:* We find that participants welcomed the integration of PGx testing into therapeutic decision-making. Public concerns about PGx testing and ancillary information specifically centered on personal implications of learning such additional information, suggesting that patient–provider discussion of the benefits and risks of testing will be necessary until public familiarity with these tests increases.

Introduction

Pharmacogenetic (PGx) testing is considered to be one of the most promising clinical applications in personalized medicine, with the potential to improve therapeutic outcomes through reduction of adverse drug responses and increased likelihood of response. In comparison to disease-based genetic tests, PGx tests are generally believed to have fewer ethical and social implications (Roses, 2000). However, they may potentially reveal disease risk information unrelated to the drug therapy question for which the test was ordered (Netzer and Biller-Andorno, 2004; Haga and Burke, 2008; Henrikson *et al*., 2008). This additional, or ancillary, clinical information may relate to disease susceptibilities, prognosis, or other drug responses.

Several studies have explored public attitudes about PGx testing, though none have considered the issue of ancillary information. Focus group studies have reported that participants preferred individualized genetic testing versus race-based medications but raised concerns about cost, privacy, and discrimination (Bevan *et al*., 2003; Almarsdottir *et al*., 2005). In addition, some members of the public were likely to be highly suspicious of the safety and efficacy of race-based drugs (Condit *et al*., 2003; Bates *et al*., 2004). Survey studies indicated public support for PGx testing, but concerns were raised about patient sovereignty, the unavailability of suitable drugs based on their genetic makeup, cost of tailored drugs, impact on health disparities, and genetic privacy (Rothstein and Hornung, 2003; The Royal Society, 2005; Rogausch *et al*., 2006). We recently reported findings from a national survey that most respondents were interested in PGx testing, particularly those that had experienced an adverse event, but shared similar concerns about privacy and confidentiality of both the test result and DNA sample (Haga *et al*., 2011a).

To gain a better understanding of the views of the general public regarding PGx testing and specifically ancillary information, we conducted a series of focus groups. In particular, we aimed to explore the public's attitudes about their interest in PGx testing, the impact of ancillary information, and sharing of PGx test results amongst healthcare professionals. These data will provide greater understanding of potential barriers to uptake of PGx testing from the patient perspective that will be critical to address as PGx testing becomes more widely available.

Materials and Methods

Study population

Participants were recruited from community locations across Durham, NC, through advertisements in community newspapers, flyers posted in public areas, and word-of-mouth.

Institute for Genome Sciences and Policy, Duke University, Durham, North Carolina.

A meal and $25 were provided as compensation for participation in the focus group. The study was approved by the Duke University Health System Institutional Review Board.

Focus group design

A moderator guide was developed to ensure consistency of the material presented and questions asked between focus groups. Questions were intended to guide participants toward formulation of informed opinions regarding PGx testing and to elicit their reasoning. A hypothetical vignette written at an eighth-grade reading level was used to illustrate potential clinical and ethical issues that may arise with PGx testing, particularly regarding ancillary information.

Focus groups

Four focus groups of the general public were held between January and March 2009 at locations across Durham. Consent was obtained from participants upon arrival. Participants were asked to complete a demographic questionnaire at the beginning of the session. Each focus group discussion was audio-recorded and transcribed.

Data analysis

Transcripts were first analyzed for accuracy and completeness prior to data analysis. We used the software NVivo 8.0 (QSR International) to partition the transcripts according to sections of discussion dictated by the moderator guide. Themes were independently identified by each author; consensus was reached on the themes through discussion amongst the authors. The themes were used to code similar responses and opinions voiced by participants in each focus group; transcripts were independently coded by two authors (S.B.H. and G.T.). Disparities in coding were resolved through discussion and reanalysis of the relevant sections of the transcript. This analytical approach allowed for comparative interpretation of concerns, issues, and opinions between groups.

Results

Characteristics of focus group discussants

Forty-five individuals participated in four focus groups of the general public. Participants were predominantly female and African-American (76%), with a median age group of 40–49 years (Table 1). Educational status ranged from a high-school education or less (13%) to a Bachelor's or graduate degree (58%). The make-up of each group represented a mix of backgrounds with respect to age, gender, race, and education.

General interest in PGx testing

Overall, participants were interested in PGx testing and recognized the immediate benefit to improve drug therapy outcomes. Many participants indicated they would be interested in testing either to predict the risk of serious adverse events (less so for mild adverse events) or to learn of their likelihood to respond favorably to a particular drug (Table 2). Several participants expressed disappointment that PGx testing might not be able to specifically determine for which side effects they would be at risk. Additional concerns included cost of the test, insurance coverage, the predictive value of the test, the time required for testing to be completed, test feasibility in an urgent care situation, and follow-up steps needed based on the test result.

Table 1. Characteristics of Focus Group Participants

	General public (n = 45) (%)
Female	34 (76)
Race	
African-American	30 (67)
White	14 (31)
American Indian/Alaskan Native	1 (2)
Asian	0 (0)
Age	
18–29 years	6 (13)
30–39	6 (13)
40–49	14 (31)
50–59	9 (20)
60–69	7 (16)
70+	2 (5)
No response	1 (2)
Education	
HS or less	6 (13)
Some college/AA	13 (29)
BA or higher	26 (58)

HS, high school; AA, associate's degree; BA, bachelor's degree.

- "I would [have] the testing done to determine the best medication—the medication that is best for you based on your genetic makeup." [Female FG#2]
- "I think I would take the test just to know more about the side effects, just to be prepared and to know my risk." [Female FG#2]
- "Right, if it comes to me or family members or whatever, and they get into a crisis like going into a death situation, who has time to try to figure out what medications is going to work on this person and what's not gonna work?" [Female FG#3]

Obligation to disclose ancillary information

In our overview about PGx testing, we defined ancillary information as "extra information such as your risk of developing a disease," and that it was most likely "not related to your current health." We then presented a hypothetical scenario about a patient who has PGx testing to predict her risk of a serious adverse effect associated with a specific asthma medication being considered. The PGx test could also reveal her risk of colon cancer.

Compared with the attitudes toward PGx testing in general, there was greater agreement of opinions between groups regarding ancillary information (Table 2). When asked whether or not they believed that physicians had an obligation to disclose the presence of ancillary risk information revealed by a PGx test, most participants agreed that they did:

- "I think it's important to create awareness, as if a person had that information. You're not saying they have the specific illness or disease, but if they have that information as opposed to not giving them that information

Table 2. Issues Raised by Focus Groups About Their Attitudes Toward Pharmacogenetic Testing and Potential Ancillary Information

Attitudes toward PGx testing	*1*	*2*	*3*	*4*
Recognized benefit in learning personal risk for side effects	X	X		X
Recognized benefit in testing to optimize effective drug selection			X	
Would reduce trial-and-error approach to treatment				X
Cost-savings		X	X	
Concern about cost/insurance coverage	X	X		X
Concern about turn-around time of testing				X
Not feasible in urgent care situation				X
Concern about test accuracy and reliability, and predictive value	X			X
Concern about needing testing for every new drug prescription			X	
Attitudes toward ancillary information	*1*	*2*	*3*	*4*
Perceived obligation of clinician to inform patient about potential incidental information prior to testing	X		X	X
Benefit of learning of incidental risk information is opportunity to take preventive measures		X	X	X
Concern about insurance implications, specifically discrimination	X	X	X	
Concern about potential need for further follow-up testing or new treatment	X		X	
Concern of psychological harms, anxiety, and stress associated with learning of incidental risk information	X	X	X	X
Would not want to know incidental risk information for untreatable diseases	X	X	X	
Would want to know incidental risk information for any type of disease	X	X	X	X
Concern about familial implications		X		
Privacy/confidentiality concerns	X		X	

PGx, pharmacogenetic.

and they find out 5 years down the road that they were at risk and they could have had some preventative measures, made some preventative measures, they could come back and say, 'why didn't you tell me?' You had the data in front of you. Had I know[n], I could have done A, B, C or D, which may not have required medication. It could have just been some lifestyle changes." [Female FG#3]

- "There are lots of people who don't know their family history and genetic tests, they may help open up some doors to conversation that about other things they have going on health-wise." [Female FG#2]

Interest in learning of ancillary information

Although the majority of participants felt that their physician should disclose the possibility of ancillary information, their desire to actually learn of that information varied between participants within each group. Some indicated that they would want to learn of any type of ancillary information revealed by a PGx test, whereas others were interested in learning of risks for diseases only with preventable actions. Ancillary information was also considered by some to be beneficial, particularly if it motivated people to adopt healthy lifestyles to prevent disease onset.

- "No matter what it was, if I was prone to get cancer or something–change your lifestyle or eating habits–and I think I would want to know especially if there is a known history in my family." [Female FG#2]
- "A lot of things that we get, especially as we get older, that are preventable, if we had just known they were coming." [Female FG#3]

In contrast, others were less inclined to want to learn of the ancillary risks because of concerns about anxiety and potential psychological harm and insurance discrimination as these issues were mentioned by almost all of the groups. The introduction of unexpected disease risk information was viewed by some to be overly burdensome.

- "I've got to worry about my insurance going up, not worrying about whether or not I'm gonna live for a few more years. I've just gotta worry about I'm gonna have to pay a few more dollars." [Male FG#3]
- "… what if the information is something dreadful that they can't do a damned thing about?.…I wouldn't want to know that, and I would certainly want to know in advance if that was a possible piece of information, especially given the insurance considerations." [Female FG#1]
- "I don't want to know if I don't have a strong family history–don't tell me. If I am coming in there for a cold, you treat me for a cold. I don't want to go out of there with high blood pressure, something else to be worried about.… I am already not feeling well, so then you gonna tell me about this? This is gonna upset me more. [When] I go in there for a physical, let me know." [Female FG#2]

If interested, participants favored receiving the ancillary risk information from their primary care provider (PCP) with whom they had an established relationship and who was familiar with their health history. Some, however, preferred to see a specialist with appropriate expertise in the particular disease for follow-up care or a genetic counselor for help with managing the new information.

- "I would like to get it from my primary care physician mainly because they have the whole picture and they know all the other things that are going on with you. But at the point where their expertise ends, I would like for them to refer me to the specialist so I am getting more information if that is what I want." [Female FG#2]
- "I think in general though, I think I would prefer a counselor in addition to a doctor just for follow-up care, especially if it is some sort of disease that could be very detrimental to hear, because one never knows." [Female FG#2]

Storage of PGx test results

Participants also recognized the value of convenient access to PGx test results and that test results should be stored somewhere easily accessible to other healthcare providers as needed. Pharmacists, in particular, may benefit from having access to a patient's PGx test results. Although participants acknowledged this, they were more hesitant regarding pharmacist access to both the PGx test results and any potential ancillary risk information.

- "I mean it would be weird you going into a pharmacy and you got a cold and she would know that there is a higher increased risk for colon cancer …" [Male FG#2]

Discussion

The promise of PGx tests to improve outcomes through tailored drug treatment based on patients' genetic risk of adverse effects and/or likelihood of drug response also comes with its own side effects, potentially revealing additional disease risk information unrelated to the drug therapy. As such, the delivery and management of PGx tests may be challenging, warranting careful consideration of stakeholder views. In this study, we explored public attitudes and interest toward PGx testing in general as well as ancillary information related to PGx testing. Similar to other reports of public attitudes toward PGx testing in general (Rogausch *et al*., 2006; Fargher *et al*., 2007; Haga *et al*., 2011a), interest among our participants was positive, but participants were cognizant of potential risks associated with testing. In particular, similar concerns related to disease-based genetic testing were raised in most focus groups, such as privacy and confidentiality and psychological harms of test results.

Given the number of African-American participants in our study, we reviewed the literature of attitudes and use of genetic testing in African-Americans, particularly of African-American women. Reports have documented the limited uptake of genetic services by African-American women (Armstrong *et al*., 2005). Discrimination is a common concern reported in studies of African-Americans, both racial and other forms of discrimination as well as concerns of privacy and confidentiality (Laskey *et al*., 2003; Bates *et al*., 2004; Sussner *et al*., 2011). Lower uptake may be due to lower knowledge and/or perceived disease risk (Halbert *et al*., 2005; MacNew *et al*., 2010; Akinleye *et al*., 2011; Long *et al*., 2011), although interest in some types of genetic testing appear high (Kessler *et al*., 2005; Long *et al*., 2011), particularly in individuals with a family history or high perceived risk (Kinney *et al*., 2001; Kessler *et al*., 2005).

In our study, we also found a high level of concern regarding about the potential for discrimination and the practical issues of cost and follow-up care. Although most participants believed that physicians had a responsibility to disclose at least the potential of ancillary risk information, they was widespread concern about the personal implications of this information, notably about managing and coping (emotionally and financially) with unexpected disease risk information. The ensuing discussions indicated that although participants recognized they may struggle with the interpretation of the result, they are able to decide, based primarily on their personal circumstances, what would be best for them regarding learning this additional risk information. Further, it was not certain whether the potential for ancillary risk information would dissuade participants from undergoing PGx testing. However, in a national public survey, we found that respondents were still interested in PGx testing even if the test could reveal ancillary disease risk information (Haga *et al*., 2011b).

This study has some limitations, which should be noted. Given the small sample size and recruitment from one region, the opinions of our focus group participants may not be representative of the general public. Thus, findings from this preliminary study will also need to be confirmed through larger studies in other geographic locales and medical specialties. In addition, responses to hypothetical scenarios are often positively biased (Persky *et al*., 2007), and therefore, further studies are also needed to test the impact of real clinical situations.

Given public familiarity and experience with adverse events and nonresponse, it is understandable that attitudes toward PGx testing have been reported to be positive. However, it remains to be seen whether the general interest will translate into actual uptake of testing. Public uptake will likely depend largely on the delivery of PGx testing, particularly on patient–provider discussions about the benefits and risks of testing in order to ameliorate concerns and/or confusion. From this pilot study, we find that participants welcomed the integration of PGx testing into therapeutic decision-making and were able to decide, primarily based on their personal circumstances, how to manage potential ancillary risk information.

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References

Akinleye I, Roberts JS, Royal CD, *et al.* (2011) Differences between African American and White research volunteers in their attitudes, beliefs and knowledge regarding genetic testing for Alzheimer's disease. J Genet Couns [Epub ahead of print]; DOI: 10.1007/s10897-011-9377-6.

Almarsdottir AB, Bjornsdottir I, Traulsen JM (2005) A lay prescription for tailor-made drugs—focus group reflections on pharmacogenomics. Health Policy 71:233–241.

Armstrong K, Micco E, Carney A, *et al*. (2005) Racial differences in the use of BRCA1/2 testing among women with a family history of breast or ovarian cancer. JAMA 293:1729–1736.

Bates BR, Poirot K, Harris TM, *et al*. (2004) Evaluating direct-to-consumer marketing of race-based pharmacogenomics: a focus group study of public understandings of applied genomic medication. J Health Commun 9:541–559.

Bevan JL, Lynch JA, Dubriwny TN, Harris TM, *et al*. (2003) Informed lay preferences for delivery of racially varied pharmacogenomics. Genet Med 5:393–399.

Condit C, Templeton A, Bates BR, *et al*. (2003) Attitudinal barriers to delivery of race-targeted pharmacogenomics among informed lay persons. Genet Med 5:385–392.

Fargher EA, Tricker K, Newman W, *et al*. (2007) Current use of pharmacogenetic testing: a national survey of thiopurine methyltransferase testing prior to azathioprine prescription. J Clin Pharm Ther 32:187–195.

Haga SB, Burke W (2008) Pharmacogenetic testing: not as simple as it seems. Genet Med 10:391–395.

Haga SB, O'Daniel JM, Tindall GM, *et al*. (2011a) Survey of US public attitudes toward pharmacogenetic testing. Pharmacogenomics J [Epub ahead of print]; DOI: 10.1038/tpj.2011.1.

Haga SB, O'Daniel JM, Tindall GM, *et al*. (2011b) Public attitudes toward ancillary information revealed by pharmacogenetic testing under limited information conditions. Genet Med 13:723–728.

Halbert CH, Kessler LJ, Mitchell E (2005) Genetic testing for inherited breast cancer risk in African Americans. Cancer Invest 23:285–295.

Henrikson NB, Burke W, Veenstra DL (2008) Ancillary risk information and pharmacogenetic tests: social and policy implications. Pharmacogenomics J 8:85–89.

Kessler L, Collier A, Brewster K, *et al*. (2005) Attitudes about genetic testing and genetic testing intentions in African American women at increased risk for hereditary breast cancer. Genet Med 7:230–238.

Kinney AY, Croyle RT, Dudley WN, *et al*. (2001) Knowledge, attitudes, and interest in breast-ovarian cancer gene testing: a survey of a large African-American kindred with a BRCA1 mutation. Prev Med 33:543–551.

Laskey SL, Williams J, Pierre-Louis J, *et al*. (2003) Attitudes of African American premedical students toward genetic testing and screening. Genet Med 5:49–54.

Long KA, Thomas SB, Grubs RE, *et al*. (2011) Attitudes and beliefs of African-Americans toward genetics, genetic testing, and sickle cell disease education and awareness. J Genet Couns [Epub ahead of print]; DOI: 10.1007/s10897-011-9388-3.

MacNew HG, Rudolph R, Brower ST, *et al*. (2010) Assessing the knowledge and attitudes regarding genetic testing for breast cancer risk in our region of southeastern Georgia. Breast J 16:189–192.

Netzer C, Biller-Andorno N (2004) Pharmacogenetic testing, informed consent and the problem of secondary information. Bioethics 18:344–360.

Persky S, Kaphingst KA, Condit CM, *et al*. (2007) Assessing hypothetical scenario methodology in genetic susceptibility testing analog studies: a quantitative review. Genet Med 9:727–738.

Rogausch A, Prause D, Schallenberg A, *et al*. (2006) Patients' and physicians' perspectives on pharmacogenetic testing. Pharmacogenomics 7:49–59.

Roses AD (2000) Pharmacogenetics and the practice of medicine. Nature 405:857–865.

Rothstein MA, Hornung CA (2003) Public attitudes about Pharmacogenomics. In: Rothstein MA (ed) Pharmacogenomics: Social, Ethical, and Clinical Dimensions. Wiley-Liss., NJ, pp 3–27.

The Royal Society (2005) Pharmacogenetics dialogue. Available at http://royalsociety.org/2004–2005-Dialogue-Pharmacogenetics/

Sussner KM, Edwards TA, Thompson HS, *et al*. (2011) Ethnic, racial and cultural identity and perceived benefits and barriers related to genetic testing for breast cancer among at-risk women of African descent in New York city. Public Health Genomics 14:356–370.

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