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Preferences for and acceptability of receiving pharmacogenomic results by mail: A focus group study with a primarily African American cohort

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Abstract

Although genetic counseling is traditionally done through in-person, one-on-one visits, workforce shortages call for efficient result return mechanisms. Studies have shown that telephone and in-person return of cancer genetic results are equivalent for patient outcomes. Few studies have been conducted with other modes, result types or racially diverse participants. This study explored participants' perspectives on receiving pharmacogenomic results by mail. Two experienced moderators facilitated six focus groups with 49 individuals who self-identified primarily as African American and consented to participate in a genome sequencing cohort study. Participants were given a hypothetical pharmacogenomic result report (positive for c.521T>C in *SLCO1B1*).

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Author Contributions

Authors PAC and ET confirm that they had full access to all the data in the study and take responsibility for the integrity of the data and the accuracy of the data analysis. All of the authors gave final approval of this version to be published and agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

1.Conflicts of Interest

LGB is an uncompensated member of an advisory committee of the Illumina Corp, receives in-kind research support from ArQule, Inc. (now wholly owned by Merck, Inc.), and honoraria from Cold Spring Harbor Press.

PAC has no conflict of interest to disclose.

KLL has no conflict of interest to disclose.

BBB has no conflict of interest to disclose.

LHE has no conflict of interest to disclose.

GAF has no conflict of interest to disclose.

SE has no conflict of interest to disclose.

ET has no conflict of interest to disclose.

2.Human Studies and Informed Consent

The National Human Genome Research Institute institutional review board approved of this study. All procedures were in accordance with the standards of the responsible committee on human experimentation (institutional and national) and the U.S. Code of Federal Regulations 45 CFR 46. Informed written consent for focus group participation was obtained from all participants involved prior to the groups.

3.Animal Studies

No non-human animal studies were carried out by the authors for this article.

An accompanying letter explained that the result was associated with statin intolerance along with a recommendation to share it with one's doctor and immediate relatives. Participants reacted to the idea of receiving this type of result by mail, discussing whether the letter's information was sufficient and what they predicted they would do with the result. Two researchers coded the focus group transcripts and identified themes. Many participants thought that it was appropriate to receive the result through the mail, but some suggested a phone call alerting the recipient to the letter. Others emphasized that although a letter was acceptable for disclosing pharmacogenomic results, it would be insufficient for what they perceived as life-threatening results. Most participants found the content sufficient. Some participants suggested resources about statin intolerance and warning signs be added. Most claimed they would share the result with their doctor, yet few participants offered they would share the result with their relatives. This exploratory study advances the evidence that African American research participants are receptive to return of certain genetic results by approaches that do not involve direct contact with a genetic counselor and intend to share results with providers.

ClinSeq: A Large-Scale Medical Sequencing Clinical Research Pilot Study (NCT00410241)

Keywords

Communication; Pharmacogenomics; Scalable; African Americans; Genetic Counseling

Introduction

With the advancements of precision medicine, more people are undergoing genomic testing. In-person and telephone appointments are still the most widely used genetic counseling methods (National Society of Genetic Counselors, 2020). However, some researchers have predicted that there is an insufficient number of genetic counselors to meet this demand (Hoskovec et al., 2018; Villegas & Haga, 2019). Geographical challenges further affect access to genetic counseling services (Penon-Portmann et al., 2020). These advances in genomics and challenges in accessing genetic counseling services call for alternatives to traditional in-person and telephone genetic counseling. Return of results is one component of the genetic counseling process that can be studied to develop less resource intensive result return strategies. Pharmacogenomic (PGx) results are a logical context for assessing these modes because they are common and clinically significant (Chanfreau-Coffinier et al., 2019; Reisberg et al., 2019) but not life-threatening. For example, statins are widely prescribed (Gu et al., 2014), and individuals with the c.521T>C variant in *SLCO1B1* have a 2-4.5x increased risk to develop myopathy from taking statin medications to lower their cholesterol (Link et al., 2008).

For post-test disclosure genetic counseling broadly (outside of the context of PGx), telephone genetic counseling has been the most widely studied alternative to in-person genetic counseling (Athens et al., 2017). In cancer genetics, return of results by telephone genetic counseling has been shown to be non-inferior to in-person genetic counseling on many outcomes including knowledge, quality of life, decisional conflict, cancer-specific distress, anxiety, and perceived personal control (Kinney et al., 2016; Schwartz et al., 2014). Telephone genetic counseling to return Alzheimer's disease results has been shown to be non-inferior to in-person results delivery in terms of patient anxiety, depressive symptoms,

and test-related distress outcomes (Christensen et al., 2018). Overall, these three studies suggest that return of results by telephone genetic counseling results in similar patient outcomes to in-person genetic counseling for some types of results and in predominantly White cohorts. Given the lack of diversity in these research studies, more research is needed to assess whether these conclusions hold true for minority populations, such as African Americans. For example, one study showed that minority women have lower uptake of telephone genetic counseling and *BRCA1/2* testing compared to White women (Butrick et al., 2015).

There are several studies on alternative modes of PGx results delivery. One randomized control trial compared returning *SLCO1B1* results by electronic health record (EHR) to not returning results. Participants with results in their EHR had improved statin reinitiation and larger decreases in low-density lipoprotein cholesterol (Peyser et al., 2018). Thus, return of PGx results through the EHR may be sufficient for promoting recommended health behaviors and beneficial health outcomes. However, some participants may prefer to receive PGx results outside of the EHR. For example, participants in the Geisinger MyCode study were varied in their responses about PGx result delivery preferences with equal proportions preferring results be mailed, placed in the EHR, or mailed and placed in the EHR (Jones et al., 2018).

A key component of successfully returning results outside of in-person genetic counseling sessions is optimizing the written materials provided about the result. For PGx results specifically, a small interview study with clinicians and patients suggested that the report should contain information on how the genotype affects current and future medication metabolism in a way that is patient-friendly (Jones et al., 2018). However, standardized wording does not exist (Haga, Mills, & Bosworth, 2014). According to Goehringer and colleagues, the report can be worded clearly without altering the meaning of the result (e.g., “positive result” conveys the same meaning as “pathogenic result” but is more readily understood) (Goehringer et al., 2018). Also where possible, wording that minimizes stigmatization should be used (e.g., “non-working” rather than “defective” or “mutant,” and “variant” rather than “mutation”) (Haga, Mills, & Bosworth, 2014). Haga, Mills, Pollak, and colleagues have also suggested result report options that adhere to federal and professional guidelines that regulate the information required in genetic testing reports while improving their comprehensibility (Haga, Mills, Pollak, et al., 2014). When developing written materials, it is essential to engage with recipients for input regarding appropriateness, acceptability and clarity.

In developing materials to communicate genomic sequencing information, it is important to have a diversity of perspectives to ensure that the outcomes are broadly generalizable. However, the participants in the aforementioned studies were mostly White. We conducted a series of focus groups with participants in a genomic sequencing cohort who self-identified as African American, Black, Afro-Caribbean or African (referred to throughout as “primarily African American participants”). We aimed to fill gaps in the literature about how these individuals want to engage in genomics research, their perspectives on return of results and the extent to which focus groups were an effective tool for gathering data on participant return of result preferences.

Study Aim

This study explored African American individuals' perspectives on a hypothetical scenario of receiving PGx results by mail. Focus group participants were asked about the appropriateness of the delivery mode and content, and their intentions to share results with their healthcare provider and relatives.

Methods

We selected focus groups to capture the details and nuances of participant perspectives. A constructivist perspective guided the study, which acknowledges the existence of multiple realities (Braun et al., 2019).

Participants

A purposive sampling strategy was used in that focus group participants were enrolled in the ClinSeq® study (NCT00410241, 07-HG-0002) and originated from the cohort of 467 mostly healthy adults who self-identified as African, African American, or Afro-Caribbean (Lewis et al., 2019). Participants previously consented to genome sequencing, return of results when available, and re-contact for future studies. The National Human Genome Research Institute institutional review board reviewed this study and approved it as human subjects research.

Focus Groups

Participants who were eligible for the focus group study lived locally, had not previously received genomic sequencing results from the ClinSeq® study and did not have a spouse or close friend enrolled in a focus group. Eligible participants were contacted one time by phone to offer participation. They were verbally consented and scheduled for a focus group on a first-come, first-serve basis. Participants were compensated with a $50 gift card. The groups were held locally either on the National Institutes of Health Bethesda campus or in community centers or libraries in the Washington, D.C. area. Six focus groups were held (FG1-FG6), one being all-female (FG2) and one being all-male (FG1), with up to ten participants in each group. The focus groups were conducted by one of two professional moderators using a guide, which asked about a variety of topics relevant to participation in a genome sequencing study. The moderators, one female and the other male, both self-identified as White and not Hispanic or Latino. A note taker (PAC), the second moderator, and a genetic counselor who served as a content expert to answer technical questions (GF) also attended each group. The note taker self-identified as a Chinese American female and the content expert as an African American female. At the commencement of the sessions, moderators disclosed to participants that they were not part of the Clinseq® study.

The findings for this study came from the final section of the focus group sessions. The focus groups explored participants' views and experiences of being involved in a genome sequencing study, motivations for joining the study and preferences for engagement and return of results prior to what is presented here (Lewis et al., 2021). Participants were given a hypothetical PGx report and summary letter (see Supplementary Material) and asked to predict how they would react to receiving this type of result through the mail, to

describe who they would share the result with, and to comment on the clarity and content of the letter. The mock Clinical Laboratory Improvement Amendments (CLIA)-certified report indicated the presence of the variant c.521T>C in *SCLOB1*. The report classified the variant as pathogenic and listed the associated condition as statin-induced myopathy. The letter outlined that the participant was at risk for having side effects to statins and that the participant should share the result with his/her family members and doctor. The summary letter and the questions asked in the guide were developed by several study authors (KLL, LHE, LGB) based on their clinical genomics expertise. The letter was one page long and written in plain language (Flesch Reading Ease 67.6; Flesch-Kincaid Grade Level 7.6) with details omitted (e.g., relative risk for side effects, how the medication is metabolized) to assess whether participants would ask for more information. Participants were given time to read through these materials, after which the moderator facilitated a discussion.

Data Analysis

Each of the sessions was audio-recorded and transcribed. The primary coder (PAC) reviewed the transcription and edited the transcripts in Microsoft Word (Version 16.35) for accuracy before importing them into NVivo 12.3.0 (QSR International Party Ltd) for coding.
The transcripts were coded using a constant comparison approach (Doody et al., 2013; Onwuegbuzie et al., 2009). The primary coder developed the codebook in consultation with the secondary coder (ET). PAC and ET coded all the transcripts using the agreed upon codebook. Where disagreements were identified, the coders discussed and reconciled codes before they were finalized. Common codes were grouped to create categories which were then expanded to generate themes (Braun et al., 2019). All living focus group members were sent a summary of focus group findings and were telephoned to briefly discuss their thoughts about the findings.

Results

Participant Demographics

From the cohort of 467 eligible participants, 179 were contacted and 60 (34%) participants were scheduled for one of six focus groups. Of those, 49 (82%) participants attended a focus group. Most focus group participants had at least a college education (75%), a slight majority were female (59%) and nine (18%) participants were using statins at the time they enrolled in the ClinSeq® study. The majority of focus group participants who responded to an open-ended survey question about racial/ethnic self-identity when enrolling in the ClinSeq® study, stated they were African American and/or Black (36/40). The remainder self-identified as African or Afro-Caribbean. Participants had been enrolled in the Clinseq® study for an average 3.4 years. At the time of their respective focus group, participants were on average 61.3 years old (ranging from 51.2 to 70.9).

Mailed letter considered acceptable for pharmacogenomic results

Many participants thought it was appropriate or “okay” (FG3P6) to receive PGx, or similar results through the mail, for example, noting, “this is exactly what I was hoping for, this is all [I] need” (FG5P5). When the moderator asked, “So how do you feel about getting

this [result] in a letter? Is that okay?" a participant responded, "I think it's okay" (FG3P6). Another noted that a letter was advantageous because it could be easily shared, "In that way I can take the letter to my doctor or whoever and share with them" (FG2P6).

In FG2, several participants agreed when the moderator asked if a letter was "enough" for returning a PGx result. However, after one participant (P2) commented that she wanted a phone call followed by a mailed letter for any kind of result and had understood this to be the ClinSeq® study policy, all went on to agree with that protocol. One participant in FG5 expressed a similar opinion, saying: "That's exactly how I would do this: a phone call saying, 'I'm going to send you this letter that you can take to your doctor.' […] So you expect it and you don't get a shock in the mail" (FG1P5). A few participants raised the idea that mail was not ideal from a privacy perspective. For example, one said that she "doesn't always get [her] mail correctly" (FG2P5). Another said he would prefer secure email for expediency and privacy but would not mind receiving results by mail.

Three groups discussed that, in contrast, it was not appropriate to send higher-risk results (e.g., "variants for colon cancer," "life-threatening") by mailed letter.

> FG4P1: The letter's effective for something on this level [i.e., the PGx result or similar], it would be fine. But I would think, I don't know if I would want to hear something more life-threatening or more or more [sic] of a health concern in the letter in the mail.

In one group, a few participants suggested that if a "life-threatening" result was sent through the mail, the materials should be eye-catching (e.g., "red," "very bold").

> FG5P5: Just something that's really gonna grab your attention immediately, you know. Don't go through the whole letter to find out. Something that just gets you right away that this is life-threatening.

Only one participant voiced that he may panic if he received a result that had any health implications. He likened receiving these materials to his experience with receiving a cholesterol blood test result.

FG4P6: I got a call from the doctor that did the [blood] test. And he said […] it showed my cholesterol was high and […] [I was] going to get more results from [sic] the FedEx. So I was panicked and like, 'Okay, what is going on?' I had this and I took it to the doctor. I was happy to know this, but it's [sic] sort of put you on panic at first.

Moderator: Okay. You didn't feel good about it, but you wanted to know.

FG4P6: Yes, I wanted to know. I needed to know.

Moderator: Would you feel the same way about this letter?

FG4P6: Yes.

Materials were sufficient with few edits

Many participants thought that the letter was enough to act on. For example, “Well, I think also the letter is very clear and I like what it says, ‘We recommend that you share this result with your doctor,’ […] And everything else is here, your contact information is at the top. It seems like it’s very self-explanatory” (FG5P1). When asked by the moderator, “For most of you, is this information clear enough? Is it clear you should share with your doctor and [know] what to do?” four participants responded, “Yes” (FG4). In another group when asked if the letter supplied enough information, a participant responded, “For something this simple, I mean a statin, you know, for a high cholesterol, that’s, this is fine. If it’s something much more serious, you know, we go back to life threatening then maybe not” (FG5P8).

Some participants said the materials did not provide enough information or were hard to understand. A few participants provided insights about what was missing and how to improve the materials. Most commonly, they wanted more information about potential health effects and next steps. Participants specifically wanted to better understand the range in severity of side effects. As one participant said, “I would like a little bit more of a delineation of what’s […] going to happen, you know, if it is mild, what's […] going to happen, you know, if it's severe. Maybe a place to go look for some additional data” (FG6P6). Others suggested providing more resources on statin-induced myopathy. Two participants said that they personally knew how to look up resources and had access to them but raised the point that this may not be the case for everyone.

Finally, a few participants suggested some minor changes to wording or tone. None of these suggestions arose in more than one group. For example, one group wanted the letter to be more directive in recommending that the report be shared with a doctor:

> FG3P2: I think the letter’s fine and the [CLIA report]. The only thing I would probably change is the verbiage. It says, ‘You should tell your doctor right away if you have side effects from taking a statin.’ I think it should be, ‘You should give this [CLIA report] to your physician.’

Would share the materials with their doctor and family

Five of the groups discussed whether they would take the result to their doctor. Some said that this would allow their doctor to use the result for medical management. One participant said, “A doctor might say, ‘Well you know, I don’t want you to take a statin because you’re at risk for having these side effects’” (FG3P1). A few participants planned to ask their doctor to clarify what the result meant.

Moderator: Would the letter leave you with any other questions […]?

FG1P7: I will be peppering the hell out of my doctor.

FG1P2: Well, I follow [FG1P7]'s comment. I think that’s really a follow up with your doctor.

However, a few participants were skeptical about whether their doctors would understand the result.

FG6P2: Okay. Well, I am looking at the report that goes to my doctor. It says, you know, you should confirm certain things before you act on them. So, I'm wondering [about] the validity of the test.

FG6P1: The doctor don't know what that mean [sic].

Only two groups discussed sharing the result with family members. In one group, the moderator asked, "What do you plan to do with the result?" and one participant said she would share the result with her family (FG3P3). In the other group, two participants wanted the study team to facilitate communication of the result to their relatives, so that their relatives could have the correct information and ask any questions. As one of the participants put it, "Well maybe they [my family] should contact NIH [National Institutes of Health] then because I don't want to give them the laymen's explanation. Especially if there's something they need to be talking to their doctor about" (FG5P3). Another participant described that these materials would be effective communication tools for sharing the result with his family. He paralleled it to his experience of sharing non-genetic health-related information and said that he would do the same with these materials.

> This is what I would expect, and I've had an experience with my family where it's been recommended that I see my doctor about certain conditions — my father had it — like I think it's just follow up. (FG5P6)

Discussion

Our study shows that, among our predominantly African American participants, most found it acceptable to receive a PGx result through the mail. This is similar to the findings of a survey study (of mostly White respondents) where most were comfortable with or ambivalent about the idea of receiving genetic testing results (e.g., predisposition, inherited, or carrier) by mail (LaRocque et al., 2015). In our study, a few participants suggested receiving a phone call ahead of the letter to simply let them know that a result would be arriving – not to provide any genetic counseling about the result. The phone call may also be used to identify individuals who want to receive the result from a genetic counselor instead, such as one participant (an outlier in our study) who anticipated that receiving health-related information would make him panic. No other participants mentioned an affective response to these results. Perhaps one reason that the majority of participants were receptive to receiving PGx results by mail is that they do not view PGx results as particularly threatening to their psychological wellbeing. Result disclosure by mailed letter has not been widely studied, though data from our exploratory study suggests that low risk results could be expediently communicated this way.

Overall, our participants described the materials as clear and sufficient for taking to their doctor and managing their health. This is in contrast to a previous study where participants made suggestions to simplify language and include a personalized risk assessment (Olson et al., 2017). In our focus groups, some participants wanted additional information, with a few suggesting more explicit descriptions for mild and alarming symptoms. Other focus group participants in our study suggested the addition of resources which echoes what was found in another study (Jones et al., 2018). Other researchers suggest the inclusion of sections

for “Citations” and “Patient Resources” in genetic reports (Davis et al., 2019). Our study is consistent with other findings in that participants wanted these pieces of information in addition to their genetic result. As major edits to our concise letter were not proposed by participants, this provides some evidence that a brief letter in plain language may be sufficient, potentially with an addendum directing recipients to further resources. Future research could use our developed letter to study the impact of receiving a low risk result in the mail, when compared to other result delivery methods.

Brief and accessible materials may also be valuable to healthcare providers. A recent meta-data analysis found that many healthcare providers have never ordered a pharmacogenomics test, feel they are not well informed about such testing, and lack confidence to use pharmacogenomic results in their practice (Veilleux et al., 2020). To address this issue, researchers are exploring genomic testing reports for non-genetics providers, with early studies demonstrating provider endorsement and effectiveness trials underway (Vassy et al., 2015; Williams et al., 2016).

Participants in our focus groups expressed their willingness to take responsibility for sharing the results with their doctor. A less resource intensive mechanism is deposition of PGx results into the EHR (Hoffman et al., 2014; Kullo et al., 2014; Rasmussen-Torvik et al., 2014). Such a method allows the result to be readily accessible to patients’ healthcare providers who use that particular EHR. However, giving the result directly to patients allows them to manage their health information and share it with providers without access to the specific EHR where the result was deposited (Adler-Milstein et al., 2017; Myrick et al., 2019). Furthermore, with a patient-initiated approach, patients are engaged with their health information, which may serve as a reminder for recommended health behaviors beyond those pertaining to the result at hand (Miller et al., 2019). Future research could expand on prior work to study the effectiveness of a combination of approaches whereby results are deposited in the EHR and provided directly to patients (Williams et al., 2018).

While most participants in our study mentioned they would take the result to their doctors, few raised the intention to share the result with their relatives. Similar to what was reported in other studies (Miller et al., 2019; Smit et al., 2016), one participant mentioned that a letter can be an effective tool to review the information at a later time and to share this information with family members. The letter used in our study was not specifically designed to be shared with family members; however, family communication tools and letters have been developed and used in other studies, which could be used in combination with our letter (Kardashian et al., 2012; van der Roest et al., 2009).

Practice Implications

The practice of returning genetic results to participants has evolved, along with participant expectations. The volume of testing has increased and genetic results are more frequently being returned by modes that do not involve in-person sessions (Phillips et al., 2018). Evidence is needed to inform which patients and/or which results are best delivered during in-person genetic counseling sessions and how to best incorporate other modes of result return . In research settings, there is a growing consensus that individual research results

should be returned (Jarvik et al., 2014; Middleton et al., 2016) and optimization of a return of result pipeline may allow projects to address barriers of time and cost (Bennette et al., 2015). Our results suggest that a brief mailed letter may be one acceptable method for returning medically actionable, low-risk results such as PGx results. Participants may be willing to take responsibility for sharing these results with their doctors. A future optimized strategy might explore the use of multiple low cost methods.

Strengths and Limitations

This study's strength lies in its use of focus groups to elucidate more nuanced perspectives that a survey may not evoke. Our participants were primarily African American, which helps fill gaps in the existing research that was conducted with predominantly White participants. This study provides timely evidence about engaging with members of these populations when improved diversity in genomics is a priority for the field. This study helped us to learn a broader range of participants' perspectives and expectations of return of result methodologies while engaging our participants in a meaningful way. Yet, the study does have several limitations. We held focus groups with a subset of our cohort and the method used may have selected for highly engaged individuals, so these results may not be applicable to the entire cohort. In general, participants had higher education than the general public and were enrolled in a sequencing study (Lewis et al., 2019), so their perspectives may not be generalizable to other study populations or the general public. Also the moderators of the groups were White, which may have affected participants' candor, although the presence of the African American content expert may have mitigated this effect.

In the activity, groups were asked to react to the idea of receiving a hypothetical PGx result through the mail. We studied intention and not actual behavior. Many participants said that if they were to receive a result like this, they would share the results with their doctor, but few talked about sharing the results with relatives. However, not all groups were explicitly asked if they would share the results with relatives.

Lastly, the section of the focus group that was analyzed for the PGx results discussion was at the end and ranged in length from 5 to 13 minutes. The time constraints may have limited the exploration of these topics. While topic saturation was indicated, our ability to confirm that data saturation was reached was limited.

Conclusions

This study demonstrates that African American participants are open to receiving low-risk results, like PGx results, by mail, which may decrease burden on resources for returning results while still promoting desired sharing behaviors. Future research could include comparing a mailed letter to other result delivery methods and returning more result types to other study populations to further assess and broaden this method's applicability.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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4. Data Availability

The data that support the findings of this study are available on request from the corresponding author. The data are not publicly available due to privacy or ethical restrictions.

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What is known about this topic

Past research has shown noninferiority of telephone to in-person genetic counseling, but this paradigm still requires substantial resources. There is a paucity of research about less resource intensive mechanisms, such as a mailed letter, for return of results, specifically in the African American population.

What this paper adds to the topic

Our findings provide evidence that African American participants are receptive to the idea of receiving low-risk genetic results by mail and would be prompted to share the information with their doctor. Future research should directly compare a mailed letter to other result delivery methods.