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"It's not like judgment day": Public understanding of and reactions to personalized genomic risk information

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Abstract

The value of genomic risk assessment depends upon patients making appropriate behavioral changes in response to increased risk leading to disease prevention and early detection. To date, few studies have investigated consumers' response to personalized genomic disease risk information. To address this gap, we conducted semi-structured interviews with 60 adults participating in the Coriell Personalized Medicine Collaborative. The interviews took place after receiving results providing genomic and other risk information for up to eight common complex diseases. We found that participants were most likely to recall results which conferred an increased risk or those of particular personal interest. Participants understood the multi-factorial nature of common complex disease, and generally did not have negative emotional responses or overly deterministic perceptions of their results. Although most participants expressed a desire to use results to improve their health, a minority had actually taken action (behavior change or shared results with their doctor) at the time of the interview. These results suggest that participants have a reasonable understanding of genomic risk information and that provision of genomic risk information may motivate behavior change in some individuals; however additional work is needed to better understand the lack of change seen in the majority of participants.

Keywords

Personalized medicine; Genomic risk; Qualitative research; Public understanding; genetic testing; direct-to-consumer genetic testing; risk assessment; behavioral change

INTRODUCTION

Personalized genomics has the potential to revolutionize healthcare, most notably in the areas of pharmacogenomics and genomic risk assessment for prevention of common complex diseases through behavior modification (Ginsburg & Willard, 2009; Guttmacher et al., 2010). Although personalized genomics has already begun to make its way from bench to bedside, or at least from bench to home computer, there are many barriers to the successful implementation of this new form of genetic testing. Among the most concerning is absence of clinical utility research on genomic testing. Clinical utility may be limited

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because the single nucleotide polymorphisms (SNPs) that have been identified to date represent only a small fraction of the genetic markers that influence common disease (Yang, et al., 2005), and the relative risk increases conferred by the identified alleles are not large enough to be considered predictive (Hindorff et al., 2009; Kraft & Hunter, 2009). In addition, with most genomic testing for common diseases occurring in the direct-to-consumer (DTC) market, the ability of the public to adequately and accurately interpret their genomic risk is questionable, with the primary risk of misinterpretation being false reassurance or undue distress (Marteau et al., 2005; Ransohoff & Khoury, 2010). Concern about the ability of the public to interpret genomic data is compounded by the risk that genomic data, delivered directly to the public, will not be shared with healthcare providers (McGuire et al., 2009) or that genomic testing will lead to an increase in utilization of healthcare resources, perhaps inappropriately (McGuire & Burke, 2008; (Heshka et al., 2008).

Despite the current limitations of personalized genomics and the concerns of the scientific and medical communities, there is considerable public enthusiasm over personalized genomics and the promises it holds including early detection of disease; efficient and more effective drug therapeutics; disease prevention through targeted behavior modification; and more appropriate disease screening (Gollust et al., 2011; Grant et al., 2009; Issa et al., 2009; McGuire et al., 2009). In fact, improving health is the main motivation cited by people undergoing genomic risk assessment (Gollust et al., 2011; Su, et al., 2011). However, to date, most studies assessing the impact of genetic risk information on health behaviors have failed to show that genetic information leads to significant behavioral or lifestyle changes aimed at improving health (Bloss et al., 2011; Kaufman et al., 2010; Marteau et al., 2010). With growing public interest in personalized genomics yet limited data concerning public interpretation of the complex risk information (Leighton et al., 2011), scientists, policy makers and ethicists have all emphasized the need for empirical research into the psychosocial and behavioral impact of personal genome assessment (Cameron & Muller, 2009; Collins et al., 2003; Fackler & McGuire, 2009; McBride et al., 2010).

To address the current dearth of knowledge regarding the interpretation and use of personalized genomic information, we conducted a qualitative study of a subset of participants in the Coriell Personalized Medicine Collaborative (CPMC), a large prospective study that offers personalized genomic, family history and other non-genetic risk estimates for selected medically actionable diseases. The current qualitative study was designed to gain a comprehensive understanding of CPMC participants' interpretation of their results, and what the impact of their results has been on their lives and on their state of health.

METHODS

Study Population

Interview participants were recruited from the larger Coriell Personalized Medicine Collaborative (CPMC) research participant pool. The study design, methods for estimating and reporting disease risk, and recruitment methods for the CPMC at large are described elsewhere (Keller et al., 2010; Stack et al., 2011). Briefly, the CPMC is a longitudinal prospective research study that aims to assess the impact that disease risk assessment has on behavior and ultimately on health outcomes. CPMC participants must be at least 18 years old, have a valid email address, attend an in-person informed consent session to consent to participate in the study, and provide a saliva sample for genomic analysis. Participants are asked to complete a series of online questionnaires about their medical history, family history and lifestyle through the study's secure web portal, as well as disease-specific outcome questionnaires following results disclosure for each condition.

Participants are invited to view their results, as well as educational materials through a secure web-based portal. Unlike direct-to-consumer companies and other observational research studies, the CPMC provides participants with estimates of their relative risk of disease based on medical history, family history, lifestyle and other non-genetic contributions to risk in addition to their relative risk due to the presence or absence of specific genetic variants (Keller et al., 2010; Stack et al., 2011). For example, a participant carrying two genetic risk variants for coronary artery disease might be told he has a relative risk of 1.7 based on genotype, a relative risk of 1.2 based on family history, and a relative risk of 2.1 based on smoking history; no combined multifactorial risk number is provided. Importantly, given the genotype frequency of the variants tested, nearly all CPMC participants receive results indicating that they are at increased risk for at least one disorder based on genotype. In addition to providing personalized results, the reports generated for participants provide general educational information including a written description and visual image of the heritability of each condition. Also in contrast to some DTC genetic testing companies, the CPMC reports results only for conditions which an outside advisory committee have deemed 'potentially medically actionable', meaning there is a specific medical or behavioral intervention that may mitigate the risk of disease or improve the disease outcome.

Recruitment

To recruit for this study, e-mails were sent to 2073 CPMC participants who had received at least one result at the time of mailing. The e-mails notified CPMC participants of an additional research opportunity, and directed them to log in to the secure CPMC web portal to learn more. Additional information about the project was posted on the "optional studies" page of the CPMC web portal, including a link to obtain more information. Individuals who requested more information received an automatically generated e-mail from the study coordinator with additional information about the study, as well as a link to access the online screening survey, administered through Survey Monkey. The online screening survey required participants to read the informed consent and confirm their desire to participate in the study before proceeding with the remaining questions. The screening survey contained questions about age, sex, educational background, racial and/or ethnic identity, marital status, number of children, health insurance coverage and contact information. All completed screening surveys were reviewed by the study coordinator to verify eligibility. To qualify for the interview, CPMC participants had to be at least 20 years of age, speak English, and must have received at least one result at the time they completed the survey. The results from the CPMC risk assessment, including genomic test results, were not made available to investigators. Interviewees were purposefully selected from among the 191 eligible respondents to ensure a heterogeneous sample by age, sex, racial/ethnic identity, and educational attainment (n=60). The study coordinator communicated with eligible participants via e-mail and phone calls to schedule the telephone interviews, and interviews were scheduled such that at least 3 months had elapsed since the participants received their first result. At the time the interviews were conducted, CPMC participants had received results for seven conditions: coronary artery disease, diabetes mellitus type II, prostate cancer, age-related macular degeneration, melanoma, hemochromatosis, and systemic lupus erythematosus.

Interviews

Interviews were conducted by two trained research assistants using a semi-structured interview guide, and averaged 40 minutes in duration. Interviewees were sent a $25 gift card after completing the interview. At the beginning of every interview, the interviewer answered any additional questions about the study and confirmed that the interviewee consented to be interviewed and audio taped. The interview guide contained direct questions

regarding: motivations for participation; the interviewee's description of their results, including how understandable they were; emotional responses to results; what changes they made to lifestyle, behavior, or healthcare management based on results, including the reasons for making changes; whether and why they chose to share their results with family members, friends, and/or health care providers, and their reactions; attitudes toward routine offering of similar testing in a primary care setting; and recommendations for the routine offering of this type of testing through primary care providers. Additional probes were used to ensure an adequate level of detail for each question. All interviews were recorded using a digital audio recorder, and the audio files were subsequently transcribed verbatim and entered into the qualitative analysis software NVivo 8.0 (QSR International Pty Ltd., Doncaster, Victoria, Australia) for coding and analysis.

Data Analysis

Data were analyzed using a phenomenological qualitative approach. Analysis of the data began by members of the research team each reading transcripts to identify domains or broad topics (for example, "recall of results"), and then categories of responses within domains (for example, "recalled all results", recalled no results" and "recalled some results"). Team members met several times to discuss these domains and categories as well as themes emerging from the data (for example, "knowledge is power"). We then developed a detailed code book to include a code for each domain and category of response, as well as codes for emerging themes. Research assistants were trained to code the transcripts using the codebook and several sessions were held to assess and resolve any inter-rater inconsistencies. After training, a subset of 3 transcripts were coded by each of the three research assistants and discrepancies in coding were discussed. An additional 5 transcripts were then coded and intercoder reliability was greater than 90%.

Formal data analysis began by retrieving coded sections of transcripts for each code and analyzing them for salient attributes. We also reviewed coded data to determine whether there were any trends or relationships in findings according participant sociodemographic factors (sex, age, etc) or other attributes. Representative quotations were selected to illustrate important themes in the data. For this paper, we are only reporting on findings relating to understanding of results, reactions to results, behavioral and lifestyle modifications based on results, and sharing results with health care providers.

The study design and all materials were approved by the internal review boards of the Coriell Institute for Medical Research and the University of Pennsylvania.

RESULTS

A total of 60 individuals were interviewed. By design, participants were selected to represent varied age groups, educational levels, sexes and races (Table I).

Result Recall, perception and understanding

Most participants (51/60) were able to recall at least one of the seven results they had received prior to the interview. Although two participants were able to recall all results, most (49/60) recalled only a few results. Of those participants who remembered only a subset of the results reported, more than half remembered their results based on an elevated risk; while a minority attributed recall to a specific disease interest based on their family or personal medical history.

> *"I only remember those two or three: heart disease, AMD, and maybe type 2 diabetes because those were ones that were at increased risk."* (CPMC #29, male)

> *"Lupus was of interest to me because at one point many many years ago I was having a number of symptoms of that…I have one risk and one non-risk."* (CPMC #112, female)

Although results included personalized risk information based on genetic variants, family history and, when possible, lifestyle, when asked what they recalled about their results, participants tended to mention their genetic result first or put more emphasis on this result as compared to other risk factors.

> *"Even though my [non-genetic] risk factor may be higher for diabetes because my BMI is higher than actually what the numbers are for my genetic risk, it's interesting because I still… I guess in my mind I focus even a little bit more on the genetic factors".* (CPMC #103, female)

When asked what they recalled about their results, about one-half of participants used adjectives to describe their level of risk (e.g., high, low, or normal). The other half of participants discussed their risk in terms of their emotional response to results (e.g., reassured, surprised, worried). More men discussed results in terms of level of risk compared to women (54%; n=13 vs. 39%; n= 14).

Although participants were given specific relative risk values for each risk factor and were also presented with a graph illustrating these numeric risks, participants appear to have been more comfortable interpreting their risk in a dichotomous fashion (elevated versus not elevated), rather than reflecting on the actual risk number provided.

> *"Well I forget exactly how it was broken down as your risk factors whether it was ethnicity or male or female but I do remember that it was a lower risk so I don't have the percentages or anything like that but I do know that it was lower."* (CPMC# 173, female)

Two participants expressed confusion about their results, either because they didn't understand how the results were presented, or because they didn't understand the term variant. Some participants did express difficulty understanding relative risk.

> *"The 1.3, I believe that means I'm 30% more likely than other people, or am I three times more likely than other people? That particular number means very little to me."* (CPMC # 92, female)

The majority of participants (40/60) appeared to understand the multi-factorial nature of the complex diseases for which results were presented, and most believe that they have some measure of control over whether they would develop a disorder, even if they were at increased genetic risk. Therefore, very few expressed fatalistic views.

> *"There's some people out there that if they were to find out that they had this, they're like "Oh my God" and they're the ones that can't go to sleep at night thinking "Oh my God! When am I gonna get this?" I think that's just telling you this is what's in your genes, but even though you may have two variant genes, there's a higher possibility for you to get it. But maybe because of their lifestyle, or lifestyle changes that you can put into effect, that might help reduce it. So I mean I think they would have to be trained on that. Yeah, it's not like judgment day."* (CPMC #91, female)

> *"When I read more of the information where it said how much is lifestyle, I think it was like almost fifty percent was lifestyle and fifty percent was genetics, that was my recollection…. It's not something that I'm just hopeless and have no choice in."* (CPMC #86, female)

Emotional Response

When participants were asked about their emotional response to the results provided, the most common emotional response was reassurance or acceptance (53/60). Some people accepted their genetic results because the results aligned with their pre-conceived notion of risk based on their family or personal medical history.

> *"There have been people in my family who have gotten heart disease or they have had diabetes. So it wasn't entirely a surprise to me that I had a risk variant for that because I know other people in my family had it…but I mean none of it was really shocking"* (CPMC #6, male)

Among those participants who expressed reassurance, common themes for this feeling were: not having "the risk factors" for the disease; a feeling that their risk was normal or consistent with the general population; a perception that their risk was low; and reassurance that their lifestyle put them at a lower risk or that they can control their lifestyle to impact their risk.

> *"It was reassuring to know that I had the non risk variants in several of them. It was reassuring to know that what I'm trying to do with my lifestyle, diet and exercise and stuff like that, no smoking, no drinking, exercising and stuff like that, puts me at a lower risk for pretty much everything."* (CPMC #150, female)

> *"I haven't had anything that striked where I should be concerned or talk to a doctor about. So that made me feel better."* (CPMC #16, female)

Some people were not reassured even though they did not carry a risk allele because they understood that the variants tested accounted for only a small portion of heritable risk:

> *"The results only account for a small fracture of the hereditary portion, …I don't know what else is there that you didn't report on."* (CPMC #12, female)

> *"I know these studies at this point are based on a select number of variants and that the whole picture is not really available to us yet."* (CPMC # 82, female)

Twenty-five percent of participants reported being worried about their results, either because they carried more than one disease-associated variant, they had a family history of the disorder, or because they were at higher risk due to lifestyle factors. More women (31%; n=11) and younger participants (age 50 and younger) (46%; n=13) were worried about their results than men (17%; n=4) and older participants (6%; n=2).

About one-quarter of respondents reported being surprised by at least one of their results. Feelings of surprise were often related to a disconnect between the participant's genetic results and their family history or their personal experience.

> *"I think the only one I was surprised [to see] was the diabetes [result], that I didn't have both [alleles] when I do have it [diabetes]."* (CPMC #86, female)

> *"So just to hear lupus was just surprising because it's nobody in my family has ever had lupus, you know, and I've heard of other people who had lupus but it was just something that was a little bit unexpected."* (CPMC #150, female)

There were a small number of participants, all non-Caucasian, who expressed skepticism about their results, noting that many of the disease-associate variants were studied only in Caucasian populations and may not be relevant to their particular race.

> *"And also these results are based on Caucasian population. My father is black and my mother is white, so I don't know how that would affect me also. So this is all basically from Caucasian studies, so it doesn't factor in minorities, so it makes me wonder how accurate is this for me?"* (CPMC #91, male)

Sharing Results with Health Care Providers

Twenty-five of the 60 participants had acted on results by sharing them with a health care provider before the interview took place. An additional 14 interviewees stated that they planned to share, but had not seen their provider since their results were returned. More older participants choose to share their results with their health care provider as compared to younger participants (53%; n=17 vs. 29%; n= 8). Participants' rationale for sharing with their physician varied. Only one person shared in order to have the physician help interpret results. Most often (60%; 15/25), results were shared so the provider could take some action or offer advice to reduce risk:

> *"I went to the eye doctor as well for the macular degeneration, and I asked her… 'is there anything I should do to prevent getting it, vitamins or things to avoid?'"* (CPMC # 11, female)

Results were sometimes shared in an effort to educate the provider about personalized medicine:

> *"Well, I felt that that was important that the doctor have a clue what I've been doing with this medical collaborative, and I figured it would be a good idea for her to really see what's going on out there in the field."* (CPMC #190, male)

Several participants felt an obligation to share health-related information with their provider:

> *"I think you should tell your doctor as much as you can about yourself as it relates to your health. And, I would hope that some doctors will find it interesting information."* (CPMC #8, male)

Of the participants who shared results with a health care provider, most said that their provider made no specific recommendations based on results; only about one-fourth indicated that their provider made some recommendation based on their results. Several of the providers recommended additional testing or consultations, such as a skin check for a participant at increased risk for melanoma, or checking lipid levels for a participant at increased risk for coronary artery disease. Two providers encouraged the use of a previously recommended medication, in one case, the participant was encouraged to take a previously prescribed Statin to reduce the risk of coronary artery disease; the second participant did not disclose the specific recommended medication. Approximately one-fourth of interviewees who shared results believed that their provider was not able to understand the results, or that the provider didn't know what to do with the results:

> *"He didn't understand enough about what he was supposed to do. It was more or less, 'Okay, we have them, we'll file them, and keep taking your medicine.'"* (CPMC #55, female)

Of the participants who did not share results with their health care providers, most chose not to share because they didn't believe that the results provided any new information that their provider needed to know.

> *No, 'cause like I said, I didn't have any, thank God, any results that triggered like "Oh, I should go see a doctor." But like if I did I would definitely talk to a doctor, one of my doctors, and go from there.* (CPMC #61, female),

A few participants chose not to share results with their health care provider because in addition to not being alarmed by results, they were able to get information they needed through the CPMC website:

> *"Had I felt that there was an alarm I would have probably gone to my medical doctor to get even more detailed information. But I feel pretty comfortable with*

what I'm being provided by in the way of explanation through video, through the other reading portions that are set up within the study". (CPMC #73, male),

Behavioral Responses to Results

In addition to sharing results with health care providers, some participants responded to results by making or planning lifestyle or behavioral changes. Approximately one-third of participants reported making a change in their lifestyle, one-third reported that they planned to change their behavior and one-third reported that they had no intentions to change their behavior. For those participants who do not plan to make any specific lifestyle changes, the most frequently cited explanations were that results were not sufficiently surprising to warrant action, that action was not indicated because the participants were not at increased risk for the conditions tested, or that participants already had a healthy lifestyle.

> *"I haven't, because I had already taken action. For instance I stopped smoking thirty years ago. I try to watch my diet. I have tried a little harder to lose weight but that really wasn't because of the results, it was because I know I need to."* (CPMC #60, female)

Those who reported making changes to their lifestyle indicated that their results provided additional motivation to maintain healthy behaviors that were already acknowledged as being important for reducing disease risk:

> *"So that did give me a little incentive to make sure I put my sunscreen on …it's kind of like, "Okay, this is real. It could happen to me."* (CPMC #172, female)

For many who reported making lifestyle changes, the results were only indirectly associated with behavioral changes because maintaining good health was already a general priority for them:

> *"Is there a direct relationship between the test and the genetic markers and some of the things I'm doing? Possibly only from an awareness perspective, a more conscious effort on my part to actually start taking care of all those things."* (CPMC # 97, male)

Several participants did report changes that they directly attributed to their genetic test results or to their participation in the CPMC. One man indicated that he was going to enroll in an NIH-sponsored clinical trial for age-related macular degeneration, and another began an exercise program.

> *"I'm looking at enrolling in a study up at NIH which is in fact on AMD… I thought this was a great opportunity since I know I've got a risk factor for it, it'll probably help them with the study. And the people who'll be seeing me are specialists in looking at AMD and so I'll be able to follow up with that."* (CPMC # 29, male)

> *"Well the one thing I changed but it was because I was filling out the survey and I felt like I was failing a test when I put that I wasn't exercising so I joined a gym… I'm like, 'Okay, I definitely have to exercise. I definitely have to have a good diet for the coronary artery disease one,' especially since I have the two risk variants which I wasn't surprised at because I have family history. That kind of thing would make me push myself more towards exercising. "* (CPMC #150, female)

One woman directly attributed her participation in the CPMC with her subsequent diagnosis of melanoma, which was discovered at an early stage and was easily and successfully treated:

> *"I was a little surprised that I had the melanoma gene…so I went to a dermatologist after that to get a full body check…I had a mole removed from my back that turned*

out to be a melanoma. Now I'm here with no melanoma because it was stage zero. I'm really grateful for having taken this test because it caught it at that stage." (CPMC # 11, female)

DISCUSSION

Although there has been significant commentary on the utility, lack of utility, potential benefit and potential harm of genomic testing (Collins et al., 2003; Guttmacher et al., 2010; Khoury et al., 2007; McBride et al., 2008; McGuire & Burke, 2008), there is only a small amount of empirical evidence to support or refute these notions. This study has generated qualitative data on individual response to genomic information delivered through the Coriell Personalized Medicine Collaborative, a key step in informing the discussion around the impact of genomic testing for disease risk.

Our finding that participants recall their results based on the presence of a genetic risk factor or based on familial inheritance is consistent with previous literature which shows that having a family history is associated with increased recall and a heightened perception of risk (Axworthy et al., 1996; Katapodi et al., 2004; Montgomery et al., 2003). In addition, the reporting of results by participants as dichotomous rather than numeric is consistent with other qualitative studies showing that participants are most likely to recall and convey their risk using categories rather than percentages (Eckert et al., 2006; O'Doherty & Suthers, 2007; Timmermans et al., 2008; Van Dijk et al., 2004; Vos et al., 2011). This implies that health care consumers tend to personalize disease risk, and determine their own risk status, taking into account not only the actual risk presented, but their own perceptions, anchors, and perceived consequences of the disease as well. It should be noted that to maintain participant confidentiality, interviewers did not have direct access to participant results. Therefore, it is not possible to determine with certainty whether participant recall was accurate or a reflection of perceived risk.

One of the primary concerns expressed by the scientific and medical communities related to personalized medicine is that people may perceive SNPs with relatively small associated risks as being deterministic, leading to undue worry or to reduced efforts to engage in healthy behaviors. Alternatively, small reductions in disease risk attributed to SNPs could lead to false reassurance and poor compliance with general population screening recommendations. Both of these fears are predicated on the assumption that the public has a poor understanding of the multifactorial causation of complex diseases, and that they will misinterpret their genomic test results. The interview data collected here suggest that most participants had a good understanding of the multifactorial nature of the complex diseases for which they received results, reflecting the high level of understanding that has been previously reported (Gollust et al., 2011; McBride et al., 2009). Although some participants had difficulty with numeracy, they appeared to understand their results enough to make reasonable decisions about acting upon them. Most participants were not overly surprised or worried by their results, and even fewer have deterministic or unreasonably reassuring views of their results.

While most people who seek genomic risk assessment are motivated by a desire to improve their health, (Bloss et al., 2011; Gollust et al., 2011; Kaufman et al. 2010, McGowan et al., 2010; McGuire et al., 2009; Su et al., 2011) research assessing the extent to which information about disease risk motivates behavior change has yielded variable results. It has been proposed that behavior change is driven by risk perception; however there is conflicting evidence regarding the validity of this association with several meta-analyses having disproven this association or found the connection to be small (Floyd et al., 2000; McCaul et al., 1996; Milne et al., 2000). These studies, however, have all focused on disease

risk without taking genetic information into account. Other studies that incorporate genetic testing and communication of genetic results in their assessments of behavior change have provided conflicting results suggesting that genetic information may increase motivation (Lerman et al., 1997), or that genetic information may serve to de-motivate behavior change (Bates et al., 2003). A recent meta-analysis by Marteau and colleagues (Marteau et al., 2010) that examined 13 studies (7 which delivered genetic results and 6 which posed hypothetical scenarios) found that genetic information has little to no effect on actual behavior change but may impact participants' reported intention to change. In our study, some participants made changes to their lifestyle that they attributed directly to their genomic test results, and a number of participants indicted that they planned to make some changes in the future.

The extent to which personalized genomic medicine is embraced by health care consumers and providers hinges on the establishment of clinical utility (Khoury et al., 2007; McBride et al., 2010). Even though most CPMC enrollees were motivated to pursue testing by a desire to improve their health, our qualitative data show that taking action, either by sharing results with a healthcare provider, or by changing lifestyle, was not a universal response to CPMC genomic testing, despite the fact that nearly all CPMC participants are given results indicating that they are at increased risk for at least one disorder. Given the poor predictive power and relatively low relative risk of the genetic variants included in the testing offered through the CPMC, as suggested by Evans, (Evans et al., 2011), participants' decisions to not act on their results may actually be appropriate. Consistent with our data, McGowan and colleagues have suggested that many early adopters of DTC genomic testing have expressed disappointment with results that provide limited additional information about health risks. (McGowan et al., 2010) Failure to act on results may therefore be based on a good understanding of the minimal contribution of the variants tested on disease causation, especially in populations who are already engaged in healthy behaviors. For those who do act, genomic information may serve as one motivating factor among many. Whether behavioral changes that were adopted in response to results to support a healthier lifestyle are sustained over time requires longitudinal study; CPMC participants will be followed to assess long-term impact of genomic risk assessment (Keller et al., 2010). Data from longitudinal follow-up of the CPMC participants should lead to an understanding of whether the responses of participants relate to the particular diseases for which results were provided, or if their impressions and reactions apply to the provision of genomic information more broadly.

Limitations

There are several limitations to the current study. All interviewees were participants in the CPMC, a research study offering genomic risk assessment. They are unique in that they had altruistic motivations to participate, and they represent early adopters of personalized medicine (Gollust et al., 2011). As with early adopters of other new technologies, this population is likely to be socially well-connected, have higher education than average, have a strong capacity for coping with uncertainty, and possess favorable attitudes toward science (Rogers, 1995). The genomic testing offered through the CPMC includes only one SNP per disease and fewer diseases than that offered through other research projects or through DTC companies. The good understanding and recall of results, as well as the lack of negative psychological response to results observed in our subjects may be attributed to the fact that results were returned for only actionable conditions and for only a small number of disorders. Furthermore, since we did not have access to participants' actual test results, we were not able to confirm whether participants recalled their results accurately. Although we chose participants for this qualitative study in order to maximize diversity according to sex, race, age and educational level, they still are not representative of the general population. We also were only able to interview participants who contacted us expressing a willingness

to be interviewed. These individuals may also differ from other CPMC participants in important ways. An additional limitation of this study is the relatively small number of participants who were interviewed. Although 60 participants did allow us to collect perceptions across a variety of demographic groups and the study team agreed that saturation had been reached, the opinions of these 60 participants should not be misconstrued as representing all possible reactions to personalized genomic results.

Practice Implications

Genomic risk assessment, offered either directly-to-consumers or through a health care provider, will be a part of medicine in the future (Guttmacher et al., 2010). The public generally holds favorable views of genomic medicine (Leighton et al., 2011; McGuire et al., 2009), and are eager to use the information gained from genomic testing to improve their health, often in conjunction with their health care provider (Gollust et al., 2011). While the early adopters of genomic medicine tend to be highly educated, as testing becomes more widespread, less well educated consumers will be offered genomic testing resulting in an increased need for patient and public education, especially in light of the complexity of the information available to consumers on DTC companies' websites (Lachance et al., 2010). Given genetic counselors' training in genetics, risk communication; the risks, benefits and limitations of genetic testing' facilitating decision-making under conditions of uncertainty; and cultural competency, there is a clear role for genetic counselors to assist patients as genomic risk assessment diffuses into clinical care (Zierhut & Austin, 2011; O'Daniel, 2010). Currently, even though genetic counselors have limited experience discussing results of genomic risk assessment with patients, they believe that they are well positioned to help patients understand and act upon genomic results (Hock et al., 2011). In addition, because physicians, especially primary care providers will have a need to keep up-to-date with regard to new advances in genomic medicine, genetic counselors also will have a role in targeted education of providers about genomic testing (Clarke & Thirlaway, 2011; O'Daniel, 2010).

Research Recommendations

Despite the value of the insights gained through this study of participants in a personalized genomic risk assessment project, there are many areas of future research that will add to our understanding of the impact of personalized genomic medicine, its utility, preferred modes of delivery, resources needed to support the integration of personalized medicine into healthcare, and how the delivery of personalized medicine can be scaled to accommodate the future delivery of whole genome sequence results. These future studies need to include not only early adopters but the public at large, and other key stakeholders such as healthcare providers and insurers.

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Table I

Participant demographics

Characteristic	N (% of total)
Gender	
Male	24 (40%)
Female	36 (60%)
Race	
Caucasian	41 (68%)
Non-Caucasian	29 (32%)
Education	
College degree or more	36 (60%)
Less than college	24 (40%)
Age (Average=48.9 years)	
≤ 50 years	28 (47%)
> 50 years	32 (53%)