Great expectations: patient perspectives and anticipated utility of non-diagnostic genomic-sequencing results

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Abstract The management of secondary findings is a challenge to health-care providers relaying clinical genomic-sequencing results to patients. Understanding patients' expectations from non-diagnostic genomic sequencing could help guide this management. This study interviewed 14 individuals enrolled in the eMERGE (*E*lectronic *M*edical *R*ecords and *G*enomics) study. Participants in eMERGE consent to undergo non-diagnostic genomic sequencing, receive results, and have results returned to their physicians. The interviews assessed expectations and intended use of results. The majority of interviewees were male (64%) and 43% identified as non-Caucasian. A unique theme identified was that many participants expressed uncertainty about the type of diseases they expected to receive results on, what results they wanted to learn about, and how they intended to use results. Participant uncertainty highlights the complex nature of deciding to

undergo genomic testing and a deficiency in genomic knowledge. These results could help improve how genomic sequencing and secondary findings are discussed with patients.

Keywords Genomic sequencing · Precision medicine · Patient perspectives · Perceived utility · Genetic counseling

Introduction

Genomic medicine is increasingly being implemented into health care and is predicted to approach mainstream care within the decade (Delaney et al. 2016; Lazaridis et al. 2014; Linderman et al. 2016; Manolio et al. 2013). Whole genome and whole exome sequencing, or genomic sequencing, has increased the frequency with which clinicians and researchers must handle the challenge of managing secondary findings. As we learn more about the genome and the use of genomic sequencing continues to grow, it will become increasingly common for secondary findings, which could affect health care management, to be identified (Henderson et al. 2013). With the incidence of significant, actionable, and secondary findings in presumed healthy adults falling between 3 and 4%, although one study shows a prevalence as high as 24% (Dorschner et al. 2013; Pillar et al. 2015), input from key stakeholders, such as patients and physicians, are needed to provide guidance on how to manage these secondary findings.

Numerous, complex factors contribute to optimizing management and communication of genomic results to patients and physicians including policy, participant preferences, and the financial and logistic burdens of returning results. Clinicians received some guidance regarding the return of secondary results when the American College of Medical Genetics and Genomics (ACMG) issued a controversial recommendation suggesting that laboratories and clinicians

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return information about variants in 56 genes to patients who undergo genomic sequencing for any reason (ACMG Board of Directors 2015). ACMG chose these genes because they relate to conditions deemed serious, highly penetrant, and potentially treatable (Green et al. 2013). This recommendation provoked concerns about patient autonomy, criteria for genes on the secondary findings list, and testing children for adult-onset conditions (Burke et al. 2013; Richardson 2014). The complexity of formulating such a gene list was reflected in the recent update, removing one gene and adding four others, resulting in a modified list of 59 genes (Kalia et al. 2017). Additionally, several international genetics and ethics societies in Canada and Europe have formed similar statements, emphasizing the patient's right to opt out of secondary findings (Knoppers et al. 2015). Guidelines on how to communicate these results to patients have still not been developed (Cassa et al. 2012). Many studies have elicited clinicians' perspectives on which results should be disclosed to patients; however, there is a need for deeper exploration of what patients actually desire (Shahmirzadi et al. 2014).

Several studies have investigated early adopter's preferences and motivating factors for receiving secondary results returned from genomic sequencing. The most common motivators were obtaining personal disease risk information, satisfying curiosity, contributing to research, self-exploration, and interest in ancestry (Linderman et al. 2016; Hall et al. 2015; Gollust et al. 2012; Facio et al. 2013; Sanderson et al. 2015). Another study surveyed the general public about what types of results they would want from genomic sequencing (Middleton et al. 2016). These individuals represented 75 countries and the majority of them had not undergone genomic sequencing indicating that they are not necessarily early adopters. In this population, over 90% of participants wanted to know about life-threatening conditions if they were preventable, carrier status, and pharmacogenomics results. The majority of these individuals also wanted to learn about life-threatening conditions that were not preventable, and overall, participants were more interested in results that confer a higher risk of developing disease. Another study in Denmark showed research participants undergoing genomic sequencing were interested in learning about all incidental findings instead of limiting the disclosure to results that would have a direct impact on medical management (Jelsig et al. 2015).

The majority of participants wanted all of their available results returned. The relationship between what motivates an individual to pursue genomic testing and their expectations of results are closely intertwined. Only a small number of studies have explored patient expectations for non-diagnostic genomic-sequencing results. Recently, researchers have begun to examine what types of results patients desire and their perceived value and utility of those results (Hall et al. 2015; Facio et al. 2013; Sanderson et al. 2015; Lupo et al. 2016). One study found that patients undergoing clinical genomic

testing expect genomic information to improve their health through pathways such as guiding personalized treatment and improved cure rates (Hall et al. 2015). While another study found that participants expected to learn information about their risks for specific types of conditions such as heart disease or cancer, they also found that participants had unrealistically high expectations for the utility of genomic-sequencing results (Gollust et al. 2012). An additional study found the majority of respondents expected that their results would have utility for health-related decision-making (Lupo et al. 2016). Participants in all of these studies were self-selected, suggesting they are information-seekers and early adopters. In addition, the vast majority of these participants were highly educated and identified as non-Hispanic Caucasian. Therefore, their expectations may not represent the attitudes of the general public or patients who are offered genomic sequencing but have not explicitly sought it out.

As tools, processes, and, ultimately, policies are developed for consenting and returning secondary genomic-sequencing results to individuals, the viewpoint of a key stakeholder, the patient, needs to be investigated further. Currently, little is known about expectations, intended use for results, and which results people expect to communicate when individuals undergo genomic sequencing for non-diagnostic purposes. This study explored patient expectations of genomic sequencing for non-diagnostic purposes in a population of individuals recruited from outpatient clinics. These participants were approached and did not seek out this testing for themselves; however, they still elected to undergo sequencing. Results from this study contribute to our understanding of patient expectations with regard to personal genomic information and will help inform how this testing should be presented to patients.

Methods

Subjects and methods

Participants were recruited from a group of patients participating in the eMERGE III study at Northwestern University (NU). Northwestern University is one of 12 sites which make up the eMERGE (*Electronic Medical Records and Genomics*) Network (Gottesman et al. 2013). As part of the eMERGE Network, Northwestern has investigated the implementation of genomic medicine into clinical care, including how to incorporate genomic information into the electronic health record (EHR) in a useable format and how this information impacts medical care. Northwestern's third phase of the study for the eMERGE Network (eMERGE III) is further investigating how patients use, value, and communicate genetic information.

Participants in eMERGE will receive sequencing results from approximately 100 genes, which include the 56 ACMG-recommended genes (ACMG Board of Directors 2015). Participants agreed to have results placed in their EHR to be used by physicians at Northwestern Medicine. Only results from genes deemed to have clinical actionability (as determined by the eMERGE Clinical Annotation workgroup) will be placed in the EHR. Participants in the NU eMERGE study are randomized to either receive all the results or have the option to decline results related to specific types of conditions (e.g., cancer, dementia, etc.).

Selection criteria: to be eligible for the NU eMERGE III study, individuals had to be patients of Northwestern Medicine or one of its affiliates and be 18 years of age or older. Participants were recruited from a variety of outpatient clinics, including cardiology and dermatology clinics, or were previous participants in a Northwestern Medicine genetic study conducted with patients from a general internal medicine clinic. Individuals were not targeted based on specific characteristics or disease states.

Participation involved a one-time blood donation for genetic sequencing, providing permission for results to be placed in the participants' EHR at Northwestern Medicine, allowing ongoing access to the EHR at Northwestern, and completion of a 10-min survey. The survey consists of items related to illness perception, health behaviors, privacy concerns, and demographics. All participants were recruited and consented by genetic counselors. Consent for eMERGE occurred during an appointment in an outpatient clinic at Northwestern Medicine. The study was briefly introduced by their provider and if they were initially interested, a genetic counselor would describe the study in detail and provide consent at the end of their visit. Consent focused on the purpose of the study, the risks and benefits of participating, the types of results that will be reported, how results will be returned to them and their Northwestern physician, and the option of follow-up genetic counseling to discuss their results. The genetic counselor also described the potential risks involved in participating including psychological risks and discrimination, as well as potential benefits and limitations. Consent lasted between 30 and 40 min. Between December 2015 and February 2016, after consenting and enrolling in NU eMERGE III, participants were offered the option to be interviewed regarding their expectations of the results. Participants were eligible for interviews if they spoke English. During this three-month period, 19 eMERGE participants were approached and 15 consented to the interview. One participant declined at the time of the phone interview. The study was approved by the Northwestern University Institutional Review Board.

Eligible participants were contacted by phone or email to schedule interviews. Up to five attempts to contact participants were made within 3 weeks of enrollment. Thirty-minute semi-structured phone interviews were conducted.

Each participant explored themes around the research questions and aims focusing on their motivation to learn specific types of results and their anticipated use of results. At the beginning of each interview, the study and consent were reviewed. All interviews were recorded using the smartphone app TapeACall Pro© and subsequently transcribed by RH via an assigned ID number. After completing the interview, participants were emailed a US\$25 Visa gift card to compensate for their time. Interviews were conducted until saturation was met for all themes identified. Analysis was performed continuously during the interview process to modify the interview guide to appropriately address aims and determine saturation.

Data collection and analysis

Data was collected using a semi-structured interview guide that was developed by the authors. The interview guide included questions pertaining to expectations and motivations for pursuing whole genome sequencing. Participants were prompted to share how they predicted to use specific types of results they anticipate possibly receiving. Throughout the interview, participants were encouraged to share their thoughts, feelings, and opinions openly. All 14 interviews were conducted by a single interviewer (RH) to keep interviews consistent.

A single investigator transcribed the interviews. Transcripts were coded and analyzed, using Dedoose version 6.2.2.1 coding software, by one investigator (RH) and manually by a second investigator (SA) via consensual qualitative research (CQR) (Hill et al. 1997). A content-analysis approach was utilized to identify emerging themes and create a codebook. The analysis consisted of identifying meaningful segments of text, assigning a code to categorize the meaning of each segment (some segments received more than one code), and combining segments and categorizing them into subthemes. Lastly, all subthemes were grouped into major themes. Frequency counts for each major theme were calculated. If a theme emerged multiple times in an interview, it was only counted once. When conflicting coding emerged, the transcript was reviewed again and discussed until an agreement was reached between the coders. This method of consensus building ensured multiple perspectives contributing to validation of the codes.

Results

Fifteen eligible individuals agreed to be contacted, but one declined to participate at the time of the interview when contacted. The remaining 14 participants were interviewed within 3 weeks from the date of enrollment into the eMERGE study. Interviews lasted from 20 to 40 min, with an average length of approximately 24 min. Participants

tended to be male, highly educated, and reported having children. The majority identified as non-Hispanic Caucasian (57%; refer to Table 1). This population is similar to outpatient Northwestern Medicine patients in terms of ethnicity (48% non-Hispanic Caucasian). Males represented a higher proportion of participants in this study (64%) compared to outpatient Northwestern Medicine patients (40% male).

Five (36%) participants self-identified as having a significant health problem at the time of the interview and nine (64%) did not. The majority of participants (57%) reported having a significant family history of a specific disease, defined as more than one first-degree relative or more than two second-degree relatives with the same or related diseases. The most common types of disease reported in the family history were cancer and cardiovascular disease. Individuals are identified by their participant number (P no.) and by their age (no.).

Content analysis revealed 17 themes that were grouped into five overarching themes. We report on three of the overarching themes: expectations for results, expectations for

communication, and uncertainty. This subset of themes contained unique information that has not yet been well described, or described at all, in existing published literature.

Expectations for results

Expectations varied from expecting results that would specifically provide more information about a current health problem to expecting the information to have broad implications for improving health care. Some participants expressed general curiosity and interest in learning information from their results as well as future results from the eMERGE study. Participants repeatedly expressed a desire to learn as much as possible about their health through the study. Many participants stated they expected to use information regarding their genetic makeup to allow them to plan and prepare for the future. Another common expectation was that the personal health information could provide reassurance about diseases the participant is not at an increased risk of developing.

A number of participants relayed that whatever information they could learn from the testing would be beneficial to them and there were no particular expectations about a specific condition as one patient explained, “I kind of believe in, you know, the more information the better. So you know, the more I can learn about any type of diseases that I may be prone to or whatever you know, the better for me. Um- I know they kind of explained it like some people get anxiety over it, but I was more along the lines of, you know, I’d rather have all the information I could” (P 4, age 34).

Many participants stated they anticipated and were interested in learning about diseases in their family. The presence of disease in multiple family members made several participants suspicious of a hereditary component or genetic etiology. In some cases, this suspicion impacted their perceived personal risk for the disease. Likewise, participants who did not have certain types of diseases in their family, like cancer, were not expecting to get results related to those diseases. The following quote illustrates one participant’s interest in learning about their own personal risk for a condition based on their family history: “I’m curious as to with all the cancers in my family, how likely am I to get that cancer and if I’m gonna get one, just any details around it” (P 13, age 43).

Several participants indicated they had no specific expectations for the type of information they would learn from their results and even doubted they would learn anything of value to them personally. After being probed, some individuals were able to come up with potential expectations but were not confident in the type of information they expect to gain. “Let me put it this way. I don’t think I have any expectations of anything right now. I’m just hoping that I might be able to find some interesting information in it and see if there’s anything in there specifically that pinpoints anything. That might be

Table 1 Participant characteristics (*n* = 14)

Characteristic	<i>n</i> (%)
Male	9 (64)
Race/ethnicity	
White/Caucasian (non-Hispanic)	8 (57)
Hispanic/Latino, White	3 (22)
Asian	1 (7)
Native Hawaiian/Pacific Islander	1 (7)
Black/African American	1 (7)
Age (years) ^a	
25–35	4 (29)
36–45	2 (14)
46–55	1 (7)
56–65	6 (43)
66+	1 (7)
Education	
Some college	2 (14)
Bachelor’s degree	7 (50)
Postgraduate degree	5 (36)
Household income (USD) ^b	
45,000–59,999	1 (8)
60,000–89,999	3 (23)
90,000–149,999	3 (23)
>200,000	6 (46)
Has children	8 (57)
Has family history of disease	8 (57)
Self-reports personal health problem	5 (36)

^a Median 50 years, range 27–70 years

^b One participant declined to report annual household income

something that not only could help me but future generations” (P 11, age 60).

Expectations for communication

A large majority of participants were enthusiastic about communicating their results, specifically with family members. Many participants expressed the importance of sharing genetic information with family as well as other members of their support network for personal reasons as well as increasing general awareness. The most common intent to communicate results was to give family members the opportunity for prevention. Participants expressed hope that awareness and early detection may be able to improve the medical outcomes for family members, particularly children. One participant shared, “Well I’d share ‘em with my wife. I’d share ‘em with my kids cause theoretically, some of the results could impact their life. They would have to address those issues from preventative standpoint and what could they do in their lives and their lifestyle to try to assure that they won’t incur the disease... And hopefully, as time goes on, the advancement of medical science there will be more cures to various diseases that will benefit everybody- including them” (P 9, age 70). Another participant stated, “I want to tell them...if it was something where that knowledge could be used to help them defy or figure it out, I would help” (P 13, age 43).

Other participants had more complex views surrounding the pros and cons of sharing this information. They stated they would not share certain results with family members in order to avoid the potential psychological burden that could accompany the information. This was particularly true when they spoke about terminal or untreatable illnesses: “... I have two daughters, and if the result was they’re going to incur disease in the next three years- that it will be terminal, I may not tell them. But it’s hard to say until you’re faced with the results” (P 9, age 70). Another participant shared, “My inclination would be to share it but I don’t know whether sharing would be wanted” (P 10, age 65).

Uncertainty

Many participants were unsure about what types of results to expect and how they might use those results. Some also stated they had not thought through how they would use the information or what they would do if they received a specific type of result. Individuals repeatedly gave an impression that their thoughts and expectations for using results were not fully developed. One patient explained, “I have no plan to use the results. It’s just see the results and then I’ll make a plan. There is no specifics... It all depends. I just think it would be fascinating” (P 6, age 57).

A minority of participants expressed feeling conflicted or unsure about what type of information they would like to learn from their results, particularly if the results are not medically

actionable. One participant shared, “I would want to know- but then I also- that would be something I wouldn’t want. Sort of a double-edged sword like that where I would want to know to be able to do something about it. My concern is not being able to do anything about it” (P 3, age 41).

Multiple participants voiced concerns, uncertainty, and sometimes skepticism about the level of risk associated with results and the associated utility for them personally. A common theme was being unsure about whether a result would translate to a diagnosis or a risk of potentially developing a disease. This was exemplified by one participant: “When you learn about ‘em, what is the risk there? Is it a minor risk? Is it just something that’s indicated as a possibility or is this something that’s a real risk and it’s something that’s high likelihood of happening” (P 11, age 60).

In some cases, the uncertainty of the information led participants to question whether they really wanted to know the results. “I would assume I’ll know at some point (laughs) because I’m also assuming this is not a given, so I’m not going to hear you’re one hundred percent going to get this disease in about eight to twelve years. It’s gonna be three percent to ten percent risk of this disease and I’d rather not know that” (P 7, age 56).

Discussion

This study explored the expectations of patients who were approached and opted-in to non-diagnostic genomic sequencing distinguishing it from previous studies consisting of early adopters who sought out genomic testing (Gollust et al. 2012; Facio et al. 2013; Sanderson et al. 2015; Lupo et al. 2016). While early adopters of personal genomic testing are proactive in utilizing genetic information to manage their health care (Gollust et al. 2012), participants in this study were approached and many of them stated they agreed to the study to help with research, not because they were actively seeking out personal genomic information. Therefore, learning the expectations of participants in this study may contribute a unique perspective. Additionally, almost half ($n = 6$, 43%) of our participants identified as having at least part of their ethnic background as something other than non-Hispanic Caucasian. This is notably more ethnically diverse than other studies exploring participants’ motivations and perspectives on genomic sequencing (Gollust et al. 2012; Facio et al. 2013; Sanderson et al. 2015; Lupo et al. 2016). This information may provide a different perspective than preferences expressed by predominantly Caucasians who actively pursued genomic sequencing for developing guidelines on consenting for genomic sequencing and returning secondary results.

Some participants in this study, although highly educated, had unrealistic expectations for the accuracy and utility of results. Many participants believed results would give them an absolute answer on whether or not they would develop a

specific disease. This differed from another study where less than one third of participants expressed unrealistic expectations (Gollust et al. 2012). However, over 45% of participants in that study worked in the medical field or shared a household with a medical professional. That distinction suggests that the finding of participants having unrealistic expectations may be more in line with the general population. Other genomic studies have also found that participants have unrealistically optimistic expectations for genomic information, meaning they had idealistic hopes in a successful outcome that were not necessarily in line with reality (Hall et al. 2015, 2012; Nelson et al. 2010; O'Rourke 2013).

Additionally, a subset of participants reported no specific expectations even though they were consented and informed that positive results will be returned. Many participants focused on their altruistic motivation for participating in this study rather than being motivated by receiving personal results. This is a common finding in genomic research studies (Gollust et al. 2012; Sanderson et al. 2015; Bollinger et al. 2012; Facio et al. 2011). Perhaps individuals who stated they had no personal expectations truly had no personal motivations for participating in the study and had not considered how impactful the results could be for them. Further research could explore whether or not committing to personal genomic sequencing that is not contributing to research has an impact on expectations, motivations, and intended use of genomic information.

The majority of participants in this study plan to communicate their results with their families, citing using the information to protect family members as their motivation. This is consistent with participants in studies undergoing hypothetical genomic testing as well as direct-to-consumer testing (Gollust et al. 2012; Sanderson et al. 2015; Bollinger et al. 2012). Another study surveying biobank members found a majority of participants would want their results disclosed to family members in the event of their death (Allen et al. 2014). Since some results are associated with non-preventable, untreatable diseases, it would be important to help patients in the future distinguish what results they want for themselves and what they would be comfortable sharing with family members, highlighting the importance of providing patients with resources to discuss disease risk with family members.

More than one participant mentioned they may not share “devastating” results with family members. To our knowledge, this theme did not arise from similar studies evaluating perspectives of early adopters or in hypothetical studies (Gollust et al. 2012; Bollinger et al. 2012). Participants in another study brought up concerns of the implications for their children, which could indirectly be related to a desire to keep devastating results from family members (Bollinger et al. 2012). This finding supports the idea that in clinical practice and in research settings, it is crucial to use anticipatory guidance to help patients devise a plan for adapting to distressing results that they would not feel comfortable sharing with family members.

Participants also expressed feeling conflicted about learning certain types of results. They voiced indecision about whether information about certain diseases would be helpful to them or if they would rather be blissfully unaware of risks. Again, perhaps these thoughts would be more completely developed if they were doing the testing for personal reasons rather than research. It is also plausible that participants chose not to think about these implications and possible outcomes thoroughly. Information seekers may say yes to receiving all results without understanding what that could mean for them medically and emotionally. Many had underdeveloped thoughts about expectations but were adamant that they wanted all of the information. By participating in this study, they were forced to contemplate how the information might impact them. One study found differences in participants thinking about potential results and actually facing them (Sanderson et al. 2010). It would be interesting to elicit perspectives from participants in this study once they have results to see if their opinions have changed.

Contrary to other studies, all participants responded that they did not consider any drawbacks to participating in this study. Similar studies found participants held concerns about the potential emotional response caused by results and consequences like insurance discrimination and loss of privacy (Gollust et al. 2012; Sanderson et al. 2015; Bollinger et al. 2012). This finding supports the idea that participants in this study had not thought methodically about possible outcomes of the testing, particularly concerning personal consequences. Declining to verbalize drawbacks, despite expressing concern or anxiety about receiving certain types of information, could also be explained by cognitive dissonance theory. This theory describes how humans strive for mental consistency because intellectual inconsistency can cause psychological stress and motivates humans to attempt to reduce the dissonance (Festinger 2010). Participants in this study voiced concern over the potential of results causing anxiety, yet stated they did not consider drawbacks to having genomic sequencing. One technique to reduce dissonance is justifying one's actions by changing the conflicting thought (Festinger 2010). This theory could possibly explain the seemingly conflicting responses of participants in this study.

These results provide unique insight into patient expectations and intended use of genomic-sequencing results. The uncertainty and unrealistic beliefs expressed by participants highlight the need to elicit the patient's perspective in order to continually manage expectations in the pre-test consent and results-disclosure sessions. Patient misunderstanding and uncertainty could lead to compliance issues in future management.

Limitations and future directions

This was an exploratory study on a specific, small population. Therefore, the results of this study are not generalizable to all patients being offered genomic sequencing; however, themes

that arose in this study could be used to develop a quantitative questionnaire to sample a broader population. Participants in this study may be biased since they were recruited through the eMERGE study, had agreed to receive genomic-sequencing results, and may have a more favorable view of genetic information than the general public. Additionally, this was a highly educated, mainly Caucasian population with high household incomes. Therefore, future studies should be completed on a larger, more diverse population to test if the findings in this study hold true.

The findings from this study draw attention to the necessity to improve the consenting process for genomic sequencing, managing expectations, and returning results. Our results raised the suspicion that the inability to describe every possible result in the consent process contributed to participants filling in their own gaps, leading to unrealistic expectations. Lastly, many participants affirmed that they had not thoroughly considered what types of results they would receive and how that would impact them, highlighting the importance of giving patients time to consider whether or not they want to pursue testing. Making a truly informed choice may also help avoid the cognitive dissonance our participants appeared to experience. Further studies could explore techniques for optimizing the consenting process and avoiding cognitive dissonance.

Unrealistic expectations of genomic results and an overestimate of the utility of genetic testing made us question if there needs to be more of a focus on genetic literacy in the general public. Other studies have identified genomic illiteracy and the ensuing communication barrier between patients and providers (Haga et al. 2012; Leighton et al. 2012; Kelly et al. 2007). Future studies could evaluate if improved genomic literacy is correlated with better understanding and more accurate expectations for genomic results. The expectations identified in this study could contribute to the development of guidelines for consenting individuals and returning results. The overarching theme of uncertainty in expectations highlights the importance in education and the ability of the patient and the health-care provider to communicate effectively in order to manage expectations.

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Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

Human and animal rights and informed consent All procedures performed in studies involving human participants were in accordance with

the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki Declaration and its later amendments or comparable ethical standards. All participants provided informed consent for being included in the study.

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