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Patients' willingness to reconsider cancer genetic testing after initially declining: Mention it again

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Abstract

Patients at risk for hereditary cancer syndromes sometimes decline clinically appropriate genetic testing. The purpose of the current study was to understand what preferences, concerns, and desires informed their refusal as well as their current level of interest in being tested. We interviewed patients who had been seen in a hereditary cancer clinic at Vanderbilt University Medical Center and had declined genetic testing. Twenty-one in-depth, semi-structured qualitative interviews were conducted. Although patients provided many reasons for declining testing, they most often cited their psychosocial state at the time of the initial invitation to participate in genetic testing as their reason for refusal. The majority (67%) said that they either would or had changed their mind about testing if/when their clinicians "mentioned it again." Patients at risk for hereditary cancer who refuse testing at the time of genetic counseling may later change their mind. In particular, if a patient declines testing around the time of a major medical diagnosis or intervention, clinicians who are providing ongoing care may want to raise the topic afresh after the patient has had time to recover from initial distress related to diagnosis or treatment. Strategies to prompt clinicians to have these conversations are suggested.

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AUTHOR CONTRIBUTIONS

CME Halverson, EW Clayton, and GL Wiesner contributed substantially to the conception and design of the work. These authors as well as BC Wessinger contributed substantially to the interpretation of the data, drafting of the work, and revising of the drafts. All authors had full access to all the data in the study, gave final approval of the current draft, and agree to be accountable for all aspects of the work.

HUMAN STUDIES AND INFORMED CONSENT

The study was approved by the Vanderbilt University Medical Center Institutional Review Board and conforms to recognized standards for ethical research.

CONFLICTS OF INTEREST: CME Halverson, BC Wessinger, EW Clayton, and GL Wiesner declare no conflicts of interest.

Keywords

Decision Making; Ethics; Genetic Testing; Hereditary Cancer; Genetic Counseling; Barriers to Genetic Testing; Psychosocial

INTRODUCTION

It is estimated that 5–10% of people who have cancer in the United States have germline pathogenic variants in cancer predisposition genes (Grant et al., 2015; J. Zhang et al., 2015; S. Zhang et al., 2011). In many cases, detecting these variants can inform individuals' care in ways that improve their long-term clinical outcomes, including increasing surveillance to permit early detection and altering management (Feero, Guttmacher, & Collins, 2010). Although at times controversial (Turna Ray, 2019), guidelines have been proposed to define the clinical circumstances under which it would be appropriate to offer testing as well as which genes should be tested in particular cases (Daly et al., 2017; Hampel, Bennett, Buchanan, Pearlman, & Wiesner, 2015). Pertinent factors typically include the nature of the cancer(s) that the patient already has as well as his or her family history. At many centers, given the complexity of genetic testing for cancer predisposition, a specialist in cancer genetics provides counseling about what tests, if any, might be appropriate and whether to proceed. These visits can coincide with a cancer diagnosis in the patient or can be linked to the discovery of a worrisome variant in a family member for unaffected patients. Most people who attend these clinics and who are ultimately found to have a clinical indication for testing decide to proceed (Butrick et al., 2015): Patients typically return to the referring provider or other healthcare provider for their ongoing care, and the cancer genetics provider does not typically follow them.

However, many people who meet current clinical criteria for genetic testing do not receive such tests (Childers, Childers, Maggard-Gibbons, & Macinko, 2017; Hann et al., 2017; Selkirk et al., 2014). Some patients may not be aware that they are at risk, at times because clinicians may not recommend appropriate testing or refer patients to specialists who could do so (Clift et al., 2015; Yushak et al., 2016). Many people are offered referral to a cancer genetics clinic but do not go (Willis et al., 2017). In addition, a small but important percentage of patients and family members who are offered clinically indicated genetic testing decline such testing after counseling (Anderson et al., 2012; Bellcross, Leadbetter, Alford, & Peipins, 2013; Finlay et al., 2008; Ropka, Wenzel, Phillips, Siadaty, & Philbrick, 2006), at which time they return to their other providers for care.

The literature suggests several factors that may be involved in decisions to forgo genetic tests. Some people refuse testing because their insurance would not cover the cost. Payers vary widely in their willingness to pay for genetic and genomic tests, particularly those that are more complex (Deverka & Dreyfus, 2014; Phillips et al., 2018). Cost, however, is not the only issue. Numerous surveys and interviews, often involving responses to hypothetical questions, suggest that people worry that results will cause them or their family members distress, put them and their loved ones at risk for employment or insurance discrimination, or compromise their privacy in other ways (Hayden et al., 2017; Kinney et al., 2006; Kne

et al., 2017; Rosenberg et al., 2016). This qualitative interview study examines the past and present motivations and needs of patients who have considered using genetic technology for their healthcare.

METHODS

Participants

A qualitative study using semi-structured interviews was conducted with patients who had declined genetic testing after cancer genetic counseling within the preceding five years in one of Vanderbilt–Ingram Cancer Center's Hereditary Cancer Clinics. Participants were identified from the IRB-approved Hereditary Cancer REDCap registry (Harris et al., 2009). (REDCap is a web-based application for managing online surveys and databases developed at Vanderbilt and is now in widespread use.) Enrolled and consented participants were interviewed between April and September 2018. The study was approved by the Vanderbilt University Medical Center Institutional Review Board, and all individuals gave their informed consent prior to their inclusion in the study.

Using purposive sampling, we sought to recruit participants from a set of 50 potential candidates. We located individuals in the registry who were diverse in terms of age, gender, race, and clinical indication. In particular, we attempted to oversample non-white candidates. Patients were required to be English-speaking and over 18. Patients who were noted to have explicitly declined for insurance coverage or other economic concerns were excluded, as these circumstances are often beyond the clinician's control. An invitation letter from one of the researchers (GLW), a clinical cancer geneticist, was sent, explaining the goals of the study, and two researchers (CMEH, BCW) followed up by telephone. A $25 online gift certificate was offered as an incentive for participation. Telephone contact was attempted up to three times. Interested individuals scheduled a telephone interview and completed an oral consent. Patients were interviewed until thematic saturation was reached (Glaser, 1978; Strauss, 1998). Each candidate was randomly assigned a three-digit identification number to be used with de-identified data. Contact and relevant healthcare data were stored in a REDCap database created for this project.

Instrumentation and Procedures

An in-depth, semi-structured interview guide (available in the Data Supplement) was designed to evaluate patients' concerns with genetic testing and reasons for declining. The interview guide was developed iteratively, directed by a review of the existing literature and the recommendations and feedback of methodologists and other skilled qualitative researchers at Vanderbilt University.

The interview guide was divided into three sections. The first explored the participant's personal medical history. The second ascertained the risks and benefits of genetic testing as described by the participant. The third asked what societal (social, educational, cultural) influences might have shaped the participants' healthcare decision making. We included a brief demographic questionnaire at the end. We made a concerted effort not to suggest the types of reasons our participants might give for declining testing and did not directly raise

any potential reasons until the very end of the interviews. Our final questions before turning to demographics were explicitly about participants' opinions on privacy in healthcare settings and their influence on their choices.

Data Analysis

Interviews were conducted by an experienced interviewer trained in qualitative methods (CMEH) using the interview guide. Interviews were audio-recorded, professionally transcribed verbatim, and de-identified. Transcripts were analyzed using grounded theory, which allows for the identification of common themes across interviews (Glaser, 1978). Two authors (CMEH, BCW) coded each transcript both for broad categories and for narrower subtopics relevant to the purpose of the study. Conflicts in interpretation were discussed and resolved by consensus on all codes. All authors discussed emerging themes and patterns found across each of the interviews; coding was refined iteratively.

RESULTS

Response Rate and Demographics

Of the 44 potential candidates located in the registry (six of the initial sample were deceased), 21 (48%) agreed to participate in our study and eventually completed an interview, at which time we determined that thematic saturation had been reached. The average interview length was 39 minutes, with a maximum length of 57 minutes and a minimum length of 28 minutes. Despite oversampling non-white individuals, the interviewees were largely white (67%) and female (95%). Only one man agreed to be interviewed. Five African Americans (24%) and two patients of Asian descent (10%) completed interviews. Eleven (52%) were in their forties or fifties. Most participants had a personal history of cancer (16, or 76%), and most of those participants had been seen in clinic with concerns about breast cancer (12, or 75%). Other indications included sarcoma, thyroid, lung, and colon cancer. Those without a personal history had been referred or sought consultation themselves due to a family history of cancer. For more demographic information, see Table 1.

"I've Got Enough on My Plate Right Now."

Participants offered many reasons why they declined genetic testing (Table 2). Many (8, or 38%) participants said that they had declined because they were personally too distraught or overwhelmed at the time of counseling. One woman stated she had said no because she did not want to think more about her cancer diagnosis and was tired of letting it control her life: "Part of me wants to get past all this, but you can't if you keep getting tested" (306). "I've got enough on my plate right now," she had told her genetic counselor, saying she didn't have the emotional resources to make yet another decision about her healthcare.

Many people mentioned that they faced emotional stress about the wellbeing of their family. "Once you have that information, you have to deal with it, and then that can spread to other family members […] That fear spreads" (220). Another woman (202), almost in tears, told us about her clinicians' suspicions that she had Li Fraumeni syndrome. She had opted not to seek a molecular diagnosis, because

> "If I had it, then there was a 50/50 chance that each one of my kids had it, [and] if you have it, there's not anything you can do […] There's a high risk of suicide with it, [and I] didn't want my children just living with that dark cloud over their heads for the rest of their lives."

She was one of the few participants who was glad never to have been tested. "If I found out that I did have it, I think I would've driven myself crazy, not for myself, but for my kids and my grandson."

Other reasons for declining testing were linked to finances, despite our sampling method that excluded patients who expressed these concerns at their clinic visit. However, even those who identified economic factors as barriers also said that their psychosocial situation at the time of their decision limited their willingness to undergo testing.

"Genetic Testing Would Be a Lot More Important Than My Privacy."

Our respondents rarely expressed reservations about the tests themselves. The most prominent of these other concerns related to privacy, and even that was limited. No one spontaneously brought up privacy as a reason to decline testing. It was only with the final prompt that we elicited discussion on the topic. No one expressed major anxieties about genetics and privacy. "Obviously there are people that have more concern about [privacy], but I don't; I don't have that kind of concern. […] I don't hide things" (212).

Promoting research was a major reason some respondents were willing to forgo privacy. "I could absolutely [not] care less about privacy […] if it's research, I don't care" (202). One woman viewed protecting privacy as antithetical to progress in medical research: "Privacy in the health places isn't appropriate" (406). Similarly, a woman in her sixties (217) told us, "To me, being part of the human race and being a good person" means putting the needs of the community ahead of one's privacy.

"Mention It Again."

Seven of our 21 participants (33%) eventually underwent genetic testing after initially declining, including all of our interviewees who had no personal history of cancer. Only one of them had pursued genetic testing on her own initiative; all the others had agreed when it was offered to them at a later time. A participant with a history of breast cancer (311) said she began to rethink genetic testing when other members of her family started developing cancers as well. However, she did not bring the topic up with her clinicians on her own, recounting, "I had another doctor, and she mentioned it again. That's how I ended up taking [the genetic test]." Without this second mention, she conceded, she would not have pursued testing herself.

Several people actively regretted not being tested when it was first suggested, and others expressed that they would be willing to proceed with testing now. In fact, the majority (67%) of those who had originally declined testing said that they would gladly have it at the time of our interviews. One woman explained that if she were asked today, "if it will help me or somebody else, I'm ready to do it. I don't want to leave others out in the cold" (108).

Another participant (220) got tested two months after our interview with her and convinced her father to get tested as well.

The issue was simply that patients did not recall anyone mentioning the possibility again. Many people told us that the last time they had discussed genetic testing with anyone was when it was initially offered – often while they were in the midst of chemotherapy or other onerous medical care or simply dealing with their diagnosis. “I wish they would bring it up,” said one of our interviewees (212), “and then I would do it.” A woman with a family history of breast cancer believed “I would be a good candidate for genetic testing” (308), but she said that no one had mentioned it to her in the four years since the first and only time she was offered it. A third participant, echoing this sentiment, told us that she had initially declined because she was overwhelmed by her circumstances when the test had been offered. “I don’t feel it to be overwhelming now” (302), she said, but no one had discussed genetic testing with her again “since I had this counseling done […] three or four years ago.”

DISCUSSION

The most important finding of this study is that the majority of our respondents who initially refused testing either went on to be tested or said that they would agree to be tested if it were offered again. This suggests that patients may not be fixed in their choices made at the time of counseling and that they may be willing to reconsider the use of genetic tests if prompted by their healthcare providers. It builds on the hypothetical responses of respondents in Ardern-Jones and colleagues’ study, many of whom were concerned that offering genetic testing at the time of cancer diagnosis would be “too much, too soon” (Ardern-Jones, Kenen, & Eeles, 2005). On the most practical level, providers should recognize that patients may continue to be concerned about genetic contributions to their health and that they may appreciate a discussion about genetic testing at a later time.

But these findings raise a larger question: To what extent is it desirable or even ethically or legally required to try to ensure that these patients have the chance to reconsider their decision? Complicating this issue is the likelihood that it may be prudent to wait until patients have had some time to adjust to their diagnosis or the diagnosis of a family member before raising the question of testing again given the reasons they gave for refusing testing in the first place – e.g., being emotionally overwhelmed at the time, the cost, and fears about testing’s impact on relatives’ psychosocial wellbeing. Waiting until a more propitious time may involve a delay of several months to a few years. Another complication is that the current workflow for oncologists and geneticists may not easily accommodate reminders and communication with patients who are no longer under their care.

Practice Implications

Given that most consultations with cancer geneticists or genetic counselors are time-limited (David et al., 2019), the question becomes how best to provide the opportunity for genetic testing at a later time. As a starting position, we suggest that a combination of clinical workflow modifications and informatic support by the electronic health record (EHR) could be developed to aid the clinician and patient in re-addressing the option of genetic testing in

the future. Workflow modifications could include instructions to the patient and provider if there are questions or a change in the initial decision. Electronic records could be modified to add genetic testing to the problem list and the need to revisit the issue (Peterson et al., 2013). In addition, informatic support with reminders could be used to identify patients with whom the provider could discuss additional testing at future appointments. Geneticists have for years written comprehensive summaries of their counseling sessions to patients and their providers (Baker, Eash, Schuette, & Uhlmann, 2002; Brown, Skinner, Ashley, Reed, & Dixon, 2016). Many already include invitations for patients to recontact them for follow-up, a practice that should be encouraged.

Recommendations for EHRs are more challenging for several reasons. While these systems hold great promise, their implementation across the country is still in its early stages (Ury, 2013). Thus, while it may not be particularly onerous to create a designated space in the medical record to include information regarding cancer predisposition genetic testing, some healthcare systems may not have resources to implement this feature given the press of other matters. Potentially more troubling, medical centers still do not always share EHRs with their patients' other providers who work in other systems or in local practices (Mandl & Kohane, 2012). Thus, these outside providers often need to develop their own notations and reminders when they receive a notification from the cancer geneticist that a patient has declined testing, especially since it may be desirable for patients to talk with clinicians with whom they have an ongoing relationship when reconsidering genetic testing.

One might also ask, however, whether the cancer geneticist or the medical oncologist has or should have an ethical or legal obligation to recontact those who have refused clinically indicated testing after counseling in order to offer them an opportunity to reconsider their decision. While EHRs could be used to remind clinicians about patients who had previously declined testing, even the most expansive arguments that clinicians have duties to patients after the completion of a clinical intervention (Otten et al., 2015; Stevens, Senner, & Marchant, 2017) should not be extended to create a duty to track down patients to re-offer a test that had previously been refused.

Another intriguing finding of this study is that even though some interviewees expressed concern about insurance discrimination, participants were not particularly worried about privacy *per se*. No one volunteered any privacy concerns spontaneously, and no one expressed these as a major worry even after specific questioning. Several of our participants explicitly did not care who knew about their genetic status or were willing to obtain that knowledge to benefit themselves or others. These findings suggest that the large body of literature that equates discrimination with privacy concerns may mischaracterize individuals' attitudes (Clayton, Halverson, Sathe, & Malin, 2018).

Study Limitations

This study does have certain limitations, which point the way to future research. As the majority of the patients interviewed for this study were seen in clinic with concerns about breast cancer or a family history of breast cancer, our sample was limited in terms of gender distribution and medical indication despite our efforts to increase diversity. It would be worthwhile to compare attitudes described herein not only with those of a more gender-

diverse group but also with views of patients who had other indications for genetic testing. Many of the patients we interviewed had been offered testing several years prior to the interview. While this time gap was integral to the discoveries presented in this article, it also means that our interviewees' responses about their thoughts at the time testing was offered are based on recall. Their thoughts at the time of these interviews, however, are pertinent to their understanding of their experience and to their current attitudes about testing. Finally, more needs to be learned about those who refuse referral for counseling in the first place.

CONCLUSIONS

We have discovered that some patients are not resolutely opposed to genetic testing for cancer susceptibility after initially declining the test. The majority of our interviewees reported that they would now be interested in testing if it were again offered even though they had not been prepared to proceed at the time of their initial visit, usually due to the press of other issues. We suggest practical steps to making this opportunity available.

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Table 1:

Patient characteristics (n=21)

Gender	Male	1 (4.8%)
	Female	20 (95.2%)
Race	African American	5 (23.8%)
	Asian	2 (9.5%)
	White	14 (66.7%)
Age at interview	≥59	8 (38.1%)
	40–59	11 (52.4%)
	≤39	2 (9.5%)
Personal history if cancer	No	5 (23.8%)
	Yes [a]	16 (76.2%)
	Breast	12 (75.0%)
	Colon	1 (6.3%)
	Leukemia	1 (6.3%)
	Lung	1 (6.3%)
	Lymphoma	1 (6.3%)
	Renal	1 (6.3%)
	Sarcoma	1 (6.3%)
	Skin	2 (12.5%)
	Thyroid	2 (12.5%)
Family history of cancer	No	1 (4.8%)
	Yes [a]	20 (95.2%)
	Breast	15 (75.0%)
	Other	18 (90.0%)
Number of years between offer of testing and interview	1	4
	2	5
	3	4
	4	5
	5	3

[a]Some participants have had multiple cancers and had family members with multiple forms of cancer. Therefore these numbers will not total 100%.

Table 2:

Reasons to decline cancer genetic testing, expressed by more than one person.

Domain	Instance (*n*= 21)	Exemplary quotes
Personal emotions	8	"If you find out that you're *BRCA* positive, that would be devastating" (306)
		"I wasn't feeling able to do it." (310)
		"I just didn't want another whammy on me at the time of going through already being diagnosed and going through treatment." (302)
Family's emotions	6	"I didn't want to hear that it was my fault that my son had [the disease.]" (502)
		"I don't want to upset [my relative]." (217)
		"For people that know that they have [the disease], we found out there was a high risk of suicide with it. […] I didn't want my children to have to deal with that, and so that was the decision we made, was to not have the testing." (202)
Insurance	6	"Of course they're not going to want to cover somebody that's due to have different kinds of cancers. [My emotional state] was the main concern at first, but the second concern would've been the insurance companies." (507)
		"I decided to get life insurance before I was tested … because I wanted to make sure I could get it if I had the gene." (503)
Lack of utility	4	"The testing would show inconclusive results because I'm Asian. There are not as many patients who are Asian who took it." (306)
		"We had an unusual amount of cancer in my family, but it didn't seem to be hereditary." (406)
		"Since none of my family, my mom, no one had it, so I was like, maybe […] it was not genetic. That's why I didn't go earlier to the doctor." (304)