The Use of a Family History Risk Assessment Tool within a Community Health Care System: Views of Primary Care Providers

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Abstract:

Primary care providers (PCPs) offered input regarding the incorporation of a family health history (FHH) risk assessment tool into a community health care system (CHCS). Sixteen PCPs participated in one of three focus groups. Perceived impediments included the lack of standard screening guidelines, effective screening tests, genetic counseling resources, and services for high-risk patients. The PCPs were concerned about their level of expertise, the cost of preventive health care, and genetic discrimination. They also were concerned about the use of a FHH tool by oncologists within the CHCS because of communication gaps between oncologists and PCPs, lack of clarity regarding follow-up and legal liability, and reimbursement issues. To integrate a FHH tool into a CHCS, PCPs will need consultation and referral services, evidence-based recommendations, and "just-in-time" educational resources. Oncologists who use the tool will need to develop a streamlined communication system with PCPs, establish clearly defined roles, and ensure patient follow-up.

Keywords: Family history | Primary care providers | Community health care system | Personalized medicine

Article:

Introduction

Over the past decade, there has been growing interest in genomic medicine—the use of genetic information to improve health outcomes (Guttmacher and Collins *2002*; Khoury et al. *2000*)—as a strategy for reducing healthcare costs and promoting disease prevention (Abrahams et al. *2005*;

Ginsburg and Willard *2009*). Of particular interest is the use of family health history (FHH) information to stratify individuals into specific risk categories for common complex diseases. The goal of stratification is to identify those people who could benefit from earlier and more frequent surveillance as well as genetic counseling (Rich et al. *2004*). Primary care providers (PCPs) are particularly well situated to use FHH to guide their medical management decisions (Bell *1998*; Collins *1997*; Greendale and Pyeritz *2001*; Kemper et al. *2010*). However, a number of barriers to the use of genomic medicine by PCPs have been identified (Suther and Goodson *2003*). These barriers include a lack of knowledge about genetics and genetic testing (Collins and Guttmacher *2001*; Harvey et al.*2007*), a lack of time to obtain a detailed family history (Blumenthal et al. *1999*; Valdez et al. *2010*), a lack of referral guidelines or tools to facilitate the use of family history information (de Bock et al. *1999*; Emery*1999*; Friedman et al. *1997*; Fry et al. *1999*; Lucassen et al. *2001*), and a lack of information about genetic services available to patients at increased risk for disease (Hayflick et al. *1998*).

Efforts have been made to address some of these barriers. For example, educational needs assessments have been conducted (Burke and Emery *2002*; Julian-Reynier et al. *2008*; Rich et al. *2004*; Trinidad et al.*2008*), and a list of core competencies in genetics for healthcare professionals has been created by the National Coalition for Health Professional Education in Genetics (National Coalition for Health Professional Education in Genetics *2007*). Educational programs have also been developed to address PCPs' lack of general genetics knowledge and improve their genetic literacy (Burke et al. *2002*; Clyman et al. *2007*). Examples include changes in medical school curricula (Korf *2002*), implementation of the Genetics in Primary Care program which was designed to improve the incorporation of genetic information into training programs for medical students and residents (Burke and Emery *2002*; Laberge et al. *2009*), and continuing medical education programs for practicing physicians (Carroll et al. *2009*; Smith and Scott *1988*).

Computer-based family history and risk assessment programs, such as the U.S. Surgeon General's tool "My Family Health Portrait," have been developed (Cohn et al. *2010*; U.S. Department of Health and Human Services *2010*; Yoon et al. *2009*) to eliminate the barrier posed by the lack of time that PCPs have to collect a detailed FHH. Some of these FHH tools also incorporate risk assessment algorithms and risk-based recommendations to further facilitate the use of family history information by health care providers (Cohn et al. *2010*; Yoon et al. *2009*). However, few studies have looked at the use of these tools within a community health care system (CHCS).

"MeTree" is an example of a computer-based family history collection and risk assessment tool that was being developed at the time of this study. Development of MeTree was undertaken as part of a larger demonstration project whose goal is to create a practice based model to facilitate the integration of genomic medicine into a CHCS. MeTree was designed to identify patients at increased risk for breast cancer, colon cancer, and thrombophilia and offer recommendations for screening and or treatment in the form of reports for both the patients and providers. These

conditions were selected because evidence-based guidelines exist to guide the medical management (e.g., to reduce risk and/or enhance screening) of at-risk individuals (NCCN *2011*).

This study was undertaken to identify potential barriers to the eventual use of MeTree by PCPs, oncologists and hematologists within a CHCS, in Greensboro, NC. The rationale for working with the oncologists and hematologists at the Regional Cancer Center (RCC), in addition to primary care providers, was that they see a subset of patients who are more likely to have familial and hereditary forms of cancer and thrombosis. Use of a family history collection tool with decision support in this setting could potentially result in the identification of a greater number of patients, and therefore families, who could benefit from earlier and more frequent screening, and possibly a referral for genetic counseling and testing.

One of the potential outcomes of enrolling patients seen at the RCC was that they would approach their primary care providers with questions about their risk for the pilot diseases before their PCPs had seen the MeTree report. This raised questions about referral practices, medical management and what PCPs would need in order to counsel and support their patients.

Given that the staff at the RCC specialize in treating patients with cancer and not following at-risk individuals, we wanted to learn 1) what information/resources PCPs would need if the FHH tool were used at the RCC, 2) the potential barriers to the use of MeTree by the RCC, and 3) what steps could be taken to circumvent anticipated problems. The decision was also made to ascertain 4) the current risk assessment practices of PCPs, 5) how they manage at-risk patients and 6) the expectation PCPs have when working with medical specialists.

Methods

Focus groups were chosen as the method of study because qualitative techniques can provide a more in-depth understanding of opinions as well as allowing for group interaction (Krueger *1994*). A semi-structured interview guide with a total of nine questions was produced with input from genetic counselors, experts in qualitative research, and healthcare providers (Table 1). Specific questions about the use of the MeTree tool at the RCC were included to explore the advantages and disadvantages of using the tool in this potentially high-risk population.

Table 1 Questions

1. What do you currently do to assess your patients' risk for common diseases?
2. How does this information affect your medical management?
3. When you refer a person with a family history of a common disorder like cancer to a specialist what are your expectations?
4. If you referred a patient with breast cancer to the Regional Cancer Center for treatment and

she was found to carry a BRCA1 mutation, what information would you like to receive to manage her on-going care?
5. What if this patient completed the MeTree tool and was found to be at increased risk for colon cancer. What information would you like to receive to help you manage her on-going care?
6. If one of your patients is found to be at high risk for cancer, how would this impact your care of their relatives in your practice?
7. What type of problems, if any, can you anticipate occurring if your patient participates in the Genomic Medicine Initiative while receiving services through the Regional Cancer Center?
8. What can we do to circumvent these problems?
9. What other questions or concerns do you have about this project?

All PCPs in the region that had referred patients to the RCC within the previous 12 months were eligible to participate in this study. They were recruited using IRB-approved letters. A total of 353 letters were mailed to potential participants. Follow-up phone calls and faxes were sent to non-responders.

Three focus groups were conducted from July 2006 through August 2006. Focus group size ranged from 4 to 6 providers. Each focus group was conducted by a single facilitator (CAC) with at least one note taker who helped distribute a form to collect demographic information from the participants, record the session and take field notes. The focus groups lasted approximately 90 min. They were conducted at a local restaurant with chairs around one large table so that everyone could see each other. The focus groups were audio-taped after participants had signed informed consent forms. Each participant was offered a meal as compensation for his or her time.

A script was read at the start of each focus group as part of the informed consent process and a description of the MeTree tool was provided. Focus group participants were informed that patients would be recruited by staff at the RCC to complete the family history tool and that both the patients and providers would receive a report listing topics for discussion regarding the patient's risk for the pilot diseases and recommendations for screening and/or treatment. Mock reports were given to each participant during the focus group. The reports included information on a 51-year-old female patient who had a family history suggestive of hereditary breast and ovarian cancer syndrome, a personal and family history of thrombosis, and a 5 year risk of breast cancer >1.66%. These indications were provided on the report as the rationale for the recommendations to refer the patient for genetic counseling, consider chemoprevention, and talk to the patient about routine colon cancer screening. References were also provided for relevant articles regarding these recommendations. These mock reports were distributed in order to give

participants a context in which to consider the type of information they would like to receive about patients who used the MeTree tool at the RCC, and the resources they would need should their patients approach them with questions about their report.

Data Analysis

The taped conversations were transcribed verbatim and reviewed for accuracy by the note taker against the audiotape and field notes taken during each session. Each statement within the transcribed data was coded by location and topic area. This allowed the investigators to not only analyze each statement in isolation but also be aware of where the statements occurred throughout the transcripts. All coded statements were then grouped according to consistent themes. Reliability and validity measures were applied. These included using: an interview guide; a single facilitator; an audiotape to record the focus groups; a note taker to document the focus groups and write down observations; and written transcripts from each focus group for analysis (Krueger *1994*). The computer program, N'Vivo©, was used for data management. Because of the small number of participants in each group, complete saturation was not achieved, but a number of themes were identified and are reported herein.

The methodology for the focus groups was approved by The University of North Carolina at Greensboro and the U.S. Army Medical Research and Materiel Command Institutional Review Boards.

Results

Sixteen PCPs (14 physicians and two mid-level providers) representing nine practices in Guilford County, North Carolina participated in the focus groups. Demographic data are provided in Table 2, excluding the two mid-level providers because neither filled out the demographic data collection form. The average age of the physicians was 52 years, with a range from 31 to 64 years. The average number of years in practice was 23 (range 4 to 33 years). All but two physicians had been practicing for over 20 years.

Table 2 Personal characteristics of respondents

Characteristics	**Total**	
	***N* = 14**	**%**
Gender		
Male	12	85.7%
Female	2	14.3%
Race		

Caucasian	13	92.9%
African American	1	7.1%
Hispanic or Latino		
Yes	1	7.1%
Age (Range 31–60 years)		
≤39 years	2	14.3%
40–49	2	14.3%
50–59	9	64.3%
≥ 60 years	1	7.1%
Board specialty		
Family medicine	8	57.1%
Internal medicine	4	28.6%
Obstetrics/gynecology	2	14.3%
Years in practice (Range 4–33 years)		
≤10 years	2	14.3%
20–29 years	8	57.1%
≥30 years	4	28.6%

Participants specialties included: eight family practice physicians, one of whom specialized in geriatrics and another who specialized in sports medicine; four internists—one was boarded in both family practice and internal medicine and one was boarded in both internal medicine and pediatrics; and two obstetrician/gynecologists. The majority of participants were Non-Hispanic ($n = 13$), Caucasian ($n = 12$) and male ($n = 12$). One participant was African American and one was Hispanic.

The themes that emerged were organized into five main categories (Table 3).

Table 3 Themes

Themes	**No. of groups**
Risk assessment practices	
Collection of family health history (FHH)	3
Impact of FHH on medical management	3
Challenges to the collection and use of FHH	
Time constraints	3
Patient error	2
Outdated information	3
Logistics and perceive utility of risk assessment tools	3
Limits to physician knowledge/experience	3
Barriers to the use of FHH in medical management	
Need for guidelines	3
Cost	3
Patient confidentiality	2
Genetic discrimination	3
Potential harms of testing	2
Patient adherence to recommendations and cascade screening	2
Challenges to the use of FHH at the RCC	
Lack of systematic communication	3
Assignment of responsibility for follow-up	3
Legal liability	3
Reimbursement	3
Systems to insure appropriate follow-up	3
Resources	

Genetic counseling services	3
Physician education	3
Up-to-date internet site and access to geneticist/genetic counselor	1
Patient education materials	1

Risk Assessment Practices

All participants reported collecting a patient's family history during their initial visit as part of their risk assessment strategy. However, the methods used (e.g., verbally vs. standardized questionnaires) and the level of detail that is collected varied significantly.

Some physicians ask targeted, disease-specific questions about common, complex conditions like heart disease, diabetes, and high blood pressure. The three main cancers inquired about are breast, colon, and prostate cancer. The rationale for asking about these three cancers is that they "…are things that we can screen for." The participating obstetricians also ask about "…genetic disorders, bleeding disorders, and pulmonary [disorders] such as cystic fibrosis," and if their patients are of Ashkenazi Jewish descent they ask about "…Tay-Sachs and Canavan disease."

Participants in two groups talked about how their management of patients changes if they have a positive family history of colon, breast, or prostate cancer, or heart disease and obesity. If patients have a family history of colon cancer, for example, participants reported referring these patients for screening earlier than recommended by the American Cancer Society. For example, one physician reported that if a patient has a first-degree relative with early onset prostate or colon cancer he would begin screening 10 years earlier than the earliest diagnosis. This physician also said that he would refer women for mammograms at age 35 if they had a family history of breast cancer. Another physician stated that family history information "does dictate some of what you do on your physical exam." Two participants commented that a positive family history makes them more insistent when talking to the unaffected family members about getting screened. To demonstrate, one physician said he tells patients with a family history of colon cancer, "You need to be checked for colon cancer. *No ifs, ands,* or *buts*!"

One physician mentioned that if a person has a family history of early heart disease, then he is more aggressive in his medical management and in treating high blood pressure and cholesterol in younger individuals. This participant is also more aggressive in his treatment of diabetic patients if they have a family history of obesity.

Challenges to the Collection and Utilization of Family History Information

Time constraints largely dictate how much family history information is collected. One participant stated, “We typically ask about parents and grandparents because that’s what most of our patients can remember in the timeframe that we need to move on with…our exam.” Other participants reported that they only ask about first degree relatives.

Participants in two focus groups talked about the fact that patients do not always provide detailed or accurate family history information. One physician commented that people were knowledgeable about family members with common diseases like breast cancer, colon cancer, and prostate cancer, but “[a]fter that, it’s hard to tell whether people really have it right.” Additionally, participants talked about the fact that “a lot of people don’t know what their parents died of,” that patients use imprecise terminology, and that patients tend to focus on diseases that are in the media, such as breast cancer, and not the more common diseases like heart disease. Other challenges to collecting FHH include the fact that patients may forget to record this information on a form because it is not the reason for their visit, and that some patients resent filling out forms and write down as little as possible.

The practice of updating a patient’s family history also varies among primary care practices and providers. Some practices use a questionnaire to update a patient’s family history. However, it is up to the patient to complete these forms, and patients do not always comply with this request. A few participants said that updated family history information is collected only “on the periodic exams,” or “if something new comes up.” Physicians in two groups said that they only ask questions if a patient complains of symptoms and they are trying to establish or rule out a diagnosis. Only one participant mentioned that he routinely updates his patients’ family histories, collecting information “every 2–3 years when I have people come in for an annual exam.”

While participants reported collecting family history, many cited reasons why this information is of limited use in assessing a patients’ risk for disease. One reason given for this limited utility is the location of the information. Unlike a problem list that is usually in the front of patient charts, the complete family history may not be located in a single place within the chart. As one physician reported, “it may not register [with him] as being significant.” Another physician said that the information is “sort of embedded [as] part of the … sequential notes” and that he “has to really dig to find… family history.”

Comments regarding the use of other risk assessment tools were mixed. Several physicians reported that they have used the NCI Breast Cancer Risk Assessment tool in the past to calculate their patients’ risk for breast cancer. However, none reported using this tool on a regular basis. As one participant put it, he stopped using this program because “[i]t took a fair amount of time to do it, and there were so few people who [had] a positive score.”

In contrast to the Breast Cancer Risk Assessment tool, a number of respondents reported using the Framingham calculator multiple times a day because they see far more patients at risk for

cardiovascular disease. One participant even stated that he has the Framingham calculator on his PDA because he uses it so often throughout the course of a day.

Participants in all three groups expressed some level of discomfort when working with patients who have a family history of breast cancer, ovarian cancer and other, less common, conditions. Some physicians commented on their lack of knowledge as a reason for their discomfort as well as the small number of patients they see who have a significant family history of these diseases. One physician said "…we look at cardiovascular [disease], diabetes, and colon cancer, and if there's a question of anything else, the patient gets referred for another opinion. We just can't possibly tackle everything."

Participants also commented on the lack of genetic counseling resources and high risk breast cancer clinics available to them. One physician refers patients with a family history of breast and ovarian cancer to "a general surgeon that does a lot of mastectomies and… [is] more educated and up-to-date on that than [she is]." Another physician said, "We need to streamline the system for accessibility to genetic counseling through our oncology service here in [town]." He also commented that he does not "think the majority of the physicians are well-qualified or know the information or where to go … to offer this essential [genetic] testing for patients."

Perceived Barriers to the Use of Family Health History Information in Medical Management

All participants agreed that physicians want a list of recommendations from the medical specialist with whom they consult when patients are identified at increased risk for specific common diseases. Physicians in all three groups commented on the absence of standard screening guidelines for common diseases. They mentioned that the effectiveness of screening tests such as early mammograms vary, and that even among experts, there are questions about the value of specific screening tests like the prostate specific antigen test (PSA) for prostate cancer and CA-125 for ovarian cancer.

The issue of cost and insurance coverage were brought up in all three groups. Participants mentioned that screening and genetic tests are often expensive, uncomfortable, and not covered by insurance. Unless insurance companies cover these costs, participants felt that it is unlikely that patients at increased risk will be able to benefit from genomic medicine. One physician thought that it will be necessary to convince third party payers to cover the cost of predictive tests and preventive medicine if genomic medicine is going to work. Fear of genetic discrimination was stated as another reason why patients may not comply with screening recommendations.

Several participants in two groups were particularly concerned about patient confidentiality and the topic of genetic discrimination was brought up in all three groups. Participants in one group talked about the "stigma" that might be attached to having a gene mutation and the possibility that patients could be "shunned from society." They also discussed the need to develop secure databases to reduce the risks associated with electronic medical records. For one participant,

these risks were sufficient to keep him from using the MeTree tool and from allowing patients to use the tool in his practice.

Participants in two groups brought up concerns about genetic testing doing more harm than good, resulting in a "genetic underclass." Participants were concerned about the rate of "false positive" tests and the potential misconceptions that patients may have about genetic tests (i.e., not making a distinction between predisposition and presymptomatic testing). One participant was concerned that patients would think that having a genetic test will rule out all genetic disorders and that screening "…will prevent [them] from getting anything." Participants also worry that patients will insist on having tests that are not medically necessary.

Concerns about the potential effectiveness of using family health history to promote behavior change were also raised in two groups. A number of physicians commented on the need to overcome "patient stubbornness," "inertia and noncompliance," if the benefits of identifying people who are at increased risk for disease are to be realized. They voiced skepticism about their patients' ability and/or willingness to change their behaviors and lifestyles. They also pointed out that "there has to be community education" and that people must be "receptive to this information" if use of a family history collection tool is going to result in better health outcomes.

When asked about at-risk family members, participants in two groups commented that they do not often take care of multiple people in a family and that there is no good way to inform other family members of their risk. Their solution was to "try to identify a family member who will get the info to other members of the family" and pass onto this individual written information they can copy and send to their relatives.

Potential Challenges to the Use of a Family History Collection Tool by the Regional Cancer Center

Focus group participants brought up a number of concerns when talking about the use of the MeTree tool at the cancer center. These included 1) the current system of communication between the oncologists and PCPs, 2) who would be responsible for the follow-up of the MeTree tool recommendations, 3) the potential legal liability that would be incurred by the PCPs if the responsibility for follow-up fell to them, and 4) the lack of reimbursement for the PCPs' time. There was also some discussion, but no consensus, about the systems that would need to be put in place if the oncologists and hematologists at the RCC were to use the MeTree tool.

Communication

Participants in all three groups expressed concern that the MeTree results would not be sent to them by the RCC because of an existing lack of systematic communication between the RCC staff and primary care providers. The PCPs pointed out that RCC staff generally communicate with the referring physician(s). As the PCPs' patients are often referred to the RCC by their surgeons, radiologists, gastroenterologists, or other specialists involved in their initial work-up,

primary care providers are often "…cut out of the loop." Unless patients specifically ask their oncologists to send reports to their PCPs, the PCPs may never receive them. PCPs also expressed concern regarding the time it takes to receive a report from the RCC when one is sent.

Participants in two groups pointed out that this lack of feedback puts them at a significant disadvantage when patients ask "What am I supposed to do now?" They expressed the need for "good guidance" in order to respond to these questions, facilitate decision making, and "…reinforce whatever recommendations are made." As one participant put it, he needs "…the most up-to-date information about what the options for testing are [and] once the testing is done, what the options [are] for addressing the results."

Responsibility for Follow-Up

Given the perceived lack of communication between the RCC staff and PCPs, participants were also concerned about who would assume responsibility for follow-up. The oncologists focus on treatment, not diagnosis or prevention. Therefore, if the MeTree tool identifies someone who is at familial or hereditary risk for cancer, participants cited a need for a system to determine who is responsible for the follow-up. Participants voiced concern about the risk of patients falling through the cracks, or the duplication of efforts on the part of the RCC and PCP. One physician said:

I'm worried about … these reports coming in and they plop on your desk, and you don't know who is responsible for them. You know, is this something that the oncologist has already taken care of? Or, are you going to have two people or two systems independently scheduling colonoscopies?

Participants were concerned that the oncologists would either send them a copy of the MeTree report, or that, patients would be instructed to bring the report with them to their next primary care appointment. If the former was implemented, participants worried they would be responsible for contacting the patient to schedule an appointment to discuss the information on the report that was not pertinent to their patient's current cancer diagnosis. In the latter situation, a number of participants pointed out that many patients with cancer are "overwhelmed with the diagnosis" and "forget their primary care physician." Or, if they do see their PCP, it is for an acute situation that may prohibit a proper discussion of the MeTree report.

Legal Liability

The issue of legal liability was raised by participants in all three groups. One participant indicated that "every piece of paper we get in the office generates some potential liability." Another participant asked, "Do I want the liability of getting a letter that says, 'This patient is at increased risk; you may want to handle this.'? And, the patient hasn't chosen to come to me for 3 years because they've been using their oncologist as their primary care doctor."

Participants wondered what liability would be incurred if they were unable to reach the patient and schedule an appointment to go over the results of the report. Their fear is that they will miss something or that a patient will be lost to follow-up if these recommendations are not addressed by the oncologists at the time the report is generated.

Reimbursement

A number of participants in each group commented on the overhead costs they would incur if they were expected to provide follow-up counseling for patients identified at the RCC as being at increased risk for cancers other than the one for which they were being treated. Participants from one practice mentioned the cost they would incur if they added to the work load of their care coordinators. Another group was concerned about the lack of reimbursement for counseling patients who have a family history of cancer. As one participant pointed out, “There are V codes that pertain to this kind of thing. And, you know how reimbursement works for V codes? Zero.” Another participant said, “So, no matter how much good information I get back [from the MeTree report]….unless [I am] compensated,…I’m going to have it on a lower priority.”

Proposed Systems to Insure Appropriate Patient Follow-up

One solution offered by participants in all three groups to address some of these problems is to divide the responsibilities for screening between the PCP and oncologist based on a patient’s risk for disease. The PCPs expressed confidence in their ability to manage patients at average risk who can follow routine screening guidelines. However, PCPs would like the oncologists to follow patients identified by MeTree as being at increased risk for disease. If the oncologists at RCC expect the PCPs to manage those patients at increased risk for disease they will need to provide the PCPs with specific instructions regarding medical management.

Requested Resources

Participants in all three groups expressed the need for local genetic counseling services. They were also interested in gaining genetic knowledge through education. One participant commented that “there’s a huge educational process that has to happen for primary care physicians surrounding genetic testing and genetics.” Nevertheless, it is unclear whether they would take advantage of formal educational opportunities. As one participant said “The huge problem is things change from year to year and we have many more problems than this—this little niche of medicine.”

The educational venues they mentioned included monthly grand rounds, a Woman’s Health Symposium at the local University geared toward primary care physicians, and lunch meetings in their office. They also mentioned that programs could be provided through the Area Health Education Center (AHEC), which is a typical place for PCPs to go for continuing education in this community. Additionally, participants in one group said that they would like access to an up-

to-date internet site. They want specific guidelines and “a set of recommendations that people can download onto their Palm Pilots.”

Another resource specifically requested by participants in two groups was access to healthcare professionals, such as genetic counselors, who will respond to their questions by telephone or e-mail. For instance, a participant claimed, “What I’m interested in is simply a resource for genetic counseling. That would be helpful. And, even a place we could call for free help to [find out], ‘Should I send this patient over or not?’.” One participant requested “information that we can hand out, give to the patients here in our community who [are] at risk for breast cancer or any other genetic disorders.”

Discussion

The findings of this study suggest a number of obstacles to the use of family history collection tools within a CHCS. While PCPs currently collect and use some FHH information in the management of their patients, time constraints, patient agendas, and the lack of a systematic strategy to update FHH information, may still impede the identification of patients who could benefit from increased screening, treatment, or genetic counseling. Even if a tool is put in place that reduces the time involved in collecting and interpreting FHH, PCPs expressed reluctance to delve into patients’ family histories because of their self-proclaimed lack of knowledge about the less common conditions, and the lack of evidence-based screening guidelines for individuals identified at increased risk for disease (de Bock et al. *1999*; Fry et al. *1999*). Focus group participants were also concerned about the cost of genetic tests, the lack of insurance coverage for preventive medicine (Friedman et al. 1997; Watson et al. *1999*), and poor reimbursement for counseling. Issues regarding genetic discrimination and privacy may also prevent the adoption and use of tools like MeTree in primary care.

Focus group participants pointed out that many patient-related barriers will need to be overcome if the benefits of genomic medicine are going to be realized. For instance, PCPs repeatedly noted that patients are not always able or willing to provide a detailed FHH. This is especially true in the case of heart disease and mental illness (Qureshi et al. *2009*), which highlights the need for community education programs that focus on the importance of collecting FHH information (Christianson et al. *2010*). Other factors that may impact genomic medicine’s success in reducing the risk for, or preventing common complex disease include the lack of effective screening tests, and patient resistance to screening tests that they perceive as painful or embarrassing. PCPs were also concerned about patients’ misconceptions regarding genetic testing. They worry that patients will not make a distinction between predisposition and presymtomatic tests, or appreciate a test’s limitations.

The lack of a system to reach out to at-risk family members is also perceived as a potential impediment to genomic medicine’s overall success in reducing the risk for disease. This is a particular problem if patients are unwilling to share the results of their risk assessment or genetic

tests with at-risk family members. In a study by Cohn et al. (*2010*), 75% of participants said they would tell their relatives if they had inherited a gene that increased their risk to develop a disease. However, when asked if they had shared their risk information with other family members, only 40% indicated they had done so (Cohn et al. *2010*).

Regarding the issue of education, the focus by participants on immediately accessible resources, such as consultation services and up-to-date information on a trusted website, suggests that "just-in-time" educational resources may be more effective than using traditional education models that deliver standardized content at a distinct time and place as suggested in the literature (Julian-Reynier et al. *2008*; Kemper et al. *2010*; Metcalf et al. *2002*; Trinidad et al. *2008*). Educational programs that allow the learner to access information when the need for it arises may be the most effective. This is especially true for PCPs who are reluctant to adopt new genetic tests, as they are unlikely to seek out formal educational programs to expand their knowledge of genetics (Metcalf et al. *2002*).

Use of Risk Assessment Tools in High Risk Populations

Based on the comments of PCPs, the use of risk assessment tools, like MeTree, by oncologists and hematologists working with high-risk patient populations needs to be undertaken with forethought and planning. In these instances, a tool designed to identify patients at increased risk for conditions not typically treated by the specialist may reveal risks that go unnoticed and are not addressed. The advantages gained by identifying patients at increased risk for common chronic diseases will be lost if the specialist simply sends a report about the specific condition back to a patient's PCP, or relies on the patient to bring it up during their next appointment. Patients in the midst of a health crisis may be relying on their specialists for all of their health care needs and not see their PCPs for extended periods of time, or patients may simply forget to discuss their risk for other diseases with their PCP when they do see them because their focus is on their more immediate health problems.

For the system to work, specialists who use a comprehensive FHH collection tool will need to review each component of the report with their patients and take responsibility for making appropriate referrals. Patients at average or familial risk for a disease can be referred back to their PCP for follow-up as long as detailed medical management guidelines are provided. The medical management of patients at high risk for disease may be best handled by specialists in high-risk clinics, although PCPs can also manage these patients if they are provided with detailed management protocols and support.

Study Limitations

A limitation of this study is the low response rate and the fact that we were unable to achieve qualitative saturation in the range of responses. Nevertheless, the topics discussed by the focus group participants were consistent with findings from the literature. Another limitation is that the participants were self-selected and may have had a greater interest in genetics than the typical

PCP. Finally, we can make no claim that focus group participants are representative of the group from which they were drawn because we were unable to collect demographic information on non-responders, and qualitative data are not intended to be generalizable to the population of interest.

However, these results will inform the development of the infrastructure necessary to support the integration of a FHH collection tool into a CHCS, and the evaluation of the efficacy of using this tool to identify patients at elevated risk for chronic conditions in primary care.

Practice Implications

Genetic counselors can assist in all aspects of development of the necessary infrastructure to ingrate FHH collection tools into primary care. For instance, genetic counselors can set up consultative services for PCPs who want to use a FHH tool in their practice. They can develop the necessary educational materials and resources for providers and their patients. Genetic counselors can also participate in the development of screening guidelines to manage patients at increased risk, and assist PCPs in developing systems to promote cascade screening of all at-risk relatives. To address the concerns associated with the use of FHH by specialists, genetic counselors can participate in the development of systems of care that allow specialists to use a collection tool like MeTree to screen high risk populations. Finally, counselors can also play an evaluative role to ascertain whether these systems work.

Conclusions

The development of family history collection tools and programs to improve the genetic literacy of PCPs are important steps in personalizing health care. Nonetheless, by themselves, such tools may be insufficient to promote the integration of genomic medicine into a CHCS.

The development of family history collection tools and educational programs for providers will only alleviate some of the many barriers to the identification of patients at increased risk for disease who might benefit from earlier or more frequent screening, treatment, genetic counseling and testing. Our focus group results suggest that PCPs who want to integrate genomic medicine services into their practice will need a supportive infrastructure including email or phone access to geneticists or genetic counselors, evidence-based guidelines to prevent or reduce a patient's risk for disease, "just-in-time" educational resources such as an up-to-date website, a system of health care providers who specialize in the care of patients at increased risk for common chronic conditions and access to genetic counseling services for their patients at risk for hereditary syndromes. Strategies to educate patients on the importance of collecting accurate and detailed FHHs, and to help patients make lifestyle and behavior changes will need to be developed. A system to promote the communication of information to at-risk relatives will also need to be developed. Misconceptions regarding genetic testing and the risk of genetic discrimination must be addressed at both the professional and community level, and discussions with health insurance companies about the value of predictive genetic testing and preventative medicine must continue

if the potential benefits of genomic medicine are going to be realized. Oncologists and hematologist who use comprehensive FHH tools will also need to establish working relationships with PCPs in the community to ensure that patients are followed appropriately.

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