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Primary care physician management, referral, and relations with specialists
concerning patients at risk for cancer due to family history

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ABSTRACT

Background: Risk stratification based on family history is a feature of screening guidelines for a number of cancers and referral guidelines for genetic counseling/testing for cancer risk.

Aims: To describe primary care physician perceptions of their role in managing cancer risk based on family history.

Methods: Structured interviews were conducted by a medical anthropologist with primary care physicians in three settings in two north-eastern states. Transcripts were systematically analyzed by a research team to identify major themes expressed by participants.

Results: Forty interviews were conducted from May 2003 through May 2006. Physicians provided a diversity of views on roles in management of cancer risk based on family history, management practices, and patient responses to risk information. They also provided a wide range of perspectives on criteria used for referral to specialists, types of specialists referred to, and expected management roles for referred patients.

Conclusion: Some primary care physicians appeared to make effective use of family history information for cancer risk management, but many in this sample did not. Increased focus on efficient assessment tools based on recognized guidelines, accessible guides to management options, and patient education and decision aids may be useful directions to facilitate broader use of family history information for cancer risk management.

MESH Key words: family history, medical history taking, family, medical genetics, risk assessment, cancer, primary health care.

INTRODUCTION

Individual cancer risk assessment can be difficult given the variety of factors associated with risk and the lack of consistent guidelines. However, risk stratification and identification of individuals at above average risk for cancer is important as these individuals may be candidates for more aggressive screening and cancer prevention strategies including chemoprevention.

Family history is a relatively simple and commonly utilized risk assessment tool. A variety of studies have demonstrated the clinical relevance of family history in cancer risk assessment. [1] [2, 3] [4, 5] Family history is a key feature of screening guidelines for breast, colon, ovarian, prostate cancer [6] [7] [8] [9] and referral guidelines for genetic counseling and testing for cancer risk. [10, 11]

Several studies have identified both facilitators and barriers to obtaining and updating family history information and using family history as a cancer risk assessment tool. [5, 12] [13]. Our previous research based on surveys of a diverse sample of primary care physicians found that they commonly used family histories in specific circumstances and had generally positive perceptions of cancer family history utility. [14] Other studies have found substantial barriers to effective application of cancer family history information including limitations of patients' family history knowledge; lack of methods to systematically and efficiently focus on the most useful information, and lack of accessible guidance for risk stratification and management. [5, 15, 16] [17].

The present study was designed to document primary care physician perceptions of their role in managing cancer risk based on family history, the perceived prevalence of notable cancer family histories among their patients, the management actions they might take for patients at higher cancer risk due to family history, criteria they use for referring such patients to specialists, and types of specialists and relationships with specialists to whom they might refer higher risk patients.

METHODS

Family and general internal medicine physicians and gynecologists were recruited from urban and suburban Boston and rural Vermont for personal interviews. In each area a partner organization facilitated sampling and recruitment from comprehensive provider lists. Urban physicians were recruited from a large health center and affiliated network of community health centers serving ethnically and economically diverse populations. Suburban physicians were recruited from a healthcare organization serving Eastern Massachusetts, but restricted to practices in suburban communities. Rural physicians were recruited from a statewide rural health network, excluding those serving one urbanized county.

The sampling goal was to maximize the chances of identifying factors relevant to use of the cancer family history for management of cancer risk. A stratified purposeful strategy was used to recruit physicians with a broad range of personal and practice characteristics; including gender, year of graduation from medical school, and practice type, size, and setting. Physicians were over-sampled for minority status. [18] Institutional review boards at the University of Vermont and other participating institutions approved the study procedures.

Implementation of interviews

Interviews were conducted in-person or by telephone by one person [AS], a cultural anthropologist with significant prior experience interviewing physicians. Interviews were organized around a semi-structured outline developed by the investigator team and reviewed by members of an expert panel; topics were selected based on reviews of related research and research team judgments about the importance of each to the objectives (Table I). Each topic was used as a starting point and explored in differing levels of detail depending on relevance to the interviewee. Interviews lasted 30-90 minutes and were audio-recorded (unless participant requested that interview not be recorded; in these few cases the interviewer took written notes).

Data processing and analysis

Interview recordings were transcribed and checked for accuracy. Formal analysis was undertaken using Atlas-ti, a software program used to organize and analyze qualitative data. Coding was done by one person [AS] in close collaboration with the investigator team. Analysis involved an iterative process of coding, display of relationship patterns, and memos. [19] Descriptive topic codes based on the interview guide were applied initially; the coding scheme was elaborated further based on themes and patterns identified from reviews of the transcripts and initial coded outputs. Independent observations were obtained from expert panel members who reviewed a subset of transcripts. Once coding was completed, data were organized and condensed, using a series of matrices to define code patterns, frequencies, and participant characteristics.

RESULTS

Forty interviews were conducted with primary care physicians between May 2003 and May 2006. The sample was 40% female and largely Caucasian (78.5%), although major ethnic groups were represented (Table II). Medical school graduation dates ranged from 1963 to 2000 (mean 1985, median 1986). Table III provides characteristics of each interviewee. Extended quotes in the following summaries are followed by the number of interview;

Physician views on their roles in management of cancer risk based on family history

Some physicians observed that discussions of cancer risk management required extra time and effort because of patient anxieties about cancer, patients' uncertainty about the value of cancer risk management, and the challenges of explaining risk. For others the immediate and often complex problems facing their patients precluded any substantial investment in cancer risk management. Difficulties in communicating with patients about cancer risk based on family history, lack of confidence in addressing these issues, and the extended period of time that might pass before any potential benefits might be realized led these physicians to consider cancer risk management based on family history as a relatively low priority.

Other physicians said that they were best prepared to serve as advisors or educators who raise awareness of cancer risk and potential benefits of risk management. These physicians described

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their role in referral to specialists as identifying appropriate patients, educating them about their risk, and persuading them to accept referrals.

Physicians who were more active in cancer risk management described their role as requiring high sensitivity to patients' individual situations. Maintaining a balance between informing patients and avoiding undue anxiety can be difficult. These physicians described different levels of engagement with cancer risk management depending on their judgments about patients' risk status and patients' receptivity to the information.

Identifying patients at risk for cancer based on family history

Physicians expressed a wide range of views on assessment of cancer risks based on family history. One group emphasized that few of their patients had cancer family histories that would affect their management or referral decisions. Their comments focused on the relatively rare patients who might have “a clear cancer syndrome”. The most common descriptions of these situations were quite general and often dramatic, e.g. “significant,” “strong,” “horrible,” “striking,” or “suspicious” family histories. One physician stated that further attention was warranted if there were “too many people and [physician] feel[s] uncomfortable”. Several physicians mentioned that “a lot of cancers” in a family history might justify a referral.

Another group took a broader view of a meaningful cancer family history. These physicians typically described cancer family histories that merited additional attention in more specific terms such as “one first degree relative with breast cancer at any age”; “a lot of cancer and early onset”; or “multiple close relatives”. A few physicians described more explicit criteria including: “mother with breast cancer at young age and Ashkenazi heritage”; and “two or three first degree relatives at young age”.

Management of patients with above average risk

Physicians taking a more active view of cancer risk management described several approaches for their patients at higher risk. Most commonly, risk information (family history of cancer) was used to encourage patients to follow standard cancer screening recommendations. This situation would prompt one physician to “follow screening guidelines extremely strictly”. This approach

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was used by some physicians with high risk patients who refused referrals for further evaluation. Greater emphasis on screening guidelines was mentioned particularly with regard to colorectal cancer screening.

“Colonoscopy is important. If there is some family history [I] use it to push patients to do screening. Same with mammography.” [10]

Few physicians recommended enhanced cancer screening, such as starting at an earlier age or screening at more frequent intervals than for average risk patients. Starting screening ten years earlier than the age of an affected family member was mentioned by several, although, as one pointed out, this response is relevant only for younger patients. Use of modified screening protocols for risk management appeared to be based more on familiarity with and accessibility of screening rather than on specific guidelines or expert recommendations. As one physician noted, this strategy is “not based on a foundation of knowledge but seems a legitimate approach” [28].

Patient resistance to screening recommendations based on risk status was described by several physicians. Some patients refused screening even when elevated risk was evident. Refusal of colonoscopy was more common than refusal of mammography.

“Some would rather die than go through [colorectal cancer] testing.” [35]

Lifestyle change and chemoprevention were mentioned infrequently. The only specific lifestyle change mentioned was smoking cessation, although the general concept of lifestyle change to complement screening enhancement was mentioned by several. Chemoprevention mentioned included selective estrogen response modifiers (SERMs) such as Tamoxifen and non-steroidal anti-inflammatory agents (NSAIDs) such as aspirin. Nearly all of the few physicians who mentioned these approaches stated that they were unlikely to recommend them on their own, leaving the issue for a specialist.

Genetic testing followed referral and counseling by a specialist most often. Just four of these physicians had ever directly ordered a genetic test. These included situations where a family member was known to have a specific genetic mutation or where patients requested the test.

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Two of these four mentioned that they would not order a genetic test or refer for counseling if the patient definitely would not consider surgical treatment as a result of a positive test.

BRCA gene testing is only significant if patient would do surgery. [16]

...discussed BRCA testing and whether she'd do oophorectomy. If no way she'd do surgery, wouldn't refer. [11]

Referral of patients to specialists

Although the need for referrals in some situations was generally (but not universally) acknowledged, most physicians described very low referral rates for assessment of cancer risk due to family history. Four physicians indicated that they have never, or rarely, referred because of a family cancer history. Eleven physicians indicated that they seldom referred to any specialist for this reason. Reasons given for low referral rates usually indicated that only the strongest family histories raised sufficient concern to justify a referral. Only six physicians indicated a significant flow of family cancer history referrals ranging from 1-10 per year.

Types of specialists to whom patients were referred also varied considerably. Specialists or referral resources mentioned included oncologists, cancer services, or cancer centers (mentioned by 8 physicians), breast care centers (4), cancer services that offers genetic counseling (4), genetic services (centers, counselors or geneticist) (mentioned by 17 physicians); and others including gynecologists, GI specialists, and surgeons. Decisions to refer to cancer specialists rather than genetics specialists appeared to be influenced by access issues, such as convenient location for patient, and familiarity with the service or the specialists.

Degrees of shared responsibilities with specialists

These physicians generally had clear views of their role with regard to referred patients. They emphasized their role as a "translator" of specialist recommendations and source of support in coping with the emotional consequences of the specialist consultation. They have knowledge of the individual patient and the patient's circumstances, and trust of the patient. They are therefore well-prepared to help them place the issues in context and assist patients thinking through implications of specialist recommendations. Several indicated that they would prepare a referred

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patient for a consultation by reviewing important areas where the patient should seek information and advice from the specialist.

However, these physicians expressed a range of views on their specific management roles in relation to specialist roles. Some indicated that they preferred to be the key health care decision maker and manager, utilizing assessments and other information from specialists. Others described a joint management process, with specialists taking responsibility for some issues and primary care physicians responsible for others (e.g. enhanced screening). A third group accepted that referred patients would be managed for cancer risk mainly by specialists. Several focused on a role limited to encouraging patients to follow through on specialist referrals and recommendations.

These physicians generally, but not always, indicated good relationships with specialists. Some mentioned that they felt free to call for advice or to see if a referral was appropriate. Others, however, described difficult relationships where communication regarding referrals was poor (in most cases these comments focused on a particular service or specialist).

A wide range of post-referral communication experiences with specialists were described. Some indicated highly satisfactory reporting of results and recommendations. Others described mixed or inconsistent experiences. A third group appeared to seldom receive or expect feedback after a specialist referral. Among those reporting strong communication links, several emphasized the importance and educational value to the primary care physician of specialist reports and related feedback.

... often doesn't get much feedback from genetic counseling referrals, just enough to know the patient has gone. [28]

...does not get much narrative on genetics from gastro docs. Very focused on results of examination. Little concern with family history or genetics. [24]

Differing patient views of cancer risk based on family history

In addition to differing views among themselves, physicians reported a wide range of perspectives among patients that posed challenges even when the physician placed a high

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priority on these issues. Patients who had some prior awareness of increased cancer risk based on family history were described as more likely to accept their physician's management recommendations. They were described as "not surprised" about increased risk, and willing to accept recommendations. Several said that most patients with positive genetic tests were likely to follow subsequent recommendations for enhanced screening or surgical management.

Physicians also described patients who were reluctant to accept cancer risk management recommendations. Some were described as not interested in preventive care or learning about their risks. Some patients actively refused referrals for further evaluation when offered.

"Some patients just want to focus on the present." [34]

"Few want to know or do something with (risk) information." [13]

"Discussion of risk depends on personality. Some don't want to know." [24]

Some patients perceived that risk based on family history was not high enough to justify the effort required by a referral. Other patients said that they would not act on any management recommendations that emerged from a referral and so did not think it was worthwhile. A few physicians described referral refusals that played out only after their conversation with the patient, such as patients who did not complete the referral or patients refusing recommendations from consultants. One physician described a patient who was tested but was reluctant to see the results.

These physicians also described patients with average cancer risk who were anxious about their risk and eager to take action such as a genetic test. Most of these patient anxieties appeared to be based on cancers among remote or non-blood family members or friends that did not significantly affect the individual's risk. Direct to consumer marketing accounted for anxieties in a few patients. Some physicians referred these anxious patients for counseling so that they can hear about their average risk from a specialist.

"Some [inappropriate] patients are very focused on getting tests." [22]

DISCUSSION

These results provide a unique account of primary care physician perspectives regarding assessment and management of cancer risk based on family history. These perspectives were drawn from a diverse group of physicians in rural, suburban, and urban regions of two northeastern U.S. states through semi-structured interviews that provided considerable latitude for expression of individual perspectives. These views represented a consistently wide range of positions on general and specific issues concerning assessment and management of cancer risk using family history information. The challenges to more effective use of family history information for cancer risk management posed by differing physician perspectives were complemented, or compounded, by a similarly wide range of patient perspectives on these issues described by physicians.

These physicians expressed differing views on their overall roles with regard to cancer risk assessment and management. Some identified multiple reasons for not making this topic a priority with their patients, listing specific and credible obstacles that reduced the likelihood that this would be an effective use of time. A second group viewed cancer risk as important enough to their patients that they attempted to identify those who may be at risk due to family history, but depended primarily on specialists to provide more focused assessments and management recommendations for patients willing to accept referrals. A third group described more active roles in cancer risk assessment and management.

Similar patterns were seen with regard to perspectives on the proportions of patients at increased risk due to family history, and the criteria used to identify higher risk patients. Some physicians limited their interest in cancer risk to the relatively rare patients with dramatic family histories that might suggest a genetic syndrome. Others described a much broader group of patients who might benefit from attention to cancer risk. These differing perspectives also were reflected in impressive differences in degree of specificity used to identify higher risk patients, ranging from the most general to highly specific, guideline-focused criteria.

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The lack of consensus on appropriate roles and approaches to cancer risk management indicated in these interviews is a both cause for concern and an opportunity for forging new approaches. For example, efficient algorithms to identify risk based on family history is an area where resource development and education can be focused to encourage and facilitate physician interest in addressing these opportunities for risk management. This sentiment was echoed by the 2009 NIH Conference Statement on Family History which recommended that improving methods to use family history in a primary care setting to identify individuals who can benefit from enhanced surveillance or referral to genetic services as an important direction for the future. [20]

Physicians who took more active roles in cancer risk management described a limited range of management practices that they might consider. The most commonly mentioned approach was simply to use family history information to encourage adherence to standard screening recommendations. Few physicians mentioned enhanced screening schedules, specific lifestyle change recommendations, other than cigarette smoking cessation, and chemoprevention. Thus, even among the physicians with the most active approaches to cancer risk management, the range of management methods was limited and may represent missed opportunities. Methods for linking cancer risk assessments with management possibilities within primary care practice appears to be another key area for resource development.

Most, but not all, of these physicians referred patients with a family cancer history to a specialist at least occasionally. Reports of low referral activity for this reason were consistent with the generally low estimates of proportions of patients at risk due to family history, and the modest level of risk management activity reported. The relatively low presence of genetic counseling services among the types of specialists to whom these patients might be referred is notable.

These physicians were clear about their role regarding referred patients. They saw themselves as a coach for the patient – preparing them for the consultation with the specialist and a translator of information – reviewing and assessing recommendations from the specialists. The relative clarity of this view of the primary care physician role in relation to the specialist role is consistent with the medical home concept and should be explored further among patients, primary care physicians, and consultant specialists.

Some physicians were clear about their expectations for communication with specialists often viewing the consultation report as an educational tool for themselves. Prior studies have focused on the barriers to use of family history demonstrating primary care physicians' lack of knowledge and/or confidence in either taking, interpreting or using a cancer family history. [21] [22] [23] [24] Our results highlight the role of the primary care physician in supporting and coordinating care for their patients in collaboration with specialists and the importance of clear and detailed communications from specialists in advancing knowledge and confidence in assessment and management skills among primary care physicians.

Variation in physician views was mirrored in a wide range of reported patient views on cancer risk management. Some patients were reported to have little or no interest in learning about and managing these risks, even when their risk might be high. Other patients were reported to have inappropriately high interest in intensive risk assessments even though they were not at increased risk. These reports suggest a need for more effective patient education methods to complement efforts to improve primary care physician resources and skills regarding cancer risk management. It may be particularly important to provide resources that help physicians communicate risk information to patients whose objective risk may high and may have a good likelihood of benefiting from further action, but who are reluctant to discuss this issue.

A previous report based on these interviews documented numerous barriers to utilization of family cancer history data, including limitations of patients' family history knowledge, lack of methods to efficiently focus on the most useful information, and lack of easily accessible guidance on use of this information for risk stratification and management [18]. These perceived limitations are generally consistent with the limited utilization of family histories reported by many interviewees in the current study. Some of these primary care physicians, however, were very well-informed about criteria for identifying patients at higher risk for cancer based on family history, and active in providing risk management effort to these patients as recommended.

A survey of 880 primary care physicians drawn concurrently from the same populations as those interviewed provided additional evidence for the feasibility of utilizing family history

information for cancer risk stratification [14]. The surveyed physicians expressed generally positive perceptions of the advantages and negative perceptions of the disadvantages of utilizing family history information. These perceptions, reported confidence in skills, and perceptions of supportive resources were independently associated with reported family history quality. The combined results (of the interviews and survey) suggest that, despite perceived barriers, some primary care physicians have the knowledge, skill, and motivation to utilize family history information effectively, and many have positive perceptions that might be catalyzed into action if provided with improved resources that address barriers.

This study has several limitations that should be considered in assessment of these results. The sample was limited to primary care physicians in two states. The urban and suburban samples were drawn from physician members of two large health care organizations; the rural sample in the second state was drawn from a more comprehensive state-wide list of primary care physicians. Membership in a large health care organization may provide more opportunities for patient referrals, although this was not evident in the data. These qualitative data were collected and analyzed systematically but are subject to risks ranging from the specific composition of the relatively small sample of participants to the coding and interpretation of the results. These interviews were conducted between 2003 and 2006 and it is possible that the application of clinical genetics has increased among primary care providers since that time, although the evidence demonstrating such change is not strong.

Conclusions

Some primary care physicians appear to make effective use of family history information for cancer risk assessment and management, but many in this sample did not. The relatively successful implementation of risk stratification guidelines based on family history by some, and generally positive perspectives reported in a prior study by a larger primary care physician sample, suggest that broader implementation of these data for cancer risk assessment and management is achievable for primary care physicians if obstacles are addressed. These challenges include increased focus on efficient assessment tools based on recognized guidelines, more accessible guides to management and referral options for higher risk patients, more consistently informative feedback from specialists to primary care physician following referrals,

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and patient education and decision aids to facilitate better informed patient utilization of cancer risk information.

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CONFLICTS OF INTEREST

The authors declare that they have no conflicts of interest.

TABLE I

Physician Interview Guide Topics

- Medical background, training, and your current practice.
- Training in taking family histories, cancer risk assessment, and genetics.
- Attitudes towards cancer risk assessment and the taking of cancer family histories.
- Collection of family history.
- Assessment and interpretation of cancer risk.
- Awareness and use of guidelines or protocols for the collection and interpretation of cancer family histories.
- Perceived institutional and patient-related barriers or facilitators for taking and maintaining family histories.
- Need for educational and support resources.

Table II
Participant Demographics

	Number	Percentage
Gender		
Male	24	60%
Female	16	40%
Ethnicity		
Caucasian	31	77.5%
Asian	7	17.5%
African American	2	5%
Years from medical school graduation		
Fifteen or less	15	37.5%
More than fifteen	25	62.5%
Practice environment		
Solo	8	20%
Group	23	57.5%
Tertiary	9	22.5%
Primary Care Specialty		
OB/Gyn	9	22.5%
Family Practice	14	35%
Internal Medicine	17	42.5%
Practice Location		
Urban	12	30%
Rural	13	32.5%
Suburban	15	37.5%
Total	40	100%

TABLE III
Characteristics of Quoted Physicians

Interview Number	Gender	Primary Care Specialty	Practice Location
01	Male	Internist	Suburban
10	Female	Internist	Suburban
11	Female	Internist	Suburban
13	Male	Family	Rural
16	Male	OB/GYN	Suburban
22	Male	Family	Rural
24	Male	OB/GYN	Suburban
28	Female	Family	Suburban
34	Female	Family	Rural
35	Female	Family	Rural

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