The primary care physician role in cancer genetics: a qualitative study of patient experience
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Background. Increased availability of genetic testing is changing the primary care role in cancer genetics. The perspective of primary care physicians (PCPs) regarding their role in support of genetic testing has been explored, but little is known about the expectations of patients or the PCP role once genetic test results are received.

Methods. Two sets of open-ended semi-structured interviews were completed with patients (N = 25) in a cancer genetic programme in Ontario, Canada, within 4 months of receiving genetic test results and 1 year later; written reports of test results were collected.

Results. Patients expected PCPs to play a role in referral for genetic testing; they hoped that PCPs would have sufficient knowledge to appreciate familial risk and supportive attitudes towards genetic testing. Patients had more difficulty in identifying a PCP role following receipt of genetic test results; cancer patients in particular emphasized this as a role for cancer specialists. Still, some patients anticipated an ongoing PCP role comprising risk-appropriate surveillance or reassurance, especially as specialist care diminished. These expectations were complicated by occasional confusion regarding the ongoing care appropriate to genetic test results.

Conclusions. The potential PCP role in cancer genetics is quite broad. Patients expect PCPs to play a role in risk identification and genetics referral. In addition, some patients anticipated an ongoing role for their PCPs after receiving genetic test results. Sustained efforts will be needed to support PCPs in this expansive role if best use is to be made of investments in cancer genetic services.

Keywords. Cancer, genetics, primary care, qualitative research.

Introduction
Developments in genetics have led to increased interest in the role of primary care physicians (PCPs) in genetic medicine. Cancer genetic services are among the most readily used by PCPs as test results have proven benefits for high-risk patients. Considerable research explores the PCP role in cancer genetics, adopting the perspective of physicians and focusing on the primary care role in identifying and referring individuals at elevated risk, while reassuring those at population risk and avoiding inappropriate referral. Little research has attended to other elements of the PCP role in cancer genetics, including the care of individuals who have received genetic test results and for individuals who are also in the care of cancer specialists. In addition, few studies investigate the patient perspective. Those that do focus on the PCP role prior to genetic testing, exploring the feasibility of family history taking in primary care, the disparate communication strategies of primary care clinicians and patients in making sense of familial risk and patients’ perspectives on the role of PCPs in providing information, referral or reassurance. In short, existing studies provide little insight regarding the PCP role for patients in receipt of genetic test results and no insight on the patient perspective in ongoing cancer genetic care. We therefore explored the PCP role as part of a larger study of the patient experience of cancer genetic services in Ontario, Canada, involving interviews.
with patients shortly after receiving test results and 1 year later.

Methods

In Canada, family physicians are the main providers of primary medical care and play an increasing role in cancer prevention and screening and in follow-up care for patients with advanced cancer. In Ontario, cancer genetics is coordinated through specialized clinics to which patients may self-refer or be referred by primary or specialist providers. Because of test limitations, cancer genetic testing is offered first to high-risk individuals who have been affected by a clinically suspicious cancer to 'search' for a deleterious mutation; predictive genetic testing is offered to individuals with no prior cancer experience only when a mutation is identified in an affected relative. In 'mutation searching', therefore, the only clinically clear result (i.e. 'clinically positive') is one in which a deleterious mutation is identified. Other possible results include identification of a variant of unknown clinical significance (one person in our sample) or a negative result—considered 'clinically inconclusive'—in which the possibility remains that the individual may actually be affected by a hereditary cancer syndrome not associated with the sought-after mutation. Thus, only for 'predictive testing' applied to unaffected relatives of an affected individual with a known deleterious mutation does the laboratory result have a readily interpretable clinical meaning: a negative result is clinically negative while a positive result is clinically positive. With ethics approval from the relevant hospitals, participants were recruited from three cancer genetics clinics in Southern Ontario. Eligible patients had received genetic test results for hereditary breast/ovarian cancer (BRCA1, BRCA2) or hereditary nonpolyposis colorectal cancer (HNPCC), were at least 18 years of age and were fluent in English. Clinicians introduced the study at the end of each result disclosure session and asked recipients whether they would agree to be referred to the study team. During the 1-year recruitment period (2006), clinicians approached most result recipients regarding the study. Patients were not approached if they received results by phone or when the clinician judged this to be inadvisable due to the patient’s emotional state. Clinicians did not keep records of those who were not approached or refused but reported that most patients were approached and were willing to be referred.

During the initial interview, patients were asked about (i) their genetic test experience; (ii) the implications of the result for the individual, their health and health care; (iii) the implications of the result for family members and (iv) the actual or potential role of their family physician—with emphasis on the PCP role in referral for genetic testing. The follow-up interview addressed items 2 through 4, focusing in particular on the PCP role post-genetic testing and sought input on the operation of the cancer genetic programme as a whole. Interviews averaged 1 hour were tape-recorded, transcribed verbatim and entered into a qualitative database (NVivo, Version 7). For the current paper, two members of the research team (FAM and JPB) abstracted transcript sections relating to the involvement of, or attitudes towards, PCPs and aligned data across the two interviews. We used a 'low inference' qualitative descriptive analytic approach that seeks to present the facts of the case in the language of participants, without aiming to represent the material in other, more theoretical, ways. Drawing on the transcripts, together with literature on the PCP role in genetics or cancer (e.g. 7, 18), and the patient experience with cancer genetics (e.g. 16, 19), we categorized sections to identify thematically coherent attitudes and expectations, identifying two phases of the PCP role and consistently specified beliefs and expectations related to these roles. Where we discuss issues raised by only one participant, we have clearly identified these as individual cases.

Findings

Participants

In total, 25 individuals (mean age 57, range 21–80) participated in a first face-to-face semi-structured interview within 4 months of receiving genetic test results in 2006 (cited as P No., sex and age at time of testing); at this time, two participants had informed their PCPs about their genetic test results. Of the initial 25, 21 participated in a second follow-up interview after ∼12 months in 2007 (cited as P No. 2, sex and age at time of testing); at this time, 11 participants had informed their PCPs about their genetic test results (three had still not met with their PCP post-genetic testing). In addition, we received written permission from all study participants to obtain a copy of their genetic test results. The majority of participants (22/25) had a prior or current cancer experience and had been in the care of cancer specialists (only one of these was released from the care of cancer specialists at the time of our second interview). Most participants (21/25) were referred for BRCA1/2 testing, with only four referred for HNPCC testing (see Table 1). In a quantitative survey of cancer genetic patients in Ontario that was pursued concurrently with our study, 50% of participants had cancer and 18% were referred for HNPCC testing, suggesting that our sample over-represents the experience of patients with cancer but is reflective of the proportions of HNPCC patients receiving care in the province.

Referral

Most of our respondents (19/25) gained access to genetic testing through cancer specialists (e.g. oncologist,
surgeon), with a further three referred through genetic services because of testing in family members. PCPs were responsible for referral for three respondents. Despite the reduced involvement of PCPs in the referral process and the challenges facing PCP participation, our respondents suggested that PCPs should possess sufficient knowledge to enable referral and a supportive attitude; furthermore, for a few respondents, PCP involvement in the care of family members amplified the importance of this role (see Table 2 for supplementary quotes illustrating shared themes across transcripts).

Patients did not expect PCPs to know a great deal about genetic services. They emphasized that PCPs could not be fully informed about all areas of specialist medicine; nonetheless, they did expect PCPs to know enough to recognize familial risk:

I mean, they don’t have to be ... proficient at it but they have to have some general idea that, ‘Okay, someone’s tested positive in your family blood relation. I’ll send you off to somebody who knows.’ And if they’re not aware of it, they brush it off to the side. [P10-2, Female, 36 years old]

Several respondents were gratified by their doctor’s knowledge and attentiveness to genetic risks: ‘Yeah, but he was the one who had enough savvy to say,'
Need for positive PCP attitudes

- The general practitioner is exactly that—a general practitioner. They can’t know everything but they should know enough to send you to a specialist. And they should know enough about certain things that they can speak intelligently, but to know all the ins and outs about everything... I wouldn’t expect that because there must be so many tests that they would have to know about. [P8-2]
- They should see [the signs] and they should be able to put the little puzzle together. [P11]

Challenges facing PCP involvement

- Oh my doctor knew about gene testing because he said to me at the time, ‘Geez, that’s too much of a coincidence to be a coincidence when you’re the third one [in your family to have had breast cancer],’ so he, they do know about it but there’s no time to discuss. He’s busy enough. [P14-2]
- It’s not that they are not sincere it’s just that they put too much on their minds, so these things skip their mind. A genetic study in a normal, healthy looking female, this would be the last thing that would come in their mind. [P25]

Need for positive PCP attitudes

- ... my first GP once said to me when I was debating about doing the breast thing, about the testing, and she said well, you know, she made some flippant comment about well, you test positive for it so, you know, you’re going to maybe get it, but she said, you know, you walk in front of a truck tomorrow and you know you’re dead anyway. So to her it was almost insignificant. [P19]
- ... actually, you know what, I had, before I even knew I had breast cancer, maybe a year or so before I knew, I had asked him whether I could go for genetic testing and he said it wasn’t really… more or less it was still in its infancy and it wasn’t really accurate so he didn’t recommend that I go. Interviewer: It could have been based on your family history, too. If he wasn’t so certain about your grandmother, that kind of thing. Yeah. Well, I don’t think it was that. I just think he didn’t want me to go for it. [P17]

PCP knowledge of family

- Interviewer: [Your PCP] was aware about your whole family history? Yes, oh definitely, yes, because I’ve been going to her for quite some time [P22]
- I was hoping that—and I never verbalized this to my doctor here who’s a female—because she knows my situation that she should be aware and be monitoring my sister more closely and be counseling her on that. Now she didn’t, I was the one that brought forward to my sister that perhaps she should speak to [their PCP] about doing intervention now, preventatively, as far as whatever tests she might need or mammograms and how they would set that up. So I sort of brought that to her, just out of concern, to make sure that she’s proactive because I certainly would want to be proactive. And you’re not going to get a 30 year old to do that. So, but, I believe that that was all set up and now she’s in a loop where every two years she’ll have a mammogram and so on. [P11]

Ongoing care

Most of our respondents were in the care of cancer specialists at the time of both interviews. Thus, the patient experience of the PCP role upon receipt of genetic test results involved the PCP role in cancer care more generally. For many respondents, this implied a limited role for PCPs, though some perceived a more sustained role once the attention of cancer specialists declined. However, expectations about an ongoing role among cancer survivors, as well as among those who had pursued predictive genetic testing, were coloured by seemingly inappropriate beliefs about the nature of appropriate care (see Table 3 for supplementary quotes illustrating shared themes across transcripts). Respondents in the care of cancer specialists generally anticipated a limited PCP role in the care arising from genetic test results, suggesting that all cancer-related care was the domain of cancer specialists. Even for a patient whose family physician was ‘very good’, had identified a rare side effect of her cancer therapy and would ‘go and he would look it up’ if there was anything he didn’t know about cancer genetics, it was not important that the PCP have genetic knowledge: ‘For the oncologist to know it, yes, but for my family doctor, no.’ [P01-2, Female, 51 years old]
PCP involvement was also limited by time constraints and was, in one case, seen to be discouraged by the providers of cancer genetic testing. As one respondent noted of a family physician who ‘knew I was doing it [genetic testing] and she thought it was a good thing, definitely’.

I haven’t seen her since I had the results. And they sort of suggested, too, that you don’t have to tell your doctor your results or your . . . [pause] . . . you know, not everybody has to know what the results are. [P22, Female, 74 years old]

Despite this, this same patient identified a PCP role in ongoing care as active cancer care declined: ‘but really, when you have an oncologist, they’re the ones that make the appointment, and they . . . until, I guess, until you get to the stage where you’re not, you know, you’re only going once a year . . . but she will be, oh yeah, she definitely will be very in tune with it and having me checked out’. [P22]

Other patients who anticipated a need for ongoing PCP care as specialist attentions declined were pessimistic because of broader challenges in the coordination of primary with specialist cancer care. Two years post-cancer diagnosis and receiving less cancer care, one respondent feared her PCP ‘didn’t know she was really my caregiver now. So I had to verbalize that to her . . . I said, “So, you’re it.” She said, “Okay, well let me look a little closer.” So, you know, it’s a communication gap.’ [P11, Female, 41 years old]

A year later and facing residual genetic risks, this respondent also lacked faith that her PCP possessed sufficient knowledge to provide adequate surveillance of familial cancer risks:

. . . who’s going to close a loop if something did come up that was pertinent to me? You know? Say I did have a recurrence and . . . or they, they know the person with this type of genetic risk factor was more likely to have . . . whatever type of cancer . . . You know, it would be nice that she’s at least on that. [P11-2]

Patient expectations about an ongoing PCP role were informed by perceptions of personal genetic risk; in several cases, however, these perceptions seemed to be incorrect. For example, the results of mutation searching left some patients facing residual risk but not all expected their PCP to anticipate the need for continued watchful care. One patient, for example, believed that her provider had good genetic knowledge, as testified to by his disinterest in her unclear negative result. Similarly, three patients in our sample pursued predictive genetic testing and received clinically conclusive negative tests results, indicating that they faced no greater than average population risk for cancer; yet their expectations of
the PCP role were inconsistent with this. Indeed, one patient was angry at her result, and at the appropriately reduced care, and reported needing to ‘somehow convince my doctor that I will want more, that I will want mammograms before I’m 50’. [P19, Female, 42 years old] One year later, she reported less anxiety but continued concern at the perceived lack of her PCP’s interest and watchful care:

Because once you’ve left the genetic testing like I don’t see anyone there now. All I’m going with is the documents, the letters that I’ve been given from the genetic testing and I can show that to my FP but she’s not that interested. [P19-2]

Discussion

Seeking to understand the role of PCPs through the eyes of patients is an important approach to understanding the delivery of cancer genetic services but little research has pursued this approach. Such a vantage point is partial regarding actual PCP behaviours and attitudes but illuminates important elements of, and constraints on, the potential PCP role.

Patients in our study endorsed a PCP role in referral for cancer genetic testing. Not all were satisfied with the care that they had received but respondents expected PCPs to understand enough about cancer genetic testing to recognize elevated risk, and where relevant, to make effective use of their detailed knowledge of some families. While few respondents relied on primary care to gain access to genetic testing, they believed that others needed this support to ensure that high-risk family histories were not simply ‘brushed off’. Thus, in addition to expecting knowledge and despite time constraints, patients expected PCPs to have a supportive attitude—to believe, as they did, that genetic testing was worth pursuing.

Patients were less certain about the PCP role upon receipt of genetic test results. Most of our respondents had experienced cancer and minimized the significance of the ongoing PCP role in any type of cancer care, including cancer genetic care. However, some respondents did identify a distinctive role comprising watchful care of residual or known genetic risks and appropriate surveillance. This role was most relevant as the attentions of specialist providers declined for those who had cancer, but it was also relevant to those who pursued predictive genetic testing. Yet an ongoing role for PCPs in cancer genetic care faced several constraints, including challenges in the coordination of primary with specialist cancer care, time and knowledge limitations, and occasional confusion regarding the implications of genetic test results for personal genetic risk and thus what qualified as ‘appropriate’ care. Notably, some patients reported expectations of their provider’s behaviour (i.e. lack of concern or continued surveillance) that did not correspond with their objective risk, as evident in the copy of their genetic test results that we obtained.

These findings enhance what is known about the PCP role in referral for cancer genetic services. As respondents noted and as the literature documents, PCPs have an important role to play in the identification of familial cancer risk, referral or reassurance, but struggle to attain sufficient knowledge to be confident, experience time and financial disincentives and sometimes lack enthusiasm, expressing concern about the ‘therapeutic gap’ between genetic testing and treatment. In addition, this study highlights an important role for PCPs in ongoing care once genetic test results are received. For cancer patients, who comprised most of our sample, this role was negotiated in relation to the well-known constraints facing the coordination of primary with specialist cancer care more generally, including challenges to effective communication and coordination between primary and specialist care, knowledge limitations within primary care and attitudinal challenges in secondary and tertiary care regarding the primary care role. In addition, the primary care role in ongoing care is further complicated where patient expectations are informed by partial comprehension of the meaning and significance of genetic test results.

In sum, the potential PCP role in cancer genetic services anticipated by patients is expansive but sustained efforts will be needed to support PCPs in all aspects of this complex and important role if best use is to be made of investments in cancer genetic services.

Limitations

The strengths of the study include a longitudinal design, with interviews post-test result and 1 year later, and focused attention to the actual and potential PCP role during both interviews. Weaknesses include a sample that, despite best efforts, is not representative of the Ontario population of cancer genetic patients. While qualitative research is not intended to be statistically generalizable to populations, and the disproportionate representation of patients with cancer enables rich insight into their expectations and experiences, it limits what can be learnt about the experiences and expectations of patients who pursued genetic testing for predictive purposes.

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Declaration

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References