Perceptions of genetic risk assessment and education among first-degree relatives of colorectal cancer patients and implications for physicians

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Background. Genetic risk assessment and education is a clinical service that provides an opportunity for individuals with a strong family history of cancer to understand their risk better, identify a screening regimen and discuss benefits and limitations of genetic testing.

Objectives. The aim of this study was to assess knowledge of and attitudes to genetic risk assessment and education among first-degree relatives of patients with colorectal cancer.

Methods. We conducted focus groups among first-degree relatives of patients with colorectal cancer to assess perceptions of genetic risk assessment and education. In the groups, we elicited reactions using two definitions of genetic risk assessment and education—one brief and one more detailed—that might be used by a health practitioner during the referral process.

Results. Findings revealed a number of misconceptions and concerns including: (i) what is required to prepare for a session and a lack of desire to collect a family history; (ii) what is involved in a session (including assuming that genetic testing is always included in a session); (iii) distrust over accuracy and possible subjectivity of information provided; and (iv) fear of the effect that participation in a session might have on insurance status.

Conclusions. The findings suggest that health practitioners should educate individuals about genetic risk assessment and education during the initial referral process. Further studies should explore how best to do this.

Keywords. Colorectal cancer, education, genetics, risk communication.

Introduction

Colorectal cancer is the third leading cause of cancer deaths in the USA. An estimated 135 400 people will be diagnosed and 56 700 people will die of colorectal cancer in 2001.\(^1\) Approximately 75% of people who develop colorectal cancer have no known risk factors for the disease. First-degree relatives (FDRs) of colorectal cancer patients have a 2- to 8-fold risk of developing the disease compared with members of the general population.\(^2\)

Cancer risk information is communicated to individuals by a number of health professionals, including physicians, geneticists, genetic counsellors, psychologists, nurses, health educators and social workers.\(^3\) However, when someone has specific questions or concerns about risk or may be dealing with a hereditary disorder, referral to a specialist trained in identifying heritable diseases may be appropriate. Genetic risk assessment and education referral is most appropriate for individuals who have a strong family history of the disease.

According to Muller, a typical assessment and education session includes: (i) accurate diagnosis of the predisposition (susceptibility); (ii) risk determination; (iii) communication of genetic and medical facts to the patient; (iv) evaluation of possible options and alternatives; and (v) follow-up and continuing support of the patient and family members.\(^4\) It also includes education regarding
risk factors, prevention and symptoms of colorectal cancer, an appropriate screening regimen and clarification of whether genetic testing is appropriate for consideration.

Genetic risk assessment and education for colorectal cancer is, in most health care settings, a fairly new clinical service. Thus, individuals may not be familiar with it, or they may have incorrect pre-conceived ideas about what it entails, including, reasons for referral, issues addressed in the sessions, insurance coverage for the service, etc. These areas of potential confusion are relevant for referring physicians who want to improve communication with and knowledge among their patients. It is important for referring physicians to be aware of issues that might (i) prevent participation or (ii) raise false hopes regarding what might be gained from a session.

We conducted focus groups among FDRs of patients with colorectal cancer to assess perceptions of genetic risk assessment and education. In the groups, we elicited reactions using two definitions of genetic risk assessment and education—one brief and one more detailed—that might be used by a health practitioner during the referral process. Findings revealed several areas of misunderstanding and concern.

Methods

Recruitment

Four focus groups were conducted among FDRs of colorectal cancer patients treated at two large, university-affiliated hospitals in St Louis and Indianapolis. Using a two-step process, we first contacted patients themselves and then contacted relatives whom patients referred. All letters and procedures were approved by the universities’ respective Human Subject’s committees. Patients were first sent a letter from their oncologist explaining the study. Within a few days of letter receipt, patients were telephoned and asked to provide the names of their FDRs (parents, siblings and children) who lived within the metropolitan area and were over the age of 25.

A total of two possible relatives per patient—only one of each gender—were then invited to participate in a focus group exploring opinions and concerns regarding colorectal cancer screening behaviours, and genetic risk assessment and education. Relatives received an introductory letter prior to telephone contact. Incentives for focus group participation included a US$ 25 honorarium and the knowledge that the resulting information would be used to help other families of colorectal cancer patients. Those who agreed to participate received a reminder letter with directions to the meeting site, details regarding meeting time and location, and a reminder call the evening before the focus group. Eighty-eight patients were contacted to yield a total of 75 relatives, of whom 22 agreed to participate and actually attended the focus groups. Other than a few participants calling to report circumstances precluding their attendance, we have no information about the six additional FDRs who agreed to participate but did not attend.

Procedures

We conducted one all-male and one all-female group session at each university site for a total of four groups. Discussions were audiotaped. Informed consent of participants was obtained before the focus group began. We did not collect data on the patients’ specific type of tumour or the number of relatives affected with colorectal cancer, and thus do not know if participants were from high-risk families.

Experienced focus group moderators (one at each site) followed a detailed moderator’s guide listing predetermined, standardized, open-ended questions exploring participants’ opinions and concerns. Although questions about screening tests for colorectal cancer were also included, this paper reviews findings concerning genetic risk assessment and education, and genetic testing.

No risk information about colorectal cancer was provided to participants before beginning the groups. Because it was unlikely that participants would be familiar with genetic risk assessment and education, we believed it was important to elicit reactions to alternative explanations of the clinical service (e.g. how the service described might affect interest in participation). Thus, we developed two definitions for use in the moderator’s guide. Each was developed by the research staff and reviewed by genetic counsellors and medical geneticists at the two sites. Reactions to each are reported in the Results.

Analysis

We first examined the focus group transcripts and identified the major themes of responses. If there was disagreement in interpreting tone or meaning from the transcript, we listened to the original tapes from the groups. For this paper, perceptions were summarized using verbatim quotes to illustrate the key themes that emerged during the discussions.

Results

Demographics

A total of nine men and 13 women participated in the groups. Table 1 summarizes their demographic characteristics. The majority of participants were over 40 years old, married, had at least a high school degree or equivalent, and 50% had an annual family income of US$ 60 000 or more.

The brief definition

The moderator’s guide was designed to evaluate interpretations of genetic risk assessment and education based
two very different definitions that might be used by a physician during the referral process. The first was:

Genetic risk assessment and education is a one-on-one meeting with a medical expert to get more information about your personal chances of being diagnosed with colorectal cancer.

After this brief definition was read, participants were asked several questions about their knowledge level and interest in genetic risk assessment and education. Initial reactions, based on this brief description, are described below.

Knowledge and interest. Participants were asked what they knew about genetic risk assessment and education. Although no one acknowledged having heard of it, many said they thought it would probably have something to do with heredity and cancer.

Based on responses to this first definition, it was unclear whether some of the participants were distinguishing between the risk assessment and genetic testing. Discussion about exact risk and ‘getting guarantees’ of whether they would develop cancer seemed to indicate some confusion about the two.

Interest level for some seemed contingent on whether information about lifestyle and prevention would be included in the process.

The expanded definition

After these issues were explored, the following, more detailed description was read:

Genetic risk assessment and education is a one-on-one meeting with a genetic cancer risk specialist in which you would discuss your personal cancer risks.

A family history of cancer can be one of the most important risk factors for cancer. In order to get the best estimate of your chances of getting a specific type of cancer, you would need to complete a family history form and try to obtain medical records on family members with cancer before meeting with the cancer risk specialist. The cancer risk specialist helps to document your family history and collect medical records.

During a genetic risk assessment and education meeting, a cancer risk specialist does three things. First, the specialist gives you information about your personal chances of getting a specific type of cancer. Then the specialist talks about your characteristics or history that might be related to your chances of getting that cancer. Finally, the specialist discusses the best ways for you to detect or prevent that particular cancer. A meeting with a cancer risk specialist takes about an hour and a half.

The moderator explored participants’ expectations about the process, as described above, and what they expected to learn as a result (Table 2).

**Preparation for the meeting.** Several individuals acknowledged that it would be useful to gather medical records on family members or at least have a family history filled out ahead of time. However, there was some confusion about what family history documentation would entail, and many voiced concern about the effort involved in collecting records.

For those who appeared uninterested in genetic risk assessment and education from the beginning of the focus group, the idea of collecting records of family members was not appealing. However, others indicated that they would not allow the time and effort necessary to collect medical records to prevent them from going forward with the process.

**Expectations about what would be learned.** Because interest in and willingness to undergo genetic risk assessment and education would presumably be influenced by expected benefits or outcomes, moderators asked participants what they would expect to ‘get’ or learn from a session. Information about screening tests (i.e. when and how often to have them) seemed, to many participants, to be an important component. However, preventive measures and specific lifestyle modifications to lower risk were the most desired information. Overall, there seemed to be distrust about whether risk information would be accurate.

This type of discussion indicated some confusion between genetic risk assessment and education, and genetic testing. In fact, it became clear in one of the women’s groups that, despite specific descriptions of genetic risk assessment and education, participants were discussing genetic testing throughout the entire series of questions.

When the moderator did ask about genetic testing, almost all participants expressed an opinion. Strong
arguments were presented both for pursuing and for not pursuing genetic testing. One woman stated that she did not want to know genetic testing results because she did not want to have to worry about it. Others felt it generated paranoia. Some only wanted to know results if the knowledge would lead to risk-lowering measures. Still others felt that they needed the definitive results that genetic testing could provide.

Moderators were prepared to ask about the topic of insurance in all groups but, in three of the four, the topic was mentioned by participants before the moderators had a chance to talk about it. It is noteworthy that the moderator asked specifically about whether FDRs thought participating in a genetic risk assessment and education session would affect insurance. However, again it was difficult to tell whether participants were responding to how they thought their insurance might be affected by participation in a genetic risk assessment and education session or genetic testing.

As with previous topics, response to this issue was widely varied. Some worried that insurance status would be negatively impacted. Members in one group exchanged stories about individuals they knew who had lost coverage after developing cancer. One of these individuals expressed concern that if her insurance company knew she had ‘the gene’, she might be dropped as well.

Others felt that insurance companies would view genetic risk assessment and education as a preventive measure. One man compared it with getting his teeth cleaned at the dentist—pay for prevention now and save money down the road. One group thought it would be best to withhold all information from insurance companies, so discrimination would not be an issue.

Table 2  Examples of concerns and misperceptions of genetic risk assessment and education

<table>
<thead>
<tr>
<th>Topic area with concerns or misperceptions</th>
<th>Illustrative quote</th>
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<tbody>
<tr>
<td>Gathering of family medical records</td>
<td>(i) “... the collection of data, that’s a big deal! I mean, are you going to be able to get medical information in regard to your relatives?”</td>
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<td></td>
<td>(ii) “I don’t know how to get my dad’s medical records from Alexander, Egypt, and London. I don’t want to do that, and I couldn’t.”</td>
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<td>Assuming genetic testing is always involved in a session</td>
<td>(i) “I’m going down to see how genetically I’m attached with colon cancer.”</td>
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<td></td>
<td>(ii) “Sounds like it’s taking a test of your DNA, and then also lifestyle factors.”</td>
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<tr>
<td>Distrust over subjectivity of results</td>
<td>(i) “They’re never going to be able to say I’m going to get it, or my chances are 99.3% of getting it. It’s always going to be their opinion, whereas you want black and white.”</td>
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<td></td>
<td>(ii) “I would like to know what their accuracy rating is.”</td>
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<td>Uncertainty regarding the effect participation would have on insurance status</td>
<td>(i) “Well I’m wondering if they might take a second look at me as a pre-existing condition. So that might be a black mark against me as far as knowing that I have a high risk... they have this on record now.”</td>
</tr>
<tr>
<td></td>
<td>(ii) “I don’t know where the law stands on this, but if they find something genetic but you don’t have an active disease process or cancer, is that discrimination?”</td>
</tr>
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</table>

Discussion

Genetic risk assessment and education is an opportunity for those with a strong family history to work with a professional better to understand personal risk, identify appropriate screening regimens and determine whether consideration of genetic susceptibility testing is appropriate. Yet, in these focus groups conducted among colorectal cancer FDRs, no participant had ever heard of it. Further, although some correctly assessed, from definitions read by the moderator, what would be involved in such assessment and education, others could not seem to distinguish between the counseling, which should educate about risk and facilitate genetic testing decisions, and genetic susceptibility testing itself.

This is an important finding for medical centres that are beginning to set up hereditary cancer clinics. Our findings indicate a need for education about genetic risk assessment and education: what it is and what it can and cannot provide. Such education may be warranted not only for individuals at elevated risk for cancer but also for the health professionals who provide their primary care. As more referrals to hereditary cancer clinics become appropriate, it will be important for health practitioners who make referrals to understand where their patients may become confused about what is involved in genetic risk assessment and education.

Findings from our focus groups with FDRs of colorectal cancer patients identified a number of areas where participants displayed a lack of knowledge. These misperceptions may act as barriers to participation and should be addressed with FDRs during the referral process so that those who might be interested in and benefit from participation would be more inclined to
follow up on a referral. These areas include: (i) what is required to prepare for a session (including the importance of family history documentation and medical record collection); (ii) what is involved in a session (and that genetic testing may not be necessary); (iii) accuracy of information provided; and (iv) the effect that participation in a session would have on insurance status.

Participants were told that they would need to complete a family history form and try to obtain medical records on family members with cancer before meeting with the cancer risk specialist. There is no national cancer registry in the USA and, because of confidentiality issues, medical records cannot be accessed unless the patient has given permission to do so. Subjects are therefore asked to contact their relative with cancer directly (or the closest living relative to the patient if the patient is deceased) so that medical records can be released. If for any reason medical records are unavailable, risk assessment and education can still take place, but it is always preferable to have detailed documentation of family history. Participants in these focus groups appeared somewhat resistant to the idea of collecting medical records and also seemed unclear about the level of detail needed for a family history. Many expressed concern that gathering a family history would be a long, time-consuming process, yielding little more information other than that which they already knew, i.e. that they were at higher risk because a first-degree relative had developed colorectal cancer. Participants may have postulated that, to delineate their individual risk accurately, their individual genes would need to be examined. If so, they may have underestimated the value of family records.

Interest level in genetic testing has been cited as ~80% in the general public,6 in FDRs of colorectal cancer patients7 and among individuals who were actually eligible for testing as determined by a familial colorectal cancer clinic.8 Assuming that this represents only a subsample of individuals who are aware of the reality of genetic testing, one could assume that this high level of awareness has created bias so that anything including the term ‘genetic’ makes people think genetic testing is involved. Referring to the process as ‘cancer risk assessment and education’ instead of ‘genetic risk assessment and education’ may help to eliminate assumptions that genetic testing is always included in a session. In fact, both medical centres involved in this study offer cancer risk assessment and education as a routine clinical practice through their Genetics departments, and both sites refer to the session as such. (Studies might also assess reactions to other terms such as ‘family’ or ‘familial’ risk rather than ‘genetic risk’.)

Distrust over the subjectivity of the ‘results’ was expressed but, again, it was difficult to tell if participants were referring to genetic risk assessment and education and assignment to a category of risk (e.g. low, moderate or high risk) or to genetic testing and a more definitive risk percentile (e.g. ‘presence of the gene means an X% chance of developing colorectal cancer’ and ‘absence of the gene means an X% chance of developing colorectal cancer’). Specifically outlining how risk classification is determined may help to clear up confusion in this area. In particular, general confusion between genetic risk assessment and education and actual genetic testing may be especially important because this misunderstanding might prevent persons from participating in the very type of assessment and education that could help them make genetic testing decisions.

In a previous study, Lynch identified three barriers to effective genetic counselling of first- and second-degree relatives of HNPCC syndrome. These included: (i) lack of education among physicians regarding the need to treat/refer high-risk relatives in addition to the patient; (ii) patients’ fear and denial; and (iii) socioeconomic and educational barriers regarding insurance coverage.9 Our population also exhibited concerns regarding insurance coverage. There was not a general consensus on how participants felt that participation might affect insurance coverage. However, ‘fear of the unknown’ seemed to be a concern for several individuals who stated that they would not be willing to risk losing coverage as a result of participating. In addition, the fact that most groups raised the issue on their own before the moderator mentioned it indicates its salience to participants. These findings suggest that information about availability of genetic risk assessment and education should include—up front—cost and insurance coverage issues.

Consideration of the implications of these findings must begin with a reminder of the study’s limitations. Our study was an exploration of topics among a small number of FDRs recruited for participation in the focus groups at each site. They can in no way be seen as representative of all FDRs. However, the fact that all participants’ relatives were treated for their colorectal cancer at a university-affiliated hospital, that the participants lived in or close to one of the large cities (St Louis and Indianapolis) in which the groups were conducted, and the fact that they were at least interested enough to attend the groups probably indicates that results among a more general population of FDRs would indicate less knowledge and interest. In other words, if there were misperceptions among this group of FDRs, we can probably assume that there are misperceptions among their counterparts in the larger population.

Finally, future studies should follow individuals through the risk assessment and education process assessing areas of concern that surfaced in this study.

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References