Concerns of women presenting to a comprehensive cancer centre for genetic cancer risk assessment

D J MacDonald, J Choi, B Ferrell, S Sand, S McCaffrey, K R Blazer, M Grant, J N Weitzel

The study used a one time exploratory methodology to collect quantitative and qualitative data. Age, personal and family cancer history with ages at diagnosis, personal health history including risk factors related to breast and ovarian cancer, and ethnicity was collected on all participants. The study questions were revised before study implementation based on pilot testing for readability, clarity, and time for completion (average of 17 minutes) by three non-health care professionals, a nurse educator, and a nurse cancer risk counsellor practising in the mid-west. The study questions were designed to ascertain perceptions about: (1) magnitude of their own personal cancer risk; (2) cancer risk in general; (3) concerns about developing cancer; (4) acceptable options for cancer prevention and risk management; (5) issues about genetic testing for cancer risk; (6) communicating risk to other family members, and (7) how women learn about cancer risk. Two versions of the questionnaire were used, one for affected women and the other for unaffected women. Space was provided throughout the questionnaire for open ended comments. Subjects were asked to rate on a scale of 0 = “no risk” to 100% = “inevitable” what they believed was their own lifetime and 10 year risk for developing breast cancer. Beliefs about general population risk for breast, ovarian, uterine, and colon cancer, the age at which a woman was at greatest risk for breast cancer, and the percentage of women with breast cancer who had a family history of the disease were also ascertained. In evaluating cancer concerns, a checklist of “never/rarely,” “sometimes,” “often”, or “all the time” was used to assess the frequency of concerning thoughts about developing cancer and how often these thoughts interfered with one’s life/daily activities. For risk management, women were asked to check a response of “yes,” “no,” “unsure”, or “not applicable” on nine questions regarding what they might be willing to do if determined by a genetic test to be at greater than 50% risk for breast cancer. Women were also asked to rank, in order of first, second, third, “would not choose”, or “not applicable” their preferences for managing breast cancer risk. Risk management choices included close surveillance, participating in a research study of a medication to reduce risk, and prophylactic surgery. Similar

Abbreviations: GCRA, genetic cancer risk assessment; COH, City of Hope; CSPP, Cancer Screening & Prevention Program; FDR, first degree relative; SDR, second degree relative; OC, oral contraceptives
Table 1 Subject characteristics

<table>
<thead>
<tr>
<th></th>
<th>Affected</th>
<th>Unaffected</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total No</td>
<td>27</td>
<td>23</td>
</tr>
<tr>
<td>Breast ca</td>
<td>23</td>
<td></td>
</tr>
<tr>
<td>Bilateral breast ca</td>
<td>2</td>
<td></td>
</tr>
<tr>
<td>DCIS</td>
<td>2</td>
<td></td>
</tr>
<tr>
<td>Ovarian ca</td>
<td>1</td>
<td></td>
</tr>
<tr>
<td>Breast and ovarian ca</td>
<td>1</td>
<td></td>
</tr>
<tr>
<td>Average age at entry</td>
<td>45, 14 (24-77)</td>
<td>41, 10 (26-65)</td>
</tr>
<tr>
<td>Age at cancer diagnosis</td>
<td>40, 10 (22-58)</td>
<td></td>
</tr>
<tr>
<td>Oral contraceptive use</td>
<td>7/27 (26%)</td>
<td>4/18 (17%)</td>
</tr>
<tr>
<td>Hormone replacement therapy use</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>&gt;1 FDR with breast or ovarian ca</td>
<td>10</td>
<td>23</td>
</tr>
<tr>
<td>Ethnicity</td>
<td></td>
<td></td>
</tr>
<tr>
<td>White</td>
<td>40</td>
<td></td>
</tr>
<tr>
<td>Mixed heritage</td>
<td>6</td>
<td></td>
</tr>
<tr>
<td>Hispanic</td>
<td>2</td>
<td></td>
</tr>
<tr>
<td>African American</td>
<td>1</td>
<td></td>
</tr>
<tr>
<td>Asian</td>
<td>1</td>
<td></td>
</tr>
</tbody>
</table>

None of the unaffected women had ever had a breast biopsy or other breast surgery. Thirty-seven percent of the affected cohort (10/27) had a first degree relative (FDR) with breast or ovarian cancer: two participants (one with ovarian cancer) had a FDR with ovarian cancer, one had two FDRs with breast cancer, and seven had one FDR with the disease. An extended family history (SDR or greater) of breast and ovarian cancers in the paternal lineage was present for one subject in the latter group. Two affected women had only a third degree relative with breast cancer. Of the unaffected women, one had a FDR with both breast and ovarian cancer, two had a FDR with ovarian cancer only, and 17 had at least one FDR with breast cancer (including two cases of bilateral disease). One woman had four FDRs with breast cancer and a SDR with ovarian cancer, and two women had two FDRs with breast cancer (table 1). Two unaffected women without affected FDRs had a more distant family history of breast and ovarian cancer. One unaffected woman who had a first degree relative with ovarian cancer developed ovarian cancer after study completion. Most subjects also had a family history of other (not breast or ovary) cancers.

RESULTS

Risk perception

The mean perception of personal breast cancer risk for unaffected women was 59% (range 5-100%) compared to the mean lifetime breast cancer risk of 17% (range 4-35%) and 22% (range 10-34%) predicted by the Gail model and Claus tables, respectively. Intra-personal comparisons were not done. Affected women were asked to recall what they thought their chance of having breast cancer was before disease onset. Responders (n=19) noted a range of 0-100% with a mean of 31% (SD 34), slightly more than half the risk reported by the unaffected group, despite a near similar risk range of 5-100%. The lowest risk was reported by the youngest women and, unexpectedly, by the women who had undergone bilateral mastectomy. Many of the women who perceived themselves as at risk (both cohorts) thought that their chance of having breast cancer was confined to the next 10 years. The 10 year breast cancer risk estimates ranged from 0-75% (mean 33%, SD 24) for affected responders (n=17) and 0-100% (mean=56%, SD 32) for unaffected responders (n=21). Interestingly, subjects were less likely to perceive themselves to be at risk for breast, ovarian, uterine, or colon cancer if they had already had cancer. Both the affected (n=22) and unaffected (n=18) groups overestimated the general population risk for
breast cancer (mean 27% and 21%, respectively, NS) and the general population risk for ovarian cancer (mean 19% and 14%, respectively, NS). Similarly, both groups thought that the general population had nearly the same risk for uterine or colon cancer and greatly overestimated risks for these cancers.

**Cancer concerns**

When asked “How often do thoughts about cancer concern you” about half in each group reported “sometimes”. Only 15% of the affected women reported “often” or “all the time” in comparison to 19% of unaffected responders (n=21). While all of the unaffected responders (n=21) had at least occasional thoughts concerning cancer, 7% (2/27) of all affected women rarely or never had thoughts about cancer recurring. Overall, thoughts about cancer rarely or never interfered with subjects’ lives or daily activities for 67% of all affected women (18/27) and 64% of unaffected responders (n=22); however, 26% of affected women and 36% of unaffected responders said that these thoughts disrupted their lives sometimes or often. One affected responder worried about cancer recurrence “all the time”.

**Breast cancer risk management**

Each subject was asked about measures they would be willing to take to manage breast or ovarian cancer risk if genetic testing determined that they were at greater than 50% risk for cancer. Nearly all participants would be willing to perform monthly breast self-examinations, have more frequent clinical breast examinations, and have annual mammograms beginning in their 30s. About 75% of both cohorts would be willing to pay out of pocket for an office visit or mammograms not covered by insurance. Eighty-six percent of the affected women and 77% of the unaffected women would be in favour of taking a hormonal medication that had no serious side effects, to reduce breast cancer risk, but were much more likely to do so if the medication acted as a contraceptive or induced menopause (if applicable). Not surprisingly, more affected (48%) than unaffected women (39%) would undergo bilateral prophylactic mastectomies to reduce risk; however, 30% of all subjects would not choose this strategy at all (table 2).

**Ovarian cancer risk management**

Women were asked to rank choices for managing ovarian cancer risk. Similar to breast cancer risk management options, close surveillance was the first choice for reducing risk of ovarian cancer for both groups despite being told that the reliability of surveillance was not proven. Subjects were told that oral contraceptives (OC) could lower the odds of developing ovarian cancer by nearly a half and that several years’ use could slightly increase the risk of breast cancer. Twenty-six percent (7/27) of affected responders had previously used oral contraceptives as did 17% (4/18) of unaffected responders. Nevertheless, few responders in either group were willing to use OC, or another medication in a research study, to decrease their chances for the disease. Subjects were told that removal of the ovaries was thought to reduce ovarian cancer risk by about 90%. Forty-eight percent (11/23) of affected responders and 25% (5/20) of unaffected responders would choose this surgery as a first choice to reduce ovarian cancer risk (table 3).

**Genetic testing issues**

Women in both groups thought that genetic testing for hereditary cancer risk was important in making health care decisions, planning for the future, and knowing if their children could be at risk, and not important for deciding about child bearing or marriage. We found that 50% of affected responders (n=26) were very concerned about possible insurance problems as a result of genetic testing, whereas all unaffected responders (n=19) were much less concerned about this, possibly reflecting insurance burdens already experienced by the affected women. Few study participants thought that the cost of testing was an important issue in the decision to undergo genetic testing, though no information about actual test cost was provided. Eighty-two percent (14/17) of affected responders thought they carried an abnormal BRCA gene, as did 67% (12/18) of unaffected responders. We estimated a 15% mean probability of mutation for the affected responders and a 13% mean probability for the unaffected responders. For reasons unknown and unexpected, 10 affected and five unaffected women did not answer this question.

**Communicating risk**

Most women thought that if they were found to have a hereditary predisposition to cancer there was a duty to inform relatives about this and that they, as opposed to health care providers, should be the ones to inform relatives of this risk.

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**Table 2** Breast cancer risk reduction options*

<table>
<thead>
<tr>
<th>Options</th>
<th>A</th>
<th>U</th>
</tr>
</thead>
<tbody>
<tr>
<td>Breast self-exam monthly</td>
<td>100% (25/25)</td>
<td>96% (22/23)</td>
</tr>
<tr>
<td>Clinical breast exam more frequently</td>
<td>96% (22/23)</td>
<td>100% (25/25)</td>
</tr>
<tr>
<td>Mammogram annually, beginning in their 30s</td>
<td>96% (24/25)</td>
<td>95% (21/22)</td>
</tr>
<tr>
<td>Self-pay for office visits or mammogram</td>
<td>73% (19/26)</td>
<td>74% (17/23)</td>
</tr>
<tr>
<td>Use of hormonal medication</td>
<td>86% (18/21)</td>
<td>78% (14/18)</td>
</tr>
<tr>
<td>Prophylactic mastectomy</td>
<td>48% (13/27)</td>
<td>39% (9/23)</td>
</tr>
</tbody>
</table>

*A: affected; U: unaffected.*

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**Table 3** Ranking ovarian cancer risk reduction options*

<table>
<thead>
<tr>
<th>Options</th>
<th>1st choice</th>
<th>Would not choose as option</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>A</td>
<td>U</td>
</tr>
<tr>
<td></td>
<td></td>
<td>A</td>
</tr>
<tr>
<td>Close surveillance</td>
<td>50% (12/24)</td>
<td>52% (11/21) 0% (0/21)</td>
</tr>
<tr>
<td>Oral contraceptive</td>
<td>6% (1/18)</td>
<td>10% (2/20) 28% (5/18) 15% (3/20)</td>
</tr>
<tr>
<td>Chemoprevention</td>
<td>20% (4/20)</td>
<td>15% (3/20) 15% (3/20) 20% (4/20)</td>
</tr>
<tr>
<td>Prophylactic oophorectomy</td>
<td>48% (11/23)</td>
<td>25% (5/20) 1% (1/23) 0%</td>
</tr>
</tbody>
</table>

*A: affected; U: unaffected.*
Affected women were more likely to talk about cancer risk with their spouses/partners than unaffected women. Both cohorts reported that upsetting relatives or recalling painful memories did not prevent them from discussing cancer risk with their family members.

**Sources of information**

While women used the Internet to obtain information regarding cancer, they viewed information from physicians and magazines, newspapers, or books as more helpful. The information they obtained had little or no effect on the anxiety level of affected responders (n=25) but the unaffected responders (n=20) were nearly equally divided on whether this information raised or lowered their anxiety about cancer or had no effect at all.

**DISCUSSION**

This pilot study sought to determine the motivations and concerns of women presenting to a comprehensive centre for genetic cancer risk assessment (GCRA), counselling, and testing. The mean age of the unaffected participants (41) is close to the mean age of cancer diagnoses (40) in their relatives and slightly younger than the mean age of the affected cohort (45). For unaffected women, being close to the age of disease onset in a family member may be a milestone accentuating cancer concerns and prompting presentation for GCRA services.

As in previous studies, we found that both unaffected women with a family history of breast or ovarian cancer and women affected with these cancers greatly overestimate their own lifetime, 10 year, and general population risk for breast cancer. Despite similar family histories, perception of personal breast cancer risk was nearly twice as high (59%) for the unaffected women as compared to perceived risk before diagnosis for the affected women (31%). For the latter group, it could be that recollection of perceived risk before diagnosis is inaccurate. Alternatively, similarly to findings of Drossaert et al, the women may have been unaware of the impact of their family history on their own cancer risk. Regarding breast cancer risk in the next 10 years, in our study unaffected women reported an average risk of 56% v 33% for affected women, perhaps reflecting a lesser sense of vulnerability for women who had already been treated for the disease. A factor women often cite as contributing to the likelihood of having breast cancer is the use of oral contraceptives or hormone replacement therapy. In this study only a small proportion of both cohorts had a previous use of either medication, so this would not be expected to account for most participants’ risk overestimation. Of note, although both groups overestimated risk for several other common cancers, the affected women thought that their risk for these cancers was lower.

In our study, cancer concerns occurred more often among unaffected women than affected women. Correspondingly, more unaffected than affected women reported having disruptive thoughts about cancer at least occasionally. Cancer concerns, therefore, appear to be more disconcerting for women who have never had the disease. In contrast, Watson et al found that 28% of unaffected women with a positive family history in a London cohort had cancer worries “frequently” or “constantly”.

About three-quarters of all the women reported that they were willing to pay out of pocket if necessary for a clinician visit or mammogram and slightly more were willing to take a risk reduction medication with no serious side effects as long as it did not act as a contraceptive or induce menopause. It had not been anticipated that the women would not want to lose fertility given that the average age of both cohorts was over 40, an age at which the majority might be expected to have completed childbearing. Although more affected than unaffected women would have prophylactic breast removal, a third of all women would not (women were told that this surgery was not proven to prevent breast cancer). For both groups, closer follow up was the first choice for risk management. Many affected women would choose to undergo oophorectomy to reduce ovarian cancer risk whereas the unaffected women would not. Surprisingly, most women were unwilling to use oral contraceptives or another medication to lower ovarian cancer risk (tables 3 and 4).

Not surprisingly, more affected women thought that they carried a genetic predisposition to breast-ovarian cancer than unaffected women. Both groups thought that they should inform their family members about a hereditary cancer risk and many of the women had discussed cancer risk with at least one family member.

Consistent with previous studies, the most reported reasons for desiring genetic testing were to make health care decisions to reduce risk, to plan for the future, and to know if their children could be at risk. However, we found that reproductive decision making was not considered important regardless of the testing outcome. Potential health care or employment discrimination was a greater concern for affected than unaffected women. Perhaps the women who had experienced cancer had more intense interaction with their health care insurers, fear of job loss because of treatment related absence, and frank contemplation of mortality and reassessment of insurance coverage. Although we found that the cost of testing was not an important factor in deciding to have or not have genetic testing, it is possible that unawareness of the cost and high SES of subjects biased their response.

This study has both similarities and differences with findings from previous studies. It should be noted that most previous studies offered confidential free counselling and genetic testing as part of a grant subsidised study. Our 50 subjects were primarily white women referred by their physicians to an urban comprehensive cancer centre for GCRA in a regular clinical setting of fee for service health care. They may not be representative of lower socioeconomic groups or other ethnic populations. The small sample size also limits generalisation of study data. To help address these limitations we have initiated a study with a much larger sample size, more geographical diversity, and measurement at multiple time-points for women seeking GCRA at COH and two of our satellite clinics.

Findings from our pilot study indicate that providing information related to individual cancer risk, risk management
strategies, genetic testing, and risk to others in the family is important in meeting the needs of women presenting for GCRA. Further research is continuing to confirm and expand upon these observations. Health care clinicians should be aware of the beliefs, values, and concerns of women undergoing GCRA and be prepared to address these issues.

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