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The psychological impact of a cancer family history questionnaire completed in general practice

EDITOR—On the basis of family history, it is possible to identify subjects at significantly increased genetic risk of breast or colorectal cancer.[1 2] Evaluation of the benefits of screening these patients to facilitate early diagnosis and treatment forms the subject of continuing studies. For colorectal cancer, the benefits of colonoscopic surveillance have been reported,[3] but for breast cancer more data are needed to confirm the value of mammographic screening.[4] At present, patients with a significant family history who seek advice from their general practitioner are likely to be referred to a cancer genetics clinic and offered screening. If further research confirms the benefits of screening for patients at increased genetic risk, effective strategies for their ascertainment in primary care will be needed. One possible method is a postal family history questionnaire sent to the patient by their general practitioner. We report elsewhere on the effectiveness of this approach.[5] An important issue is whether this method of ascertainment raises anxieties, particularly among the majority of patients who do not have a significant family history. The collection of cancer family history information constitutes a form of screening. There is a large body of evidence that health related screening can have unintended adverse effects, the most studied of which is raised anxiety, particularly among those found to be at an increased risk.[6] As knowledge of the genetic component of common diseases increases,[7] more patients may be asked to provide information about their family history. It is therefore timely to consider whether such a task may inadvertently raise general levels of anxiety or worries about the disease in question. To our knowledge, there have been no previous studies of the psychological consequences of screening using a postal questionnaire to obtain information about relatives affected by cancer. The purpose of the present study was to determine the psychological impact of completing a cancer family history questionnaire and receiving an assessment of personal genetic risk of breast or colorectal cancer.

General anxiety was assessed using the six item, short form of the state scale of the Spielberger State-Trait Anxiety Inventory (STAI).[8] This yields a single score ranging from 20 to 80. The mean for the adult population is 36. Worry about cancer was measured using the shortened version of the Cancer Worries Scale,[9] which assesses (1) people's perceptions of their own chances of developing cancer, (2) their frequency of cancer related thoughts, (3) the frequency with which they perceive their mood to be affected by such thoughts, and (4) the frequency with which such thoughts affect the performance of their daily tasks.

The participants in this study were patients completing a cancer family history questionnaire as part of a separate study to evaluate its use in general practice.[5] For that study, patients aged between 35 and 65 years registered with a single general practice in Cambridge were invited to participate. They were sent an information sheet explaining that the purpose of the study was to identify the small minority of patients whose family history would put them at sufficiently increased risk of breast or colorectal cancer to warrant the offer of screening to facilitate early diagnosis and treatment. A consent form and short questionnaire to measure baseline levels of general anxiety and worry about cancer were also enclosed. Those wishing to participate were asked to complete and return the consent form and the baseline measure. They were then sent the family history questionnaire (for details see http://www.jmedgenet.com). On the basis of their responses, the majority of patients were judged not to be at significantly more than the population risk of breast or colorectal cancer (lower risk group). These patients were sent a letter telling them that, on the basis of their stated family history, their personal risk of developing breast or colorectal cancer was below the level at which extra screening tests would be recommended. A small number of patients were assessed to be at potentially increased risk where one of the following applied: (1) their family history as reported met local screening criteria for breast or colorectal cancer (table 1) or (2) their family history approached screening criteria so closely that it was considered advisable to check crucial details such as age at onset in relatives, or (3) the information provided on the questionnaire was ambiguous or incomplete and there remained a possibility that the screening criteria might be met, or (4) their family history did not meet screening criteria but suggested an increased risk to the GP assessor. Almost all of these patients were

Table 1 Criteria used to define increased genetic risk sufficient to warrant referral and the offer of screening

For breast cancer, females with one of the following:	
(1)	Three first or second degree relatives with breast or ovarian cancer
(2)	Two first or second degree relatives with breast cancer diagnosed under 60 years of age or ovarian cancer at any age
(3)	One first degree relative with (i) breast cancer diagnosed under 40 years of age, or (ii) bilateral breast cancer, or (iii) male breast cancer
For colorectal cancer, one of the following:	
(1)	A first degree relative plus two other relatives with colorectal cancer and (i) one case diagnosed under 50 years of age, and (ii) one case a first degree relative of the other two, and (iii) at least two generations affected
(2)	A first degree relative with colorectal cancer diagnosed under 45 years of age

Table 2 General anxiety and cancer worries at baseline and follow up for paired responses in (1) patients assessed not to be at significantly increased risk (lower risk group), (2) patients with potentially significant family histories subsequently shown not to be at significantly increased risk (false positive group), (3) patients confirmed to be at significantly increased risk (higher risk group)

	Mean scores (95% CI)					
	Lower risk group		*False positive group*		*Higher risk group*	
	Baseline	*Follow up*	*Baseline*	*Follow up*	*Baseline*	*Follow up*
Speilberger State-Trait Anxiety Inventory						
General anxiety	35.8 (34.6–36.9) n=427	35.1 (33.9–36.2)	34.8 (26.6–42.9) n=7	34.3 (24.1–44.5)	36.3 (31.7–40.9) n=18	38.9 (32.8–45.0)
Cancer Worries Scale						
Perception of own chances of developing cancer	2.95 (2.88–3.01) n=534	2.83 (2.76–2.91)	3.55 (3.19–3.90) n=11	3.27 (2.53–4.01)	3.56 (3.18–3.94) n=25	3.40 (2.99–3.81)
Frequency of cancer related thoughts	1.63 (1.57–1.69) n=564	1.61 (1.55–1.67)	1.73 (1.29–2.16) n=11	1.64 (1.09–2.18)	1.96 (1.59–2.33) n=25	1.76 (1.36–2.16)
Effect on mood	1.21 (1.17–1.26) n=560	1.22 (1.17–1.26)	1.45 (1.10–1.81) n=11	1.27 (0.84–1.71)	1.29 (1.10–1.49) n=24	1.25 (1.03–1.47)
Effect on performance of daily tasks	1.12 (1.08–1.15) n=562	1.11 (1.08–1.15)	1.00 * n=11	1.09 (0.89–1.29)	1.17 (1.01–1.33) n=24	1.08 (0.96–1.20)

CI = confidence interval.
*All respondents scored 1 for this item.

interviewed but in a few cases minor uncertainties were resolved over the telephone. Most were confirmed to be at significantly increased genetic risk (higher risk group) but a minority were deemed not to be at increased risk (false positive group). An explanation of their final risk assessment was given to all these patients, usually at personal interview (all patients assigned to higher risk as a result of the study were informed of this at interview), but in a few cases by telephone or letter. Patients in the higher risk group who had not previously received genetic advice were offered referral to the cancer genetics clinic. All participants were asked to complete a follow up anxiety and cancer worries questionnaire four to six weeks after feedback of their personal risk. In the group of patients referred for genetic advice, this measure was completed after the consultation at which referral was offered but before the appointment at the cancer genetics clinic. Statistical comparisons were made using the Wilcoxon signed rank test for paired data and the Mann-Whitney U test for independent samples. The Cambridge Local Research Ethics Committee gave ethical approval for the study.

The effective practice population for the study of the cancer family history questionnaire was 2265 patients. A total of 666 patients (29%) completed that questionnaire and are the participants in the present study. They differed from the practice population in terms of both gender and age.[5] A total of 62.2% were women, compared to 50.2% of the practice population (χ^2=37.8, p<0.001) and a lower proportion were aged 35-44 (30.2% compared with 40.7%, χ^2=30.3, p<0.001).

A total of 604 patients (91%) returned baseline and follow up measures of anxiety and cancer worry. The gender and age distribution of respondents did not differ significantly from the study population. Paired responses were obtained from 568 patients assessed not to be at significantly increased risk of breast or colorectal cancer (lower risk group) and 36 patients judged to be at potentially increased genetic risk on the basis of their family history questionnaires. The latter group comprised 25 patients who were subsequently confirmed to be at significantly increased risk (higher risk group) and 11 deemed not to be at significantly increased risk after further investigation of their family history (false positive group). Some patients failed to answer all six items of the Spielberger State-Trait Anxiety Inventory or all four items of the Cancer Worries Scale. Table 2 gives the numbers of valid paired responses in each patient group.

The scores for general anxiety and cancer worries at baseline (before completion of the family history questionnaire) and follow up (four to six weeks after receipt of their risk assessment) for all three groups are shown in table 2. In the lower risk group, the only difference in paired responses between the two time points assessed was in patients' perceptions of their personal risk of developing cancer, which showed a small reduction (p<0.001). For the other two groups there were no differences in paired responses for general anxiety or cancer worries. For both the higher risk group and the false positive group, baseline responses showed that their pre-existing perception of their risk of developing cancer was higher than that of the lower risk group (p<0.001 and p=0.003, respectively). For the false positive group, the frequency with which cancer related thoughts affected their mood was also higher (p=0.02).

The results of this study suggest that completion of a cancer family history questionnaire and receipt of an assessment of personal genetic risk for breast and colorectal cancer does not make patients more anxious or worried about cancer. This conclusion is based on a substantial number of subjects, but should be tempered by the fact that only a minority of practice patients returned the family history questionnaire and constituted a self-selected group.

Responses to the Cancer Worries Scale showed that most patients rarely worried about their risks of developing cancer either before or after the study. Indeed, receipt of information that their personal risk was below the level at which extra screening tests would be offered was associated with a small but significant reduction in perceived risks of developing cancer. This raises the question of whether this knowledge could influence health related behaviour. For example, it might reduce the incentive to participate in health related activities, such as attendance for routine mammographic screening or eating a fibre rich diet. This effect has been reported for other forms of screening[10 11] and merits further investigation.

For patients assessed to be at potentially increased risk on the basis of their family history questionnaire, baseline responses showed that their pre-existing perceptions of their own risk of developing cancer were significantly higher than those of other patients. This suggests that many

of these patients already understood the implications of their family history. For patients confirmed to be at significantly increased risk and advised accordingly, their follow up responses showed that they continued to perceive themselves at increased risk with no significant change from baseline. There was no indication that completion of the cancer family history questionnaire and subsequent discussion of their cancer risk exacerbated existing concerns. For the small group of patients assessed not to be at significantly increased risk after further evaluation of their family history, their baseline responses showed that before the study they too perceived themselves to be at increased risk on account of their family history. After being advised that their personal risk of developing cancer was below the level at which extra screening tests would be recommended, their responses at follow up show a mixed reaction. Four patients regarded themselves at lower risk than before, three saw themselves at increased risk, and four were unchanged. The numbers are too small to draw firm conclusions, but suggest that at least some of these patients still regarded their family history as putting them at somewhat increased risk.

Much recent work on the psychological impact of genetic screening has focused on the impact of DNA testing[12] where a positive result usually implies a much greater than population risk. There is evidence that, in women undergoing predictive DNA testing for breast cancer, a positive result has little impact on general levels of anxiety or depression.[13 14] One possible explanation for this is that these women already perceived themselves to be at high risk and were understandably anxious before testing. The result did not, therefore, alter their psychological status appreciably. The present study assessed anxiety in patients participating in a questionnaire survey designed to identify subjects at moderately increased risk with a view to offering extra mammographic or colonoscopic screening rather than DNA testing. Again, no significant change in psychological well being was detected and there was evidence that those at increased risk already perceived themselves at risk, although they may not have sought advice, while those who perceived themselves to be at lower risk had their view endorsed by the process.

If improving knowledge about familial cancer risk is to benefit all patients and not just the better informed, it will be necessary to develop effective ascertainment strategies in primary care. The results of this study suggest that it should be possible to do this without increasing anxiety either in those at increased genetic risk or in those at no more than the population risk.

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Attitudes to genetic testing for breast cancer susceptibility in women at increased risk of developing hereditary breast cancer

EDITOR—The localisation of the two breast cancer susceptibility genes *BRCA1* and *BRCA2* made possible the use of mutation detection as a susceptibility test for people who wish to learn whether they carry a risk conferring mutation.[1–4] Several studies have assessed attitudes to genetic testing for breast cancer susceptibility,[5–11] most of which involved either community samples or women with just one first degree relative with breast cancer. The objective of our study was to assess attitudes to genetic testing for breast cancer susceptibility in a large sample of women at high risk of developing hereditary breast cancer on the basis of family history. The majority of women included in our sample (80%) had a family history consistent with a dominantly inherited predisposition to breast cancer (lifetime risk of 1 in 4 to 1 in 2),[12] and the remainder (20%) was at moderately increased risk of developing breast cancer (lifetime risk of 1 in 8 to 1 in 4).[12]

The findings reported here are based on a sample of 461 unaffected women with a family history of breast cancer. Women who approached one of 14 familial cancer clinics and six associated outreach clinics in five Australian states between November 1996 and January 1999 were eligible for participation. Women were considered ineligible for study participation if they had a previous diagnosis of ovarian or breast cancer, were unable to give informed consent, or had limited literacy in English, since data were collected using self-report questionnaires. The study was approved by 16 institutional ethics committees.