An evaluation of needs of female *BRCA1* and *BRCA2* carriers undergoing genetic counselling

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Abstract

Background—The discovery of the breast and ovarian cancer susceptibility genes *BRCA1* and *BRCA2* has improved our ability to counsel women at increased risk of developing breast and ovarian cancer. The objective of our study was to identify the needs of women who have undergone genetic counselling and testing for *BRCA1/2* and to determine the impact of receiving a positive *BRCA1/2* result. This is the first study to report on a large group of women who have received positive *BRCA1/2* mutation results.

Methods—Questionnaires were distributed to 105 women who had received pre- and post-test genetic counselling for a positive *BRCA1/2* result at the University of Toronto or at McGill University in Montreal, Canada between the years of 1994 and 1998. The questionnaire items included patient motivation for seeking genetic services, information needs, screening and prophylactic surgery practices, satisfaction with access to services and support, the desire for a support group, and overall client satisfaction.

Results—Seventy nine female carriers were surveyed. The majority of the respondents (77%) were satisfied with the information they received during the genetic counselling process. Women with a previous diagnosis of cancer indicated that they needed more information relating to cancer treatment compared to women without cancer ($p=0.05$). Nineteen percent of the women felt they needed more support than was received. Fifty eight percent of the women reported that their screening practices had changed since they received their result. Young women (below the age of 50) and women with no previous diagnosis of cancer were most likely to have changed their screening practices. Nearly two thirds of the respondents said they had considered prophylactic surgery of the breasts or ovaries. Twenty eight percent of the women had prophylactic mastectomy and 54% had undergone prophylactic oophorectomy. Women with an educational level of high school or more were more likely to have undergone prophylactic bilateral mastectomy than those with less education ($p=0.07$) but were less likely to undergo prophylactic oophorectomy ($p=0.0007$).

Conclusion—These findings have a direct impact on the counselling and risk management of female *BRCA* mutation carriers. Age, education, and a previous diagnosis of cancer are important determinants in a woman's decision making after receiving positive genetic test results.

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Keywords: genetic counselling; *BRCA1*; *BRCA2*; cancer genetics

The discovery of the breast and ovarian cancer susceptibility genes *BRCA1* and *BRCA2* makes it possible to counsel a subgroup of women at increased risk of developing breast and ovarian cancer.^{1,2} These advances prompted the establishment of cancer genetics clinics in North America and western Europe, which provide genetic counselling and molecular testing to at risk subjects. The estimated risk of developing breast cancer associated with a *BRCA1* or *BRCA2* mutation is 56% to 80% up to the age of 70 and the risk of developing ovarian cancer ranges from 16% to 40% by the age of 70.^{3,4}

To date, little is known about the impact of genetic testing on women who receive a result indicating that they carry one of the high risk mutations. Areas of interest include surveillance behaviour, information needs, psychological adjustment and distress, and emotional responses. Based on experience from other adult onset conditions (such as Huntington's disease), concern has been raised about possible adverse psychological effects of *BRCA* genetic testing, particularly for women who receive a positive result. This concern has prompted a number of investigators to study predictors of adverse psychological effects and potential means of minimising them.^{5–14} Biesecker *et al*⁵ found that common reactions to a positive result included disbelief, anger, despair, relief, and ecstasy. Lerman *et al*⁶ studied patient outcomes associated with *BRCA1* testing and concluded that some high risk subjects may benefit psychologically if extensive counselling is provided from the onset of the testing process. These studies suggest that with appropriate counselling, women are better able to deal with positive test results.

One of the goals of health care professionals who provide genetic counselling and genetic testing for hereditary breast and ovarian cancer is to promote cancer prevention practices. To achieve this goal, high risk subjects are presented with options including screening and prophylactic surgery. However, numerous studies have shown that behavioural change is

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not easily achieved.^{5 6 15-17} Lerman *et al*⁶ found *BRCA* mutation carriers to be reluctant to adopt new surveillance and prevention practices and concluded that genetic testing may not lead to anticipated reductions in cancer morbidity or mortality.

The experience of patients undergoing genetic counselling and testing for breast cancer has been examined.¹⁸⁻²² Most studies are based on hypothetical scenarios about anticipated feelings and behaviour among clients before undergoing genetic testing. However, when women are told that they actually carry a *BRCA1/2* mutation, their attitude and decisions about screening and preventive surgery may be different. There have been three family based studies to date that present data on men and women who received test results.^{16 27 29} Lynch *et al*¹⁶ described reactions to disclosure of test results from 14 families with 78 *BRCA* mutation carrier males and females. This descriptive study found that over one third of those who tested positive reported anger, sadness, or guilt. Smith *et al*²⁷ described the experience and psychological distress of 87 male and 125 female members of one family who underwent predictive *BRCA* testing, including 47 females who were determined to be *BRCA* mutation carriers. At one to two weeks follow up, the study measured greater distress for female carriers who were the first to be tested and those whose tested sibs were non-carriers. Wagner *et al*²⁹ recently reported findings on 35 *BRCA* families and 90 family members in Austria, both carriers and non-carriers, and concluded that carrier status was not associated with increased depression symptoms in mutation carriers.

We have conducted a large survey on women who tested positive for a *BRCA1* or *BRCA2* mutation. The purpose of this study is to determine their level of satisfaction with the genetic counselling and testing process, and to identify unmet needs of female *BRCA* mutation carriers.

Methods

STUDY POPULATION

Eligible subjects were identified from familial breast cancer clinics at the Women's College Hospital, University of Toronto and McGill University, Montreal. Patients received *BRCA1/2* mutation results between November 1994 and November 1998.

Eligible subjects included women who had received genetic counselling and testing and were found to have a mutation in *BRCA1* or *BRCA2*. One hundred and seven eligible subjects were identified from 70 families. All women had received full pre-test and post-test genetic counselling. Of 107 eligible women, two were excluded because of language and comprehension barriers. Questionnaires were mailed out between June 1998 and December 1998. Of the total 79 respondents, 52 received genetic counselling and testing services at the Women's College Hospital in Toronto, 17 at the McGill University hospital clinics in Montreal, and 10 were counselled at other centres in Canada and the USA with testing done in

Toronto. The respondents included 46 women affected with cancer and 33 unaffected women. Two respondents resided in the United States.

GENETIC COUNSELLING SERVICES

Genetic counselling services at the Women's College Hospital of the University of Toronto and at McGill University Hospitals comprise appointments with a genetic counsellor and either a geneticist or oncologist. Genetic testing is offered as part of a research programme at these institutions. Patients are usually referred to the genetics clinic by other health care professionals, such as oncologists and general practitioners. The clients include subjects both affected and unaffected with cancer. Patients at the familial cancer programmes are uniformly provided with a minimum of one pre-test counselling session and one test disclosure counselling session (median=two sessions). The number of counselling sessions provided is not dependent on the client's cancer status. Comprehensive information about all available management options for the consultand and their relatives are discussed, including screening, prophylactic surgery, and chemoprevention. Genetic counsellors are responsible for arranging referrals to other specialists for consultation and screening. The programmes have identified local and regional health care specialists, knowledgeable about the unique needs of *BRCA* mutation carriers. Many of these patients participate in other research projects and trials. Table 1 summarises the standard procedures at these familial cancer clinics, similar to procedures at other research facilities in North America.

Men and women at these familial cancer programmes are provided with a minimum of one pre-test counselling session and one disclosure counselling session.

PROCEDURES

All study procedures have been approved by the Institutional Review Boards of Women's College Hospital and Montreal General, Royal Victoria and Jewish General Hospitals in Montreal. Eligible women received a letter explaining the study, an invitation to participate, a consent form, and a questionnaire. If the woman wished to participate, she returned the completed questionnaire to the investigators. Women who did not return their questionnaire were reminded with a telephone call and offered participation. Fifteen women opted to complete the questionnaire by telephone interview.

QUESTIONNAIRE DESIGN

The questionnaire consisted of 37 items assessing patients' motivations for seeking genetic services, information needs, screening and prophylactic surgery practices, access to services (that is, health care referrals), support resources, desire for a support group, and overall satisfaction. Participants were given the option of obtaining further follow up with the genetic counsellors. A separate questionnaire was included with the first 20 mailings to validate the appropriateness of the items for

Table 1 Genetic counselling services at the Women's College Hospital in Toronto and McGill University

	Contents of action
<i>Referral</i>	Referral by self, physician, family member, or from research programme Family history is obtained by phone or via mailed questionnaire
<i>Appointment 1</i> Genetic counsellor and geneticist or oncologist	Draught of pedigree is reviewed with patient Procedure of familial cancer clinic explained Expectations and limitations of DNA testing in research programme explained Management options are discussed if patient is found to be at high risk
<i>Appointment 1 or 2: pre-test</i>	Family history is confirmed by pathology obtained Objective risk assessment is provided (based on Claus model) Possible adverse complications of DNA testing discussed (eg, insurance) Blood is drawn with appropriate consent
<i>Appointment 2 or 3: disclosure</i>	Face to face disclosure of test results Possible family implications are discussed Screening and prophylactic treatment options are discussed Written referrals to specialists (eg, gynaecologist)
<i>Referrals to specialists</i> <i>Follow up</i> Genetic counsellor or research associate or nurse	By appointment (offered after 3 months of disclosure) By phone

assessing patient's needs using the items identified. These questionnaires are available upon request.

ANALYSIS

Patients were divided into those with no previous diagnosis of cancer and those with a previous diagnosis of cancer. Education levels were divided into high school graduation and less or greater than high school. Age was analysed as both a continuous and categorical variable. When analysed as a categorical variable, age was divided into less than 50 years and greater than or equal to 50 years of age. Pearson's chi-square test was used for nominal data and Student *t* tests and Mann-Whitney tests were used for comparison of continuous variables. Logistic regression was used for multivariate modelling. The significance level was set at 0.05, two sided.

Results

A total of 105 eligible women from 70 different families were mailed a questionnaire, including 71 women from the University of Toronto and 34 from McGill University. The women were white and of French-Canadian, Ashkenazi Jewish, European, or Hispanic ethnicity. Sixty four questionnaires were returned by mail and 15 were completed by telephone. Analysis was performed on a total of 79 completed questionnaires (17 from McGill and 62 from the University of Toronto).

There were 26 non-respondents (24.8% of questionnaires mailed). Seven women declined to participate (via returned consent form indicating refusal to participate) and 19 women did not respond to the invitation to participate in this study. The mean age of the non-respondents was 52.6 years (range 39 to 65 years), compared to 50.4 years in respondents (range 24 to 81 years) ($p=0.41$). Seventy seven percent of the non-respondents had a previous diagnosis of cancer, whereas 58% of the respondents had a personal history of cancer. The average level of education for the respondents was some college or university, 11% had less than high school education, and 14% had a postgraduate degree. The education level for non-respondents was not known.

Table 2 Sources of referral to genetic services ($n=79$)

Source of referral	No	%
Self	12	15.2
Friend	1	1.3
Family member	35	44.3
Canadian Cancer Society	0	0
Family doctor	3	3.8
Gynaecologist	4	5.1
Oncologist	9	11.4
Other	15	19

Table 2 summarises the sources of referral. Forty four percent of respondents were referred to a genetics clinic by a family member. Fifteen percent were self-referred and 11% were referred by an oncologist. Forty three percent of respondents belonged to a family with a known *BRCA1/2* mutation. Of these, 62% (21/34) were referred by family members.

Fifty eight percent of respondents had a previous diagnosis of cancer. Of these, thirty seven had unilateral breast cancer, three had bilateral breast cancer, and six had ovarian cancer. Other reported cancers were colon, uterine, melanoma, and cervical cancers. The median year of cancer diagnosis was 1990 (range 1961 to 1998). The mean number of breast cancer cases in the families in first or second degree relatives was 3.0 (range 0-7) and the mean number of deaths from breast cancer was 1.1 (range 0-5). The mean number of ovarian cancer cases in the families was 1.2 (range 0-3) and the mean number of deaths from ovarian cancer was 1.0 (range 0-3).

The mean time between respondents receiving mutation results and completing the questionnaire was 17.3 months.

The top three reasons for seeking genetic services were "to learn about personal risk of cancer" (34.2%), "to learn about children's risk of cancer" (16.5%), and "to learn about family's risk of cancer" (11.4%) (table 3). Among women with children, personal risk was still cited as the principal reason for undergoing genetic testing.

Open ended questions were used to assess the patients' emotional responses to their positive mutation test result. When asked to describe initial feelings upon receiving *BRCA1/2* results, responses included: "not surprised", "relieved", "angry", "sad", "devastated",

Table 3 Reason for seeking genetic testing

Reason	1st reason (n=79)		2nd reason (n=76)		3rd reason (n=68)	
	No	%	No	%	No	%
Educational information	6	7.6	1	1.3	1	1.5
Learn about personal risk for cancer	27	34.2	10	13.2	2	2.9
Learn about children's risk for cancer	13	16.5	20	26.3	5	7.4
Learn about family's risk for cancer	9	11.4	12	15.8	16	23.5
Learn about the risk of developing cancer if you have a cancer gene	4	5.1	13	17.1	8	11.8
To obtain screening and surveillance recommendations	3	3.8	7	9.2	10	14.7
To obtain genetic DNA testing	5	6.3	5	6.6	10	14.7
To support research	7	8.9	5	6.6	10	14.7
My doctor's recommendation	2	2.5	2	2.6	1	1.5
My family's recommendation	3	3.8	1	1.3	5	7.4

“shocked”, “concerned about children”, and “guilty for passing gene on to my daughters”. These responses were all cited by multiple respondents. Subjects were also asked if feelings and emotions had changed since first learning about being a carrier of *BRCA1/2*. Responses given by more than one woman included: “I now realise what it really means to have the gene”, “I am more realistic about the future”, and “I am more concerned about how my children will be affected”.

INFORMATION NEEDS

Seventy seven percent of women (59/77) were satisfied with the information they received during genetic counselling. Twenty one percent (16/77) said there was additional information that they would have liked, but did not receive, including information on cancer treatment. This included information on preventive surgery, chemoprevention, and new screening modalities (for example, MRI). In the study sample there were no significant differences in information needs based on age or education (table 4), but there was a significant difference by cancer status. Women affected with cancer were more likely to state that there was information they needed but did not receive at the result disclosure session than were unaffected women ($p=0.05$). There were no differences in information needs based on centre (Toronto *v* Montreal) ($p=0.40$). There were also no differences in information needs based on the time since the subject had received a positive mutation result ($p=0.95$).

Twenty eight percent of women felt they had not received enough information about prophylactic surgery during their genetic counselling sessions. Eighteen percent of the respondents felt they did not receive enough information about cancer screening during their genetic counselling sessions. There were no significant differences in need for information about prophylactic surgery or screening based on education, age, or cancer status (table 4).

The great majority of women (95%) shared the information they received, including their test result, with other family members. One third of the women indicated that the experience of genetic testing had affected either family or personal relationships. There was a significant difference in this response based on whether the woman was the proband or has another family member who received predic-

tive testing ($p=0.02$). Proband was more likely to state that their personal or family relationships had been altered as a result of the testing process. Of the 26 women who had indicated that their family relationships had changed as a consequence of the genetic testing, two did not indicate how these had changed. Twelve stated that their family relationships were strengthened by this information: “we value each day more and each other”; “(genetic testing) has brought the issue to the surface to be discussed openly and honestly”; “(I am) not waiting to do things any more with my kids”. Twelve women stated that genetic testing had a negative impact on their family or personal relationships: “my husband is even more distant”; “my daughter is terrified”; “my preoccupation with the test results and its implications has affected my availability to my family (spouse and children)”.

SUPPORT RESOURCES

The genetic counsellor was cited as the most frequent source of psychosocial support, followed by a family member and spouse (table 5). The genetic counsellor is the primary contact for each woman in the genetics clinics. This may explain the importance of the genetic counsellor in providing psychosocial support. The majority of women (84%) did not want help or support in sharing the genetic information (their test result) with their family. Nearly one third of women (23/76) would have liked their families to have been more involved in the genetic testing process.

Nineteen percent of women felt they needed more support than was received. There were no significant differences in whether women required more support according to cancer status, age, or education. Half (39/77) of the women said they were interested in being referred to another health care professional after they had received their test result. The most common request was for a gynaecologist (22/39), followed by a breast specialist (7/39). When asked if they were successful in obtaining the referral, 74.4% said yes.

Participants were asked about the need for a support group for *BRCA1/2* mutation carriers and about their desire to participate in one. Over two thirds (68.4%) felt a support group was necessary for *BRCA* mutation carriers, but only 34.2% said they wished to participate. Women with an educational level beyond high

Table 4 Patient needs and outcomes measured

Needs assessed	Variable	Response "yes" (%)	p value	Adjusted p value
<i>Information needs</i>				
Needed more information	Age	<50 y	17.5	0.39
		≥50 y	25.3	
	Education	High school or less	12.5	0.20
Needed more information about screening		> High school	25.5	
	Cancer status	Cancer	28.9	0.05
		No cancer	10.0	
Needed more information about prophylactic surgery	Age	<50 y	87.5	0.21
		≥50 y	76.5	
	Education	High school or less	88.0	0.37
Needed more information about prophylactic surgery		> High school	79.6	
	Cancer status	Cancer	81.0	0.70
		No cancer	84.4	
Needed more information about prophylactic surgery	Age	<50 y	71.8	0.316
		≥50 y	75.0	
	Education	High school or less	86.4	0.09
Needed more information about prophylactic surgery		> High school	67.3	
	Cancer status	Cancer	75.0	0.70
		No cancer	71.0	
<i>Support resources</i>				
Needed more support	Age	<50 y	25.6	0.11
		≥50 y	11.4	
	Education	> High school	20.8	0.62
Opinion that a support group is necessary for carriers		High school or less	16.0	
	Cancer status	Cancer	19.6	0.93
		No cancer	18.8	
Opinion that a support group is necessary for carriers	Age	<50 y	77.5	0.45
		≥50 y	69.7	
	Education	> High school	75.5	0.67
Personal interest in a support group		High school or less	70.8	
	Cancer status	Cancer	71.4	0.56
		No cancer	77.4	
Personal interest in a support group	Age	<50 y	46.1	0.17
		≥50 y	30.0	
	Education	> High school	50.0	0.0089
Personal interest in a support group		High school or less	17.4	
	Cancer status	Cancer	28.2	0.034
		No cancer	53.3	
<i>Screening practices and prophylactic surgery</i>				
Screening practices changed	Age	<50 y	66.7	0.08
		≥50 y	47.2	
	Education	High school or less	48.0	0.23
Screening practices changed		> High school	62.3	
	Cancer status	Cancer	41.3	0.0004
		No cancer	81.2	
Considered prophylactic surgery	Age	<50 y	80.5	0.002
		≥50 y	45.2	
	Education	High school or less	34.8	0.0002
Considered prophylactic surgery		> High school	79.6	
	Cancer status	Cancer	56.8	0.06
		No cancer	78.6	
Had prophylactic mastectomy (bilateral or unilateral)	Age	<50 y	34.8	0.33
		≥50 y	39.4	
	Education	High school or less	18.8	0.07
Had prophylactic mastectomy (bilateral or unilateral)		> High school	45.0	
	Cancer status	Cancer	51.6	0.02
		No cancer	20.0	
Had prophylactic oophorectomy	Age	<50 y	45.5	0.36
		≥50 y	64.0	
	Education	High school or less	92.9	0.0007
Had prophylactic oophorectomy		> High school	40.5	
	Cancer status		NA	NA

*Adjusted by education.

†Adjusted by age.

school without a previous diagnosis of cancer were more likely to be interested in taking part in a support group than those with less education ($p=0.009$) or without cancer ($p=0.034$).

Table 5 Sources of support ($n=74$) *

Source	No	%
Doctor (eg, oncologist, family doctor, etc)	17	23
Genetic counsellor	41	55.4
Nurse	4	5.4
Psychologist†	0	0
Family	39	52.7
Spouse	33	44.6
Friend	8	10.8
Other	7	9.5

*More than one source could be given.

†At the time, psychologists were not directly involved in genetics clinic.

HEALTH CARE PRACTICES

Screening

More than half of women (45/78) reported their cancer screening practices had changed since they learned of their *BRCA1/2* carrier status. Women younger than 50 years old ($p=0.08$) and women with no previous diagnosis of cancer ($p=0.0004$) were most likely to have increased their cancer screening practices. The majority of women (63/75) did not have any difficulties obtaining regular screening. Table 6 summarises the self-reported screening practices of respondents for breast cancer and ovarian cancer.

Prophylactic surgery

Nearly two thirds of the respondents (47/72) said they had considered prophylactic surgery

Table 6 Screening practices at follow up (as reported by respondents)

	Yes	Frequency	No
Regular BSE	72.4% (53/76)	Monthly 65.5% (36/55)	27.6% (21/76)
Regular CBE	86.8% (66/76)	Semi-annually 60.6% (40/66) Annually 21.2% (14/66)	13.2% (10/76)
Regular mammogram	87.3% (48/55)†	Semi-annually 10.4% (5/48) Annually 79.2% (38/48)	12.7% (7/55)
Regular pelvic exam	30/40 (75.0%)‡	Semi-annually 13/30 (43.3%) Annually 11/30 (36.7%)	9/40 (22.5%)
Regular pelvic ultrasound	26/39 (66.7%)‡	Semi-annually 12/26 (46.2%) Annually 9/26 (34.6%)	13/39 (33.3%)
Regular CA125 blood test*	22/40 (55.0%)‡	Semi-annually 12/22 (54.5%)	10/40 (25.0%)

*8/40 (20.0%) did not know about CA125 blood test.

†Excluding those with bilateral breast cancer or prophylactic double mastectomies.

‡Excluding those with oophorectomy (for ovarian cancer, prophylaxis, or other reason).

either of the breasts or ovaries. Women below the age of 50 were more likely to have considered prophylactic surgery (80.5%) than women aged 50 years and above (45.2%) ($p=0.002$). Among those with no previous diagnosis of cancer, women aged less than 50 years were more likely to have considered prophylactic surgery (86.4%) than older women (50%) ($p=0.054$). Similarly, among women with a previous diagnosis of cancer, younger women (<50 years) were more likely to have considered prophylactic surgery (73.7%) than older women (44%) ($p=0.049$). Women with an educational level above high school were more likely to have considered prophylactic surgery (79.6%) than those with an educational level of high school or below (34.8%) ($p=0.0002$).

Twenty two of the 76 participants (29%) had either contralateral or bilateral prophylactic mastectomy. Sixteen of these women (73%) had prophylactic mastectomy following the genetic test result disclosure, whereas six women had prophylactic mastectomy before receiving results. Sixty four percent (14/22) of women who had prophylactic mastectomy completed the questionnaire less than 18 months after receiving their genetic test results. Of the 37 women who had unilateral breast cancer, 40.5% had contralateral prophylactic mastectomy following the initial diagnosis. Of women with no previous breast cancer, 15.4% (6/39) had prophylactic bilateral mastectomy.

The number of breast cancer cases and breast cancer deaths in the family were not found to be related to the decision to have prophylactic mastectomy. Women who had a prophylactic mastectomy had a mean of 2.4 breast cancers in the family and 1.1 deaths from breast cancer, whereas carriers who did not have prophylactic mastectomy had a mean of 3.2 breast cancers and 1.3 deaths from breast cancer in the family ($p=0.211$ and $p=0.712$, respectively). Similarly, the number of ovarian cancer cases and ovarian cancer deaths in the family were not found to be related to the decision to have prophylactic oophorectomy. Women who had prophylactic oophorectomy had a mean of 1.2 ovarian cancers in the family and 1.1 deaths from ovarian cancer, whereas carriers who did not have prophylactic oophorectomy had a mean of 1.0 ovarian cancers and 0.8 deaths from ovarian cancer in the family ($p=0.353$ and $p=0.229$, respectively).

The rates of oophorectomy were higher. Among women who had no previous diagnosis

of ovarian cancer, 53.6% (30/56) had prophylactic bilateral salpingo-oophorectomy. Fifty percent of women who had prophylactic oophorectomy completed the questionnaire less than 18 months since receiving their genetic test result. There was a statistically significant difference based on educational level; those with an educational level of high school or less (92.9%) were more likely to have a prophylactic oophorectomy than those with an educational level above high school (40.5%) ($p=0.0007$). When educational level was adjusted for age, there was still a significant difference ($p=0.01$). These results included all women who had undergone a prophylactic oophorectomy before and after genetic testing. When eight women who had undergone a prophylactic oophorectomy before genetic testing were excluded from the analysis, education level was still significantly associated with prophylactic oophorectomy ($p=0.026$).

SATISFACTION

Overall, 81.3% (61/75) of women believed that they were receiving adequate medical care in the prevention of breast and ovarian cancer. Over one third (33.8%) felt the need to visit the genetic counsellor again after receiving their test result. An overwhelming majority (92.3%) said they would recommend testing for *BRCA1/2* to other women in their situation. The respondents were asked to rate their overall satisfaction with their experience of undergoing genetic testing on a scale of one to five, one indicating extremely dissatisfied and five indicating extremely satisfied. Nearly 65% (51/79) said they were very or extremely satisfied. Only three patients were very or extremely dissatisfied, including one respondent who believed she was denied disability insurance based on her genetic test result.

Patient satisfaction was also analysed according to the length of time since the mutation result was received. Thirty eight women had received mutation results greater than or equal to 18 months, and 41 women had received results less than 18 months ago. There was no difference in patient satisfaction based on time since receiving mutation result ($p=0.95$).

Discussion

In this study, we surveyed the needs of women undergoing genetic counselling and predictive testing in two familial breast-ovarian cancer clinics in Canada. The reasons cited by the participants for seeking genetic counselling were similar to those of previous reports on motivations for having *BRCA1/2* genetic testing.^{16 22 23} The significant proportion of participants (34/79) who underwent predictive testing for a known family mutation may explain why the most common reason for having genetic testing was “to learn about personal risk of cancer”, while in the other studies concern about risk of cancer to children was the primary motivation.^{16 23}

One of the goals of cancer genetic counselling is to promote surveillance for early detection of cancer among *BRCA* mutation carriers.²⁴ The majority of women did not have

any difficulties obtaining screening. This may be because of the universal access to health care in Canada. There may be financial concerns with health care for US citizens. The potential difficulties to be encountered in the Canadian health care system may relate to waiting time and access for rural communities. We found that younger women (below the age of 50) and women with no previous diagnosis of cancer were most likely to change their screening practices following genetic counselling and testing. It is possible that older women are more likely to be involved in cancer screening protocols based on recommendations currently in place for the general population. It has been shown that women do not necessarily adhere to recommended screening or preventive options discussed when they are seen for results or follow up.⁶ Adherence to surveillance measures, such as breast self-examination and mammography, appears to be influenced by levels of anxiety, intrusive thoughts about cancer, psychological distress, education, employment, and age.¹⁵⁻²⁵ A high degree of adherence to recommended mammographic screening guidelines has been reported among women at high risk for breast cancer.¹⁷ In contrast, in a group of *BRCA* carriers previously studied, only 39% reported receiving recommended mammographic screening.⁶ Table 6 describes the self-reported screening practices of our group of patients. The majority of women in all categories reported receiving regular screening for early detection of breast and ovarian cancer. The high adherence observed here may represent errors in recollection and, in future studies, it would be preferable to request the date of last screening or compare attendance for scheduled appointments at a clinic.

Nearly two thirds of the respondents (65.3%) said they had considered prophylactic surgery (either mastectomy or oophorectomy). The option of prophylactic surgery is routinely presented to women as a method of risk reduction during the pre-test genetic counselling sessions at the University of Toronto and McGill University familial cancer clinics. The content of this discussion has not changed considerably for the women who considered this option; in fact, McGill University researchers were counselling women on prophylactic surgery using linkage analysis before the cloning of *BRCA1* in 1994. Women below the age of 50 and those with an educational level above high school were more likely to have considered this option. Wagner *et al*²⁹ recently described how 50% of female carriers in Austria would undergo prophylactic oophorectomy (53% affected and 46% non-affected carriers). Previously, Lerman *et al*¹⁰ reported that 31% of female *BRCA* mutation carriers considered prophylactic mastectomy and 48% considered prophylactic oophorectomy. However, at six months post-result disclosure, Lerman *et al*¹⁰ determined that only 1% had undergone prophylactic mastectomy and 2% had undergone prophylactic oophorectomy. In contrast, among our study respondents, 28% had prophylactic mastectomy and 54% had prophylactic oophorectomy on follow up inter-

view. This increased frequency of prophylactic surgery in our study may be explained by a longer follow up period among our study subjects. The difference may also reflect temporal changes in attitudes to prophylactic surgery. Our physicians and patients may be more accepting of the effectiveness of prophylactic surgery in the prevention of breast and ovarian cancers. Recent findings by Hartmann *et al*²⁸ have suggested that prophylactic mastectomy offers at least an 80% reduction in risk of breast cancer in high risk women. This knowledge may influence the recommendations about prophylactic surgery that are given to *BRCA1/2* mutation carriers. The differences between our findings and those of Lerman *et al*¹⁰ may also reflect a difference between the health insurance systems in Canada and the United States. In Canada, prophylactic surgery (including breast reconstruction) is covered by the universal provincial health insurance plans. At present, concerns about health care coverage are minor in Canada, but possible discrimination in life insurance, disability insurance, and long term care insurance are areas of concern in both countries.²⁶ One respondent was dissatisfied with the genetic testing experience and would not recommend it to others because she believed that she was denied disability insurance on the basis of her mutation carrier status. Further research is needed in the area of genetic information and insurance discrimination in Canada.

Women with a previous diagnosis of breast cancer were more likely to opt for prophylactic surgery (of the contralateral breast) than were healthy women (table 4). Carriers with an educational level above high school were more likely to have had a prophylactic mastectomy compared to less educated women, although the association was not statistically significant ($p=0.07$). In contrast, women with an educational level above high school were less likely to have undergone prophylactic bilateral oophorectomy than less educated women ($p=0.0007$); this effect remained after adjusting for age ($p=0.01$). We believe that surgical removal of the breasts has a greater impact on a woman's self-image than oophorectomy. Level of education may be associated with the relative importance that a woman places on self-image and on the consequence of surgical early menopause. This may explain the effect observed for educational levels. For the carriers in this study, the number of breast or ovarian cancer diagnoses or deaths in the family were not found to be related to the woman's decision to proceed with surgery.

Lerman *et al*¹⁰ concluded that those who opt for genetic testing might represent a psychologically vulnerable subgroup of high risk women. In our study, probands were more likely than their relatives to report that their personal or family relationships had been affected by the genetic counselling process ($p=0.02$). This probably represents the added anxiety of being the initiator of the genetic testing for the family or "gate keeper" role in disclosing this information to other relatives at risk. One third of women stated that their

family or personal relationships had changed since they received their mutation result. There was an equal distribution of positive and adverse effects on family relationships. Although 12 of the women indicated that genetic testing had a negative impact, the majority of the women would recommend genetic testing to other women in their situation. This suggests the value of genetic test results to the subject at risk for cancer, as was described by the women in this study. As previously stated, the primary motivation for undergoing genetic testing in this population was to learn about personal risk for cancer (table 3). The information that a person receives from genetic testing has potentially life altering consequences, including both positive and negative effects on relationships. Health care professionals may wish to make clients and their families aware of these effects and offer support when necessary.

Lynch *et al*¹⁸ reported that those who are told they have a germline mutation express a variety of reactions including acceptance because the results are not a surprise to them, relief from anxiety with the removal of uncertainty about their genetic risk status, a positive attitude with regard to prevention, and feelings of sadness or even anger. Similarly, our respondents reported a wide range of emotional reactions. We agree with Lynch *et al*¹⁸ that genetic counsellors should be responsive to all of these emotions.

Overall, 81% of respondents indicated they did not need additional support. Younger women (below the age of 50) were more likely to report a need for greater support, although this was not statistically significant ($p=0.11$). Audrain *et al*²⁰ assessed the preferences for the content and process of genetic counselling and testing for breast-ovarian cancer susceptibility among women at high risk. Younger women (below the age of 50) were more likely to request a discussion of emotional reactions to testing and emotional support in post-test counselling. Women with higher levels of education and no previous diagnosis of cancer expressed a greater interest in taking part in a support group (table 4).

Bleiker *et al*²² found high levels of satisfaction with clinical genetic services received at family cancer clinics in The Netherlands. Communication of information regarding the possible impact of genetic testing on daily life and psychosocial support during and after the process of genetic counselling were identified as areas that needed attention. Client satisfaction is often cited as a measure of success in patient-physician communication, but this item is inherently flawed. The fact that clients take the time to complete a questionnaire is itself an indication of satisfaction, in that dissatisfied clients would not commit their time and effort to answer a physician survey. In our study, a relatively high response rate of 82.3% (79/96) was achieved and nearly 65% (51/79) of respondents said they were very or extremely satisfied. An overwhelming majority (92.3%) of the women said that they would recommend genetic testing to other women in their situation. This, combined with high satisfac-

tion and response rates, suggests that counselling is effective in helping a woman throughout the genetic testing process. Based on our respondent who believed she was denied disability insurance based on her genetic test result, it is important for health care professionals working in the genetics field to communicate clearly the current knowledge of employment and insurance implications and potential discrimination based on a positive *BRCA* mutation result.

Genetic counsellors and other health care professionals must also be sensitive to the emotional sequelae of disclosing positive mutation results to women, particularly in the period following disclosure, and must attempt to meet the needs of female carriers as shown in this study. Equally important is the effect of the woman's genetic test result on her family relationships as recorded in one third of our respondents. These findings have a direct impact on the counselling and risk management of *BRCA* carriers. Further work is under way to assess the effectiveness of a group counselling intervention for *BRCA* mutation carriers.

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