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Evaluation of the Needs of Male Carriers of Mutations in *BRCA1* or *BRCA2* Who Have Undergone Genetic Counseling

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To date, the concerns of men at risk of inheriting a *BRCA1* mutation or a *BRCA2* mutation have received little attention. It had been anticipated that few men would be interested in predictive testing when a *BRCA* mutation was identified in their family. However, these men are often affected emotionally by diagnoses of breast cancer in their relatives and may themselves harbor fears that cancer will develop. Male carriers of *BRCA1/2* mutations are at increased risk of development of cancers of several types, including those of the breast and prostate. We conducted an evaluation of the needs and experiences of 59 male carriers of *BRCA1/2* mutations followed at either the University of Toronto or Creighton University. We assessed their motivations for seeking genetic counseling and testing, involvement in family discussions of breast and ovarian cancer, risk perception, changes in cancer-screening practices, and overall satisfaction with the genetic-counseling process. The principal motivation for seeking genetic counseling was concern for their daughters. The majority (88%) of men participated in family conversations about breast and ovarian cancer, and 47% participated in conversations about prophylactic surgery. Most men believed that they were at increased risk of development of cancer (prostate, breast, colorectal, and skin cancers). However, fewer than one-half (43%) of the men with no previous diagnosis of cancer stated that their prostate cancer–surveillance practices had changed after they had received genetic test results. More than one-half (55%) had intrusive thoughts about their cancer risk. Although levels of satisfaction were high, practitioners should be aware of (*a*) potential pressures influencing men to request predictive testing, (*b*) the difficulties that men encounter in establishing surveillance regimens for breast and prostate cancer, and (*c*) the general lack of information about men's particular experiences in the medical community.

Introduction

In Canada and the United States, genetic counseling is available to individuals at increased risk of breast and ovarian cancer. These individuals usually have a family history of cancer and may be offered screening for *BRCA1* (MIM 113705) and *BRCA2* (MIM 600185) mutations. Once a mutation has been identified in the family, predictive genetic testing may be offered to at-risk relatives, including males. Estimates of the cumulative lifetime risk, to age 70 years, of development of breast cancer associated with a *BRCA1/2* mutation are 28%–87% in females (Ford et al. 1994; Hopper et al. 1999; Warner et al. 1999), and the risk of development of ovarian cancer is 16%–60% (Ford et al. 1994; Struewing et al. 1997). Although females face greater risks, male carriers of *BRCA1/2* mutations have an elevated risk of development of breast, prostate, and other cancers (Ford et al. 1994; Struewing et al. 1997; Breast Cancer Linkage Consortium 1999). Germline *BRCA1/2* mutations have been associated, in both sexes, with elevated risks for cancers at multiple organ sites, including colon/rectum, pancreas, gall bladder, bile duct, and stomach cancers and cutaneous malignant melanoma (Ford et al. 1994; Breast Cancer Linkage Consortium 1999; Moslehi et al. 2000).

The experiences of patients undergoing genetic testing for breast and ovarian cancer have been examined (Kelly 1992; Bleiker et al. 1997; Audrain et al. 1998; Stadler and Mulvihill 1998; Lynch et al. 1999). Most studies are based on hypothetical scenarios about anticipated feelings and behaviors among clients prior to genetic testing. To date, attention has focused on the women in these families, because they are at greater risk of development of cancer; little is known about the impact of genetic testing on men who receive a positive result.

Concern has been expressed over possible adverse psychological effects of *BRCA* genetic testing, particularly for those who receive a positive result. This con-

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cern has prompted a number of investigators to study predictors of adverse psychological effects—and potential means of minimizing them (Biesecker et al. 1993; Hoskins et al. 1995; Richards et al. 1995; Botkin et al. 1996; Lerman et al. 1996, 1997*a*, 1997*b*, 1997*c*, 1998; Dudok deWit et al. 1998). Only two family-based studies have examined the reactions of men who received test results (Lynch et al. 1997; Smith et al. 1999). Smith et al. (1999) observed that male carriers of mutations experienced greater distress if they were the first of their siblings to be tested.

In the present article, we present data on 59 male carriers of *BRCA1/2* mutations who were asked about their motivations for testing, their involvement in family discussions of breast and ovarian cancer, their changes in cancer-screening practices, and their overall satisfaction with the genetic-counseling process. To date, this is the largest sample of male carriers of *BRCA* mutations who have undergone genetic counseling and testing. The purpose of this study was to identify unmet needs and to describe men's experiences with genetic counseling and testing and to compare this information with the experience of the female carriers of *BRCA* mutations in these families.

Subjects and Methods

Study Population

Eligible subjects were identified from the familial breast and ovarian cancer clinics of the Sunnybrook and Women's College Health Sciences Centre (Women's College Hospital Campus) of the University of Toronto and of Creighton University in Omaha. Patients were those who, during the period June 1995–July 1999 (mean September 1997), had received a positive result for *BRCA1/2* mutation. Eligible subjects included men who had received genetic counseling and testing and who had been found to have a *BRCA1/2* mutation. *BRCA1/2* mutations were believed to be deleterious and resulted in a truncated protein.

Sixty-nine subjects (53 from Creighton University and 16 from the University of Toronto) were identified. Questionnaires were mailed during April–October 1999. The mean time from disclosure to questionnaire completion was 2.2 years. Two of the men identified were later excluded (one was deceased, and one was elderly and had hearing loss). From the remaining 67 eligible men, 44 questionnaires were returned by mail. An additional 15 (25%) of the men completed the questionnaire by telephone interview conducted by a research assistant from the University of Toronto. In total, 59 questionnaires were available for analysis—15 from the Women's College Hospital in Toronto and 44 from Creighton University.

Genetic-Counseling Services

Genetic counseling is offered at Women's College Hospital of the University of Toronto and at Creighton University as a clinical service and within a research program. Genetic counseling includes consultation with either a genetic counselor or an oncology nurse and with either a geneticist or an oncologist. All available management options for the consultand and his or her relatives are routinely discussed, including screening, prophylactic surgery, and chemoprevention. Genetic counselors and nurses are responsible for arrangement of referrals to other specialists, for consultation and screening. Men and women in these familial cancer programs are provided with a minimum of one pretest counseling session and one disclosure counseling session.

Procedures

All study procedures were approved by the Institutional Review Boards of Women's College Hospital in Toronto and of Creighton University. Eligible men received a letter explaining the study, an invitation to participate that was accompanied by a notice that they might be contacted by telephone, a consent form, and a questionnaire. If a man wished to participate, he returned the completed questionnaire to the investigators. Men who did not return their questionnaire were contacted by telephone and were asked to participate by telephone interview.

Questionnaire Design

The questionnaire consisted of 40 items assessing patients' motivations for seeking genetic services, information needs, screening practices, emotional reactions, access to services (i.e., health-care referrals), support resources, desire to take part in a support group, and overall experience in genetic counseling. The majority of questions were of the multiple-response type, with additional space for the respondents to explain their answer choices. Open-ended questions related to clients' opinions or emotional reactions. The questionnaire is based on a questionnaire designed for female carriers of mutations and described elsewhere (Metcalfe et al. 2000). These questionnaires are available on request.

Analysis

Data analysis was performed by SPSS statistical package 10.0. Responses were divided between unaffected men and those with a previous diagnosis of cancer. Education levels were divided into two groups: (1) high school graduation or less and (2) more than high school. Age was analyzed as a continuous variable and as a categorical variable. When it was analyzed as a categorical variable, subjects were divided into two groups:

those <50 years of age and those ⩾50 years of age. Either Pearson's χ^2 test or Fisher's exact test was used for nominal data, and Student's *t*-tests were used for comparison of continuous variables. The significance level was set at .05 (two sided).

Results

Study Subjects

Sixty-seven male carriers of *BRCA* mutations were eligible. The overall response rate was 88%, and 59 questionnaires were available for analysis. Fifteen respondents participated by telephone interview. Four men did not return the questionnaire (no contact was possible by telephone); three men were lost to follow-up, and one man refused to participate. The 59 respondents were from 31 different families. Forty-one men were carriers of *BRCA1* mutations, and 18 were carriers of *BRCA2* mutations. Twelve men (20%) had a previous diagnosis of cancer (table 1). All subjects were white and of either Ashkenazi Jewish or other European descent, except for one man of Pakistani origin. The mean age of the respondents was 53.8 years (range 26–83 years); 49% of men were age <50 years, and four men were age >75 years. More than one-half of the men had a mother diagnosed with either breast cancer or ovarian cancer. The majority (78%) of the men had one or more daughters; 25 men had one daughter, eight men had two daughters, 9 had three daughters, and 4 had four daughters. The mean age of the daughters was 26.2 years. Ten men (17%) had a daughter who had been diagnosed with either premenopausal breast cancer or ovarian cancer. The characteristics of mothers and daughters of the men are given in table 2. The majority (43/59) of respondents had some postsecondary education. Thirty-three men (56%) had received either a college or technical school diploma (12/59) or a university degree (21/59).

Table 1

Cancer Sites Reported among 12 Male Carriers of *BRCA1/2* Mutations

Cancer	No. of Sites Reported		Age(s) at Diagnosis (years)
	BRCA1 (n = 41)	*BRCA2* (n = 18)	
Prostate		1	76
Colon	2		65, 76
Breast	1	2	53, 58, 70
Testicular		1	44
Melanoma	1	1	36, 46
Basal cell	1	1	38, 50
Sarcoma	1		33
Overall	6 (15%)	6 (33%)	53.8

Table 2

Characteristics of Mothers and Daughters of Male Carriers of *BRCA1/2* Mutations

	BRCA1	*BRCA2*	Total
	No. of Subjects		
Mothers:			
Breast cancer	9	8	29%
Ovarian cancer	12		20%
Breast cancer and ovarian cancer	3		5%
Died because of either breast cancer or ovarian cancer	23	3	44%
Daughters:			
Breast cancer	4	4	14%
Ovarian cancer	1		2%
Breast cancer and ovarian cancer	1		2%
Died because of either breast cancer or ovarian cancer	1	1	3%
	Mean Age at Diagnosis (years)		
Mothers	45.3	52.6	
Daughters	33.7	38.2	

Reasons for Seeking Genetic Counseling

Approximately one-half of the men stated that the primary reason for seeking genetic counseling was concern for their families (14/59) or children (16/59), in particular for their daughters (fig. 1). Ten men indicated that their primary motivation for seeking genetic counseling was to learn about their own personal risk for cancer; 16 men cited this as a secondary reason. No significant associations were observed between motivations for testing and cancer status (P = .56), age (P = .27), education (P = .21), or daughter's cancer status (P = .12). Four men cited their primary reason for seeking genetic testing as being "my family's recommendation." When asked who had referred them for genetic counseling, the majority (49/59) of men responded that a family member had referred them. Forty-one men (70%) had a female relative (sister, cousin, mother, daughter, niece, or aunt) who had initiated the genetic-counseling process for the family; 11 men (19%) initiated the testing process themselves.

Support Needs

Genetic counselors were cited as the main sources of psychosocial support (14/59 cases), followed closely by doctors, spouses, and family members. Only two men (3%) felt that they needed more support than was received. The proportion of men who felt that they needed

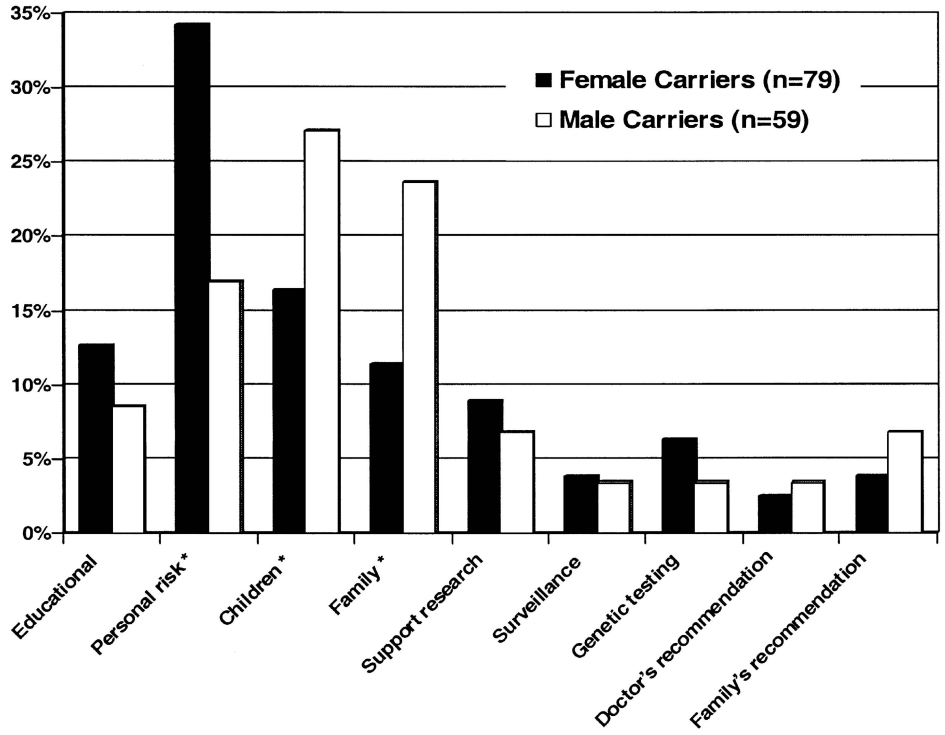

Figure 1 Primary reason for seeking genetic counseling, according to male and female carriers of *BRCA1/2* mutations. Black bars represent the percentage of respondents among female carriers of *BRCA1/2* mutations ($n = 79$); white bars represent the percentage of male carriers of *BRCA1/2* mutations ($n = 59$). Significant differences were observed between male and female respondents, for personal risk, children's risk, and family's risk as the primary motivations for seeking genetic counseling and testing. Asterisks (*) indicate that Pearson's χ^2 $P = .015$ between three categories and that Fisher's exact $P = .005$ between two categories (the categories of "children" and "family" were combined).

more support showed no significant differences within any category—cancer status ($P = .46$), age ($P = .92$), education ($P = .66$), whether they had daughters ($P = .62$), and daughter's cancer status ($P = .86$). Eight men felt that they needed an appointment for counseling after the disclosure of test results. Three men requested referrals to other health-care professionals who were knowledgeable about breast cancer genetics. Fifty-three percent (29/55) felt that a support group was necessary for both male and female carriers of *BRCA* mutations, and 63% (33/52) of the men indicated that a support group was particularly necessary for female carriers of mutations. Twenty-nine percent (16/55) expressed interest in participating in a support group specifically for male carriers of mutations. Topics recommended for discussion included how to communicate information to family members, the general perception that breast cancer is not a man's disease, and advice on lifestyle modifications designed to reduce cancer risk.

Risk Perception

The majority (36/45) of unaffected men (i.e., those with no previous diagnosis of cancer) believed that they were at increased risk of development of cancer. Two men believed that they were at significantly increased risk of development of cancers related to their occupation (firefighting and farming). The nine men who believed that they were not at increased risk of development of cancer were all unaffected. Two men with a previous history of cancer stated that they were at increased risk of development of all types of cancer. More than one-half (52%) of the respondents indicated that they had an increased susceptibility to prostate cancer. One-third of male carriers of *BRCA2* mutations specified increased susceptibility to breast cancer, whereas 22% of male carriers of *BRCA1* mutations specified increased susceptibility to colorectal cancer. The organ sites specified by the unaffected men are shown in figure 2. No significant differences in risk perception were observed within any category—cancer status ($P = .18$), age ($P = .76$), education ($P = .23$), whether the men had daughters ($P = .32$), and daughter's cancer status ($P = .44$). However, 97% (29/30) of men who had a mother diagnosed with either breast cancer or ovarian cancer stated that they were themselves at increased risk of development of cancer, compared with 70% of men

without an affected mother ($P = .007$). Similarly, 96% (23/24) of men who had had a mother die of either breast cancer or ovarian cancer stated that they were at increased risk of development of cancer. Fifty-five percent of the men indicated that they had intrusive thoughts about their increased risk of development of cancer, including nearly one-half of the unaffected men (table 3).

Surveillance Practices

Most (41/58) male carriers of mutations believed that they were receiving adequate care for the prevention of prostate cancer. Nine men (16%) felt that they had not received enough information about cancer surveillance. Six men remarked on their own delay in implementing a consistent surveillance regimen. One man from the United States felt that, because of health-insurance coverage limitations, he was not receiving adequate medical care. The majority of male carriers of mutations thought that genetic counseling provided them with adequate information about screening (85%) and about risk of development of breast and/or ovarian cancer in their female relatives (88%). Only 43% of all unaffected men reported that they had altered their cancer-surveillance programs after learning of the results of screening. Alteration of cancer-screening practices in unaffected men was not dependent on age ($P = .25$) or education ($P = .11$). The screening practices of the men prior to receipt of genetic-test results was not known.

Table 3

Risk Perception among 47 Unaffected Male Carriers of *BRCA1/2* Mutations

Characteristic	No. of Subjects		Total
	BRCA1	*BRCA2*	
Risk perception:			
Not at risk	7	2	20%
Elevated risk	26	10	80%
Intrusive thoughts:			
Absent	17	7	52%
Present:	17	5	48%
Daily	1	1	4%
Regularly	2	1	7%
Sometimes or randomly	3		7%
Monthly	1	1	4%
Annually ("on my birthday")	1		2%
Rarely	6	1	15%
Not sure	1	1	4%

Adherence to recommendations following prostate cancer screening with prostate-specific antigen (PSA) was reported by approximately one-half of the men. The majority (78%) of the men who underwent PSA testing reported being screened on an annual basis. Digital rectal examination (DRE) was reported by a similar number

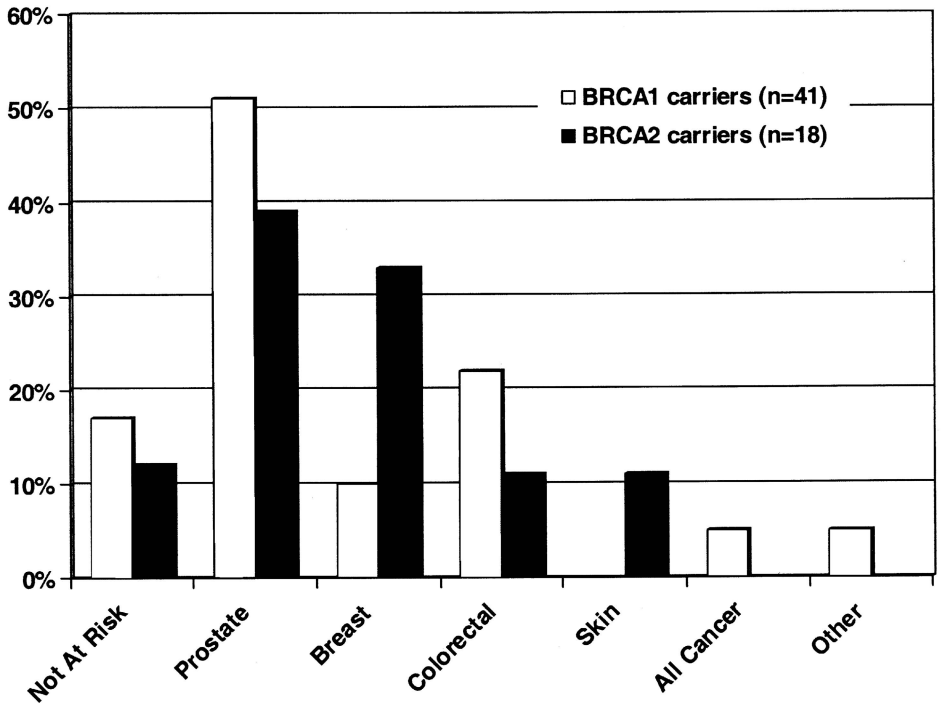

Figure 2 Percentage of male carriers of *BRCA1/2* mutations who reported risk of development of specific cancers (cumulative responses). Black bars represent the percentage of male carriers of *BRCA2* mutations ($n = 18$); white bars represent the percentage of carriers of *BRCA1* mutations ($n = 41$).

(59%) of carriers of mutations. Men ⩾50 years old were more likely to have undergone PSA screening than were men <50 years old ($P < .0001$), whereas this age effect was not significant for DRE ($P = .084$). Six men indicated that they had experienced difficulties in setting up a prostate cancer–surveillance regimen with their physician: "My urologist says PSA doesn't tell you anything"; "I was told it was not necessary for men under 50"; "My doctor was reluctant because of my (older) age." Table 4 summarizes the surveillance practices of the men with no previous cancer diagnosis. Respondents were requested to provide the date of their last visit for PSA and DRE. The mean date of last PSA was September 1998 (range 1997–2000), whereas the mean date of last DRE was April 1998 (range 1994–2000). When compared with the mean date of questionnaire completion (December 1999 [range June 1999–June 2000]), these dates suggest that male carriers of mutations are indeed attempting to maintain annual PSA screening for prostate cancer, although DRE appears to be scheduled slightly less regularly.

All three men with a previous breast cancer diagnosis reported having had clinical breast examinations and having performed regular breast self-examinations. Two affected men performed self-examinations on a daily basis (i.e., "every time I shower"). In contrast, only seven men (15%) with no previous diagnosis of cancer performed breast self-examinations, including four carriers (25%) of *BRCA2* mutations (table 4). Thirteen men (22%) specified that they either were involved in other cancer-screening programs, such as those for colorectal cancer or skin cancer, or had an annual physical examination with their general practitioner.

Table 4

Surveillance Practices among 47 Unaffected Male Carriers of *BRCA1/2* Mutations

Surveillance Practice	No. of Subjects		Total
	BRCA1	*BRCA2*	
Since learning of result:			
Not changed	16	9	53%
Changed	18	2	43%
To be arranged	1	1	4%
Frequency:			
PSA:			
Ever	19	6	56%
Semiannually	1		2%
Annually	11	6	36%
Biannually	3		6%
To be arranged	3	1	9%
DRE:			
Ever	19	6	53%
Semiannually	2		4%
Annually	9	4	28%
Biannually	2	1	6%
To be arranged	3	1	9%
Clinical breast examination			
Ever	1	4	11%
Semiannually	1	1	4%
Annually		3	6%
Breast self-examination:			
Ever	3	4	15%
Rarely	1	1	4%
Monthly	1	1	4%
Twice monthly		2	4%
Weekly	1		2%
Colonoscopy, ever	3		6%
Fecal occult blood, ever	2		4%
Skin examination, annually	1	1	4%
Physical examination, annually	2		4%

Emotional Responses

The men expressed a variety of reactions to their positive result (table 5), including relief of anxiety when uncertainty about risk status was removed (2/59), concerns about personal cancer risk (5/59), disappointment (6/59), and feelings of sadness (2/59) or fear (3/59) (table 5). Almost one-quarter (14/59) of the men felt that the genetic testing confirmed what they had always suspected. For nine men with daughters, the concern was immediately transferred: "This means my daughter needs to be tested." Two men with daughters experienced guilt as a consequence of learning their carrier status. One man described how he felt a renewed sympathy and understanding for his sisters who had tested positive, and he commented on how his risk was minimal in comparison.

Discussions with Relatives

The great majority (56/59) of men discussed their genetic-test result with a family member. Five men indicated that they would have liked assistance in sharing with their families the information about genetic testing. Only two men wished that their families had involved them more in the initial genetic-testing process, and 12 (21%) respondents stated that they wished that their families would be more involved in the genetic-testing process. The majority (52/59) of men had been included in past family discussions of risk of development of breast and/or ovarian cancer. Less than half (28/59) of the men participated in conversations about prophylactic surgery for reduction of risk of development of breast and/or ovarian cancer. Ten men stated that their family relationships had changed since they had received the results of their *BRCA* test. The majority (7/10) described how their family relationships had been strengthened by this information: "brought family closer," "heightened awareness and concern for family members," "helped communication in family, greater understanding," and "I am more considerate of daughter's choice to have preventive surgery." One man stated that, as a result of

Table 5

Descriptions of Initial Feelings When Subjects First Received *BRCA1/2* Test Result

Theme	Paraphrased Response	No. of Responses	No. of Men With	
			Cancer	A Daughter/A Daughter with Cancer[a]
Family	Concern for daughters or children	9	0	9/1
	Guilt	2	0	2/1
	Concern for sisters	1	0	1/0
Personal risk	Concern for self	5	0	2/0
General concern		3	0	1/0
Neutral	Not overly concerned	7	0	7/2
Not surprised	Confirmed what I'd always suspected	14	3	10/5
Acceptance		5	1	2/0
Relief of anxiety	Relief for myself and my family	2	2	2/0
Disappointment		6	0	5/0
Unexpected		1	1	0/0
Anxiety	Nervousness, fear, stress	3	1	3/0
Sadness		2	1	1/0
Shock	Sense of alarm	3	0	3/0
Existential	Sense of mortality	2	0	1/0
Can't remember		1	1	1/1

[a] Either breast cancer or ovarian cancer.

his genetic-test result, he is less likely to have additional children.

Satisfaction with Genetic Counseling

Men were asked to rate their overall satisfaction with the genetic-counseling process, on a 5-point scale (1 = extremely dissatisfied; 5 = extremely satisfied). The mean response for satisfaction was 4.2. All but two respondents indicated that they were at least "satisfied" with the genetic-counseling process. Four men (7%) stated that information was missing during their genetic-counseling appointment at the time of either DNA testing or disclosure of results. The topics specified were risk of development of colorectal cancer and the health-care professionals' limited awareness of male breast cancer. One man remarked that the genetic counselor should have included a discussion of both how general practitioners or other physicians may not be aware that men may get breast cancer and how the male breast is often neglected during physical examination. This man had an unsatisfactory experience with his general practitioner after he had detected a lump in his breast by self-examination. Men >50 years old were more likely than younger men to say that information was missing from the genetic-counseling session ($P = .045$). There were no differences in the need for additional information based on cancer status ($P = .13$), education ($P = .97$), whether the men had daughters ($P = .26$), or the daughter's cancer status ($P = .11$).

Two men from the United States expressed concerns about insurance implications, both for themselves and for their children. One man was thankful but thought that the counseling itself was excessive. Two men (3%) were not sure whether they would recommend genetic testing to other men in their situation, and two men (3%) would not recommend testing. For the 54 men who would recommend predictive testing to other men in their situation, there were two main reasons or themes for the recommendation—namely, awareness and family ("to be aware and take necessary precautions," "knowledge is power," "important for men with daughters," and "to inform offspring of their situation").

Comparison with Female Carriers of BRCA *Mutations*

We compared data on male respondents ($n = 59$) with those on female carriers of *BRCA* mutations ($n = 79$); the study on female carriers of mutations has been described in detail elsewhere (Metcalfe et al. 2000). Male carriers of mutations were less likely to have had a previous diagnosis of cancer than were female carriers of mutations ($P < .0001$). Men were more likely than women to be referred by a family member for genetic counseling ($P < .0001$). Figure 1 depicts the primary reasons for seeking genetic counseling, as indicated by male and female respondents. There was a statistical difference observed between male and female respondents in the three most frequently cited motivations ($P = .015$). Table 6 compares the responses of male and female carriers of mutations, with regard to specific items assessed during genetic counseling.

Discussion

To date, attention has focused on women in families with *BRCA* mutations, because they are believed to be at greater risk of development of cancer. Our study inves-

Table 6

Comparison of 59 Male and 79 Female Carriers of *BRCA1/2* Mutations

Item Assessed in Genetic Counseling	Men	Women	*P*
More information needed	7%	21%	.032
More support needed	3%	19%	<.0001
Support group needed	53%	68%	.05
Support group for females	69%	68%	.38
Personal interest in support group	23%	34%	.038
More information on surveillance needed	16%	18%	.2
Surveillance practices changed	44%	58%	.11
Surveillance practices changed (among unaffected)	43%	81%	.002
Mammography or PSA on a regular basis	59%	70%	.76
Receiving adequate care in cancer prevention	77%	81%	.77
Result was shared with family	95%	95%	1.0
Help needed in sharing information with relatives	9%	15%	.3
Wished family was more involved	21%	31%	.26
Feelings or psychological state changed	29%	53%	.007
Family or personal relationships changed	18%	33%	.04
Overall satisfaction (5-point scale)	4.19	4.02	.32[b]

[a] For comparison (Pearson's χ^2).
[b] By *t*-test.

tigated the perspectives of men who underwent genetic counseling and who were found to be carriers of a germline *BRCA1/2* mutation. These men were motivated to undergo genetic testing and therefore may be better able to assimilate genetic-risk information than are those who declined testing. Our study describes the men's reactions ⩾6 mo after receipt of the results of genetic testing, which allows time for the men to adapt to the news of their positive result.

Dudok deWit et al. (1996) reported on the psychological findings on four men from families with breast cancer who had undergone predictive testing and who had reported difficulties with the genetic-counseling process. In particular, the men exhibited avoidance behaviors and had a tendency to either miss appointments or withdraw from testing altogether. McAllister et al. (1998) concluded that men from families with breast cancer are affected emotionally by their female relatives' diagnoses of breast cancer and that their level of distress is associated with the number of daughters.

The reasons that the male carriers of mutations commonly cited for seeking genetic counseling were similar to those that other studies have reported for women (Lerman et al. 1995; Bleiker et al. 1997; Lynch et al. 1997). However, men's motivations appear to differ in priority. More than one-half of the male carriers of *BRCA* mutations were tested for the sake of their children or family. In contrast, female carriers of mutations reported that their primary reason for seeking counseling and testing was to learn about their own risk (Metcalfe et al. 2000).

Compared with men, women were more likely to report that their family or personal relationships had changed as a consequence of genetic testing and to feel that a support group is necessary for carriers of mutations; women were also more likely to want to take part in a support group. Consistent with the gender differences in interest in support groups (Fobair 1997) is the finding that men who were interested in support groups had several motivations, including a desire for more information (e.g., strategies for reduction of risk of development of cancer), because they either felt misunderstood at home or experienced a sense of loss. Other topics specified by male carriers of *BRCA* mutations who were interested in support groups included the general perception that breast cancer is not a man's disease and strategies to communicate risk information to family members.

Communication with male relatives in families with *BRCA* mutations has been described by female relatives as particularly difficult because the discovery that men can be predisposed to a "female" disease is counterintuitive (Green et al. 1997). This perception is not limited to family members; several respondents experienced difficulties with health-care practitioners in this regard. Familial cancer clinics may facilitate communication of risk-management information to general practitioners who care for male carriers of *BRCA* mutations. Female family members are not necessarily well-informed of the risks to males, and a number of women described frustration in their attempts to explain the situation to their brothers (Green et al. 1997). Contrary to previous observations (McAllister et al. 1998), the majority of our respondents participated in family discussions of breast and/or ovarian cancer. However, fewer than one-half of the men participated in family conversations about prophylactic surgery. This may be explained by the inherently gender-specific issues and risk-management con-

siderations; however, the number of women in each family who had considered prophylactic surgery was not available.

The majority of men were referred by a family member. It is of concern that four men reported that their primary reason for seeking genetic testing was "my family's recommendation." The response may reflect some degree of coercion. It is important for practitioners in familial cancer clinics to be aware of potential pressures—either for or against testing—that may exist within families and to help promote autonomous decisions.

In 1997, early guidelines for carriers of *BRCA1/2* mutations stated that there was insufficient evidence to recommend or discourage prostate cancer screening (Burke et al. 1997). Since then, several studies have reported that male carriers of *BRCA1/2* mutations are at a significantly increased risk of development of several types of cancer—in particular, prostate cancer. Prostate cancer risk is the most consistent finding for male carriers of *BRCA1/2* mutations in families with cancer, and the relative-risk range is 3.33–7.33, or an estimated 16%–35%, to age 65–70 years (Ford et al. 1994; Struewing et al. 1997; Breast Cancer Linkage Consortium 1999; Moslehi et al. 2000). However, several studies of the common *BRCA* mutations in unselected Ashkenazi Jewish men with prostate cancer have failed to confirm an increased risk (Lehrer et al. 1998; Nastiuk et al. 1999; Vazina et al. 2000). Germline *BRCA1* mutations have also been associated with elevated risks of colorectal, pancreatic, and male breast cancers (Ford et al. 1994; Borg et al. 2000; Moslehi et al. 2000). Although the risk of male breast cancer is increased for carriers of *BRCA1* mutations, the risk is greater for male carriers of *BRCA2* mutations and is estimated as being <10% until age 70 years (Easton et al. 1997). There is also an increased risk of development of pancreatic, stomach, bile-duct, and gall-bladder cancers and of cutaneous malignant melanoma, in both males and females who harbor *BRCA2* mutations (Breast Cancer Linkage Consortium 1999). Among the 59 male carriers of *BRCA* mutations, the cancers were breast (3 cases), colon (2), cutaneous malignant melanoma (2), basal cell carcinoma (2), prostate (1), testicular (1), and sarcoma (unspecified type) (1).

The majority of men in this study harbor fears of development of cancer. These fears were strongly associated either with diagnosis of breast or ovarian cancer in the mother or with the mother's death. Fifty-five percent of male carriers of mutations, including almost one-half of those with no previous cancer diagnosis, suffered from intrusive thoughts about their risk of development of cancer. The major sites of susceptibility specified were prostate, breast, colon, and skin. Pancreatic cancer was not included among these sites. With the exception of pancreatic cancer, these sites are consistent with the information presented to male carriers of mutations during genetic counseling at the University of Toronto and at Creighton University.

Consistent with previous findings for female carriers of *BRCA* mutations (Lerman et al. 2000; Metcalfe et al. 2000), more than one-half of male carriers of mutations did not adhere to the screening guidelines recommended after disclosure of genetic-test results. More attention is needed to promote cancer-screening recommendations—particularly for prostate cancer and breast cancer—to male carriers of *BRCA* mutations who are undergoing genetic counseling.

The most consistent recommendation given to male carriers of *BRCA* mutations at the University of Toronto and Creighton University clinics pertains to prostate cancer surveillance, with annual PSA and DRE beginning at age 40 years. There is no standard recommendation for breast cancer screening in male carriers of *BRCA* mutations, although men are advised to seek medical evaluation for any breast mass or change (Burke et al. 1997). At the University of Toronto and Creighton University familial cancer clinics, breast cancer screening, consisting of annual or semiannual clinical breast examinations and monthly breast self-examinations, is recommended routinely for male carriers of *BRCA2* mutations and, recently, has been recommended also for male carriers of *BRCA1* mutations. Nonetheless, only 25% of male carriers of *BRCA2* mutations have annual clinical breast examinations or perform breast self-examinations. Screening mammography is not usually recommended for males at risk. Tamoxifen is used as an adjuvant treatment for male breast cancer (as it is for female breast cancer), on the basis of tumor characteristics; but tamoxifen has not yet been proposed as a chemopreventive agent for males, and its use is not routinely discussed. Similarly, surgical prevention of breast cancer (i.e., prophylactic mastectomy) is not offered, because, at the present time, there are no data to support this type of risk reduction in males. Clinical skin examinations for the early detection of melanoma are included in the discussion of cancer surveillance for families with *BRCA2* mutations. Although the data are inconsistent, families with *BRCA1* mutations are informed of the possible risks for colorectal cancer (Ford et al. 1994), and screening by colonoscopy, at intervals of 3–5 years, is recommended.

In general, the information provided to male carriers of *BRCA* mutations during genetic counseling was felt to be sufficient. Men may have intrusive thoughts regarding unresolved grief about past and future losses—and guilt about passing on a potentially lethal gene to their children. Greater attention may be needed for male carriers of mutations >50 years old, particularly with regard to surveillance for prostate cancer and

for breast cancer. Other issues of particular focus would include insurance considerations and an increase in awareness by their primary-care physicians. Further work is necessary to explore the reasons for noncompliance in recommended surveillance for prostate cancer in male carriers of *BRCA* mutations.

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