

Health Insurance and Discrimination Concerns and *BRCA1/2* Testing in a Clinic Population¹

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Abstract

The discovery of the breast cancer genes *BRCA1* and *BRCA2* has afforded those who seek breast and ovarian cancer risk counseling the option of genetic testing. Concerns about cost, confidentiality, and the potential for discrimination, however, may prevent some women from pursuing genetic testing. To determine the impact of these concerns on BRCA testing, we studied a cohort of 384 patients presenting *de novo* to a Breast and Ovarian Cancer Risk Evaluation Program, between January 6, 1997 and March 13, 2000. Of the 184 individuals who were themselves primary candidates for testing, 106 (58%) underwent *BRCA1/2* sequencing. Of the 78 eligible patients who declined testing, 48 cited concerns about cost and insurance discrimination as their reason. On the basis of the number of positive results ascertained in the tested group, we estimate that approximately half of patients declining testing because of insurance coverage concerns would be positive for a BRCA mutation. We were unable to document any experiences of test result-based discrimination, although there were other negative insurance-related experiences. We conclude that in a high prior-risk clinic population, approximately one-quarter of patients eligible for BRCA testing may decline because of concerns about cost, confidentiality, and discrimination. Our research provides evidence that these fears may be discrepant with the actual experiences of patients in high-risk clinics.

Introduction

In most areas of medical research, a lapse of time exists between the acquisition of information and its integration

into general public knowledge and widespread practice. Reasons for this delay are multifactorial and complex. They can include interactions with regulatory agencies, assimilation of knowledge by professionals and the public, proximity to services, and acceptance of the consequences of seeking services (1, 2). Low-income, minority, and uninsured women may find specific socioeconomic barriers to obtaining health-related data and education (3, 4). In the current age of flourishing genetic research and information, new patterns of factors might contribute to the gap between the technologies available and those using them in a clinical setting. Important among these factors are ethical and legal concerns about the confidentiality of genetic information and its manipulation, the potential for discrimination, including loss of health and other insurance benefits, and the psychological consequences of genetic information.

Breast cancer research is not excluded from the impact of these concerns about genetic information. Each year in the United States, >180,000 new cases of breast cancer and 44,000 deaths from breast cancer are reported (5). Women who have a mutation in either a *BRCA1* or *BRCA2* gene are estimated to have a 50–80% risk of developing breast cancer in their lifetime (6–8). Given estimates that these two genes account for 10% of all breast cancer cases (9–13), and that 1 in 400 women carries an alteration on one of these two genes (14), >100,000 women/year in the United States alone are potential candidates for *BRCA1/BRCA2* analysis.

Many studies have analyzed aspects of cancer genetic services, such as recruitment (15), effectiveness of counseling strategies (16), and patient characteristics associated with seeking testing (9, 17–20). This research revealed a number of important findings: (a) the majority of women who undergo counseling and testing are interested not only in their personal risk for developing cancer but also in the cancer risk for their immediate family members, especially their children (9, 17–19, 21); (b) the public is generally uninformed about basic principles of genetics and confidentiality issues, thereby complicating processes of informed consent for genetic tests (22, 23); and (c) fear of discrimination is one of the most common reasons given for declining *BRCA1/2* testing, even overshadowing fear of a positive result (9, 17, 19, 23, 24). In a study examining factors associated with decisions about *BRCA1/2* testing in a clinical population, 47% of those who declined testing responded that fear of health insurance discrimination was a very important factor in their decision (25). Most of these studies have focused on subjects from research populations who were not responsible for payment for their testing or clinical services. It is not clear whether, in the move from research to practice, these concerns are more or less prevalent.

The first aim of the present report was to determine whether concerns about cost, confidentiality, and the possibility of insurance discrimination for patients and their families are major barriers to pursuing genetic testing for hereditary breast

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and ovarian cancer in a clinical setting, particularly for PTCs⁴ (3). These concerns are highly publicized in the lay media and thus deserve further attention (26–28). Second, we sought to understand why and how these concerns affect the uptake of BRCA genetic testing by eligible clinic patients. More specifically, we wanted to estimate the number of PTCs who would not learn of their mutation status because of their cost, confidentiality, and discrimination concerns, thereby potentially affecting their cancer prevention and screening options. Finally, we wanted to report the experiences of patients as they interacted with insurers about BRCA genetic services. To fulfill these goals, we performed two studies. In Study 1, we answered our first and second aims by examining attitudes toward and preferences about BRCA genetic testing with information obtained through genetic counseling sessions and chart reviews in women who visited a high-risk breast cancer clinic. We then conducted follow-up telephone interviews with a subset of the above population (Study 2) to explore in more detail fears about discrimination, confidentiality, and cost and to investigate patients' experiences with their insurance carriers. Our subjects were patients presenting for clinical risk assessment services to a specialized breast and ovarian cancer risk evaluation clinic. We hypothesized that concerns about insurance discrimination, cost of testing, and confidentiality of genetic information are significant obstacles to uptake of BRCA1/2 genetic testing services and genetic information.

Materials and Methods

Study 1: Patients' Attitudes toward and Preferences about BRCA Genetic Testing from Genetic Counseling Session and Chart Review

Eligibility Criteria and Clinical Evaluation. The cohort for this portion of the study consisted of 384 patients seen for their initial risk evaluation between January 6, 1997 and March 13, 2000 in one of two Breast and Ovarian Cancer Risk Evaluation clinics: at the University of Michigan (Ann Arbor, MI) or at Spectrum Health System (Grand Rapids, MI). From this group, PTCs were ascertained (Fig. 1A). A patient was considered a PTC and therefore eligible for this portion of the study if they met one or more of the following criteria: a history of breast and/or ovarian cancer in one or more close relatives or a known BRCA cancer gene mutation in their family and no other available person in their family that was a better candidate for initial BRCA1/2 testing in that family. Patients were excluded from the study if they did not have enough information in their files for the analysis of this portion of the study ($n = 5$) or underwent a nontraditional consult ($n = 33$; *e.g.*, a discussion of results from testing performed at another center). All eligible and consenting PTCs ($n = 184$) were competent, English-speaking adults. PTCs had a family history sufficiently suggestive of a hereditary breast/ovarian cancer syndrome as determined by the evaluator (most patients in this cohort were evaluated before the publication of the National Cancer Center Network guidelines; Ref. 29). The patients underwent a comprehensive evaluation and clinical consultation that included risk assessment, education in cancer genetics, discussion of BRCA1/2 testing, physical exam, and the formulation of a risk management plan.

The subjects who underwent clinical genetic testing signed informed consent, and Myriad Genetics Laboratories (Salt Lake City, UT) performed the BRCA 1/2 analysis. Patients were

charged ~\$225 for the initial cancer risk assessment consultation and, if applicable, were required to pay Myriad Genetics directly for any testing that they pursued. At the time of the study, Myriad Genetics offered three types of DNA analysis of the BRCA genes. Analysis of a single mutation site cost patients \$395. Patients were billed \$450 for analysis of the three BRCA mutations (185delAG, 5382 insC, and 6174delT) commonly found in individuals of Ashkenazi Jewish descent. The most complex DNA analysis consisted of full sequencing of the *BRCA1* and *BRCA2* genes, which cost \$2,400. Patients had the option of involving their insurance provider to procure payment for the initial consultation and/or for testing.

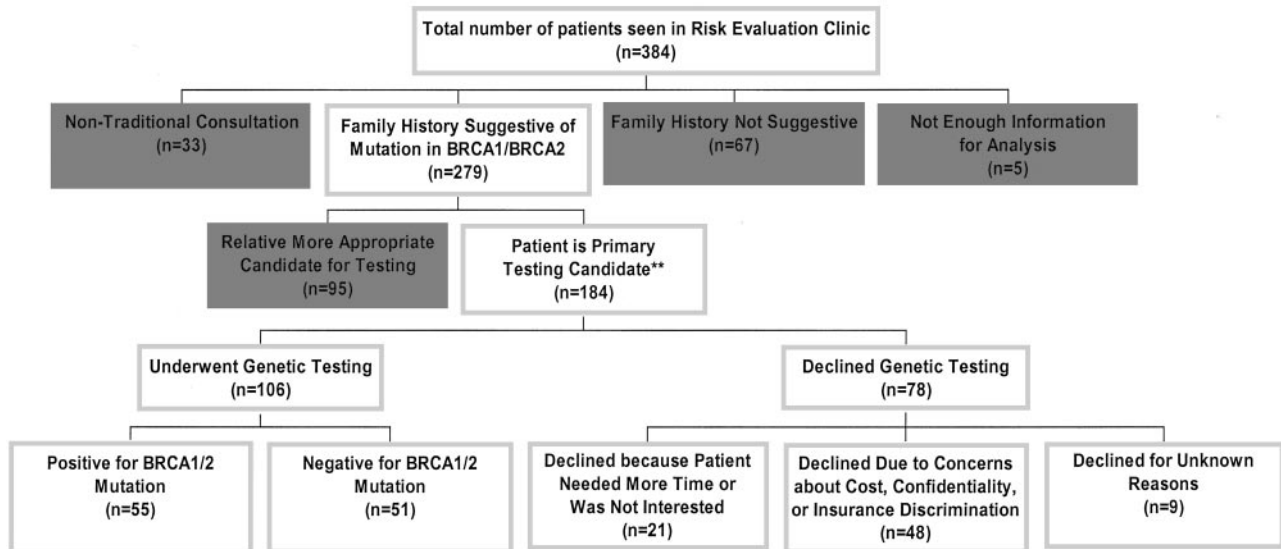
Data Collection. Data collection from patient chart review was retrospective for subjects seen before June 14, 1999 and prospective for subjects seen on or after this date. Data obtained from chart reviews had been documented during the counseling and assessment comprising routine care in both the Spectrum Health and University of Michigan clinics. It is standard practice for genetic counselors and physicians to discuss issues of cost, confidentiality, and discrimination as they facilitate patients' decision making regarding BRCA testing. Patients who did mention cost, confidentiality, and insurance concerns often volunteered the information during discussions of payment for clinical services and/or spontaneously during the counseling session. In both the Grand Rapids and Ann Arbor clinics, patients eligible for BRCA analysis were specifically asked to share their reasoning for or against testing during a discussion about their testing preferences. The genetic counselors then recorded their responses (including the rank of the reason if more than one is listed) in chart notes and reiterated them in a summary letter sent to each patient after her or his visit. Patients were explicitly invited to contact the clinic to rebut any of the contents of the summary letter.

Since 1997, there have been three genetic counselors between the two clinics (one at Spectrum Health and two at the University of Michigan); the same oncologist with experience in cancer genetics (S. D. M.) has reviewed all 384 patients in our cohort. One researcher (E. A. P.) was responsible for reviewing the chart and patient letters and recording the reasoning behind the patient's decision making. The researcher also collected the following information on each of the eligible subjects: name, age, sex, date of initial visit, insurance provider, race, education level, family income, cancer diagnosis (invasive or noninvasive), diagnosis of nonmalignant breast disease (*e.g.*, lobular carcinoma *in situ* and atypical hyperplasia), age at diagnosis, and testing results (Table 1). For some of the analyses in this report, PTCs were divided into groups based on disease status. Patients with either a diagnosis of cancer and/or a nonmalignant breast disease were assigned one group; patients with no diagnosis were assigned to another group. Patients signed a separate informed consent for the collection of medical history, socioeconomic, and insurance information.

Because of the open-ended nature in which patient decision making was explored in the genetic counseling sessions at both clinics, patients' concerns about cost, confidentiality, and discrimination were not easily separated. Indeed, it is often extremely difficult to discern among these issues as they affect patients in a clinical setting, *e.g.*, a patient may wish to pay out of pocket for BRCA testing but may not perceive herself to have sufficient resources and may thereby rely on her insurance provider to cover the payment. Therefore, she would be exposing her testing decision and possibly her results to a third party, the insurance carrier. In this example, the cost of testing is

⁴ The abbreviation used is: PTC, primary testing candidate.

A Patients Seen in High Risk Clinic January 6, 1997 – March 13, 2000*



*Data collected from genetic counseling session as well as chart review.

**A patient was deemed to be the primary testing candidate if they had a diagnosis of cancer and/or there was no other person in their family with a cancer diagnosis that was willing or able to be the primary testing candidate for that family. Thus the category "Relative more appropriate candidate" encompasses many patients whose family history is consistent with a mutation in a cancer susceptibility gene, but who themselves are not the most informative candidate for initial BRCA1/2 testing.

B Patient Recruitment for Post-Counseling Telephone Survey

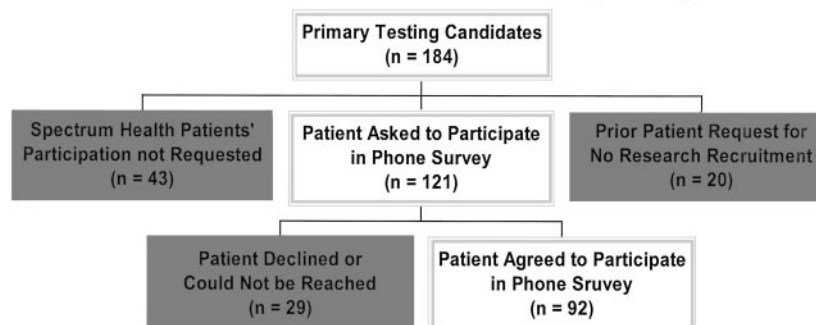


Fig. 1. A and B, patients seen in high-risk clinic from January 6, 1997 through March 13, 2000.

intertwined with both confidentiality and potential future insurance discrimination.

Analysis. We compared those who "pursued testing" to those who "declined testing" and those who "pursued testing" to a subgroup of patients who "declined because of insurance/discrimination concerns." The subgroup of "declined because of insurance/discrimination concerns" was defined as those PTCs who considered fear of insurance discrimination, loss of confidentiality, and/or monetary concerns as their primary reason for not pursuing BRCA1/2 testing. Comparison of mean age between groups was performed using a pooled *t* test. Comparison of sex, education, and income levels between groups was analyzed using a χ^2 test. χ^2 tests were also performed to determine the effect of disease status on both testing

decisions and mutation status. For comparisons between those who did and did not have a diagnosis of cancer or related breast disease, we had 92% power to detect a difference of 0.2 with an $\alpha = 0.05$. For comparisons between those who tested positive or negative for a mutation in one or both of the *BRCA* genes, we had 87% power to detect a difference of 0.3 at $\alpha = 0.05$.

Study 2: Follow-up Study of Insurance and Confidentiality Concerns

Eligibility Criteria. Of the 184 PTCs who met our inclusion criteria for Study 1, 121 (67%) University of Michigan clinic patients were asked to participate in the follow-up study of insurance and confidentiality concerns (Fig. 1B). Twenty eli-

Table 1 Subject characteristics for each decision group

	All primary testing candidates	Pursued testing	Declined testing	Subgroup declined because of concerns
Age (mean)	44.7	45.4	43.6	44.1
Race ^a				
White	180	105	75	46
Non-White	4	1	3	2
Sex				
Females	171	94	77	47
Males	13	12	1	1
Highest education				
≤High school	21	11	10	7
>High school	64	35	29	18
>College	68	39	29	17
No info given	31	21	10	6
Family income				
<\$60,000	48	23 ^b	25	19 ^b
≥\$60,000	90	55 ^b	35	18 ^b
No info given	46	28	18	11

^a Differences by race were not assessed because of small numbers in non-White cells.

^b $P < 0.05$ for differences between pursued and declined because of concerns groups (χ^2 testing).

gible subjects who requested previously not to be involved in further research were not asked to participate, and the 43 patients at Spectrum Health were not included for logistical reasons. The 121 potential subjects were sent a letter of introduction to the study followed by a telephone solicitation. On the subject's verbal consent, the telephone interview proceeded. Of the 121 patients approached for this portion of the study, 92 (76%) completed the telephone interview, 21 (17%) could not be reached for participation, 5 (4%) declined participation, and 3 (2%) were deceased.

Data Collection. The PTCs who agreed to participate in the telephone survey were interviewed 3–29 months after their initial visit in the clinic. The survey instrument included three types of questions. The interviewer first asked subjects specific close-ended questions about payment for BRCA1/2 testing and related genetic services, *e.g.*, subjects responded with a “yes” or “no” to questions, such as, “Did your insurer cover the cost of the consultation? Was there any part of the consultation that the insurer did not cover?” Participants were then asked open-ended questions about their expectations for insurance coverage, actual insurance coverage (if they sought coverage), and other experiences with insurers. Finally, all subjects, whether they pursued or declined testing, were asked to rate on a Likert scale of one (not important) to five (very important) the importance of three testing related issues: (a) insurance payment for testing; (b) keeping BRCA testing results confidential from insurers; and (c) keeping BRCA testing results confidential from family members. Subjects who added comments about discrimination, cost, or confidentiality during this portion of the survey had these comments noted by the interviewer.

One researcher (E. A. P.) conducted all of the chart reviews, telephone surveys, and coding of data. This researcher categorized subject responses to open-ended questions about insurance expectations and experiences. These categories included such expectations as insurance reimbursement for all medically appropriate services, reimbursement for only the consult portion of BRCA genetic services, and no reimbursement at all. The researcher also categorized subjects' responses about their experiences with obtaining reimbursement for

BRCA genetic services. These responses included experiences of having difficulties in procuring coverage for BRCA-related screening procedures, expending effort to obtain the maximum reimbursement for genetic testing, and no problems related to coverage of BRCA services.

Analysis. Because the distribution of responses about importance was, usually, biphasic (Fig. 2), we used χ^2 tests of dichotomized importance responses in our analysis. A factor was considered “unimportant” if the person rated it three or less on the five-point Likert scale and “important” if it was rated four or five. We compared the importance responses by income ($\geq \$60,000$ and $< \$60,000$), disease status, and by the decision to pursue or not pursue BRCA testing. Unpaired *t* tests were used where appropriate to compare mean responses and confirm the χ^2 analysis. For comparisons between those who did and did not involve their insurance provider in payment for BRCA1/2 testing, we had 87% power to detect a difference of 0.2 with an $\alpha = 0.05$. For comparisons between those who did and did not encounter difficulties obtaining insurance coverage for BRCA1/2 services, we had 84% power to detect a difference of 0.3 at $\alpha = 0.10$. For comparisons between those who did and did not have a diagnosis of cancer or related breast disease, we had 82% power to detect a difference of 0.3 at $\alpha = 0.05$.

Results

Testing Decisions. Of 184 PTCs, 106 (58%) chose to undergo BRCA1/2 genetic testing (Fig. 3). The remaining 78 (42% of PTCs) declined genetic testing. Chart reviews revealed that, of the 78 patients who declined testing, 48 (62%) declined because of concerns about insurance discrimination, cost, and/or confidentiality, 21 (27% PTCs) declined testing because they needed more time to decide or were not interested for other reasons, and the final 9 (12%) declined testing for unknown reasons. Of the entire PTC population, 26% (48 of 184) declined BRCA1/2 testing because of concerns about cost, confidentiality, and/or insurance discrimination.

Prevalence of Disease and Mutations. We hypothesized that there would be a larger proportion of cancer patients among the pursuers of testing, because disclosure of the patient's disease status to insurance companies would have already occurred, thus potentially diminishing the “discriminative” potential of a positive mutation. We found, however, that approximately half of the patients in both groups, those who underwent genetic testing for BRCA1/2 and those who declined, had a diagnosis of cancer or related breast disease. When divided into groups of those with ($n = 96$) and those without ($n = 88$) a diagnosis of cancer or related disease, there was no difference in the frequency of genetic testing (cancer diagnosis 52%, no diagnosis 48%, $P = 0.33$). Thus, having a cancer diagnosis did not increase the likelihood of obtaining a BRCA1/2 test in our clinic population. We also hypothesized that there would be a higher rate of positive test results (*i.e.*, a mutation that is deleterious or of unknown significance) in the group of subjects with a diagnosis of cancer or related breast disease than in those subjects with no diagnosis. Of the 106 PTCs who underwent BRCA1/2 sequencing, 55 (52%) were positive for a mutation in one or both of the *BRCA1/2* genes. Within the tested group, there was an equivalent proportion of positive test results for those with (26 of 52) and those without (29 of 54) cancer or related breast disease ($P = 0.64$).

Expectations for Insurance Coverage. PTCs (92) who completed a follow-up telephone interview were asked about their expectations of insurance coverage. They were asked to consider genetics-related services outside of the clinical visit and

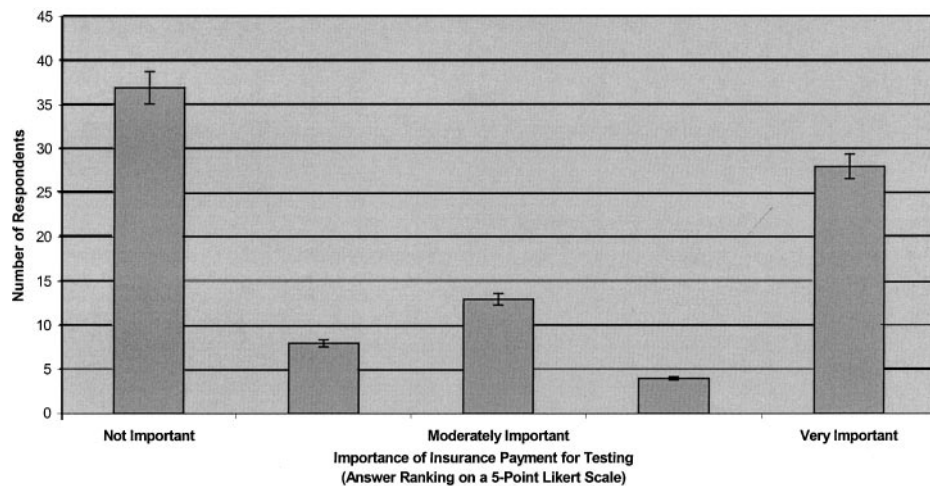


Fig. 2. Illustration of the biphasic answer pattern that was typically provided by Study 2 respondents. For each of the three questions asked about BRCA genetic testing decision making, >72% of the responses were at the extreme ends (*i.e.*, not at all important or very important) of the five-point Likert scale.

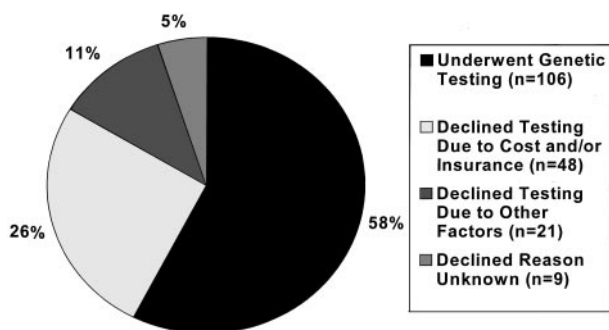


Fig. 3. Proportion of PTCs who accepted and declined BRCA genetic testing. Those who declined (~62%) did so because of concerns about cost, confidentiality, and discrimination.

Table 2 PTCs' expectations of insurance coverage for BRCA1/2-related services

	Percentage
Anything medically appropriate	29%
No idea/no expectations	24%
Consult only	15%
Consult and testing only	14%
Covers nothing	9%
Other	9%

BRCA1/2 testing, including associated risk management options, such as surgeries and chemoprevention (Table 2), as well as the BRCA1/2 test and risk evaluation consultation. The largest group (29%) responded that they expected their insurer to provide coverage for anything that was medically appropriate, with allowances for their expected copays and deductibles. The second largest portion (24%) did not have any particular knowledge or expectations. Half of the group who reported "no knowledge" volunteered the information that their uncertainty resulted from their unwillingness to inquire about coverage for specific services because of the potentially discriminatory effects of such written or telephone inquiries, *e.g.*, when asked

about her coverage for BRCA-related services, one respondent replied that she was "too scared of discrimination to even discuss the topic" with her insurance provider. Others stated that they simply did not know their insurance provider's policy on coverage of preventative services. The remaining respondents (47%) had a wide range of answers, including but not limited to no coverage at all, coverage for the initial consult only, and coverage for consult and surgery but not for the genetic test.

Insurance Experiences of PTCs. Respondents to the telephone interview were also asked to discuss their insurance experiences for BRCA1/2 testing or related services. Of this group, 15% (14 of 92) had chosen to pay for all of their services privately, intentionally not involving their insurance providers (all of these patients were privately insured). All of these patients stated that they chose the private pay option because of fear of future insurance and/or employment discrimination for themselves and/or their children. PTCs interviewed (38%, 35 of 92) did not report any problems at all in obtaining coverage for the services that they requested of their insurance providers. In many of these cases (23 of 35), however, PTCs requested that their insurer pay for only certain portions of their genetic services (*e.g.*, consult and surgery only but not testing).

The remaining 47% of interviewed patients reported efforts they made to attempt to procure payment for testing services. These efforts included time spent on the phone talking with insurance representatives, writing letters to the insurance companies requesting coverage, and time spent obtaining and coordinating letters of medical necessity from the oncologist, primary care physician, gynecologist, and/or surgeon, *e.g.*, one respondent's insurance provider declined coverage for prophylactic oophorectomy because it was billed under the diagnosis code "family history" and was excluded as a preexisting condition. Her insurance company later agreed to cover the surgery after letters of medical necessity were submitted to them, and the diagnosis code was changed. In another case, a woman <35 years old who reported problems securing reimbursement for mammography expenses obtained a letter of medical necessity from her primary care physician to procure coverage.

We hypothesized that fear of discrimination may keep testing candidates from involving their insurance companies in

Table 3 A. PTCs' involvement of insurance providers for BRCA½ genetic testing and related risk management services^a

	Involved insurance	Did not involve insurance
Pursued testing	43	10
Declined testing	35	4

	Involved insurance, no difficulties	Involved insurance, encountered difficulties
Pursued testing	16	27
Declined testing	19	16

^a $\chi^2 = 1.291$, $P = 0.256$.

^b $\chi^2 = 2.275$, $P = 0.132$.

payment for genetic services. We found no differences, however, between those who did and did not decide to undergo genetic testing in their involvement of insurance providers in payment for testing or related services ($P = 0.26$; Table 3A). We also explored the possibility that the pursuit of testing may have an effect on the amount of difficulties the subjects had in attaining coverage for genetics-related services (consultation, surgery, and screening). Although the difference was not found to be statistically significant, a greater percentage (63 versus 46%) of those who pursued testing encountered difficulties with insurers than those who declined testing but involved their insurers in other aspects of their genetic services care (Table 3B). Those who requested coverage (55%) for BRCA genetics-related services had difficulty beyond normal billing procedures in obtaining it; however, none of the PTCs interviewed were eventually denied coverage for BRCA-related services because of their family history being considered a preexisting condition. Some clients were denied coverage for other policy-specific reasons (e.g., their insurance plan did not cover office visits).

The Importance of Insurer Payment and Result Disclosure in Decision Making for Testing. PTCs interviewed by telephone were asked to rate, on a scale of one (not important) to five (very important), the importance of three testing-related issues (Table 4): (a) having an insurance provider pay for most or all of the cost of testing; (b) keeping results of BRCA testing confidential from the insurance provider; and (c) keeping BRCA testing results confidential from family members. The majority (62%) thought that having their insurance provider pay for all or most of the cost of testing was not an important factor in their decision whether to have BRCA analysis. In contrast, the majority (58%) of patients claimed that having their results kept confidential from their insurance provider was an important factor in their decision for or against pursuing genetic testing. Respondents were asked to rate how important it was that the risk evaluation clinic keeps information about the PTC's consult and any genetic results that may ensue confidential from their family members. It was emphasized that this was a hypothetical question, as currently, the clinic keeps this information strictly confidential unless authorized by the patient in writing to release the information to another party. Respondents (73%) said that keeping genetic information from their families (both immediate and extended) was of very little importance. The remaining 27% felt that it was important for the clinic to keep their genetic information confidential from their families; within this group, only 2 patients volunteered

Table 4 PTCs' importance ranking of payment and release of testing results

	Not important ^a (n) ^b	Important (n)
Insurance pay for most or all of the cost of testing	62% (57)	36% (33)
Testing results kept confidential from insurance provider	41% (38)	58% (53)
Clinic informs family of results/consult information	73% (67)	26% (24)

^a Not important was defined as a response of ≤ 3 on a five-point Likert scale.

^b Percentages do not sum to 100 exactly because of a few unusable responses.

that they chose not to share their genetic testing information with anyone at all.

Effect of Income on Testing Decision Making. We hypothesized that income level would affect decisions about testing because of the cost of the services. At the time of the study, full BRCA1/2 sequencing analysis cost \$2,400, and the typical charge for the initial clinic visit was ~\$225. Overall, approximately two-thirds of our respondents had an annual family income $> \$60,000$. We expected that the lower income respondents would consider having insurance pay for testing an important factor in their decision making. The data analysis showed that there was indeed a significant difference based on income in the importance attached to insurance coverage for testing between the two income groups ($P = 0.003$; Table 5). In contrast, there were no significant differences between the higher and lower income groups for the importance attached to test result confidentiality from insurers ($P = 0.73$) and family ($P = 0.26$).

Effect of Disease Status on Decision Making and Concerns about Confidentiality. We compared those with a diagnosis of cancer or related breast disease (such as lobular carcinoma *in situ* or atypical hyperplasia) to those without such a condition, hypothesizing that this might have an effect on the importance attached to insurance coverage for decisions about BRCA1/2 testing. We found no significant difference between the two disease status groups' rating of the importance of insurance coverage. The majority of both groups felt that having insurance pay for testing was not an important factor. We also hypothesized that patients who had been diagnosed with cancer or a related breast disease would be less likely than unaffected subjects to give a high importance rating to keeping results of BRCA testing confidential from insurance companies, as it was probable that their insurers were already aware of their cancer diagnosis. Contrary to our hypothesis, a majority of subjects in both disease status groups (affected and unaffected) expressed that having the results of BRCA1/2 testing kept confidential from their insurance provider was an important factor influencing their decision on whether or not to undergo testing. Similarly, no significant difference was found between disease status groups on the importance of keeping the results confidential from their families.

Discussion

In this study, we set out to investigate the hypothesis that concerns about insurance discrimination, cost of testing, and confidentiality of genetic information are significant factors in uptake of BRCA1/2 genetic testing services and genetic information. In our cohort, we demonstrated that a quarter of all PTCs eligible for and interested in testing reported the reasons

Table 5 Importance rankings of payment and confidentiality issues by PTCs based on annual family income level

	Family income <\$60,000, proportion rating as important ^a (n) ^b	Family income ≥\$60,000, proportion rating as important (n)	χ^2 (P)
Insurance pays most or all of the cost of testing	62% (13)	25% (13)	8.55 (0.003)
Testing results kept confidential from insurer	52% (11)	56% (29)	0.12 (0.728)
Testing results kept confidential from family	38% (8)	25% (13)	1.25 (0.263)

^a Important was defined as a response of >3 on a five-point Likert scale.

^b Nineteen of the respondents did not provide economic information and were not included in the analysis.

for declining testing were cost, confidentiality, and/or discrimination worries.

About two-thirds of our cohort had family incomes >\$60,000/year, probably reflecting both the quicker diffusion of knowledge about advances in medical science among the wealthier strata in the United States and, perhaps, assumptions about the economic burden of genetic services among referring physicians. Those with annual incomes <\$60,000 were more likely to consider insurance payment for services important in their decision to pursue or decline BRCA testing. Interestingly, those who opt to pay privately for genetic testing tend to believe they have a greater control over who has access to the information that they were tested, as well as over the test results. Many testing candidates who perceived themselves without sufficient resources to pay privately for the sequencing opted to seek reimbursement from their insurance providers. For these individuals, health insurers could, in principle, request access to the results of the testing as a stipulation for payment of risk management procedures, such as prophylactic oophorectomy, prophylactic mastectomy, and increased surveillance. Those who cannot afford to pay out of pocket for testing and have concerns about cost, confidentiality, and/or discrimination may be more likely to underuse BRCA genetic testing.

An important question we sought to answer in our clinical services is the following: how many individuals who carry a deleterious mutation do not learn about their status because of cost, confidentiality, and/or fear of discrimination barriers? One-quarter of our PTCs cited concerns about cost, confidentiality, and/or fear of discrimination as their reasons for declining testing. On the basis of the number of positive BRCA1/2 mutation results in our tested cohort, we estimated that, in our programs, 52% of patients declining testing because of discrimination, cost, and/or confidentiality concerns would be positive for a BRCA mutation. In our study, this number was 14% of the entire PTC cohort of 184 patients. Thus, unless there are systematic differences in BRCA status between those who do and those who do not report concerns about cost, confidentiality, and/or discrimination, ~14% of individuals eligible for testing do not learn of a positive BRCA mutation status because of cost, discrimination, or confidentiality concerns. Conversely, we estimate that 13% of PTCs fail to learn about a negative test result, which, depending on the mutation status of the family, may modify risk management and/or avoid unnecessary heightened screening practices and/or surgeries. The prevalence of disease is approximately equivalent in our populations with and without these concerns. This finding adds indirect evidence that there are no such systematic differences in mutation status based on the above concerns. The percentages are probably a gross underestimate of those who avoid genetic testing for these reasons, as our subjects were only those who had already decided to seek clinical consultation, a sample that was less diverse and wealthier than the general population. We surmise that many potentially eligible women and men are not even

evaluated because of geography, income, lack of knowledge, referral bias, and the cost and discrimination fears documented in our study, among other reasons.

One of the goals of genetic research is the dissemination of information from the scientific and academic community into the general population. Although referral sources were not studied *per se*, it is notable that within the study group of 384 patients seen in the clinic between January 6, 1997 and March 13, 2000, 279 (73%) had a family history sufficiently suggestive of a deleterious BRCA1/2 mutation to warrant offer of genetic testing, according to present medical practice (29). The high proportion of appropriate referrals is multifactorial; there appears to be significant awareness by the public and physicians in the State of Michigan of factors correlating with hereditary breast and ovarian cancer syndromes. It probably also reflects underuse of such services, particularly for lower income, uninsured, and minority women.

Several groups have sought to understand the factors that enter into patients' and physicians' decisions about BRCA-associated genetic services. Geller *et al.* (17) found that 34% of the physicians in their study underestimated the importance of discussing the risks of insurance discrimination with their patients. In their study, oncologists were more likely than other specialists to feel that the potential for loss of insurance was an important issue to discuss with patients (17). Because of increased education efforts by individual physicians, physician organizations, and foundations, physician awareness about insurance concerns may be increasing. Indeed, Matloff *et al.* (30) recently surveyed cancer genetics specialists and found that over three-fourths of the providers would not submit the charges for genetic testing to their own insurance carriers because of fears of genetic discrimination for both themselves and their children. The fear of insurance discrimination is therefore not limited to patients as seen in our study but is also present in genetic specialists, a population with presumed overall higher awareness and education about genetics and the law.

The question of whether these fears are discrepant with the actual practices of insurance providers is slowly being answered. In their extensive study of the effects of laws restricting the use of genetic information by health insurers, Hall and Rich (31) found that some of their insurance agent and insurance provider subjects responded that having a mutation in BRCA1/2 might have minimal relevance in health underwriting. They further reported that although the genetic counselors they interviewed could give examples of patients having trouble obtaining coverage for their counseling, testing, or preventative measures, none could furnish specific examples of overt health insurance discrimination (31). The data in our study, although still sparse, also support their conclusion. Although nearly half of the PTCs who chose to involve their insurers made efforts beyond normal billing procedures to have their BRCA1/2 genetic services covered, none of the patients were eventually denied insurance coverage for risk prevention services on the

basis that family history was considered a preexisting condition. Thus, in agreement with Hall and Rich, our tested cohort incurred no specific discrimination in insurance coverage because of genetic testing results when they sought coverage for those services. Whether they might experience later discrimination (*e.g.*, inability to change insurer) was not addressed.

An important aspect of our data concerns the fact that fear of discrimination because of genetic testing results equally affected those with a diagnosis of cancer or breast disease and those who did not. In our interviews, a major concern raised was not only the proband's fear of personal discrimination but his/her fear of discrimination for children and relatives. This heightened fear of potential discrimination for insurance coverage and employability of relatives because of the proband's genetic testing results is equally common in both affected and unaffected individuals; therefore, it appears to be an important area where educational and legal efforts may be focused, *e.g.*, in the state of Michigan, Senate Bill No. 589 is designed to protect health insurance members, applicants, and their dependents from having to: (a) undergo genetic testing before issuing, renewing, or continuing a policy, contract, or certificate; or (b) disclose whether genetic testing had been conducted, the results of genetic testing, or genetic information.⁵ Because it appears that the law would preclude insurers from requesting information about genetic testing results as a condition for coverage, it would be important to educate patients and practitioners further on this specific protection.

Our report has the following limitations. The major limitation is that we are likely to underestimate the true proportion of those at high prior risk who are concerned about cost, confidentiality, and discrimination, because an unknown number of those who are most concerned may not make and/or keep appointments with a cancer risk evaluation clinic precisely because of such concerns. This report was also limited in that the cohort studied is not representative of the general population in either ethnicity or socioeconomic status. We were also unable to tease apart the specific effects ascribed to cost, confidentiality, and fear of insurance discrimination; future research in clinical or community populations may be able to separate these variables. Further, Study 2 was limited by a 17% loss of participation because of attrition. It is possible that the wide range of time that elapsed between the initial visit and the telephone survey may have affected response uptake, as well as the responses themselves. Participants responding retrospectively may not have accurately reported their feelings and experiences surrounding the initial visit, or their responses may be skewed by events in the months intervening between the clinic visit and telephone interview. Although seemingly small, our sample size is appropriate for this area of study and comparable with the sample size of other studies. The sample size does not, however, allow us to firmly draw conclusions from some of our negative findings, and thus, additional clinical studies of this nature are important future steps. A methodological limitation was the use of only one researcher not blinded to the question posed for coding all of the charts.

Concerns about cost, confidentiality, and/or discrimination were found to be the primary reasons for declining BRCA1/2 testing in the population of individuals who were considered reasonable candidates for testing according to current medical practice. Although this project explored the nature of the interactions between patients and their insurance providers and

found no cases of overt discrimination, longer follow-up time is required to evaluate whether loss of confidentiality or discriminatory practices ensued over time after genetic testing. The study reaffirms the need for cancer genetic service providers to be aware of the importance of assisting the patient in weighing the medical aspects of genetic testing, together with its ethical and legal implications for themselves and their relatives. A significant number of patients eligible for testing in a clinical setting are likely to decline when they consider these variables.

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