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Commercialization of *BRCA1/2* Testing: Practitioner Awareness and Use of a New Genetic Test

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Abstract

It was our purpose to determine the characteristics of practitioners in the United States who were among the first to inquire about and use the *BRCA1* and *BRCA2* (*BRCA1/2*) genetic tests outside of a research protocol. Questionnaires were mailed to all practitioners who requested information on or ordered a *BRCA1/2* test from the University of Pennsylvania (UPenn) Genetic Diagnostics Laboratory (GDL) between October 1, 1995 and January 1, 1997 (the first 15 months the test was available for clinical use).

The response rate was 67% of practitioners; 54% (121/225) were genetic counselors, 39% (87/225) were physicians or lab directors. Most physicians were oncologists, pathologists, or obstetrician/gynecologists, but 20% practiced surgery or internal or general medicine. Fifty-six percent (125/225) had ordered a *BRCA1/2* test for a patient; most of the rest had offered or were willing to offer testing. Of those who had offered testing, 70% had a patient decline *BRCA1/2* testing when offered. Practitioners perceived that patients' fear of loss of confidentiality was a major reason for declining. Nearly 60% of practitioners reported that their patients had access to a genetic counselor, but 28% of physicians who ordered a *BRCA1/2* test reported having no such access, despite the GDL's counseling requirement. The proportion of physicians reporting no access to genetic counselors for their patients increased from 22.4% in the first half of the study to 50% in the last half. Many practitioners have an interest in *BRCA1/2* testing, despite policy statements that discourage its use outside of research protocols. Practitioner responses suggest that patient interest in testing seems to be tempered by knowledge of potential risks. An apparent increase in patient concern about confidentiality and inability to pay for testing could indicate growing barriers to testing. Although most practitioners reported having access to counseling facilities, perceived lack of such access among an increasing proportion of practitioners indicates that lab requirements for counseling are difficult to enforce and suggests that an increasing proportion of patients may not be getting access to counseling.

Keywords

genetic testing; *BRCA1*; *BRCA2*; test use; genetic counseling

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INTRODUCTION

The *BRCA1* and *BRCA2* (*BRCA1/2*) genetic tests for predisposition to breast and ovarian cancer were among the first cancer genetic tests to become available for clinical use [Hubbard and Lewontin, 1996; Kolata, 1996; McCullough, 1995; Zoler, 1995]. *BRCA1/2* testing remains controversial among clinicians, patients, ethicists, and policy-makers [American Cancer Society, 1996; Hubbard and Lewontin, 1996; Koenig, 1996; Kolata, 1996; National Advisory Council for Human Genome Research, 1994; Stephenson, 1995; The American Society of Human Genetics, 1994; The National Breast Cancer Coalition, 1995] because of: (1) uncertainty about the predictive value of the test [Healy, 1997] and frequency of *BRCA* mutations [Newman et al., 1998]; (2) uncertainty about the effectiveness of treatments for, or preventive measures against, breast and ovarian cancer (especially in young women or women with strong family histories of breast or ovarian cancer) [Burke et al., 1996]; (3) concern about the psychosocial impacts of testing [Biesecker and Brody, 1997]; (4) questions about the availability or adequacy of counseling in some clinical settings [American Society of Clinical Oncology, 1996; National Action Plan on Breast Cancer, 1996; National Advisory Council for Human Genome Research, 1994; The American Society of Human Genetics, 1994; The National Breast Cancer Coalition, 1995]; and (5) a lack of knowledge about genetics among some patients and health care providers [Hofman et al., 1993].

In contrast, others believe that information about genetic status should be made widely available to help people make important personal and medical decisions, despite the absence of definitive knowledge about test efficacy or medical management strategies [Schulman and Stern, 1996; Skolnick, 1996].

Although patient demand could be an important factor in the use of the *BRCA1/2* test, practitioners are gatekeepers of the technology, strongly influencing how and when the test will be used. However, practitioners from different specialties are likely to differ in their attitudes towards testing and subsequent preventive or treatment strategies [Belanger et al., 1991; Houn et al., 1995; Long, 1993; Mulvihill et al., 1982; Tarbox et al., 1992], and these differences could have important implications for how the testing is presented to and affects patients. For example, medical geneticists and genetic counselors have generally adopted an [Bosk, 1993; Caplan, 1993; Fraser, 1974; West, 1988]. In contrast, practitioners such as internists or surgeons might consider it negligent not to give patients recommendations about genetic testing [Caplan, 1993; Geller and Holtzman, 1995; Geller et al., 1993], or believe it unnecessary to obtain elaborate, written informed consent for a genetic test. Therefore, if testing becomes increasingly used by practitioners who are unfamiliar with genetic testing, patients might be less likely to be drawn into the decision-making process.

Furthermore, the quality and quantity of informed consent procedures and genetic counseling may differ widely among practitioners. Although practitioners with specific training in genetics are not the only professionals who can provide adequate counseling for genetic testing, there is evidence that non-geneticists are less knowledgeable than geneticists about mode of inheritance, calculation of genetic risks, and other skills important to the use of genetic tests [Hofman et al., 1993]. It is unlikely that practitioners who cannot refer patients to other professionals for genetic counseling services will be able to spend as much time providing this service as would a genetic counselor.

The passage of *BRCA1/2* testing from the research to the clinical realm allowed us to study the evolution of the commercialization of a genetic test (the University of Pennsylvania was the first organization in the United States to begin offering the *BRCA1/2* test for clinical purposes

[i.e., to patients not participating in a research protocol], in October 1995). No previous studies have focused on the characteristics or role of the practitioner in the use of such testing.

The purpose of this study was to obtain information on the practitioners in the United States who were among the very first to inquire about or use the *BRCA1/2* test in clinical settings (i.e., outside of a research protocol). Key variables included: (1) practitioner characteristics and practice settings; (2) why *BRCA1/2* tests are offered, and practitioners' perceptions of reasons for patients declining testing; (3) practitioners' attitudes towards genetic testing; and (4) patients' access to genetic counseling. These results are the baseline for a three-year longitudinal study to track changes in practitioner characteristics as the commercialization of *BRCA1/2* genetic testing evolves.

METHODS

Sampling and data collection

Our target sample was all practitioners in the United States who requested information on or ordered a *BRCA1/2* test from the University of Pennsylvania (UPenn) Genetic Diagnostics Laboratory (GDL) between October 1, 1995 and January 1, 1997, the first 15 months in which UPenn offered the test for clinical use. Data was collected from a one-page questionnaire mailed to practitioners, with a single follow-up mailing to non-responders after six weeks. Mailings were sent out to new practitioners as they entered our sample pool (approximately once a month). The study was approved by the institutional review board of UPenn (Committee on Human Subjects Research).

Questionnaire

The questionnaire was designed to collect information on: (1) practitioners' demographics, degrees and professional board certifications, academic affiliation, and type of clinical setting; (2) how practitioners first learned about *BRCA1/2* testing, reasons for requesting information about the test; (3) whether the practitioner had ordered a *BRCA1/2* test, and reasons for doing so or not doing so, whether the practitioner had ever had a patient who declined *BRCA1/2* testing, and if so, why; and (4) a description of genetic counseling resources available to the practitioner's patients.

Testing policies and advertisement

During the study period, GDL tested only patients with a positive family history of breast and/or ovarian cancer suggestive of dominant inheritance (early-onset and bilaterality [Ford et al., 1994]). Testing was only performed through a physician or genetic counselor, and genetic counseling was a requirement. Test costs were \$260 to \$680, depending on the number of mutations screened. GDL did not directly advertise test availability, but articles about test availability appeared in local [McCullough, 1995] and national [Behen, 1996; Cooke and Ochs, 1996; Kolata, 1995] lay publications, as well as in professional newsletters aimed at clinical pathologists [Titus, 1996] and internists [Zoler, 1995].

Analysis

Analyses included descriptive statistics about practitioner characteristics, as well as analytic statistics to test the following associations: (1) that specialties and availability of counseling facilities of practitioners changed over time (comparing the first half of the study with the last half); and (2) that there is an association between test ordering and availability of counseling facilities. For analysis of categorical data, we used the chi-squared test (or two-tailed Fisher exact test where appropriate). For tests of significance, $\alpha = 0.05$ (two-tailed).

RESULTS

Response rate and practitioner characteristics

Three hundred and thirty-four health care practitioners requested information on or ordered a *BRCA1/2* test from the GDL between October 1, 1995 and January 1, 1997, and all were sent questionnaires. The response rate was 67% (225/334). Practitioners were primarily genetic counselors (54%) and physicians (39%) (Table I). Two thirds of physicians identified themselves as either oncologists or pathologists/cytopathologists (Table II). However, substantial proportions of physicians indicated that they practiced obstetrics/gynecology, or general or internal medicine. The average age of genetic counselors was 38 years (range 26–67). Ninety-seven percent were female, and 93% identified themselves as white. The average age of physicians was 48 years (range 34–71); 41% were female, and 93% identified themselves as white.

We performed contingency table analysis to examine whether the proportion of practitioners in each of the identified specialties differed between the first 7 months of the study and those contacting the GDL in the last 8 months. We found no significant differences (data not shown).

Practice setting and counseling facilities

We determined the proportions of practitioners that characterized their practice settings as private practices, academic research institutions, oncology centers, and/or genetic clinics (Table III). Categories were not mutually exclusive. Not surprisingly, 62% of genetic counselors identified their practice settings as a genetics clinic. Over two thirds (69%) of physicians worked in private practices.

We asked practitioners to describe the personnel available to their patients for genetic counseling, and coded responses into the following categories: no genetic counselor, at least one genetic counselor, a multi-disciplinary team (i.e., at least one genetic counselor plus one or more other practitioners, including a psychologist or psychiatrist, an oncologist, obstetrician-gynecologist and/or other specialist). The extent of available facilities varied widely (Table IV). Although nearly 60% of responding practitioners indicated that their patients had some access to counseling facilities, 12% indicated having none. Of physicians, 28% reported having no counseling facilities available (Table IV). We could not determine the extent of counseling facilities for 29% of practitioners because the respondent provided the name of the facility, but not a description of the personnel or services. Because most of the named facilities were large academic research institutions with genetics clinics, most of these facilities probably had at least one genetic counselor available to patients referred there.

We conducted contingency table analysis to determine whether the extent of counseling facilities was associated with either of three factors: (1) having referred a patient for testing; (2) time of contacting the GDL (i.e., in the first half of the study or the last half); or (3) practice setting (i.e., were practitioners in private settings more or less likely to have access to counseling facilities than practitioners in other settings?). For these analyses, we examined the proportion of physicians reporting no available counseling facilities, dichotomizing the counseling facilities variable into no genetic counselor vs. at least one counselor, multi-disciplinary team, or indeterminate (because most indeterminates probably had at least one counselor available). Table V shows the results of these three analyses. We found that the proportion of practitioners reporting no available counseling facilities did not differ significantly between those who had referred patients for testing and those who had not. However, we did find that practitioners who contacted GDL in the last half of the study (compared to those contacting GDL earlier) and practitioners in private practice were significantly more likely to report having no access to a genetic counselor for their patients.

Of physicians contacting the GDL in the last 8 months of the study, 50% reported having no counseling facilities available, compared with 22% in the first 7 months ($P = 0.03$, Fisher exact test).

Awareness and use of *BRCA1/2*

Most practitioners reported becoming aware of testing through journals (54%), professional meetings (47%), and colleagues (29%), but some reported learning about the test from the lay media (11%), patients (4%), or other sources (10%). (Percentages do not add up to 100% because responses were not mutually exclusive.) Fifty-six percent (125/225) of practitioners indicated that they had ordered a *BRCA1/2* test from the GDL. An additional 26 practitioners said they had offered at least one patient the test but none had yet accepted. Thus, two thirds of practitioners (151/225) had offered a *BRCA1/2* test to at least one patient. Of the 74 practitioners who had not offered a test, the main reasons for not doing so were lack of appropriate patients (32/74; 43%) or because the practitioner was awaiting completion of testing or counseling protocols (19/74; 26%). Only 6/74 (8%) said they were unsure whether they would offer the test. Thus, most of those who had not yet ordered a test were prepared to or intending to do so. Fifteen percent of practitioners who ordered a test tested a patient for reasons not consistent with GDL requirements (e.g., because of patient request, without family history of cancer).

Patients' reasons for declining testing

Forty-seven percent (106/225) of all practitioners (or 70% [106/151] of the subset who had offered testing) indicated that they had at least one patient who declined *BRCA1/2* testing. 102 practitioners responded to the question "Why did they decline?" Given four response options for the practitioner's perception of the primary reason the most recent patient declined testing, 68% of practitioners (69/102) indicated concern about confidentiality, 52% (53/102) indicated fear of results, 42% (43/102) indicated inability to pay, and 29% (30/102) indicated other reasons. These response options were chosen based on the most frequently given responses in pilot testing. Reasons most often cited in the "other" category were that the test results would not change the patient's course of action (11%; 11/102), the patient did not think the test was definitive enough (6%; 6/102), and because the patient felt at insufficient risk to pursue testing (6%; 6/102). (Percentages do not add up to 100% because responses were not mutually exclusive.)

DISCUSSION

Practitioner characteristics and practice setting

A wide variety of practitioners are interested in or have ordered *BRCA1/2* tests from GDL. Genetic counselors and physicians specializing as medical geneticists represent a little over half of the practitioners requesting *BRCA1/2* tests or information from the GDL, indicating that this group is predominantly practitioners specifically trained in the interpretation of genetic test results. A substantial proportion of physicians in our sample are in specialties that generally do not have direct contact with patients (i.e., pathology or cytology), and probably are ordering tests at the request of other practitioners. Together, internists, practitioners of general medicine, and pediatricians formed one quarter of our sample, indicating that this genetic test is starting to penetrate the primary care sector. If any changes in the proportions of practitioners from particular specialties occurred during our 15-month study period, they were not large enough to be detected in our sample.

As would be expected, most practitioners interested in *BRCA1/2* testing worked in settings that would likely serve patients with a family history of cancer, such as genetics clinics and oncology centers. The high proportion of physicians in private practice indicates that use of

the test is not confined to academic research centers where *BRCA1/2* testing was first offered under research protocols.

Counseling facilities

Most practitioners (both those who had referred a patient to GDL for a test and those who had not) had access to some sort of counseling facilities for their patients. However, a small, but growing, proportion of practitioners reported not having such access or were not aware of how to obtain such services. Although we cannot distinguish between lack of counseling facilities and lack of knowledge about counseling facilities, both possibilities may result in lack of access to counseling for patients. The proportion of practitioners reporting no access to counseling services grew significantly over the 15-month time period of our study. The growth may be due in part to the increase in practitioners in private practices, who are less likely than other practitioners to have access to counseling facilities, and whose proportion grew significantly over the study period. Thus, the availability, amount, and quality of counseling for *BRCA1/2* testing may be decreasing as time goes on.

Given the policy statements of professional and patient organizations that stress the necessity of counseling for *BRCA1/2* testing, and GDL's requirement for access to genetic counseling, the trends we observed are of concern. Our findings also point out the difficulty that a genetic testing laboratory can have in enforcing counseling or informed consent requirements from a distance; similar problems with policy implementation have been observed in other areas of medical practice [Hogle, 1995].

Awareness and use of *BRCA1/2* testing

As with other new technologies, a high percentage of practitioners reported learning about the *BRCA1/2* test through professional contacts and publications. This mode of awareness of new technologies may be a reflection of our sample, which is limited to practitioners who contacted a university laboratory. This finding may also reflect a general bias in self-reports by practitioners about awareness of new medical technologies towards professional, rather than lay sources [Avorn et al., 1982]. Therefore, this finding is likely to underrepresent patients, advertising or the general media as sources of information for practitioners.

According to practitioners, patient acceptance of testing was by no means unanimous. Almost as many practitioners have had a patient who declined testing as have had a patient who accepted. Demand for testing, even among those with strong family histories of breast or ovarian cancer, may therefore be lower than predicted by previous studies in hypothetical situations [Chaliki et al., 1995; Lerman et al., 1996; Lerman et al., 1995]. Although our data do not provide the actual rate at which patients decline *BRCA1/2* testing, they do show that declining *BRCA1/2* testing is not uncommon. Our findings are consistent with other studies on acceptance of *BRCA1/2* testing [Doksum et al., 1997; Lerman et al., 1996; Loader et al., 1997; Wiesner et al., 1997], genetic tests for cystic fibrosis, [Bekker et al., 1993], and Huntington's disease [Bloch et al., 1989; Craufurd et al., 1989; Quaid and Morris, 1993].

Practitioners believe that concerns over test result confidentiality is a major factor in declining *BRCA1/2* testing. This suggests that patients are aware of privacy risks created by genetic testing [Billings et al., 1992; Wulfsberg et al., 1994] and are willing to forgoe acquisition of genetic information because of these risks. Indeed, patients might have become aware of these risks through discussions with their practitioners themselves, or through other sources. Whereas the severity of these risks remains to be demonstrated definitively, it seems that awareness among patients currently offered *BRCA1/2* testing about some of the potential problems of genetic information is high.

In addition, practitioners believe that fear of test results is also a major cause of declining testing, suggesting that a significant proportion of patients do not want the additional knowledge gained from a genetic test for hereditary, adult-onset cancer, and are fearful of this knowledge. This result is consistent with other findings that have shown that some people seek genetic knowledge, while others prefer to avoid it [Codori and Brandt, 1994; Decruyenaere et al., 1995; Mastromauro et al., 1987; Tibben et al., 1993]. A smaller, but substantial proportion of patients who decline testing appear to do so because they cannot afford it. This situation may change as the technology becomes more widespread and as more insurers cover the service.

Conclusions

A wide variety of practitioners appear to be interested in and aware of the availability of *BRCA1/2* testing for clinical purposes from a university laboratory. At the same time, patient interest may be tempered by knowledge of potential risks of testing. In these early stages of test availability, interest in and use of testing among practitioners seems to be primarily coming from those with genetics training and access to counseling facilities. The significant increase in practitioners without such access, especially among those who do not have experience in genetic testing, could have a negative impact on informing patients of risks and benefits of testing. We will be able to compare our data on the early users of the *BRCA1/2* test with those who use the test in the future in order to determine (1) how practitioner characteristics, access to counseling facilities, and patient concerns about testing evolve, and (2) whether this evolution is proceeding in a way that is likely to enhance or detract from patients' abilities to receive comprehensive information and to exercise an informed choice about this genetic test.

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TABLE I

Practitioner Profession

	Respondents (N = 225) n (%)
Genetic counselor	121 (54)
Physician or lab director	87 (39)
Nurse	12 (5)
Clinical or research fellow	5 (2)

TABLE II

Physician Specialties

	Respondents (N = 87) n (%)
Oncology	29 (33)
Pathology or cytology	29 (33)
Obstetrics/gynecology	19 (22)
Internal or general medicine	15 (17)
Surgery	9 (10)
Pediatrics	7 (8)
Medical genetics	6 (7)

* Responses do not add up to totals because they were not mutually exclusive.

TABLE III

Practice Setting

	All practitioners (N = 225) n (%)	Genetic counselors (N = 121) n (%)	Physicians (N = 87) n (%)
Private practice	65 (29)	5 (4)	57 (66)
Academic research institution	52 (23)	29 (24)	18 (21)
Oncology center	47 (21)	28 (23)	14 (16)
Genetics clinic	96 (43)	75 (62)	13 (15)

* Responses do not add up to totals because they were not mutually exclusive.

TABLE IV

Counseling Personnel Available to Patients

	All practitioners (N = 198 respondents) n (%)	Physicians (N = 74 respondents) n (%)[*]
No genetic counselor	23 (12)	21 (28)
At least one genetic counselor	58 (29)	3 (4)
Multi-disciplinary team	59 (30)	9 (12)
Indeterminate	58 (29)	41 (55)

* Responses do not add up to 100% because of rounding error.

TABLE V

Association of Factors With Availability of Counseling

	Referred patient (N = 196 respondents)		Time period (N = 198 respondents)		Practice setting (N = 198 respondents)	
	Referred	Have not referred	Months 1-3	Months 8-15	Private	Not private
No genetic counselor	9 (10%)	14 (13%)	14 (9%)	9 (24%)	15 (27%)	8 (6%)
All other *	77 (90%)	96 (87%)	146 (91%)	29 (76%)	41 (73%)	134 (94%)
Total n (%)	86 (100%)	110 (100%)	160 (100%)	38 (100%)	56 (100%)	142 (100%)
P value (Fisher exact test)	0.7		0.02		0.0001	

* At least one genetic counselor, multi-disciplinary team, or indeterminate.