Attitudes and Practice of Genetic Counselors Regarding Anonymous Testing for BRCA1/2.

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Abstract:

Patients and clinicians alike view anonymous testing as a potential way to avoid perceived risks of genetic testing such as insurance and employment discrimination and the potential loss of privacy. To assess their experience with and attitudes towards anonymous testing for BRCA1/2, genetic counselors were invited to complete an internet-based survey via the NSGC Familial Cancer Risk Counseling Special Interest Group (FCRC-SIG) listerv. A majority of the 115 respondents (70%) had received requests from patients for anonymous BRCA1/2 testing at some point in their careers and 43% complied with this request. Most counselors, however, encountered such requests infrequently, 1–5 times per year. Although genetic counselors do not generally encourage anonymous testing and over a third of respondents feel it should never be offered, a substantial subset support its use under specific circumstances. In general, a strong consensus exists among counselors that anonymous testing should not be offered routinely. In light of the current legislative landscape, it is of note that a substantial proportion of respondents (42.7%) cited the threat of life insurance discrimination as a reason for pursuing AT, and fewer cited health insurance (30.0%) or employment discrimination (29.1%) as justifications. Since there exists no federal legislative protections against discrimination by life insurance companies, it makes sense that genetic counselors were more responsive to this issue as opposed to the threat of discrimination in health insurance and employment.

Keywords: genetic counseling | human genetics | clinical psychology | public health | ethics | health insurance | anonymous testing

Article:

Introduction

Offering BRCA1/2 genetic testing to individuals with a suspicious personal or family history of breast and/or ovarian cancer has become standard of care (Trepanier et al. 2004). Genetic testing allows mutation carriers who have greatly elevated risks (up to an 85% lifetime risk of breast cancer and 50% risk of ovarian cancer) to avail themselves of prophylactic surgery and other risk-reducing measures (NCCN 2007). Patients who are concerned about the potential for insurance or employment discrimination and loss of privacy will sometimes request their BRCA1/2 genetic testing to be performed anonymously. “Anonymous testing” (AT) is defined as the process by which a clinician and patient employ a pseudonym for the patient in order to exclude third parties, such as insurance companies, from accessing the test results (Visintainer et al. 2001).

Fear of genetic discrimination, defined as insurance or employment discrimination based specifically on genetic testing results, can serve as a motivator for the choice to pursue AT. In his seminal 2000 study, Mark Hall found that the fear of discrimination swayed patients against testing for adult-onset conditions, despite the presence of legal protections (Hall and Rich 2000). Similarly, in a more recent study of primary care patients, 40% did not consider genetic testing for hemochromatosis to be prudent given the possibility of health insurance discrimination (Hall et al. 2005).

Patients and clinicians involved in cancer genetic testing experience similar concern about genetic discrimination. In a 2002 study of 184 patients undergoing BRCA genetic testing, 58% felt it was important for the results to be kept from their insurance company (Peterson et al. 2002). Likewise, a 2000 survey of 163 cancer genetic counselors revealed that only 24% would submit a claim to their insurance company were they to undergo genetic testing themselves, and 25% would use an alias in their own testing (Matloff et al. 2000).

Recently, several legislations have been passed in response to the risk of genetic discrimination. Forty-three states have passed legislation protecting against health insurance discrimination (Genetic and Health Insurance State Anti-Discrimination Laws 2008) and 35 states have passed laws preventing employment discrimination based on genetic information (State Genetics Employment Laws 2008). In addition, the federal Health Insurance Portability and Accountability Act (HIPAA) of 1996 offers some protection against genetic discrimination since it aims to protect all health information equally (Genetic Discrimination 2008). Broader federal legislation was passed by Congress. The Genetic Information Nondiscrimination Act, or GINA, was signed into law on May 21, 2008 (Hudson et al. 2008).

The issue of genetic discrimination has been regularly discussed in cancer genetic counseling sessions. GINA protects patients from discrimination in health coverage or employment based on genetic information. Health insurers or health plan administrators are prohibited from using genetic information for making decisions about coverage, rates, or preexisting conditions. Employers may not use genetic information for making decisions about hiring, firing, promotion or terms of employment (Genetic Information Nondiscrimination Act of 2008 (GINA) 2009).

Despite these protections, people are still at risk for discrimination in the life, disability and long term care insurance arenas. Moreover, there is stigma attached to being positive for a gene mutation and privacy is also an issue with genetic testing (Roche and Annas 2006). According to a 2003 survey of 337 cancer genetic counselors, 96% “always” or “almost always” discuss the possibility of discrimination, including the topics of health insurance, life insurance, employment, and social discrimination. While specific strategies for dealing with the possibility of genetic discrimination vary, 32% of counselors polled in this study list AT as one way to avoid problems from this potential threat (Pfeffer et al. 2003).

Anonymous testing has been previously studied in Huntington disease. It has been estimated that 1.9% of testing for Huntington disease is done anonymously (Visintainer et al. 2001). However, before HIPAA was enacted in 1996, it was estimated that one out of every five people seeking testing for Huntington disease requested anonymous testing (Mehlman et al. 1996). Anonymous testing for Huntington disease include problems establishing report and psychosocial assessment as well as questions about informed consent and scheduling appointments (Burgess et al. 1997; Visintainer et al. 2001). Others have described benefits of AT such as alleviating stress in patients by giving them control over their own medical information (Mehlman et al. 1996) and that by having the option of anonymous testing, more individuals may be willing to get tested (Burgess et al. 1997).

In contrast to Huntington disease, there is a paucity of research regarding the use of AT for the BRCA1/2 genes. Specifically, no studies have identified how frequently patients and clinicians work together to coordinate AT for BRCA1/2. In addition, genetic counselors’ attitudes toward AT for BRCA1/2 have not been formally assessed. Thus, the purpose of this study was to determine the prevalence of AT for BRCA1/2 and genetic counselors’ attitudes towards this practice.

Methods

Sample and Procedures

The study was approved by the Institutional Review Board of the University of North Carolina at Greensboro, and was performed in two phases. Phase I of the study was qualitative in nature and carried out via telephone focus groups. Focus groups were used so that a small group of counselors could discuss, react, and respond to the experiences of each other in an in-depth fashion. Phase I study participants were solicited for participation in two ways. Approximately a month prior to the focus groups, fliers were posted at the 2005 Annual Education Conference of the NSGC. An announcement about the focus groups and an invitation to participate was also posted on the listserv for the Familial Cancer Risk Counseling Special Interest Group (FCRC-

SIG) of the National Society of Genetic Counselors (NSGC), which, at the time of the posting was composed of approximately 400 genetic counselors that specialize in cancer genetic counseling. Potential participants were asked to submit their contact information, their availability, and whether they had tested anyone anonymously for BRCA1/2. Respondents were then organized into three focus groups based on availability. Although participation in the focus groups was not limited to counselors who had done anonymous testing for BRCA1/2, the groups aimed to have at least one member who had this type of experience. Those selected to participate were sent an email message that contained information about the focus group schedule, the questions for the focus groups, and a consent form. Participants were required to fax the signed consent form to the principle investigator prior to the initiation of their focus group.

Telephone focus groups were accomplished using a conference call service and were moderated by the principal investigator (TA) and a second member of the project team (LS) who acted as an assistant moderator. Three focus groups with three participants each were scheduled. However, two of the three groups included only two participants for a total of seven participants in this phase of the study. Each participant was given the list of questions for the focus groups prior to participating. Participants used only their first names during the focus groups to identify themselves, and were given the option of using a pseudonym if they had concerns regarding their identity being uncovered.

Each focus group lasted approximately one hour and was digitally recorded. One member of the project committee also took notes during the focus groups. The tapes were transcribed and the responses were categorized and examined for themes. Phase II of the study consisted of an anonymous online survey to obtain quantitative data about the prevalence of anonymous testing for BRCA1/2, the proportion of counselors in favor or opposed to anonymous testing for BRCA1/2, and the percentage of counselors who desire formal guidelines. An invitation to participate that included a link to the online survey was posted on the FCRC-SIG. Of the approximately 400 members of the FCRC-SIG who received an invitation to participate in the survey, 117 (~29%) completed at least a portion of the survey and 110 completed the entire survey. Individuals that participated in the focus groups were excluded from the survey. Topics for the survey were derived from the focus group data.

Instrumentation

Focus group questions for Phase I of the study were developed based on review of relevant literature and are listed in Appendix A.

The on-line anonymous survey used in Phase II of the study was developed by the authors to assess the experience with and attitudes towards anonymous testing for BRCA1/2 among cancer genetic counselors. Survey development was informed by the results of the three focus groups conducted in the Phase I of the study to assure that the questions addressed the relevant issues

identified by the focus group participants. The survey was pilot tested on local genetic counselors and genetic counseling students with experience in the subject.

The survey, which was designed to take approximately 5–10 min to complete, reflects response themes identified by analysis of the focus group transcripts. The survey questions are listed in Appendix B.

Data Analysis

For Phase I of the study, telephone focus group recordings were transcribed and examined for themes. Major categories identified were: a) experience with and response to requests for anonymous testing, b) opinions about anonymous testing for BRCA1/2, c) pros and cons of anonymous testing for BRCA1/2, d) circumstances under which anonymous testing is acceptable, and e) potential utility of guidelines for anonymous testing for BRCA1/2.

Statistical analysis for the Phase II of the study was performed using Statistical Package for the Social Sciences (SPSS) version 14. Responses were coded and chi-square analysis was done on individual questions to look for significant differences among responses. Pearson's chi-square was also used to compare participant responses to different questions to look for significant differences among responses from one question to the next and to identify trends.

Questions which allowed for multiple responses were coded, and quantities of responses from each participant were quantified. The averages of these responses, excluding the response category "I do not agree with any of these choices", was determined. Normality of each was determined using a sample K-S test, and then the means were compared using a paired samples t-test. For the chi-square tests, the t-test, and the K-S test, a p value of less than 0.05 was considered significant. A p value of less than 0.10 was used to determine significance for normality.

Results

Phase I

A total of seven genetic counselors participated in the focus groups. All were currently practicing cancer genetic counseling and came from a wide range of geographic locations in the United States. Their time practicing cancer genetic counseling ranged from 5 months to 10 years. Three were currently practicing in a university setting, three in a private practice, and one in a non-profit hospital. Four had tested a patient anonymously; however all of them had experience with patients requesting anonymous testing. Focus group participants discussed many issues relevant

to anonymous testing including their own experiences and their response to theoretical situations. The major topics discussed, how the counselors responded, and some relevant quotes are presented in Table 1.

Table 1

Summary of Responses from Focus Group Participants

Responses	**Quotes**	
Feelings about AT for BRCA1/2	• Would discuss issues relating to AT first (7/7)	I don't have a problem with it.
	• Opposed to AT for BRCA1/2 (5/7)	I don't feel like that's up to me to decide so that if the patient chooses to do it. I would be willing to facilitate.
	• Would do it if patient wished (6/7)	
Benefits	• Protection from discrimination such as instances not protected by HIPAA or someone with previous history of genetic discrimination (4/7)	I think maybe it's not a long-term solution, but it definitely helps with the development of the relationship in the beginning. Later on, when you discuss the results, and you've been working with them for a long time and they trust you, maybe they will be a little bit more willing to relinquish the control. But at their first visit with you, they might not be willing to give up the control at that point.
	• Psychosocial benefits (e.g. help with rapport, patient well-being) (2/7)	
Drawbacks	• Concerns about information not getting back to pertinent health care providers (4/7)	… issues with the patient have to be discussed regarding her medical management, and so if her testing is done anonymously and perhaps her doctor doesn't feel confident that he knows that it's her result. The doctor may feel uneasy treating her based on a result that [doesn't] have her name on it.
	• AT gives false illusions (3/7)	I think [you have to] be really careful about, for lack of a better term, tricking the patient into having testing, because if they're under the illusion that they can go through all of this and be anonymous and then all of the sudden, "hey, no you really should tell your insurance company". That could be an issue in itself. I think it's just important to be very straightforward with what might happen eight weeks from now when the results come back.

Responses	Quotes	
Feelings about anonymous testing for HD vs. BRCA1/2	• No respondents were against AT for HD	I think HD is certainly something that can have a tremendous impact in life and is not really anything that you can help with medical management. So in that case I can really understand people being afraid of insurance discrimination and being afraid of employment discrimination.
	• More value than for BRCA1/2 (5/7)	
Non-genetic AT	• Approve of AT for HIV—because of social stigma (2/7)	I think again, it's the medical management issue and that, as long as the doctors have the information they need to act on... I don't know, I just, I'm not philosophically against it, I just don't think from a practical standpoint it would work very well.
	• Think that AT for HIV is generally not practical (4/7)	
Scenarios when AT is acceptable	• Military individuals (2/7)	…there's been some talk about discrimination with the military, and I would have to do more research if I ever had a patient in that situation as to what the real risks were. But if those things turned out to be possible or true, then I think that might be a circumstance where potentially it would be acceptable.
	• Long term care insurance (1/7)	
	• Confidentially issues (3/7)	
Feelings about NSGC guidelines for AT	• Against them (2/7)	I think really has to do with where you are located and until we have the same laws across the board, I feel that, again, any sort of guideline would not be able to cover every situation and in that case it just might make things more confusing than need be
	• Not against them, but feel unnecessary (3/7)	
	• Would be acceptable if they dispelled fears (1/7)	
	• None stated necessary	

The four focus group participants who had performed anonymous testing were asked to discuss their cases, especially any which went particularly well or poorly. In all cases described by the focus group participants, patients cited genetic discrimination as their reason for doing testing anonymously, with one who had a past problem with employment discrimination. The counselors were asked their personal opinions about anonymous testing. If a patient wished to have anonymous testing, all of the focus group participants said that they would first discuss

issues concerning anonymous testing with the patient. This was something heavily stressed by some participants:

“Well we have a policy where we offer it, but we really try to give the patient all the information. Once we talk with them, we find that most patients don’t want to do it in the end because of insurance covering the back end of things, if they come back positive, all the different screenings and prevention methods.”

While 5/7 focus group participants stated that they do not like anonymous testing at all, 6/7 said they would do anonymous testing for BRCA1/2 if the patient asked for it, while the other counselor has a departmental policy against offering anonymous testing. Of the five counselors who stated that they did not like anonymous testing for BRCA1/2, most cited anonymous testing as having a low value:

“I personally don’t really like anonymous testing. Obviously, I would never tell the patient that, and I’ve been happy to facilitate that if someone really requests that. But I guess I don’t really see much value to it… for me, I see the risk of discrimination is the release of the results to the insurance or employer or whatnot. I think the main issue is paying for the test verses putting it through your insurance because if you pay for it and you get it under your own name, it’s still probably not likely to get back to your insurance company. And the risk of it getting back to your insurance company when you pay for it, in my opinion, is quite low.” –Ar

The two counselors who approved of anonymous testing felt that it was the patient’s decision:

“I don’t have a problem with it. I think, while the issues with the patient have to be discussed regarding her medical management, and so if her testing is done anonymously and perhaps her doctor doesn’t feel confident that he knows that it’s her result. The doctor may feel uneasy treating her based on a result that [doesn’t] have her name on it. I don’t feel like that’s up to me to decide so that if the patient chooses to do it. I would be willing to facilitate.” –J

Because of the past research about anonymous testing for Huntington disease, participants were asked their feelings about anonymous testing for Huntington disease to compare to that of BRCA1/2. No participants stated feelings against anonymous testing for Huntington disease. Five of the participants stated that because of the severity of Huntington disease, anonymous testing may have more value than that for BRCA1/2:

“I think with something like Huntington’s disease, I definitely would see a little bit more value to it considering that it is 100% penetrant and, not that I have vast experience working with Huntington’s or other conditions with a similar outcome, but I do think it is different and I do see more potential for discrimination with conditions like Huntington’s.” –Ar

“I think HD is certainly something that can have a tremendous impact in life and is not really anything that you can help with medical management. So in that case I can really understand

people being afraid of insurance discrimination and being afraid of employment discrimination." –M

Phase II

Demographics of the participants in the online survey are shown in Table 2. Years in practice ranged from less than a year to over 30 years in practice, and years in cancer genetic counseling ranged from less than a year to over 20 years of practice. For settings in practice, 63/117 (53.8%) practiced in a university setting, 14/117 (12.0%) in private practice, and 40/117 (34.2%) under the category of "Other". Some of the responses in the "Other" category include community and non-profit hospitals, HMOs, and cancer institutes.

Table 2

Professional Characteristics of Respondents

Genetic counselor responses (*n* = 117)	**Number (Percent)**
Years in cancer genetic counseling	
<1 year	6 (5.1)
1–5 years	72 (61.5)
6–10 years	28 (23.9)
11–15 years	8 (6.8)
>15	3 (2.6)
Setting	
University	63 (53.8)
Private	14 (12.0)
Other	40 (34.2)

Approximately 70% of respondents (80/115) have received requests for AT. Of these, 88.7% (71/80) receive such requests 1–5 times per year. The second highest response to the frequency of requests for AT was for “Never” accounting for 30.4% of responses.

Participants were asked how many times they have helped a patient facilitate AT. Just under half of respondents (43.4%, 49/113) stated that they had facilitated AT for BRCA1/2 at least once in their careers. About half of those who had helped a patient obtain AT (55.1%, 27/49) had tested patients anonymously 1 or 2 times.

Participants were asked how well their experiences with AT have gone and whether they would assist a patient in performing AT again. Among respondents who had performed AT, 48.0% perceived the experience as problematic and 52.0% reported that the testing had gone well. Although nearly half of respondents reported that their experience with AT was “somewhat” or “very” problematic, a majority of respondents (86.0%, 43/50) who had already performed AT at least once would still facilitate it again.

Respondents that had never facilitated AT for BRCA1/2 ($n = 64$) were asked why they had never done so (Fig. 1). Nearly half (46.9%, 30/64) stated that after a discussion of genetic discrimination, the patient requesting AT opted not to proceed with AT, a response significantly more common than any other reason ($p < .001$). Eight of the 64 respondents that had never facilitated AT (12.5%) reported that they had a policy against doing AT. Of these, 7 responded that they practice in a university setting and 1 had responded “Other” to the question about practice setting.

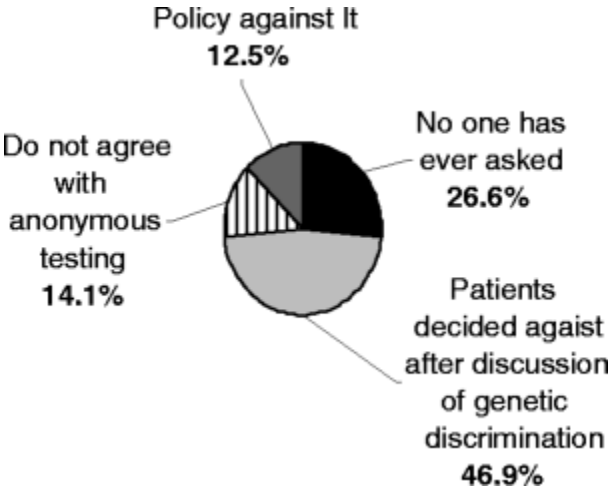

Fig. 1

Among the Genetic Counselors Who Have Never Helped Facilitate Anonymous Testing ($n = 64$), Their Reasons for not Doing so.

To assess the overall attitude toward offering AT, participants were asked to choose from four responses about the circumstances in which AT should be offered (Fig. 2). Only three

respondents (2.7%) felt that genetic counselors should routinely offer AT for BRCA1/2. All three of these reported they had been working as a genetic counselor for less than 5 years; two reported working in the university setting, with the third reporting “Other.” The remaining respondents were split evenly among three other positions with regard to AT: “As a matter of policy it should not be offered (35.7%, 40/112)”; “We should only offer it in specific circumstances” (27.7%, 31/112); “We should offer it if a patient requests it” (33.9%, 38/112). Given that much of the scholarship regarding AT in the past has focused on Huntington Disease (HD), participants were also asked about circumstances in which AT should be offered for this disorder and responses were compared with answers given in the context of AT for BRCA1/2. As shown in Fig. 2, there was a significant difference in counselors’ responses to anonymous testing for HD vs. BRCA, with counselors expressing more comfort with HD anonymous testing than BRCA (Pearson chi square = 98.9354, $p < 0.0001$).

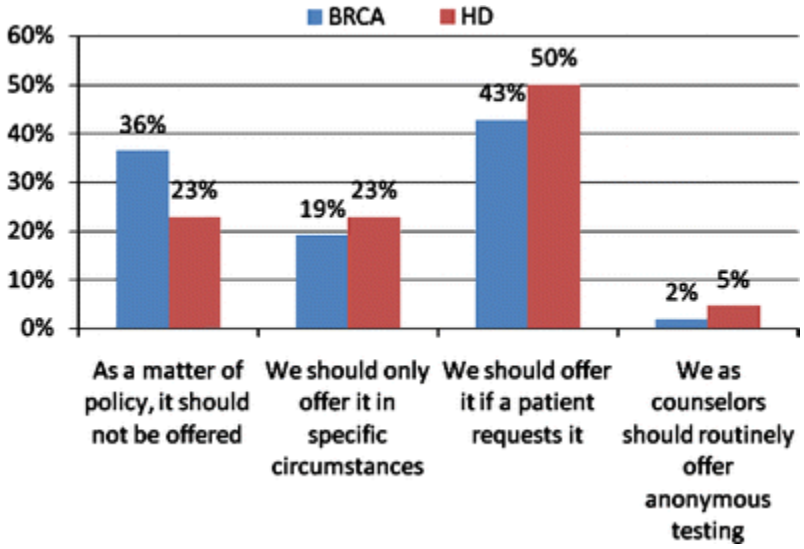

Fig. 2

Comparison of Genetic Counselors’ Attitudes Regarding the Offering of Anonymous BRCA Genetic Testing Versus Huntington Disease Testing (n = 110).

In an attempt to assess genetic counselors’ attitudes toward AT, respondents were offered a list of reasons to perform or deny AT. Responses are shown in Figs. 3 and 4. Respondents were generally inclined to agree with statements that discourage AT. Among the reasons used to justify offering AT (Fig. 3), only one rationale was selected by a majority of respondents “If the patient might not otherwise have testing,” (52.7%, 58/110) followed closely by “The patient is absolutely insistent on AT” (49.1%, 54/110).

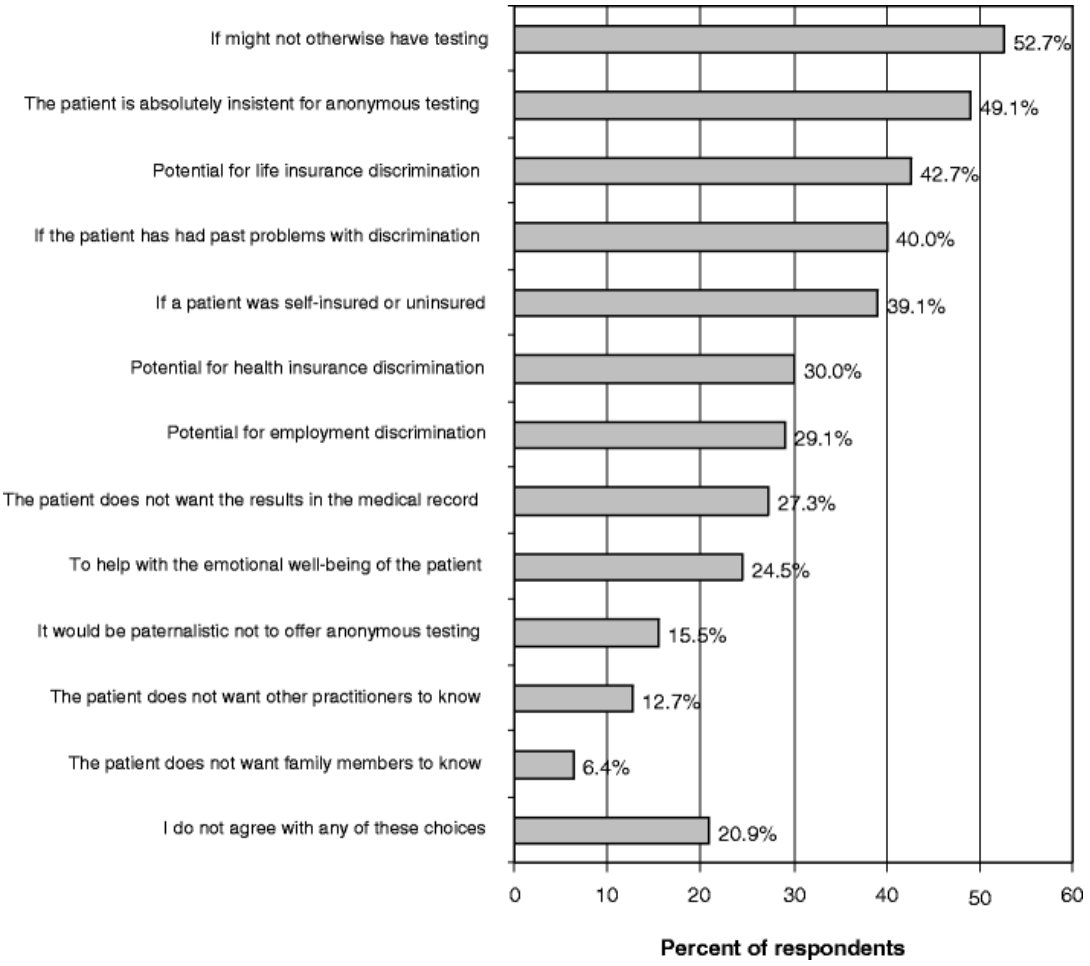

Fig. 3

Arguments for Anonymous BRCA Genetic testing. Counselors were Asked to Choose all the Responses with Which They Agreed (n = 110).

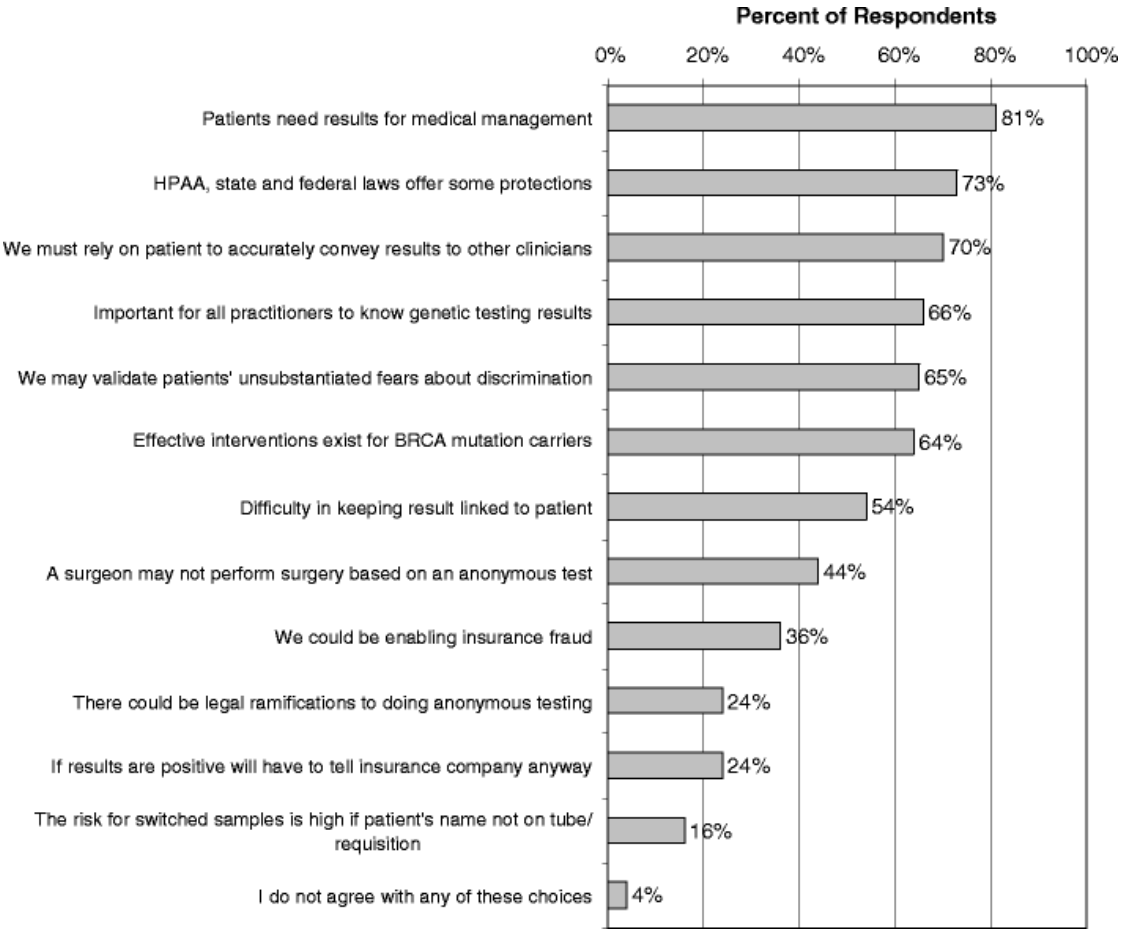

Fig. 4

Perceived Downsides of Anonymous Testing. Counselors were Asked to Choose all the Responses with Which They Agreed (n = 110).

Participants were asked if they would welcome NSGC guidelines for AT, with three different scenarios for what the guidelines would say including; if the guidelines strongly discouraged AT, if the guidelines left the decision of AT up to the clinician, and if the guidelines strongly encouraged AT. Responses were split, with no significant difference ($p = .450$), over whether or not counselors would welcome guidelines which discouraged anonymous testing for BRCA1/2. Respondents nearly unanimously (93.6%, $p < .001$) stated they would not be happy with guidelines that strongly encouraged offering anonymous testing. A significant number of respondents (62.7%, $p < .001$) would welcome a statement if it left judgment about anonymous testing up to the clinician. Participants were also asked if the NSGC should have a policy statement about anonymous testing. Among respondents, 58/110 were against NSGC developing a policy statement about anonymous testing and 52/110 are in favor of such a statement. There was no significant difference between these responses ($p = .705$).

Discussion

Genetic discrimination can be an important source of concern for patients in genetic counseling. Even now that protective legislation in the form of GINA exists, such concerns will likely remains an issue since GINA only addresses the risk for discrimination in health insurance and in the work place. The risk of discrimination remains real to patients in the realm of life and disability insurance. Additionally, the concern over the potential loss of privacy and stigma attached to having a gene that places one at risk for a life threatening condition lingers. The results of the current study demonstrate that a substantial proportion of respondents (42.7%) cited the threat of life insurance discrimination as a reason for pursuing AT, and many fewer cited health insurance (30.0%) and employment discrimination (29.1%) as justifications. Since there are currently no protections against discrimination by life insurance companies, it makes sense that genetic counselors were more responsive to this issue as opposed to the threat of discrimination in health insurance and employment.

AT can be seen as one mechanism for dealing with such concerns. In this study we found that a majority of genetic counselors that responded to our survey have received requests for AT (69.6%) and about half (43.3%) have performed AT. In spite of this cumulatively significant prevalence, the actual practice of AT appears to be limited, given that 55.1% of those counselors who had facilitated AT had done so only once or twice. While this practice is not occurring in the majority, it still is significant in that it is happening. Visintainer et al (2001), in their study of anonymous testing for Huntington disease, stated that if anonymous testing was also occurring with other conditions besides Huntington disease, there needs to be a dialog in the genetics community about it.

Counselors expressed disparate attitudes about AT for BRCA1/2. Over a third of counselors felt that AT should never be facilitated. While most of the other counselors could envision circumstances in which AT would be reasonable, there is virtual unanimity that AT should not be a matter of policy, with only 3 of 112 genetic counselors supporting the routine offering of AT. The variety of responses appears to reflect both the willingness of counselors to try to accommodate patients' requests as well as counselors' desire to keep the frequency of this method of testing low.

In an effort to illuminate some of the different reasons which may motivate a counselor to offer or discourage AT, we offered respondents the opportunity to select reasons for and against AT. The responses seem to reflect both the high value genetic counselors place on BRCA1/2 testing and their desire to help patients obtain such information. Given that the most common reasons for AT were if the patient would not do testing otherwise or is absolutely insistent on AT, it appears that many genetic counselors are willing to facilitate genetic testing rather than having the patient forego genetic testing altogether. Medical management based on genetic testing results may still be available for individuals who undergo AT, however, by seeking to avoid disclosure of testing results to third-party payers, patients would be concealing the very

information that such payers would demand in order to pay for medical interventions such as risk-reducing surgery or enhanced surveillance.

In contrast to the tally of respondents' reasons supporting AT, in which most responses were chosen only by a minority of counselors, most of the reasons against AT were each selected by a majority of counselors, presumably reflecting their general discomfort with AT. The three arguments against AT most frequently selected reflect issues related to medical management and the fear the AT will impede proper care.

It is of interest that 64.5% of respondents felt that by offering AT we were validating patients' unsubstantiated fears about genetic discrimination and 72.7% of counselors were reassured by existing protections, including state and federal legislation including HIPAA and now GINA. These data suggest that a substantial number of counselors may feel the risk of genetic discrimination is inflated and that counselors derive at least some reassurance from the existence of those legal protections against discrimination that do exist at present.

Genetic counselors that participated in this study are almost evenly split in their opinions about the need for guidelines for anonymous testing for BRCA1/2. When asked under what conditions they would welcome anonymous testing guidelines for BRCA1/2, the only clear consensus was that very few of the respondents would welcome guidelines stating that AT for BRCA1/2 should be offered by all genetic counselors. Respondents were also almost evenly split on the issue of whether the NSGC should develop a policy statement about AT testing for BRCA1/2. In light of the responses to this survey, it does not appear to be appropriate to recommend that the NSGC develop such guidelines or policy statements. While guidelines may not be ideal for anonymous testing for BRCA1/2, a discussion about it should occur in the genetics community as per Visintainer's suggestion.

Finally, over two decades of experience with pre-symptomatic testing for Huntington Disease and the dramatic nature of that condition have served to make it a common focus of studies that address various features of genetic testing and counseling. However, as genetic testing expands to encompass many disorders, we must be careful not to over-generalize lessons learned from HD. Indeed, in the current study, when counselors were asked about their attitudes towards AT, they were more inclined to offer it in the context of HD as opposed to BRCA testing. This difference likely reflects the fact that no therapies or preventive strategies exist for HD, while preventive strategies with documented benefit exist in the realm of breast cancer risk. As genetic testing becomes part of mainstream medicine and is commonly applied to situations in which it will impact therapy or care, it is likely that the appeal of AT will decline.

There was general agreement between the conclusions drawn in the focus groups and those of the survey. For the potential benefits for AT; the focus groups mentioned protection against discrimination for those not covered by HIPAA as well as those who had previously experienced genetic discrimination. As shown in Fig. 4, the potential for insurance discrimination and past

problems with discrimination were selected as arguments in favor of AT by 43% and 40% of survey respondents respectively. Focus group participants discussed medical management issues, including the information not getting back to the provider, as arguments against AT. That patients need the information for medical management was the most frequent argument against AT among the survey respondents (Fig. 4).

This study was done prior to the passage of the Genetic Information Nondiscrimination Act (GINA) of 2008. With the enactment of GINA, new protections against genetic discrimination for insurance and employment have been made (Coalition for Genetic Fairness 2008; Hudson et al. 2008). Given the expanded coverage given against heath insurance and employment discrimination (Coalition for Genetic Fairness 2008; Hudson et al. 2008), it can be assumed that for AT because of these reasons should decrease. However given that GINA does not cover life, disability, and long term care insurance, nor does it cover individuals in the military (Coalition for Genetic Fairness 2008), requests from individuals for AT due to these reasons will probably stay the same.

There are several limitations to this study. First, since we asked genetic counselors to retrospectively estimate the rate of AT over their careers, the data we obtained are approximations. Further study needs to be done to accurately quantify the actual frequency of AT for BRCA1/2 testing. Second, the modest response rate from the FCRC-SIG listerv may have skewed our data by eliciting more responses from genetic counselors who had strong feelings about AT. Third, given the substantial portion of respondents who chose “other” as their practice setting, the choices that were provided were not sufficient, and we were not able to make accurate correlations between practice setting and experience with AT. Finally, since we only surveyed genetic counselors, we cannot make any inferences about the prevalence of anonymous BRCA1/2 genetic testing that might occur in conjunction with other clinicians such as primary care physicians, gynecologists or oncologists. Future studies will be needed to address more precise estimates of the prevalence of AT as well as nuances in counselor attitudes and circumstances in which they feel AT could be done.

Conclusions

Although genetic counselors clearly do not generally encourage AT and over a third of respondents feel it should never be offered, it is equally clear that a substantial subset support its use under specific circumstances. While GINA provides wider coverage against discrimination than has been available in the past, there are still gaps in this coverage. Due to these gaps, genetic counselors will continue to confront the issue of AT.

Appendix A

Focus group questions

1. Tell us your first name, if you are currently practicing cancer genetic counseling, and how long you have been (or were) practicing cancer genetic counseling.

2. Do you practice in a private or university setting?

3. Have you ever tested someone for BRCA1/2 anonymously by using a pseudonym or a number in testing to conceal the patient's identity?

4. Has anyone ever requested anonymous testing for BRCA1/2 from you?

5. Have ever had any experiences with anonymous testing sessions that went particularly well or poorly? Can you tell me about those experiences?

6. How did those patients pay for their testing?

7. How do you feel about anonymous testing for BRCA1/2?

    a. What do you feel are the benefits?

    b. The drawbacks?

8. What circumstances do you feel are acceptable to anonymously test for BRCA1/2?

a. Which are not?

9. Are there other conditions, such as Huntington's disease, where you feel anonymous testing is more or less acceptable?

10. Are there any other medical tests where you feel anonymous testing is acceptable (e.g. PSA)? Are your feelings based on whether or not the test is genetic? Why?

11. The following are scenarios in which patients may as for anonymous testing. What do you think of their cases?

a. Anne is a healthy 39 year-old who has never had cancer. She is a self-employed artist and has an individual health insurance policy. She has a family history of early-onset breast cancer. She is asking for anonymous testing because she is afraid of insurance discrimination.

b. Brenda is a 40-year-old woman recently diagnosed with breast cancer. She is contemplating bilateral mastectomy and would like testing to help her decide. She wants to pay out of pocket for anonymous because she doesn't want the result in her medical record.

c. Sue's mother had breast cancer two years ago and was found to have BRCA mutation. Sue, who is 45, is healthy, but doesn't want anyone to put the presence of this mutation into the medical record. For this reason, she is planning to have anonymous testing, and is not planning to tell her GYN who does her breast exams.

d. Terry works for a construction company that requires yearly physicals. She has a family history of breast cancer but wants to keep her testing anonymous so her employer won't lay her off due to the risk of breast cancer.

12. Would you welcome guidelines for anonymous testing for BRCA1/2?

Appendix B

Survey Questions

1. Years in practice (open ended)

2. Years in cancer genetics (seeing patients) (open ended)

3. University, private practice, other?

In this survey, we'll define anonymous testing as using a pseudonym or number on test request form.

4. In your practice, how often do patients request anonymous testing for BRCA1/2? (Regardless of whether you offer anonymous testing)

a. never

b. 1–5/year

c. 6–15/year

d. 16–50/year

e. 51+/year

5. Do you currently offer anonymous testing for BRCA1/2?

a. Routinely offer

b. Offer if requested

c. Offer only if patient is adamant

d. Refuse to offer/never offer

6. Have you ever helped a patient undergo anonymous testing for BRCA1/2? Yes/No

For participants that responded "Yes" to question 6:

7. In your opinion how well has this worked?

a. Very well

b. Well

c. Somewhat problematic

d. Very problematic

8. Would you do it again?

9. When was the last time you offered it? 0–1 year, 1–5 years, 5–10 years, 10+year

10. How many times have you helped a patient undergo anonymous testing?

a. 1

b. 2

c. 3

d. 4

e. 5–10

f. 10+

For participants that responded “No” to questions 6:

11. Why not?

a. No one has ever asked

b. After discussing genetic discrimination the patients have decided against anonymous testing

c. Don’t agree with anonymous testing

d. Have a policy against doing anonymous testing

12. What is your general feeling about anonymous BRCA1/2 testing?

a. As a matter of policy, it should not be offered

b. We should only offer it in specific circumstances

c. We should offer it if a patient requests it

d. We as counselors should routinely offer anonymous testing

13. What is your general feeling about anonymous Huntington disease testing?

a. As a matter of policy, it should not be offered

b. We should only offer it in specific circumstances

c. We should offer it if a patient requests it

d. We as counselors should routinely offer anonymous testing

14. Does your institution/clinic have formal guidelines regarding anonymous testing?

Yes/No

15. If so, what are they? (open-ended)

16. Here are some arguments for anonymous testing for BRCA1/2. Check the reasons you feel justify anonymous testing.

a. Potential for health insurance discrimination

b. Potential for life insurance discrimination

c. Potential for employment discrimination

d. If might not otherwise have testing

e. If a patient was self-insured or uninsured

f. It would be paternalistic not to offer anonymous testing

g. The patient does not want the results in the medical record

h. The patient does not want other practitioners to know

i. The patient does not want family members to know

j. To help with the emotional well-being of the patient

k. If the patient has had past problems with discrimination

l. The patient is absolutely insistent for anonymous testing

m. I do not agree with any of these choices

17. Here are some arguments against offering anonymous testing for BRCA1/2. Check those you agree with.

a. The risk for switched samples is high if patient's name not on tube/ requisition

b. Difficulty in keeping result linked to patient (i.e. if it gets separated from chart)

c. Patients need results for medical management (too expensive to pay for out of pocket.)

d. Important for all practitioners to know (radiologists, oncologist doing CBE, GYN)

e. By offering anonymous testing, we may validate patients' unsubstantiated fears about discrimination and HIPAA protects the medical record, including genetic testing results

f. With anonymous testing, there's no way to communicate with other health care providers so we must rely on patient to accurately convey results correctly

g. Effective interventions exist for those who test positive for BRCA1/2 mutations

h. A surgeon may not perform surgery based on an anonymous test

i. We could be enabling insurance fraud

j. If results are positive, will have to tell insurance company anyway

k. There could be legal ramifications to doing anonymous testing

l. HIPAA & state and federal laws offer some level of protection against discrimination

m. I do not agree with any of these choices

18. Would you welcome a policy statement from NSGC regarding anonymous testing for BRCA1/2?

a. If it strongly discouraged it? Yes/No

b. If it encouraged decisions about it up to clinician's judgment? Yes/No

c. If it stated all counselors should offer it? Yes/No

19. Should NSGC submit a policy statement about anonymous testing for BRCA1/2? Yes/No

20. After taking this survey, what is your general feeling about anonymous BRCA1/2 testing?

a. As a matter of policy, it should not be offered

b. We should only offer it in specific circumstances

c. We should offer it if a patient requests it

d. We as counselors should routinely offer anonymous testing

References

Breast Cancer Risk Reduction. National Comprehensive Cancer Network (NCCN) Guidelines, v1, 2007. Available at:www.nccn.org/professionals/physician_gls/PDF/breast_risk.pdf.

Burgess, M. M., Adam, S., Bloch, M., & Hayden, M. R. (1997). Dilemmas of anonymous predictive testing for Huntington disease: privacy vs. optimal care. *American Journal of Medical Genetics, 71*, 197–201.

Coalition for Genetic Fairness. (2008). How Does GINA impact ME? February 1, 2009 fromhttp://www.geneticfairness.org/ginaresource_impact.html#1.

Genetic and Health Insurance State Anti-Discrimination Laws. (January 2008). National Conference of State Legislators. Retrieved February 22, 2008 from http://www.ncsl.org/programs/health/genetics/ndishlth.htm.

Genetic Information Nondiscrimination Act of 2008 (GINA): Information for researchers and healthcare professionals. (April 2009). *National Human Genome Research Institute*. Retrieved July 17, 2009, from http://www.genome.gov/Pages/PolicyEthics/GeneticDiscrimination/GINAInfoDoc.pdf.

State Genetics Employment Laws. (January 2008). National Conference of State Legislators. Retrieved February 22, 2008 from http://www.ncsl.org/programs/health/genetics/ndiscrim.htm.

Genetic discrimination. (January 2008). *National Human Genome Research Institute*. Retrieved February 22, 2008, from http://www.genome.gov/10002077.

Hall, M. A., & Rich, S. S. (2000). Patients' fear of genetic discrimination by health insurers: the impact of legal protections. *Genetics in Medicine, 2*(4), 214–221.

Hall, M. A., McEwen, J. C., Barton, J. E., et al. (2005). Concerns in a primary care population about genetic discrimination by insurers. *Genetics in Medicine, 7*(5), 311–316.

Hudson, K. L., Holohan, M. K., & Collins, F. S. (2008). Keeping pace with the times—the Genetic Information Nondiscrimination Act of 2008. *New England Journal of Medicine, 358*(25), 2661–2663.

Matloff, E. T., Shappell, H., Brierley, K., Bernhart, B. A., McKinnon, W., & Peshkin, B. N. (2000). What would you do? Specialists' perspectives on cancer genetic testing, prophylactic surgery, and insurance discrimination. *Journal of Clinical Oncology, 18*(12), 2484–2492.

Mehlman, M. J., Kodish, E. D., Whitehouse, P., et al. (1996). The need for anonymous genetic counseling and testing. *American Journal of Human Genetics, 58*, 393–397.

Peterson, E. A., Milliron, K. J., Lewis, K. E., Goold, S. D., & Merajver, S. D. (2002). Health insurance and discrimination concerns and BRCA1/2 testing in a clinic population. *Cancer Epidemiology, Biomarkers & Prevention, 11*, 79–87.

Pfeffer, N. L., McCarthy Veach, P., & LeRoy, B. S. (2003). An investigation of genetic counselors' discussions of genetic discrimination with cancer risk patients. *Journal of Genetic Counseling, 12*(5), 419–438.

Roche, P. A., & Annas, G. J. (2006). DNA testing, banking and genetic privacy. *New England Journal of Medicine, 355*(6), 545–546.

Trepanier, A., Ahrens, M., McKinnon, W., et al. (2004). Genetic cancer risk assessment and counseling: recommendations of the National Society of Genetic Counselors. *Journal of Genetic Counseling, 13*(2), 83–114.

Visintainer, C. L., Matthias-Hagen, V., Nance, M. A., et al. (2001). Anonymous predictive testing for Huntington's disease in the United States.*Genetic Testing, 5*(3), 213–218.