Behavioral Risk Factors among Women Presenting for Genetic Testing

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Abstract

Considerable research attention has been given to the impact of genetic testing on psychological outcomes. Participation in genetic testing also may impact on health behaviors that increase the risk of cancer and other chronic diseases. The purpose of this study is to describe behavioral cancer risk factors of women who requested genetic testing for breast and ovarian cancer susceptibility (BRCA1, BRCA2). Before participation in a genetic testing program, 119 women completed a series of questionnaires designed to assess their health behaviors, perception of risk, and depressive symptomatology. Eight percent of participants were current smokers, 27% did not engage in at least moderate exercise, 46% did not regularly protect themselves from the sun, 39% did not consume at least five servings of fruits and vegetables per day, and 9% drank at least one alcoholic beverage per day. Poisson regression analysis revealed that age was the only predictor of behavioral risk profiles, with older women having fewer cancer risk behaviors. These patients who presented for genetic testing generally had better health behaviors than the general population. However, given their possible high-risk status, these patients should consider further improving their preventable cancer risk factors and, in particular, their diet, sun protection, and physical activity levels. Inclusion of behavioral risk factor counseling in the context of the genetic testing process may be an important opportunity to reach this at-risk population.

Introduction

The influence of heritable gene mutations on cancer risk has been well-documented. A significant amount of research attention is being devoted to identification of genes that are responsible for the development of cancer and to evaluating strategies for testing and informing at-risk populations regarding their risk status. This work in genetics represents a new direction for cancer prevention efforts, which have traditionally focused primarily on prevention and the reduction of behavioral risk factors.

Genetic testing for a variety of genes thought to be related to cancer is now available in the context of research protocols and through commercial testing facilities. A considerable amount of recent research has focused on the consequences of genetic testing, including the impact of testing on psychological functioning. Several studies have examined psychological distress among participants in genetic testing for breast cancer (1–3), as well as the impact of individualized breast cancer risk counseling on psychological outcomes among women with a family history of breast cancer (4, 5).

To date, much of the focus on sequelae of genetic testing has been concentrated on psychological outcomes, and indeed the development of interventions to ameliorate any adverse psychological effects is a very important area of study. One area of potential concern that has received relatively little research attention is the impact of participation in genetic testing on health behaviors that are modifiable and are known to increase overall cancer risk (e.g., smoking, diet, physical activity, alcohol consumption, and sun exposure; Refs. 6 and 7). If behavioral risk factors are not incorporated into the genetic testing process, concern about modifiable risk factors could be reduced by virtue of their absence from the discussion of risk factors. Thus, it is possible that individuals who have modifiable risk factors for other cancers may be inclined to underestimate the impact of those behaviors on their total cancer risk. All participants in genetic testing, regardless of their carrier status, could benefit from engaging in risk reduction practices that have been shown to reduce risk for other cancers and chronic diseases. In particular, women who are mutation carriers need to take their behavioral risk factors into account when making decisions about treatment options. For example, mutation carriers may be able to reduce their risk of breast and ovarian cancers through prophylactic surgery (8) and through the use of chemopreventive agents such as tamoxifen (9) and raloxifene (10). In making decisions to use such procedures, women need to consider how these treatment strategies will affect their risk for other chronic illnesses such as cardiac disease and osteoporosis. Women who have health behaviors that increase the risk of adverse consequences associated with prophylactic treatments must carefully consider the costs and benefits of such treatments in relation to their overall health status.

It is important to begin to understand the interaction between behavioral and genetic risk factors for cancer. Little is currently known about the prevalence of behavioral risk factors of individuals who present for genetic testing or the number and types of risk factors this population may possess. The purpose of this study is to describe the behavioral risk factor profiles among women who requested genetic testing for breast and ovarian cancer susceptibility (BRCA1, BRCA2) as part of the Dana-Farber Cancer Institute’s Cancer Risk and Prevention Program. A primary goal of this study is to describe the potential for health behavior change and subsequent reduction in preventable risk in individuals who are presenting for genetic testing.

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Materials and Methods

Program Description

Genetic testing for germ-line mutations in breast cancer susceptibility genes (BRCA1, BRCA2) is offered to adults who meet one or more of the following criteria: (a) there is a documented germ-line mutation in a cancer susceptibility gene in at least one family member; (b) the family history is suggestive of a germ-line mutation in either the BRCA1 or the BRCA2 gene, and candidate family members have a minimum 10% chance of having the mutation; and/or (c) the candidate family member has cancer and a family history that is suggestive of a germ-line mutation in a cancer susceptibility gene. Eligible subjects were identified and invited to participate on the basis of a visit to the Dana-Farber Cancer Institute Cancer Risk and Prevention Clinic, self-referral, or as a result of prior involvement in other research efforts. Mutation analysis of BRCA1 and/or BRCA2 was performed at Clinical Laboratory Improvement Amendment Act (CLIA)-certified laboratories.

The overall testing program includes a pretest genetic counseling session, a results disclosure session, and a follow-up visit at 2–3 months after disclosure. All study sessions are conducted by a genetic counselor and medical oncologist, with the exception of the third visit, which, at the request of a participant who tests negative, can be conducted by phone with the genetic counselor. In the event that a participant exhibits signs of psychological distress, a psychologist conducts a clinical assessment and makes recommendations as to the most appropriate approach for continued participation. Genetic counseling is an integral part of each visit and focuses on issues relevant to predictive testing, including a review of the family pedigree, a thorough discussion of the limitations of testing, potential results, appropriate medical surveillance, cancer risk management options, and individual and familial implications of both positive and negative results. Health behaviors are not typically discussed in the context of genetic counseling, although compliance with surveillance recommendations is reviewed. All in-person visits take place at the Dana-Farber Cancer Institute and incorporate semistructured interview guides, both to lend uniformity to the sessions and also to serve as a data collection tool. This study was approved by the Dana-Farber Cancer Institute’s human protection committee, and all participants signed a written consent form.

Participants who consent to the study are asked to complete a battery of instruments that assess their baseline knowledge of genetic testing, attitudes toward testing, motivations for determining their gene status, and routine health surveillance and risk factors for cancer. Questionnaires are sent to participants before their pretest counseling session, and they are instructed to bring the completed forms with them to their first appointment. This study presents baseline data focusing on risk factor prevalence.

Measures

Participants completed a number of questionnaires related to their demographic and personal characteristics and health behaviors. For each health behavior, current status was assessed, as was motivation for changing that risk factor. The target risk factors were selected because they confer the greatest impact on overall chronic disease morbidity and mortality.

Demographics and Medical History. Participants provided detailed information on educational, employment, and income status, as well as medical history related to risk factors and surveillance practices. Standard survey items for assessing demographic characteristics were used. Participants were asked about the frequency of participation in cancer screening tests, including breast self-exams, mammography, pelvic ultrasounds/exams, and CA-125 tests.

Smoking Status. Current smoking status was assessed using standard National Cancer Institute definitions; a positive smoking history was defined as having smoked at least 100 cigarettes in one’s lifetime. Current smokers were defined as those who had smoked any cigarettes in the previous 7 days. Readiness for changing smoking behavior was also assessed using the Stages of Change algorithm (11), which categorizes individuals into five stages of readiness to change: (a) Precontemplation includes individuals who report that they are current smokers and are not seriously thinking about quitting smoking in the next 6 months; (b) Contemplation includes current smokers who are seriously thinking about quitting smoking in the next 6 months; (c) Preparation includes current smokers who are intending to quit smoking in the next month, and who have tried to quit in the past year; (d) Action includes individuals who report that they are not currently smoking and that they have quit smoking within the past 6 months; and (e) Maintenance includes former smokers who report that they have not smoked for at least 6 months.

Physical Activity. The physical activity assessment was a modified version of the Paffenbarger Activity Questionnaire (12) and focused on lifestyle-oriented physical activity (e.g., blocks walked, stairs climbed). Motivational readiness for physical activity was measured using a standard algorithm that categorizes individuals similarly to the smoking algorithm described above, based on intention to adopt the target behavior within the next 6 months (13).

Nutrition. The fruit and vegetable screener (14), which was developed as part of the National Cancer Institute’s 5-A-Day for Better Health Program, was used to assess fruit and vegetable intake. We considered using a more extensive assessment of dietary intake. However, the respondent burden related to questionnaire completion in the Predisposition Testing Program is quite significant; therefore, we opted to selectively assess fruit and vegetable consumption, which is related to risk for a variety of cancers. The stages of change algorithm for fruit and vegetable consumption characterized intention regarding change in intake, similar to the smoking algorithm described above.

Sun Protection. Sun protection was measured with a 12-item scale that assessed regular participation in a variety of sun protection practices (e.g., use of sunscreen, staying out of sun between 10 a.m. and 2 p.m.; Ref. 15). The stages of change algorithm characterized intention regarding and participation in regular sun protection practices.

Alcohol. Participants were asked to categorize approximately how many times a week they had drank alcohol during the last month and to quantify how much they typically consume on each drinking occasion. Average daily alcohol intake for the preceding 30 days was then calculated for each individual and used as a proxy for typical alcohol consumption. A cutoff of more than 2 drinks/day (e.g., 60 drinks/month) was used as the level of alcohol consumption that was likely to increase risk. (16). Stage of readiness to change was not assessed for drinking behavior because of the low prevalence of alcohol consumption in this sample.

Behavioral Risk Profile. Although individual behavioral risk factors make an important impact on cancer risk, the presence of multiple risk factors may have a synergistic effect on risk (17–19). Thus, in addition to assessing individual risk behav-
ior, we evaluated the presence of multiple risk factors using a continuous measure designed to evaluate the relationships between the target risk factors (19, 20). The multiple risk factor index is based on current smoking status (smoking), consumption of fewer than five servings of fruits and vegetables per day, not engaging in at least moderate regular exercise, inconsistent practice of sun protection, and drinking an average of more than two alcoholic drinks per day. Subjects were assigned a score of 1 if they had each risk factor or a score of 0 if they did not have that risk factor. These categorizations were based on what is generally accepted in the field as the minimum requirements for risk factor reduction and/or cardiorespiratory benefit. The individual risk factor scores were then summed to yield a continuous multiple risk factor assessment.

**Depression.** Mood has been found to be related to the prevalence of cancer risk behaviors; therefore, we felt it was important to assess the impact of mood on health behaviors in this population. Depressive symptomatology was assessed using the CES-D (21), a widely used symptom rating scale that has adequate test-retest reliability in general populations as well as among those presenting for predictive testing (22). The range of possible scores on this measure is from 0–60; higher scores reflect more depressive symptoms. For the regression analyses that use CES-D score, individuals were considered to possess depressive symptomatology if they were over the standard cutoff of 15 recommended by Radloff (21).

**Risk Perception.** Risk perception is another potentially important modifier of cancer risk behaviors (23). Participants were asked to estimate their overall lifetime risk of developing any cancer (or another cancer, for those with a cancer history) on a scale of 0–100% and to provide a categorical lifetime risk estimate, where the response choices included “very high,” “high,” “average,” “low,” and “very low.” For the regression analyses that examined the relationship between risk perception and cancer risk behaviors, percentage lifetime risk estimates were dichotomized into categories of either up to 50% or greater than 50%.

**Data Analysis Plan.** The goal of the analysis was to examine the prevalence of each of the target cancer risk factors among women who were presenting for genetic testing and to examine predictors of cancer risk factor profiles among the participants. Univariate analyses, using χ² and ANOVA were used to examine the prevalence of the target health behaviors among the study population and to examine the relationship between health behaviors and patient characteristics, including cancer history and depressed mood. Poisson regression was then used to model predictors of the behavioral cancer risk profiles. Estimated regression coefficients are equal to the log of the relative ratio of mean numbers or risk factors for a unit increase in a covariate. Thus, the relative ratio will be greater than 1 if subjects who have higher scores on a covariate also tend to have a greater number of cancer risk factors. Similarly, relative ratios of less than 1 occur if subjects who have higher scores on a covariate also tend to have fewer cancer risk factors.

Data were available from 119 subjects from 106 families. Because people from the same family tend to behave similarly, responses from family members might not be independent. Estimates of variance were therefore corrected for possible dependencies using the generalized estimating equations approach described by Liang and Zeger (24). All models were fit using the SAS procedure PROC GENMOD (25).

Potential predictors of the number of behavioral risk factors included subjects’ age, education (college versus no college), cancer status (cancer versus no cancer), CES-D score (depressed versus not depressed), and risk perception (up to 50% lifetime risk versus greater than 50% lifetime risk). All hypothesis tests were two-tailed and were declared statistically significant if the P was less than or equal to 0.05. Furthermore, all of these procedures were used to test whether there was any association between a covariate and the behavioral risk profiles.

**Results**

**Univariate Analyses**

**Demographics and Medical History.** The sample was comprised of 119 women. The average age of participants was 46 years (range, 26–84 years); all of the participants were Caucasian, reflecting the clinical population from which they were drawn. Seventy-one percent of the sample were married, 11% were single, and 11% were separated or divorced; 75% of the participants had children (range, 1–5). Seventy-eight percent of participants had a college or advanced degree, and 64% were employed at least part-time. Annual household income was at or above $50,000 for 68% of the sample; 98% had health insurance, and 64% had life insurance. Sixty-three percent of the sample had a previous cancer history; among these women, 79% had a diagnosis of breast cancer, 15% had ovarian cancer, and 6% had been diagnosed with both breast and ovarian cancers. Fifteen percent of the sample met the criteria for depression established by the CES-D. The difference in the frequency with which cancer patients and unaffected participants met clinical criteria for depression was not statistically significant (19% versus 9%; P = 0.16).

Sixty-four percent of respondents provided lifetime cancer risk estimates in excess of 50%. Eighty-eight percent classified their lifetime cancer risk as “very high” or “high,” 9% reported “average” lifetime cancer risk, and 3% reported “low” or “very low” risk estimates. As expected, participants with cancer histories provided higher lifetime cancer risk estimates than those without cancer (P = 0.008); however, the range of estimates was similar between the groups (25–100% for affected participants versus 11–99% for unaffected participants). Categorical risk estimates did not differ significantly, with 53% of participants with a cancer history versus 37% of unaffected participants rating their lifetime risk as “very high,” and 36% of cancer patients versus 50% of unaffected participants rating their risks as “high.”

**Health Surveillance Practices**

Health surveillance practices in the cohort were largely consistent with recommendations for women with increased breast and ovarian cancer risks: (a) monthly breast self-exam; (b) annual/semiannual clinical breast exam; (c) annual mammography after the age of 25 years; and (d) annual/semiannual pelvic ultrasound and CA-125 measurement. Sixteen percent of participants reported having had prophylactic surgery; eight women had prophylactic mastectomy, seven women had prophylactic oophorectomy, and two women had both surgeries. Two others reported prophylactic hysterectomy with removal of the ovaries. Three of those who had an oophorectomy did not have a history of cancer; all of the other women who underwent prophylactic surgery had cancer. An additional 14 women (12%) reported total abdominal hysterectomies that were not prophylactic in nature; all but one of these women had a cancer history.

Among the 44 women without cancer histories, 46% reported that they performed a breast self-exam at least monthly. Forty-four percent had clinical breast exams at 3- or 6-month intervals, and 51% had clinical breast exams annually. Nine
percent had mammography every 6 months, and 83% had mammography once a year. Those who did not report having regular mammography were all over the age of 25 years. Among women with intact ovaries, 24% reported having pelvic exams every 6 months, and 70% had them annually. Twenty-seven percent had ultrasounds every 6 months, and 40% had ultrasounds once a year. Sixteen percent reported analysis of CA-125 levels twice a year, and 42% reported annual CA-125 screenings.

Those with a cancer history were more vigilant about their health surveillance. Sixty-one percent of those who had not had prophylactic and/or therapeutic mastectomies reported breast self-exam at least monthly. Sixty-seven percent had clinical breast exams every 3 or 6 months, and 31% had annual clinical breast exams. Eighteen percent had mammography at 6-month intervals, and 77% reported annual mammography. The majority of women whose ovaries remained intact reported having pelvic exams at 3- or 6-month intervals (32%) or annually (43%), although some had them every 6 months (11%) or annually (28%); 31% reported having annual CA-125 screenings, whereas 19% had them every 3 months, and 12% had them every 6 months.

**Health Behaviors**

**Smoking.** Forty-five percent of the sample had smoked at some point in their lifetime; the vast majority (59%) of former smokers had quit for more than 5 years. The prevalence of current smoking was 8%, likely reflecting the high educational and socioeconomic status of this sample. There was relatively little difference in smoking prevalence between women who had a previous cancer diagnosis (8%) and those who were unaffected (7%).

Motivational readiness to change was examined using ex-smokers and current smokers because the smoker sample size was quite small. Among the ever smokers, 9% were in the earliest stages of motivation to change, 7% were in preparation, 7% were in action, and 77% were in maintenance (see Table 1 for stage distribution of current smokers and the entire sample).

**Physical Activity.** Seventy-three percent of the sample reported engaging in at least some moderate or vigorous exercise; 32% reported exercising vigorously at least three times per week, and 54% reported moderate physical activity at least five times per week. There were no significant differences in either moderate or vigorous exercise patterns based on cancer history. There was a trend for younger participants to engage in more regular vigorous physical activities ($P = 0.06$).

Among those who did not report engaging in regular moderate physical activity, the majority (84%) were in the contemplation and preparation stages for change (see Table 1). Among those who were not engaging in regular vigorous physical activity, the majority (85%) were in precontemplation and contemplation.

**Sun Protection.** Ninety-eight percent of the sample had fair or medium skin tone, and 89% reported getting at least a slight burn upon first exposure to summer sun when not wearing sunscreen. Forty-four percent of the sample reported experiencing at least three blistering sunburns in their lifetime, placing them at high-risk for developing skin cancer. There were no significant differences in consistent sun protection use between those who had a cancer history (58%) and those who did not (48%).

Fifty-four percent of participants reported that they consistently protect themselves from the sun; 34% of the sample reported that they always wear sunscreen when out in the sun for more than 15 min. The majority of participants who were not engaging in sun protection were in the precontemplation stage of change (see Table 1).

**Nutrition.** Fifty-one percent of the sample reported following the recommended five or more servings of fruits and vegetables per day; among those who did not, the majority were in the contemplation stage of readiness to change (see Table 1). There were no significant differences in fruit and vegetable consumption based on cancer history.

**Alcohol.** On average, participants reported consuming nine alcoholic drinks in the last month (range, 0–80 drinks). Ninety-one percent of participants reported drinking less than once per day. Seven percent consumed 1–2 drinks/day, and only one participant had more than 2 drinks/day on average. There were no differences in prevalence of problem drinking based on cancer history.

**Behavioral Risk Profiles**

Calculation of participants’ behavioral risk profiles provides a way to evaluate overall behavioral risk. Twenty-six percent of the sample had none of the targeted behavioral risk factors,
Cancer risk perception (lifetime percentage risk) was explored by modeling the risk perception scores using dummy variables including a category for subjects whose risk perception scores were unknown. Results were run separately using lifetime percentage risk and categorical risk as the estimate of cancer risk perception, and no differences in the results were found. The only significant association with the behavioral risk profile was age (see Table 2), with older subjects having fewer cancer risk behaviors.

### DISCUSSION

Among this sample of affected and unaffected women who were seeking genetic testing for breast and ovarian cancer, less than 10% were current smokers, over one-third did not regularly engage in physical activity, almost one-half did not regularly protect themselves from the sun, and over one-third did not consume at least five servings of fruits and vegetables per day. Population-level data suggest that in the general population, 23% of women are smokers (26), 35% report being very likely or likely to always use sun protection (27), about 30% of women consume at least five servings of fruit and vegetables each day (28), and about 16% engage in regular vigorous activity. These results suggest that patients who present for genetic testing have substantially better health behaviors than women in the general population. However, this study also revealed that between one-third and one-half of women in this sample had cancer risk behaviors that may further increase their risk. This is particularly important, because the socioeconomic status of the sample was high, and therefore it could be expected that their behavioral risk factor profiles might be better than those found among lower-income populations (29–35).

The prevalence of a substantial number of behavioral risk factors in this sample is of concern. In contrast, the health surveillance practices in this cohort were largely consistent with recommendations for women with increased breast and ovarian cancer risks (36). It should be noted that there was no relation-
health behaviors, although these behaviors might improve overall health and reduce risk for a number of cancers and other chronic conditions. It may be particularly important for mutation carriers to practice optimal health behaviors; as successful treatment strategies for reducing the risk of breast and ovarian cancer are identified, the risk of other common cancers and diseases will become of greater concern for carriers. As genetic testing moves into the commercial sector and primary care physicians become more involved in referral and follow-up related to testing, additional opportunities for discussing lifestyle risk factors with patients will be available. It is important to note that among those not currently engaging in the recommended behaviors, motivation for change was relatively low (40, 41). This suggests that successful intervention strategies for targeting this population will include motivational as well as action-oriented skills-based strategies.

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