

Note from the Editor

After a year-long gestation, we are pleased to present the first Special Issue of *Cancer Epidemiology, Biomarkers & Prevention*, entitled “Psychosocial Aspects of Cancer Genetic Testing: Findings from the Cancer Genetics Studies Consortium.” This topic was selected to highlight the growing importance of cancer genetics and behavioral interventions in the reduction of cancer morbidity and mortality among susceptible populations. The success of this effort is due to the dedication of the Associate Editors for the Special Issue, Deborah Bowen, Andrea Farkas Patenaude, and Sally Vernon.

Planning has begun on a second Special Issue of the journal, featuring the work of the Cooperative Family Cancer Registry for Breast Cancer Studies, an NCI-funded multinational resource for researchers worldwide. We welcome feedback from our readers regarding this and any future Special Issue.

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Introduction

Psychosocial Issues in Cancer Genetics: From the Laboratory to the Public

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Introduction

This special issue of *Cancer Epidemiology, Biomarkers & Prevention* represents diverse conceptual approaches to studying ethical, legal, and social issues surrounding cancer genetic testing. The papers in this issue represent the first joint report of studies emerging from the Cancer Genetics Studies Consortium (CGSC). These studies, funded jointly by several of the National Institutes of Health, address a range of psychosocial issues that arise as we move from the development of genetic mutation tests in the laboratory towards the use of genetic testing as a component of modern medicine. The ultimate goal of cancer genetic testing is to improve cancer prevention efforts in susceptible populations through targeted chemoprevention, improved surveillance and screening, and dissemination of information about potential benefits of prophylactic surgery. Together these studies increase our understanding of behavioral and psychosocial aspects of genetic testing for cancer and point to the gaps in our knowledge base to lead us to future treatments and prevention activities.

We reflect on the commonalities in approaches and findings as well as on the areas of divergence that emerge in these papers. We also summarize some of the policy implications and suggest areas in need of additional research. The CGSC is composed of 12 multidisciplinary teams of researchers who have come together annually since 1994 to discuss methodologic and conceptual issues, review relevant literature and

preliminary findings, and identify common research questions and issues that arose in the conduct of these studies. Disciplines represented in the CGSC include clinical and molecular genetics, epidemiology, medicine, psychology, sociology, nursing, anthropology, statistics, ethics, and law. This collaborative effort was greatly encouraged by the allocation of funding at the federal level for behavioral studies of the ethical, legal, and social implications of advances resulting from the Human Genome Project, as discussed in the Foreword to this issue. The funding for the research reported here was the direct result of the decision to advance social and psychological understanding of the human impact of genetic testing at the same time that more basic research was supported.

Common and Divergent Findings and Approaches

There are several common findings among the papers presented here. Many of the studies measured interest in obtaining genetic testing or in receiving test results. However, “interest” or “intention” was defined and measured in different ways across studies. Depending on the population studied and on the definition and measure used, the proportion of self-reported interest in genetic testing ranged from 45% (1) to over 80% (2–4). Consistent with other published studies in this area (5), uptake of cancer genetic testing was considerably lower when it was actually offered to high-risk groups (6–7). Measures of interest reported here included: *a*) a rating in response to a hypothetical question on a questionnaire, “How interested are you in being tested for . . . ?” (2); *b*) willingness to attend counseling sessions as a possible precursor to testing (7); *c*) willingness to involve an affected relative in testing (7); *d*) willingness to give a blood sample for possible future use in genetic testing (8); and *e*) intention to learn one’s test results after having given blood for testing (4). These studies begin to define why some people are motivated for testing [e.g., for family benefit (4) or altruistic reasons (7)] and why others are inhibited from seeking testing despite high medical risk (7). In general, knowledge of being at increased genetic risk does not immediately translate into a

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desire to take all possible actions to increase understanding of that risk.

Given the high level of self-reported interest, these findings underscore the importance of consumer education as cancer genetic testing finds its way into clinical medicine. Counseling and/or educational efforts should include educating individuals about their medical risk status, providing accurate information about current test characteristics of genetic mutation testing for cancer risk, and encouraging regular screening and other self-care behaviors among persons at all levels of risk. However, the studies reported here also show that demographic, psychological, personality, and cultural characteristics affect decision making about genetic testing. For example, Lerman et al. (8) found that genetic counseling did not diminish motivation for *BRCA1* testing in low-to-moderate risk women, indicating that more emphasis in counseling or education sessions may need to be placed on reducing psychological anxiety and distress.

Several studies point to the predominance of psychological variables rather than medical factors in influencing individuals' decisions about genetic testing. Perceived risk and worry about cancer were common predictors of both interest and of uptake across studies (1–4, 6, 7). These findings contrast with the lack of importance accorded by patients to quantitative assessment of future cancer risk, often defined as family history or high scores on a risk algorithm (2, 3). In contrast, medical and genetic professionals make decisions about the appropriateness of testing for an individual on the basis of medical factors (e.g., family history, risk analysis). This discrepancy may lead to difficulties in communication between patients and providers about the appropriateness of testing. These findings strongly suggest that emotions and self-perception combine in a range of ways to affect what had been thought to be a largely medical decision.

Several of the papers studied race or ethnicity and culture as a predictor of interest or uptake. These studies in general found that race/ethnicity or cultural identity was a significant predictor of interest in testing (1, 2, 8). In general, groups outside the dominant culture (e.g., African Americans, lesbians) expressed less interest in testing. In the one study evaluating genetic counseling for breast cancer risk (8), African American women were less likely to complete an educational intervention and were less likely to provide a blood sample than were Caucasian women. Differences in cultural subgroups in use of medical tests and innovations have been shown previously in mammography uptake (9), in participation in clinical treatment trials (10), and in the use of other preventive services and care options (11). Understanding the interrelationships among objective risk, subjective risk, cultural values, and intention to seek genetic testing will be important in developing appropriate educational materials for members of cultural minorities and in maintaining access to services for members of diverse cultural groups.

An important commonality that emerged from several of the papers is the complexity of family structure and interaction patterns as they relate to cancer genetic testing. In one study (11), reactions to learning the results of one's mutation test were dependent on gender and on the carrier status of siblings. In another study of *BRCA1* testing (7), attendance at a counseling session varied by whether the invitee had a daughter. Family support predicted interest in genetic mutation testing (1). In the study reported by Peterson and colleagues (3), a relatively high proportion of individuals did not provide contact information for a first-degree relative. Finally, the complexity of developing a tool for measuring family-related constructs

(12) indicated both the challenge of doing research in this area and the need for such efforts. Many assumptions are made about the importance and influence of family dynamics during genetic counseling and testing for cancer risk, but most of these assumptions have not been evaluated. The role of family dynamics needs to be studied systematically in order to conduct counseling and testing in ways that maximize psychological adjustment of family members.

One of the differences among the papers is the composition of the study populations, which showed considerable variation in cancer risk. Investigators studied patients with cancer (4), individuals with strong family histories (6, 11, 12), first-degree relatives of cancer patients (1, 3), invitees to a high-risk clinic (7–8), and groups recruited from the general population (2). This diversity underscores the need to better understand the determinants of interest in and the reactions to genetic testing in well-defined groups, including cancer patients, persons with high and low familial risk, and persons with no known cancer risk factors. The description of the recruitment process varied considerably from study to study, as did the description of retention in prospective studies. Persons were recruited in a variety of ways from many different sources. In prospective studies, participants could withdraw at different points during the study. For example, in studies that offered genetic testing (3, 4, 6, 11), eligible subjects could decline initial participation, decline to be tested after an initial counseling session, decline to learn their test results, or decline follow-up interviews after disclosure of test results. Inclusion of flow diagrams showing the number of eligible persons approached for the study and their status during the course of the study improves clarity, aids in interpretation of the results, and facilitates comparison across studies. A systematic description of recruitment and retention should be reported with as much rigor as we report other findings if we are to increase our understanding of the demographic and psychosocial characteristics of persons who decline and accept genetic testing.

Why Are These Findings Important?

The studies reported here provide data that can help guide clinical medicine and, ultimately, public health practice for cancer genetic counseling and testing. Collectively, these findings strongly support the view that a strictly educational approach to genetic counseling and testing for cancer is not sufficient. A counseling process that takes into consideration an individual's demographic, psychological, and cultural profile is necessary to facilitate informed decision making and maximize patient satisfaction with potentially irreversible decisions. The findings also raise an issue for health care policy makers. As genetic testing becomes more integrated into clinical practice, it will be even more important to understand the characteristics of those who do and do not seek cancer genetic counseling and testing and the impact of testing on subsequent preventive health behaviors like cancer screening. If we understand what individuals want to know about cancer genetics and what, if anything, they are willing to change about their behavior in order to reduce their cancer risk, we will be better able to advise health care professionals and those who plan genetic services about how to structure and offer services in ways that reduce, rather than increase, a patient's anxiety. By understanding more about those who do and do not wish to acquire genetic knowledge, we can ensure that programs of mandatory testing are available that minimize psychological distress. Better understanding of the cultural biases and the role of psychological factors such as denial or resistance will enable us to develop

educational materials and programs that speak to patients literally and figuratively in the languages they understand. Being able to measure processes that are relevant to the flow of genetic information through families and to understand the ways in which familial roles affect motivation and use of genetic testing will permit the development of effective, tailored educational materials and counseling methods. In a similar vein, understanding that health care professionals and patients may use different parameters in considering the appropriateness of testing will help us improve professional education about patient decision making for genetic testing. As a consequence of this increased understanding, our ability to provide truly informed consent may improve.

Areas for Future Research

Research in this area is still in its early stages; as such, these articles represent the state of the art in research and at the same time point out the gaps in our knowledge. Future research should be conducted on a range of study populations, e.g., defined by age, risk status, type of cancer. Research also should include diverse endpoints, including quality of life, participant-family interactions, patient-provider relationships, and effects on health and health behaviors. To fully inform prospective consumers, we must be able to offer accurate data about how distressing or helpful participation in genetic testing is and who is most at risk for adverse psychological effects. The research should be longitudinal in nature, to allow for the assessment of both short and long-term effects. Finally, we should begin to assess the incorporation of this new technology into the health care delivery system as it becomes appropriate, including the effects on providers and medical practice, payment for services, and treatment and prevention options and choices.

Issues Raised for Future Research Activity

From these projects and from future efforts, we will obtain needed answers to research questions that can be used to guide clinical medicine and public health policy on genetic testing. The field of psychosocial research in cancer genetics must develop simultaneously with molecular biology and must take into consideration advances in genetic knowledge. As our knowledge about human genetics increases, its potential impact on human behavior also increases. These papers point to a need to increase psychological training for those providing genetic services and the need to incorporate psychological services into genetics clinics. Conversely, increased knowledge about human behavior in the face of advancing genetic possibilities will maximize the utility and application of scientific progress. The recently established National Cancer Institute's Cancer Genetics Network may provide an infrastructure through which population-based interdisciplinary studies of psychosocial issues related to cancer genetic testing can be conducted.

One issue that these papers highlight is the absence of a

national standard for what constitutes an appropriate level of risk for cancer genetic testing. Individual health groups, such as Kaiser Permanente, are creating policy to address this gap, generally approving testing only in the presence of strong family history. Genetic testing is available publicly, however, through primary care providers and oncology offices. Some companies have marketed genetic tests directly to the public. The lack of medical consensus and clarity about appropriate use of testing, and public demand fueled by such publicity, by anxiety about cancer, and by easy access may lead to confusion or inappropriate use of genetic mutation testing. Clearly defined criteria for inclusion in a study population and careful consideration of the generalizability of the results is particularly important in psychosocial studies of genetic testing.

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BLOOD CANCER DISCOVERY

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