Controversies in Communication of Genetic Screening Results for Cancer: A Report from the American Society of Preventive Oncology's Screening Special Interest Group (ASPO's 33rd Annual Meeting, March 8 to 10, 2009, Tampa, Florida)

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Introduction

Genetic screening for cancer susceptibility for many cancer sites has become routine for high-risk populations, particularly breast, ovarian, and colon cancer. Such genetic tests hold great promise for informing tailored cancer prevention and risk reduction recommendations. Heightened consumer awareness of “cancer genes” and tests to detect them has escalated interest and demand for cancer risk assessment, generating a pressing need to identify effective, efficient methods of communicating complicated genetic information to patients and their potentially at-risk relatives. As more is learned about cancer genetic susceptibility, population-level demand for genetic screening will necessitate considering new modes and methods for effective genetic risk communication other than traditional in-person genetic counseling. Optimal outcomes of predictive genetic testing for cancer include adherence to preventive or risk-reducing strategies, accurate communication of test results to at-risk relatives, maximization of psychosocial well-being, and ultimately, reduction of morbidity and mortality associated with hereditary or familial cancers. This will require the effective communication of risk information tailored to patients’ translation of knowledge into perceptions of cancer risk, and the biopsychosocial factors that mediate and moderate that translation into risk-reduction behaviors.

Two very important issues in the effective communication of genetic screening results include (a) the mode of communication and (b) the age of subjects who receive risk information. Thus, the purpose of this session was to review the state of knowledge and to identify the future research necessary to inform policy and practice about testing for, and communication of, cancer risk information to adults and minors.

The example used in this session was BRCA1/2 testing, given its widespread use at the population-level in adults.

Optimal Method of Communication of Genetic Test Results: In Person, Telephone, Web?

The American Society of Clinical Oncology and other professional societies recommend that predictive genetic testing be paired with pretest and posttest counseling (1) to optimize patients’ informed consent, understanding, and processing of the implications of test results (1, 2). Given the complexity of genetic information, the potential for false reassurance, psychosocial sequellae, and limited interventions to modify identified risk for cancer, communication of predictive genetic information has traditionally been conducted in-person by health care professionals trained in clinical genetics. Future demand for cancer risk assessment and predictive genetic testing might surpass the availability of cancer genetic specialists, suggesting a need for alternative, more efficient methods of service delivery (1, 3).

Historically, prenatal genetic counselors incorporated telephone communication of teratogenic information into counseling services (4, 5), suggesting one model to extend genetic services. Telephones, computers, and audio-visual equipment could be used to provide genetic services in which geographic or socioeconomic factors or consumer demand have limited access to in-person services (3, 6-11). Interviews with patients and providers suggest that they perceive similarly the potential advantages and disadvantages of telephone disclosure of genetic risk information (12). Genetic counselors and patients most frequently cite patient convenience and lack of nonverbal communication as key potential advantages and disadvantages of telephone communication. Although 98% of genetic counselors had provided some telephone disclosure, their comfort varied widely by test result; 33% reported experiences that made them question telephone disclosure. Only 50% of patients (ages 22-70 years) awaiting BRCA1/2 test results were interested in receiving their results by telephone (12). Of those interviewed after receiving their test results in-person (ages 21-84

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year), only 35% reported that they would have been interested in receiving their results by telephone. Like genetic counselors, patient interest varied widely by test result, with those receiving positive results least likely to favor telephone disclosure (12).

Systematic evaluation of telephone/internet versus in-person communication of components of genetic services has been limited to small select populations, or women who self-selected their mode of communication (13-15). Other technologies, such as Web-enabled platforms and video conferencing, may overcome some perceived limitations of telephone communication, such as providers’ and patients’ loss of visual cues (12). However, using novel technologies might limit, rather than improve, access for some, including underserved patients (16). Thus, randomized studies of the mode of pretest counseling and/or disclosure of genetic test results assessing their psychosocial and behavioral outcomes (e.g., screening, risk reduction behaviors, and communication to family members) must be conducted before these procedures become fully incorporated into clinical care models (12, 13). Further, evaluation of biopsychosocial factors that mediate and moderate psychosocial and behavioral outcomes will inform the development of communication interventions tailored to optimize patients’ outcomes.

Communication of Genetic Risk Information to Minors: Helpful or Harmful?

Although patients often share genetic information with at-risk adult relatives (17-21), there is an ongoing debate over the value of sharing genetic risk information with, and offering genetic testing to, at-risk minors (22-28). For example in breast/ovarian cancer, risk reduction options for mutation carriers (prophylactic surgeries, heightened surveillance, and/or chemoprevention) are generally not recommended until age 25 years, and thus, genetic testing is not currently recommended until age 25 years (29, 30). There is no known medical benefit to communicating risk to at-risk minors. Virtually all professional societies recommend against offering BRCA1/2 testing to children under age 18 years (1, 31-36). Yet, professional recommendations neither preclude, nor recommend against, the communication of familial risk to children and adolescents. There is evidence that at least 50% of parents share their genetic test results and information about familial risk for cancer with minor at-risk children (37-39). Offspring understanding and biopsychosocial responses to early communication of familial risk remain understudied/unknown (40-42), although a small study of adult offspring who learned of their risk during adolescence or early adulthood suggests that many offspring may understand the parent communicated risk, and may modify their health behaviors (that is, stop smoking, improve diet, increase physical activity; ref. 42).

Recent evaluation of semistructured interviews with 163 parents who had BRCA1/2 testing and had an offspring under age 25 years at testing reveals that the majority of parents (66%) share their test results with at least one offspring. Sixty-two percent of offspring (201 of 323) learned of their parent’s genetic test result. Communication was associated with older child age (P < 0.001) and parent having a negative test result (P = 0.056). Few parents reported offspring anxiety or distress (19%) following disclosure (38 of 201 offspring), and fewer reported that their child did not understand (11%) or expressed denial, fatalism, or skepticism (3%; ref. 43). Thus, existing and emerging data support further evaluation of the content of parental communication (that is, what parents share), and direct evaluation of offspring understanding and perceptions of risk and performance of health behaviors, to better understand the risks and benefits of early disclosure and nondisclosure of genetic risk throughout child and adolescent development.

Many health and risk behaviors that affect the morbidity and mortality of youths and adults begin in (e.g., tobacco and alcohol use, and sexual activity) or become established in (e.g., diet and exercise) adolescence (44-46). Thus, early communication of hereditary risk might create a “teachable moment,” an opportunity to facilitate health behaviors and eliminate risk behaviors in adolescence and throughout the life span (42, 46). Additionally, there are ongoing breast cancer awareness education programs in secondary school and college, reaching girls at high risk and population risk for breast cancer (47, 48). What children, adolescents, and young adults understand of cancer as a genetic disease and predictive cancer genetic testing; what they perceive of the relevance of this information for themselves; how they respond to and use this information; and how those processes change into, during, and out of adolescence has not been well described. Further understanding of how genetic susceptibility may interact with environmental determinants even early in life will be crucial in the effective communication of genetic screening results, particularly in minors when healthy life-style choices are typically patterned (18, 49, 50). Thus, this research has the potential to effectively extend the prevention of breast/ovarian cancer and other adult-onset diseases, reducing morbidity and mortality throughout the life course and across the population.

Recommendations for Future Research

Increased availability and heightened consumer awareness of “cancer genes” and testing has increased consumer interest in, and demand for, hereditary cancer risk assessment and thus identified a pressing need for providers and policy makers to ensure effective, efficient, ethical methods of communicating complicated genetic information to patients and their potentially at-risk relatives. Optimizing adherence to risk management strategies, psychosocial well-being, and communication to at-risk family members requires an understanding of the patients’ translation of that information into personalized
perceptions of disease risk and intervention benefit. Further research evaluating the risks and benefits of novel methods of providing genetic services and early communication of genetic risk to minor offspring is needed to inform guidelines about effective, ethical delivery of genetic services for cancer susceptibility.

Given the likelihood of increased use of genetic testing by all ages, increased demand for the service of genetic counselors requires more research into effective, efficient, and ethical communication. In addition, even if guidelines recommend against genetic screening of minors for disease susceptibility for adult-onset cancers such as breast/ovarian cancer, many minors are aware of the role of genes in their cancer risk. Thus, effective, ethical communication of cancer risk, risk reduction, and cancer prevention strategies to minors requires thoughtful multidisciplinary research to inform policy and practice.

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