Psychosocial aspects of genetic testing

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Purpose of review

With rapid advances in genetic testing for disease susceptibility, behavioral medicine faces significant challenges in identifying likely patterns of use, how individuals interpret test results, and psychosocial and health impacts of testing. We review recent research on these psychosocial aspects of genetic testing for disease risk.

Recent findings

Individuals exhibit limited sensitivity in their perceptions of genetic risk information, and mental representations of disease risk appear to guide testing perceptions and behavioral responses. Motivations to undergo testing are complex, and efforts to develop decision aids are underway. Findings on psychological and behavioral impacts of genetic testing vary markedly, with some evidence of minimal or positive effects and other evidence indicating negative consequences that may be undetectable using common measures of general well being. Recent evidence suggests that genetic risk information can motivate health behavior change. Research demonstrates wide-ranging influences of testing on family dynamics, and use of genetic testing with children is of increasing concern.

Summary

More research is needed to determine how to structure health communications and counseling to motivate informed use, promote positive responses, and optimize behavior change. Given the ramifications of genetic information for families, personalized genomics will demand a shift toward a family-based healthcare model.

Keywords

cancer, family communication, genetic testing, psychological and behavioral outcomes, risk perception

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Introduction

Advances in medical genetics promise to transform healthcare, particularly through the availability of genetic tests for disease susceptibility. Knowledge of the psychosocial ramifications of genetic testing is needed to develop health communications about genetic tests, counseling services, and interventions for helping individuals and their families cope with genetic risk information. In this article, we review recent research on psychosocial aspects of genetic testing for susceptibility to chronic diseases. These issues include comprehension of genetic risk information and factors influencing testing decisions, adjustment and behavior change following testing, family communication and dynamics, and use of genetic testing with children.

The promises and perils of genetic testing: an overview

Developments in genetic testing are motivated by expectations that genetic information will be used by healthcare providers and individuals to facilitate healthcare decisions, personalize treatments, and motivate protective behaviors. Yet, there are concerns about the extent to which individuals make informed decisions about their use by taking into account likely negative consequences and limits in treatments, comprehend the meaning of test results, and respond adaptively to testing. Moreover, genetic risk information carries significant ramifications for family members, and so medical practice will need to adopt a more familial focus to healthcare. Understanding how families communicate genetic risk information, contribute to testing decisions, and react to test results will guide the evolution of a more family-based healthcare model.

Our review is complemented by three recent reviews and commentaries on psychosocial aspects of genetic testing. Scheuner *et al.* [1••] provide an overview of psychosocial outcomes of genetic testing, consumer information needs, and challenges in integrating genetic testing services into clinical practice. Hay *et al.* [2•] present a theoretical framework for anticipating and addressing the psychosocial challenges in public health genomics,

with the aim of guiding research and interventions for enhancing informed use and adaptive responses. Matloff and Caplan [3] describe trends in direct-to-consumer advertising of genetic testing for BRCA1 and BRCA2 mutations conferring high breast cancer risk, in which they highlight the need for policy to guide standards for accurate marketing, dissemination of information by public health sources, and continuing education for healthcare professionals.

Understanding genetic risk information

Owing to the limited penetrance of most mutations conferring disease risk, most genetic assessments provide probabilistic rather than deterministic information about disease development. Of critical interest is identifying how individuals perceive and respond to probabilistic information about genetic risk, and how cognitions (e.g. beliefs about the disease) and affect (e.g. worry and anxiety) influence perceptions of genetic risk information. Evidence indicates that individuals have limited sensitivity to genetic risk information. Rather than perceiving differences between tests conferring different risk levels (e.g. 40 versus 70% risk), individuals tend to extract the 'gist' of the probability as simply indicating low, moderate, or high risk [4•]. Interestingly, perceptions exhibit a '50% blip' effect such that appraisals of worry and distress induced by mutations are greater for tests conferring a 50% risk (and thus high ambiguity) than for tests conferring 40% or less risk or 60% risk. These nonlinear patterns of risk perceptions and affective appraisals in response to genetic risk probabilities suggest biases in interpretations of genetic risk information and a need to understand how individuals perceive and mentally represent genetic risk.

Recent research on mental representations of disease risk has been guided by the Common-Sense Model (CSM) [5], which delineates key attributes (i.e. beliefs about identity, timeline, control, consequences, cause, and coherence) and how they interact with disease-related affect to guide responses. For example, a study of individuals with genetic risk for venous thrombosis revealed how representations guide responses to genetic risk [6]. Beliefs about illness identity (identifying symptom experiences associated with venous thrombosis), coherence (beliefs that one has a clear understanding of venous thrombosis), and treatment control (beliefs that venous thrombosis cannot be controlled through medical treatment) independently predicted higher risk appraisals, whereas beliefs about identity and consequences of venous thrombosis independently predicted higher worry.

Longitudinal studies also provide insights into the role of representational beliefs in genetic testing experiences [7]. For example, evidence suggests that causal beliefs may mediate genetic testing effects on protective behavior [8•]. Because genetic testing may strengthen beliefs in genetic causes of a disease (e.g. heart disease), individuals with mutations may be motivated to use biologically based protective measures (e.g. medication use or surgery) rather than behavioral measures (e.g. dietary changes or physical activity). Consistent with this hypothesis, a study of individuals randomized to receive either genetic or nongenetic diagnoses of familial hypercholesterolemia revealed that genetic causal beliefs were associated with greater perceived effectiveness of medication for reducing risk which, in turn, predicted better medication adherence; in contrast, behavioral causal beliefs were associated with greater perceived effectiveness of dietary practices in reducing risk. These and other findings identifying the contents and influences of genetic risk representations can assist in developing counseling and health communications so that they address aspects of risk that are meaningful to individuals and thus promote informed decisions about testing and adaptive responses to tests.

Making decisions about genetic testing

Research assessing the motivations influencing genetic testing decisions confirms that a desire to increase one's certainty of disease risk is a key motivator for testing [9••]. Other important motivators include desires to find out if one's children have increased risk, to further research on genetic testing, to know if more screening is needed, and to act on the recommendations of a genetic counselor or doctor [9••]. Counterbalancing these motivations are concerns over losing health insurance and the psychological consequences of testing [10,11]. Such concerns are not unwarranted; for example, many who have tested positive for risk of Huntington's disease report concerns about discrimination from insurers and employers as well as social stigma from others [12].

The complexity of genetic testing decisions has prompted efforts to develop decision aids. Of particular promise are computer-based, usually web-based, interactive information packages that allow tailored searches for information. Internet-based decision aids may be particularly useful in light of the need to assist consumers purchasing genetic tests through private, Internet-based companies. A few such packages now exist [13], but more programs are needed to address the demand created by the wide variety of genetic tests now available and the need for culturally appropriate versions for use worldwide.

Adjustment and behavior change after testing

The psychological and behavioral impact of genetic testing is a continuing concern in the research area. Studies have yielded mixed results. Empirical evidence

from a systematic review [14•] and an independent study [15] shows that carriers of mutations for breast and ovarian cancer, hereditary nonpolyposis colorectal carcinoma (HNPCC), and Alzheimer's disease experienced only short-term or no increases in general anxiety or depression, even 3 years after testing. High rates of appropriate screening were also observed for both carriers and noncarriers. Conversely, a prospective study [16] found that carriers of mutations for long QT syndrome (a life-threatening but treatable cardiac arrhythmia syndrome) experienced heightened levels of disease-related anxiety for the 18 months following the receipt of test results. Partners of carriers (relative to partners of noncarriers) also reported greater anxiety over the 18 months.

Recent qualitative studies shed some light on these empirical inconsistencies, as they revealed complex and nuanced responses to genetic testing that may not be detected using standard indices of distress and adjustment (e.g. measures of general anxiety and depression). For example, individuals who underwent testing for HNPCC described feelings of guilt and injustice, fear of cancer when new symptoms arise, use of emotion regulation strategies such as denial and immersion into work activities, difficulties in communicating the news to relatives, and uncertainty and worries about the future [17••]. Individuals receiving negative results also reported difficulties adjusting from a self-identity as someone with likely genetic risk to a self-identity of someone without genetic risk. Complex ramifications for family dynamics were also observed, suggesting that mutation-negative individuals may need additional psychosocial support (potentially with a family systems focus) in coming to terms with their status [18].

Genetic risk information can yield psychological benefits, with individuals often reporting such changes as a greater appreciation of life and adoption of realistic goals [17••,19•]. Potential benefits are also indicated by a trial evaluating the impact of information about genetic determinants of obesity on responses by individuals to an obesity management consultation [20••]. Relative to the consultation without genetics information, the consultation with the genetics information was viewed as more beneficial, had no negative effects on self-efficacy or body weight over the next 6 months, and led to greater improvements in negative mood for those reporting a family history of obesity. In contrast, the consultation without the genetic information led to greater improvements in negative mood for individuals reporting no family history. These findings suggest the utility of tailoring obesity management consultations according to family history so that the genetics information is consistent with one's representation of familial risk.

Aside from general trends in reactions to genetic testing, some individuals clearly are at greater risk for distress than others [7]. A prospective study, guided by the CSM, assessed risk representations of individuals undergoing genetic testing (for BRCA1/2 or HNPCC mutations) and found that representational beliefs, particularly coherence and genetic-based causal beliefs, were linked with coping and distress [21]. Assessments of representations may aid in identifying individuals who have difficulty coping with test results and identify cognitive appraisals that may be targeted by counseling and psychoeducational communications.

A key premise to promoting genetic tests is that results will motivate protective behaviors and, in turn, reduce morbidity and mortality. To date, findings on the behavioral impact of genetic testing remain limited, but the evidence base is growing. For instance, a trial assessing the impact of genetic testing for Alzheimer's disease showed that, among individuals undergoing the genetic test, changes in behavior thought to be protective (e.g. diet, exercise, and vitamin use) were greater among those with the genetic risk than among those found not to have the genetic risk [22•]. In another trial, smokers receiving results indicating high genetic risk for lung cancer reported greater motivation to quit smoking and smoked fewer cigarettes over the following week relative to the smokers who did not receive genetic testing [23•]. Although the sample was small and group differences disappeared after 2 months, this exploratory study demonstrated that this test is feasible and acceptable for larger randomized controlled trials.

The need for evidence-based counseling and information services providing risk assessment, education, and support has been recognized since the advent of genetic testing; yet, few empirical evaluations of such interventions have been reported. A Cochrane review identified only three trials assessing genetic risk assessment services for individuals with perceived risk of breast cancer [24•]. Although these trials provided promising support for the efficacy of genetic counseling, the paucity of studies underscores the need for further trials to identify the optimal structure and contents of these services and their impact on behavior, health, and well being over time. A descriptive study of a 1-day retreat provides initial evidence that it may be an efficient means for providing such support [25]; however, a controlled trial is needed to fully ascertain its efficacy.

Family considerations and communications

In light of the familial dynamics involved with managing genetic risk, research is extending beyond the individual to consider the impact of genetic testing on the family. Research questions include when and how genetic testing information is communicated within the family and how genetic knowledge influences the lives of family

members. In the arena of testing for BRCA1/BRCA2 mutations, individuals have reported strong inclinations to communicate results to their family, with 98% intending to tell some relatives and 63% intending to tell all relatives [26•]. Sharing results with children can pose considerable difficulty, however. In one study, only one-half of BRCA mutation carriers disclosed the results to their children, and disclosure was more likely as children got older [27•]. Mothers expressed a strong need for (currently lacking) resources to aid in decisions about disclosure to children, with preferences for literature and meetings with a family counselor or other parents who had undergone testing [28•].

Other studies highlight the strain placed on family members by knowledge of genetic disease risk. Poignant evidence is provided by reports from parents of individuals with 22q11DS, who are at risk for heart defects, psychotic illness (particularly schizophrenia and bipolar disorder), and other disorders [29]. Knowing about the predisposition created considerable anxiety for parents, particularly owing to the risk of psychiatric disease, the burden of uncertainty about whether any conditions will develop, and worries about stigma due to the psychiatric risks. Of particular concern to parents was that most learned about the psychiatric risks not from their doctors but from the Internet and other nonmedical sources. Such information is critically important to parents eager to facilitate early diagnosis and treatment.

The stress of having a parent with genetic risk for disease is illustrated by qualitative research involving adolescents with a parent diagnosed with Huntington's disease [30•]. These adolescents described themselves as living under the shadow of Huntington's disease, waiting and watching its progression in their parent and other relatives, and feeling overwhelmed from taking adult responsibilities. They reported extensive deliberations about whether to get the genetic test; decisions about future careers, education, and having partners and children were seen as daunting in light of their risks of Huntington's disease.

Models of communal coping, which identify the linked and interactive support processes within families coping with a shared threat, provide a conceptual framework for evaluating family responses to genetic risk [31••]. In a study of communal coping, sisters with BRCA1/2 mutations were found to share similar levels of perceived risk and worry, though not for ovarian cancer. These women also reported similar levels of distress and somatization. Sisters who shared high levels of emotional support from family reported lower anxiety and somatization relative to those who did not. These findings suggest the potential utility of interventions for improving emotional support within families.

Genetic testing of children

Genetic testing of children is receiving increased empirical attention. Guidelines generally recommend that genetic testing for children should be limited to tests detecting conditions that can be medically managed soon after testing. This recommendation is made in light of the psychosocial consequences that may follow from information of risk for an incurable condition, including loss of self-esteem, anxiety, overindulgence or scapegoating by relatives, and problems with discrimination and insurance.

Genetic testing for childhood-onset diseases may confer benefits through targeted surveillance, possibly resulting in early diagnosis and treatment; however, early genetic testing may also result in parental distress. For example, one study found that mothers with newborn infants with genetic risk for type 1 diabetes reported greater worry and rumination up to 1 year later relative to mothers whose infants had low genetic risk or were not tested [32••]. Further research is needed to determine whether the heightened worry influences monitoring for disease, health behaviors, and interactions with the child and other family members.

Genetic testing for familial adenomatous polyposis (FAP), a bowel cancer predisposition syndrome, is recommended from the time of puberty and so is being offered to adolescents. Although adolescents undergoing testing have reported benefits (e.g. relief from uncertainty, greater ability to plan for the future, improved family relationships, and clarity about life priorities), they also identify negative consequences (e.g. distress over illness risk, witnessing distress in parents, friendships being affected by negative mood, and feeling distant from family members) [33•]. These and other findings on the range of testing benefits and harms for minors can assist in developing counseling protocols that help them anticipate their likely reactions and make informed decisions.

Given the rise of genetic tests for a multitude of conditions, studies assessing anticipated interest in genetic tests are important for identifying potential demand and use. For example, genetic tests for propensity of obesity are anticipated and already private companies are marketing genetic and nutritional profiling for obesity risk that can be used on children. One issue to consider is the ages at which children should be tested and then informed of the results. In one study, parents of obese children were presented with hypothetical scenarios about genetic tests for obesity [34]. Most parents (80%) felt that testing should be made available for children. Of these, 87% believed that the child should always be informed of the results, the average optimal age being 10 years.

Future research directions

Behavioral medicine faces many challenges in keeping pace with the rapid advances in genomic discoveries and genetic testing. Emerging research directions include explorations of responses to testing for multiple health conditions [35••]. Given the difficulties in understanding and managing information about genetic risk for a single condition, genetic testing for multiple conditions will create even greater challenges in terms of comprehension and adaptive responses. Another research direction is to identify strategies for communicating information about the complex relationships between genes, behaviors, and environmental factors. As risks conferred by most mutations are influenced by environmental and behavioral factors, individuals must understand how their behavior and environment can be altered so as to reduce their genetic risk. Finally, with unregulated genetic tests being sold through the Internet and marketplace, research and policy must develop and disseminate effective communications about genetic testing, interpretation of results, and recommendations for action.

Conclusion

Recent research on psychosocial aspects of genetic testing for disease risk has provided insights into the key areas of factors influencing comprehension of genetic risk information and testing decisions, adjustment and behavior change following testing, family communication and dynamics, and use of genetic testing with children. These findings can inform efforts to develop a more family-based healthcare system that can address the ramifications of genetic testing within families.

References and recommended reading

Papers of particular interest, published within the annual period of review, have been highlighted as:

- • of special interest
- •• of outstanding interest

Additional references related to this topic can also be found in the Current World Literature section in this issue (pp. 247–249).

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