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Future Health Applications of Genomics:

Priorities for Communication, Behavioral, and Social Sciences Research

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Abstract

Despite the quickening momentum of genomic discovery, the communication, behavioral, and social sciences research needed for translating this discovery into public health applications has lagged behind. The National Human Genome Research Institute held a 2-day workshop in October 2008 convening an interdisciplinary group of scientists to recommend forward-looking priorities for translational research. This research agenda would be designed to redress the top three risk factors (tobacco use, poor diet, and physical inactivity) that contribute to the four major chronic diseases (heart disease, type 2 diabetes, lung disease, and many cancers) and account for half of all deaths worldwide. Three priority research areas were identified: (1) improving the public's genetic literacy in order to enhance consumer skills; (2) gauging whether genomic information improves risk communication and adoption of healthier behaviors more than current approaches; and (3) exploring whether genomic discovery in concert with emerging technologies can elucidate new behavioral intervention targets. Important crosscutting themes also were identified, including the need to: (1) anticipate directions of genomic discovery; (2) take an agnostic scientific perspective in framing research questions asking whether genomic discovery adds value to other health promotion efforts; and (3) consider multiple levels of influence and systems that contribute to important public health problems. The priorities and themes offer a framework for a variety of stakeholders, including those who develop priorities for research funding, interdisciplinary teams

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engaged in genomics research, and policymakers grappling with how to use the products born of genomics research to address public health challenges.

Introduction

The promise that genomic discovery offers for positive clinical and public health outcomes is being heralded in high-profile public arenas.^{1,2} This acclaim is due in part to the advances in technology that allow genomic scientists to do more—faster and better at less expense^{3,4}—and to the findings from genome-wide association studies (GWAS) that are identifying an expanding list of gene variants associated with numerous common diseases. As these results appear with increasing frequency in prominent scientific journals,^{5,6} expectations are growing that personalized genomics will enable the creation and delivery of individualized prevention and treatment interventions and widespread public health benefits.^{7,8} In contrast to the heightened focus on genomic discovery, however, far less attention has been given to defining the communication, behavioral, and social sciences research required to translate genomic information into public health applications.⁹ Thus, a sizable gap exists between the scientific investment made in genomic discovery and the translational research needed to satisfy the understandably high public expectations for the health benefits such a discovery might deliver.

Defining the Translational Research Gap

For the past decade, the CDC's Office of Public Health Genomics has tracked the number of scientific publications that address the translational research gap.^{10,11} "Translational research," as defined for this report, "seeks to transfer basic science discoveries (bench) into clinical applications (bedside), prevention, and public health interventions (trench)" and, in turn, to transfer knowledge "about effective interventions of all types into successful policy and program initiatives across multiple levels of the healthcare system and society."¹² As Figure 1 illustrates, the number of publications reporting results of human genomic discoveries (e.g., gene–gene and gene–disease associations) far exceeds the number that describe research aimed to transfer these discoveries into clinical applications (e.g., evaluation of genetic tests).¹¹

A principal reason for the relative dearth of translational research is that scientific stakeholders disagree about whether genomic discovery is ready for such research. Some genetic scientists believe there is too little understanding of the complexity of the underlying biology of gene–gene and gene–environment interactions to support translational research.¹³ Scientists in epidemiology and public health contend that the small and as yet imprecise effects revealed by research on gene–gene and gene–disease associations will contribute very little to existing metrics for disease risk prediction and clinical decision making.^{14,15} Some also argue that, given limited resources, emphasis should be placed on applying effective interventions that address environmental exposures rather than pursuing genomics-informed solutions.¹⁶ Social and behavioral researchers, too, have been skeptical of translational research based on concerns that the research will shift the focus of health promotion away from known and powerful environmental, social, and behavioral influences.¹⁷ Similarly, caution has been recommended by some medical professionals^{18,19}

who find insufficient evidence that genomics-based personalized medicine will promote positive health outcomes.^{20,21}

Addressing the Gap with an Interdisciplinary Approach

To address some of the issues that underlie the translational research gap, the National Human Genome Research Institute convened a 2-day workshop in October 2008 that brought together a group of 50 scientists from the U.S. and the United Kingdom, representing a broad range of disciplines including public health; communication, behavioral, and social sciences; genetics; epidemiology; medicine; and public policy. The group was asked to recommend research priorities to evaluate potential public health applications of genomic discoveries with a focus on communication, behavioral, and social sciences research.

Asked to assume the role of “trailblazers” rather than “translators” in their approach to framing critical research priorities,²² group members were encouraged to anticipate advances in genomics and to assume that their recommendations would be carried out by interdisciplinary research teams of the future (nihroadmap.nih.gov). Discussions over the 2 days culminated in the identification of three priority areas that need translational research focused in communication, behavioral, and social sciences, and three crosscutting themes to consider in carrying out such research (Table 1). These priorities and themes are intended to provide a framework for a variety of stakeholders, including those engaged in setting priorities for research funding, interdisciplinary teams engaged in the translation of genomics research, and, ultimately, policymakers grappling with how the products born of genomics technology might address public health challenges.

Group members reviewed the current landscape of genomic research and the status of its translation into clinical and public health practice. Acknowledging the importance of rigorous studies of the clinical utility of genetic information, the group nevertheless cautioned that there were limits—and even risks—to enforcing a linear path from discovery to translation (“bench to bedside to trench” model) in which basic science discoveries are refined in highly controlled circumstances and perfected before being applied in clinical, community, or other public health settings. In this model, basic research in genetics has traditionally been the domain of bench science, whereas translational research has been the realm of communication, behavioral, and social sciences.

The concern about this divided mission is that basic research approaches will be unable to anticipate critical challenges and opportunities that come with translation of genomics information into public health practice.^{23–25} The group recommended that cross-disciplinary and multilevel collaborations be fostered among genomic discovery and dissemination researchers. A lack of collaborative, interdisciplinary, translational research explains the often premature, inefficient, and sometimes even harmful adoption of new technologies. Negative outcomes can occur when the powerful “push” forces of scientific discovery are matched with market “pull” forces without the mediating presence of a translational evidence base.²⁶ For example, in the case of direct-to-consumer personalized genomic profiles,²⁷ a better understanding of how people comprehend the limits of these profiles might have informed the marketing of these products, and perhaps even enabled

polymakers to engage sooner in a public debate about their risks and benefits. Instead, translational researchers are now playing catch-up to gather evidence about whether these tests will offer individual- or population-level health benefits or instead lead to negative consequences as has been observed with other aggressively marketed, yet not fully evaluated, technologies such as whole-body imaging.

Setting a Proactive Agenda

Attendees agreed that the answer to whether personalized genomics adds value to existing public health promotion efforts is largely unknown, and that if broad public health benefits are to be derived from genomic discovery, then translational research must be elevated to a more prominent place. Further, in order to reduce the possibility of misapplying new knowledge, the group endorsed an “inverse planning” model.²⁴ This model is characterized by an early and ongoing bidirectional exchange of knowledge between discovery and translational research. In this exchange, the acceptability, efficacy, effectiveness, cost effectiveness, and predicted public health impact of basic science findings are evaluated early and iteratively throughout the discovery process. For example, translational research could be conducted using early prototypes such as a representative set of genetic susceptibility tests that embody important characteristics of future tests—not necessarily the full set of markers envisioned for future use. These prototypes could be evaluated in comparison to other medical and public health interventions and used to assess their uptake, personal decision making, and patterns of patient–provider communication, all important to inform downstream clinical and public health policies.

The group agreed that priority should be given to research that targets the top three risk factors: tobacco use, poor diet, and physical inactivity, which contribute most to the four major chronic diseases (heart disease, type 2 diabetes, lung disease, and many cancers) that together account for half of all deaths worldwide (www.3four50.com). In the discussions, poor dietary behaviors and physical inactivity were used as examples of how genomic discovery might add value to current public health efforts to reduce obesity.²⁸ Although there is evidence of the importance of several psychological and environmental factors in the development and maintenance of a healthy diet and appropriate levels of physical activity,^{29–31} interventions on the whole have yet to produce the long-term improvements at the individual or population level needed to prevent or treat obesity.³² It is an open question as to whether—and for whom—these interventions can be improved by applying the knowledge gained in translational genomics research based on the heritability of these unhealthy behaviors or of obesity itself,^{33,34} in order to suggest appropriate behavioral innovations to help treat them.

As the group discussed, such research also might lend insight into where best to target limited health promotion resources or to focus policy development. For example, Khoury and colleagues³⁵ have suggested that family history and genetic risk assessments could be used to stratify populations into differing levels of obesity risk. Could these tools aid efforts to address macro-environmental contributors, such as the concentration of fast-food outlets in low-income areas, the relatively higher price of quality foods such as fruits and vegetables, and national marketing campaigns of high-calorie foods? Perhaps awareness of

genetic risk could galvanize genetic identity groups (those at risk) in ways that prompt community engagement and empowerment to influence social environmental exposures with an impact beyond those of current community organizing strategies.

To help close the translational research gap, the group outlined several priority areas and crosscutting themes. Table 2 frames examples of the research questions found at the nexus of these priorities and themes.

Priority Areas in Communication, Behavioral, and Social Sciences Research

Public Understanding and Use of Genomic Information

Genetic information is complicated, and explanations of causative factors in disease development are becoming increasingly complex. For example, what is the best way to convey to the public concepts such as pleiotropy, in which a single gene variant can have multiple disease manifestations or even be protective for one disease and increase risk for another? Will explanations about gene–environment processes, in which environments turn on or off gene expression in some individuals, be accepted equally by different constituencies, and how might this new knowledge influence life choices and behaviors? Research also is showing differences in public interpretation of concepts such as gene–environment interaction. Some interpret this concept as meaning that behavior is the major contributor to health outcomes; others see the two as working together in an additive relationship (i.e., genes plus behavior).³⁶ These interpretations will likely be sensitive to differences in how the information is conveyed.³⁷ However, it is unclear how this understanding will influence uptake and use of, for example, genetic testing. Well-documented tendencies of individuals and groups toward positively or negatively biased interpretations of risk communication also will likely influence understanding and application of genomic discoveries.^{38–40}

Advances such as full-genome sequencing products from which individuals can obtain a complete description of their genome⁴ are anticipated in the public arena. These products will provide ever-larger amounts of personal genomic information with unknown implications for health outcomes. To date, little consideration has been given to whether and how such information might be used by individuals, healthcare providers, program planners, or policymakers. Thus, research on the applications of genomic discovery should give priority to understanding these groups' expectations, perceived benefits and harms, and capabilities of full-genome sequencing and other future genome products. For example, communication research such as surveillance of web-based discourse; population attitudes; developments in professional training; credentialing developments; and mass media promotions could be used to monitor trends, recognize milestones in public awareness, and assess uptake of and response to new genomic information.

Direct-to-consumer marketing of genetic tests also offers a context for several research questions. What systems and tools (e.g., web-based, print-based, numeric or descriptive risk communication) are needed to help targeted populations interpret and use genetic information that is based on multiple gene variants for multiple health conditions? Further,

what might be the intended and unintended uses of these tests for social groups based on age, ethnicity, geographic locale, and literacy level?

Communication theory suggests that the medium of delivery (newspaper, TV, Internet, healthcare providers) and characteristics of both the communicator and the recipient will influence individual, community, and social responses. The Internet, now a well-accepted source for highly customized health-related information and social networking, will likely predominate as a communication channel for genomic information. However, the difficulties of evaluating the quality and accuracy of information on the Internet are well known, and this is compounded for esoteric subjects such as genetics that are laden with unfamiliar jargon and complicated concepts. What will individuals and society need in order to develop the genetic literacy sufficient for ensuring balanced consideration of current and anticipated genomic discovery?³⁸ Social context, often characterized by close family and friends, as well as interpersonal communication, also influences interpretations of mass communication.⁴¹ Because family members share genetic profiles and environmental exposures, social context becomes particularly relevant for understanding the effects of genomic communication.

Empirical evidence may contradict popular assumptions about how different constituencies will receive and use technologies. For example, most early speculations assumed that public uptake of information about genetics would lead to simple and overly deterministic associations between genes and health.^{42,43} Research on lay understanding of health has shown that most Western populations realize that health is a product of variable inputs of both personal behavior and familial inheritance.^{36,44–51} Evidence supports the public's nuanced understanding of the role of genetics in health that varies based on the character of the disease,^{46,49} the family's experience with the disease,^{52,53} the character of the treatments available, the pathways by which the genes act,⁵⁴ and the communication approach used to present genetic risk and behavioral options.³⁷ Research to identify these human factors more precisely is essential if genomic discoveries and their applications are to be most useful and least harmful. Appreciating the import of these human factors need not wait on understanding the specificity of the biological information conveyed in genetic tests.

Potential for Genomics to Improve Risk Communication and Health Behavior Interventions

Some predict that the potential to personalize disease prevention and treatment afforded by genomics will be more powerful than other biomarker feedback, such as blood pressure or lipid levels.⁷ Moreover, personalization is being advocated as a potent motivational tool for promoting behavior change to modify lifestyles and for adopting new medical therapies. However, these predictions are based on studies of families carrying single gene mutations strongly associated with disease risk, or on self-selected populations, potentially limiting their external validity.⁵⁵ Screening behaviors have been the primary outcomes explored with these high-risk families, with appropriate shifts in screening following feedback regarding mutation status.⁵⁶ But the influence of genomic information on health behaviors that require sustained changes, such as those related to tobacco use, diet, and exercise, have not been studied, except when the healthier behaviors occurred serendipitously.⁵⁷ It is reasonable to hypothesize that genomic information alone may not improve these behaviors given the

remarkable consistency found in psychological responses to genetic tests—increased negative emotions immediately following testing that taper off in a 12-month time frame.^{56–65}

Studies exploring genetic testing for common disease susceptibility, associated with only slight to moderate increases in risk, have relied almost exclusively on hypothetical scenarios in which individuals are asked to imagine testing and, in turn, predict the impact it might have on their health behaviors.^{66–72} Future research needs to evaluate the effect of providing actual test results to individuals or groups in different testing scenarios such as clinical or direct-to-consumer settings.⁷³ Behavioral research suggests that individual risk communications as a singular intervention strategy will be slow to yield the sustained improvements in health habits (for example, physical activity) that are necessary to reduce disease risk.⁷⁴ Thus, with epidemics such as obesity, genomic discoveries likely will become one part of an overall systems approach to achieve individual- and population-level improvements in health outcomes.

Social and psychological theory can be used to frame these research questions and hypotheses, and accepted gold-standard behavioral outcomes can be used as end points. Consideration should be given to the mechanisms through which genetic communication might influence health behaviors. Genetic-risk feedback might have its greatest influence on intermediate factors such as emotional responses (e.g., fear or worry), motivation to seek formal interventions, self-confidence to adhere to an intervention (self-efficacy), or beliefs that interventions will or will not be effective (response efficacy). Whether and how genetic information might add positively to existing interventions or produce counterproductive responses is still largely unknown.

The opportunity genomics may offer for influencing behavior change beyond the individual is largely unexplored. Genomic risk information represents shared risk that may influence communication flow within social networks⁷⁵ and stimulate behavior change and improved health outcomes within a network such as a household or family. Interdependence theory suggests that providing genomic information could increase behavior change in the broader kinship network by fostering “communal coping.”^{76–78} In this model, interventions that increase awareness of genetically based risk for disease within a family, provided in tandem with formal behavior change interventions, might influence family members’ cognitive and emotional interpretations of what this information means for the broader kinship network. These interpretations could motivate cooperative problem-solving efforts wherein members of the kinship network cooperate in making lifestyle changes.⁷⁸

Health promotion interventions often target social arenas such as worksites, schools, churches, and housing developments. How these interventions might make use of genetic information in promoting health and wellness in these settings has not been explored. The potential for discrimination based on genetic profiles⁷⁹ suggests the need for group-level interventions that are not based solely on individualized communication of genetic information. Risk communication approaches could be tested that convey depersonalized risk communication, such as proportions of the group holding particular gene variants, with the aim of motivating group improvements in health behaviors to achieve collective benefits

(e.g., reduced healthcare premiums). These settings offer a natural context for exploring the associations between interpersonal models and system-level applications of genomic discovery.

Using Genomics and Other Emerging Technologies to Identify New Behavioral Intervention Targets and More-Sensitive Intervention Outcomes

Genomics discoveries are providing new insights into the biology underlying disease development and the acquisition and maintenance of health behaviors.^{80–85} Insight into the genetic underpinnings of behavioral phenotypes, such as eating in the absence of hunger⁸⁴ or physiologic responses to exertion,⁸⁵ could expand current conceptualizations of behavioral moderators, mediators, and outcomes in ways that yield more-sensitive and more-specific measures of environmental influences and intervention impact.

Using these discoveries to influence health behaviors might be modeled on pharmacogenomics research to apply genome-wide association study findings related to the cytochrome p450 family of genes and its influence on drug metabolism.⁸⁶ These discoveries are showing some promise for improving drug-dosing regimens and reducing adverse drug reactions. A question for communication, behavioral, and social sciences translational research is whether it is possible to identify common genetic underpinnings (e.g., dopamine receptor genes) of behaviors associated with disease risk that can help explain one of public health's most vexing problems: the co-occurrence of multiple risk behaviors such as tobacco use, poor diet, and physical inactivity.

Advances in population genetic research enabled by GWAS are also providing information about the population distributions of genetic variants associated with common diseases. Results to date suggest that the prevalence of some gene variants such as the MYH9 locus of chromosome 22, a variant strongly associated with the occurrence of complications of type 2 diabetes (such as end-stage renal disease), differs across racial and ethnic groups.⁸⁷ Although such gene–disease associations support the relevance of ancestry in health disparities, there is similarity in genetic makeup across racial and ethnic groups, reinforcing the role of environment in these disparities.^{88,89} It is therefore important to consider how such knowledge might be used to inform public policy, improve health service delivery, or educate healthcare provider groups in ways that discourage racial and ethnic profiling in health contexts and reduce entrenched health disparities.⁹⁰

Crosscutting Themes for Translational Research in Communication, Behavioral, and Social Sciences

The Need to Anticipate Directions of Genomic Discovery

Workshop participants felt that a high priority should be placed on anticipating the direction of genomic discovery 5, 10, and 25 years hence, including predictions that genomic products will become less and less expensive, and that this decreased cost will move genomic testing away from a single-nucleotide polymorphism–based marker model to a full-genome sequence model. Also likely are the discovery and application of other biomarkers based on gene expression, as well as proteomic and epigenetic profiles.⁹ Today, the relative paucity of

validated genetic markers and of confirmed association with effective interventions places personalized genomics outside the mainstream of medical care. The increasing ability to expand the scope of genomic testing, steady improvements in knowledge of the association of genomics with disease, and demonstrations of effective interventions may change this. The impact of widespread access to such testing at low cost, although difficult to predict, will almost certainly influence the healthcare delivery model⁹¹ and may prove to be a truly disruptive technology.⁹²

The Importance of Scientific Open-Mindedness

Another crosscutting issue identified by the group is the need to frame genomic translational research questions agnostically; and not to simply assume that genomics has value for public health. Rather, there is a need for rigorous and objective evaluation of the advantages and disadvantages of the application of genomics technology. This point of view aligns with recommendations that comparative effectiveness research elucidate both the positive and the negative medical and social costs of medical technologies so as to inform the evidence supporting medical care and public policy.⁹³

The Importance of Systems Thinking and Transdisciplinary Collaborations

Genomic knowledge is evolving and is being shaped in the context of broad social, cultural, and political forces. Because genetic variants do not act in isolation, personalized genomics has inherent environmental dimensions. Evaluations of its efficacy will require multilevel study designs that include biological and pharmacologic factors; the social and cultural context of people's lives; and influences of the built and natural environments.⁹⁴ Traditional settings for health-related research will not suffice for the complexity of multilevel gene–environment research. Although rhetoric to use more complex models abounds in social science literature, actual implementation of research based on ecologic and systems models is rare. Although other new approaches may be employed (e.g., grid computing), as of yet they are more familiar to researchers in other scientific domains than to communication, behavioral, and social scientists.⁹⁵

The Exposure Biology Program of the Genes, Environment and Health Initiative (GEI), launched by the NIH in 2006, has as its overarching aim a mission to promote multilevel research to understand how environmental exposures and genetic variation may interact to cause disease. The initiative (www.gei.nih.gov) is an ambitious effort to develop and validate an array of tools such as wearable biosensor dosimeters to measure real-time physiologic responses, light-sensing headsets to monitor circadian rhythms, wireless skin patch sensors for stress, and GPS-based devices that will enable precise assessments of environmental exposures in natural settings. Together with existing measures such as neighborhood-level assessments (e.g., NifETy⁹⁶) these technologies promise exciting new directions for translational research in genomics.

Conclusion

There is a need for research in how to optimize the fruits of genomic discovery for individual and population health, and this research should not progress linearly from “bench

to bedside to trench” but rather should be interactive and bidirectional. Communication, behavioral, and social scientists should collaborate with genomic researchers and other relevant scientists to help pose practical and unbiased research questions that have relevance for individuals, communities, healthcare providers, and those in the public health and policy arenas. Genomic discovery will continue to etch its influences on public understanding and perception of health and disease. A concurrent, thoughtful, and proactive agenda is needed in genomic translational research if we are to understand and shape the applications of future genomic discovery for maximum public health benefit.

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Figure 1.
Publications related to human genetics and genomics, PubMed and HuGE Navigator, 2001–2009

Table 1

Areas of emphasis for genomic translational research

Priority research areas
Public understanding and use of genomic information
Potential for genomics to improve risk communication and health behavior change
Using genomics and other emerging technologies to identify new behavioral intervention targets and more sensitive intervention outcomes
Crosscutting themes
The need to anticipate directions of genomic discovery
The importance of framing research questions based on the assumption that genomics innovation may or may not add value to either individual or population-level health outcomes
The importance of systems thinking and ecologic or multilevel modeling, and transdisciplinary collaborations

Table 2

Examples of translational research questions at the nexus of research priorities and crosscutting themes^a

Crosscutting themes			
Assertions about genomics	Research priority	Anticipate directions of discovery	Value added to existing approaches
Public lacks literacy skills to evaluate the limits and usefulness of new multigene tests	1. Public understanding and use of genomic information	How might information provided by multigene risk assessments and whole-genome scans be organized and presented to maximize understanding given the limits of information for populations with varied literacy?	How might Internet-based decision support related to new genetic tests be used to educate the public about the contributions of genetics and environment to common health conditions?
Public will overestimate the role of genetics in health outcomes in ways that diminish perceived importance of health behavior and environment		How might a lifespan approach be used to update risk assessment in step with aging and associated changes in risk behaviors (e.g., onset of overweight) and environmental exposures (e.g., relocation)?	Can education about gene-by-environment interactions be used to increase the salience of current and future behavioral and environmental risk factors for individuals?
Genomic risk information will enable personalized prevention in ways that improve primary prevention interventions	2. Improve risk communication and health behavior change interventions	How do explanations of personal, genetically based behavioral phenotypes (e.g., low metabolic response to physical activity) influence individuals' behavior change confidence and motivation?	Can interventions that capitalize on shared patterns of genetic risk within families be more effective in promoting behavior change than existing individual-based interventions?
Target groups will interpret genetic risk messages in biased ways that undermine likelihood of behavior change (e.g., fatalism, optimistic biases)		Will personal genetic testing that indicates genetic factors that make it difficult to change specific health habits negatively influence motivation and success at behavior change?	Can genetic risk profiles (e.g., based on prevalence of genetic variants in workplaces, schools, housing developments) deactivate individual-level biased responses to risk information?
Understanding genetic underpinnings of disease will enable better understanding of gene-by-environment interactions	3. Identifying new behavioral intervention targets and more sensitive intervention outcomes	How can ubiquitous technologies (e.g., ecologic momentary assessment, geotagging) be used to validate new genetic findings (e.g., eating in the absence of hunger) and reliably measure eating patterns associated with short- and long-term weight outcomes?	Do behavioral and genomic risk factors cluster within neighborhoods and communities, enabling targeted intervention at the household, neighborhood, and community levels?
Social divides in access and trust related to genetic information will accentuate current health disparities		How might information about gene-disease associations based on variants that are over-represented in specific racial or ethnic populations be used to redress health disparities?	Can information about ancestry and its association with health outcomes be used to attract hard-to-reach target groups to become involved in health promotion programs?

^a See text for elaborations on research priorities and crosscutting themes.