Advancing the Post-Genomic Era Agenda: Contributions from Public Health

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Abstract
As the science and practice of genomics increasingly turns attention to its role in common chronic illness prevention and treatment, contributions from public health become more visible. Integration of genomics into health care and public health is often framed by principles such as equity and utility, and the accompanying need to translate findings into interventions. This paper will discuss the ways the discipline of public health is in position to lead the way in promoting these principles. This can only occur with the support of an educated workforce and informed consumer base. Recommendations from the Secretary’s Advisory Committee on Genetics, Health, and Society address the issues of education and training these groups, and are summarized.

Introduction
As we settle into the second decade of this century, a shift is occurring in the science and practice of genomics. Although human genomic discoveries continue to be made at a fast pace, society is increasingly directing attention toward how to assure benefits of the discoveries are used to improve the public’s health, with a special focus on common chronic illness.

We can examine the discourse around genomics to gauge if this shift signals a fundamental change and whether we are launching a ‘post-genomic era.’ Although it is still possible to find predictions like ‘genomics will be to the 21st century what infectious disease was to the 20th century’ [1], talk in the early days of the Human Genome Project that was characterized by references to Holy Grails and tsunamis is being replaced with a more tempered rhetoric. Now we hear language that evokes images of evolution rather than revolution, characterized by prediction of ‘a progression whereby advances in genetics are integrated into medicine and public health, in a considered and gradual way, accompanied by the necessary social and ethical debate’ [2]. If we are at a place where consideration of the role of genomics within society becomes an equal priority as new scientific or technologic discoveries, it only follows that the key actors in this endeavor will also shift.

Public health practitioners are well positioned to lead efforts for an appropriate integration of genomics into health care and public health. These professionals are scattered throughout the health care system. Some have job descriptions that include genetic or genomic responsibilities, some do this in addition to other work, and still others could be doing more, but lack training, support,
and/or opportunities. There is acknowledged need to examine and expand existing roles and responsibilities, but there also may be value in re-thinking where integration occurs. Is the primary care practitioner’s office the best place for all clinical services? One might imagine with health care reform our system could be integrated with the walls between public health and primary care more porous to allow for population health interventions within public health settings. The field of public health occupies a unique position within society as a trusted institution that is in position to promote a public health genomics agenda, including participating in the ethical and social debates that surround it.

The promise that genomic discoveries will lead to interventions that promote the public’s health is subject to debate around a number of issues. One is whether genomics is likely to bring health equity. Another concerns the need for and the difficulty in establishing evidence of utility. A third is the balance between discovery and translation, which is as much about resource allocation as it is public will. Public health practitioners are a diverse lot with wide-ranging skills and knowledge that prepare them to address these issues in collaboration with the communities they serve. As such they represent an untapped reservoir for moving genomics forward and promoting the principles of equity, utility, and translation.

Examination of each of these principles provides an opportunity to study the impact of genomics on society. This paper will discuss the richness of the discipline of public health that makes it a key stakeholder in this endeavor. Advancing genomics for this next stage can only occur with the support of an educated workforce and an informed consumer base. Recently released recommendations from the Secretary’s Advisory Committee on Genetics, Health, and Society address this need and are summarized in this paper.

The Path toward Equity, Utility, and Translation

The three aims of equity, utility, and translation are interdependent and synergistic. It is not realistic to work for one while ignoring the others. Applying genomic science and technology towards the cause of reducing health inequities fits into the larger paradigm of health disparity research. The elimination of health disparities among racial and ethnic minority and medically underserved communities continues to be priority in the US [3]. Not surprisingly, genomic supporters are challenged to demonstrate how resources devoted to genomics address disparities in health versus serving to widen the technology gap between rich and poor [4]. These questions reach beyond national borders, and in 2000 the former Director of the World Health Organization stated that there is potential for genomic science to improve global health, but ‘the specific challenge is how to harness knowledge and have it contribute to health equity’ [5]. Unanswered questions include whether genomic determinants for disparities exist, if current technologies will help find them, and how likely will there be equal access and utilization of appropriate services [6]. An alternate measured response suggests that genomics, with its focus on biological differences, can make a positive addition to the collective efforts made in eliminating health disparities globally [7].

Part of the challenge in assuring equity is demonstrating that genomic science discoveries are useful for individuals and society. The clinical utility of a genetic test is often measured by its ability to prevent or ameliorate mortality, morbidity, and disability. Although progress in this area has been slower and more disappointing than initially predicted, research will continue to identify the impact of genomics on these important outcomes. Risk-benefit frameworks that include additional approaches such as decision science and stakeholder perspectives are being developed in hopes that they may accelerate the utilization of practice-based evidence [8]. At the same time, there is recognition for the value of personal or social utility, such as consumer desire for information that has an impact on their families [9]. The overall goal is to identify those genomic tests that pose low risk and offer demonstrated utility, while discouraging use of tests that provide little benefit or pose significant health risks compared with traditional care. Beyond health risks, there are other considerations regarding premature use of predictive tests. The benefits of preventive strategies need to be assessed, and potential for discrimination must be addressed – in employment and insurance, and to prevent the development of a genetic underclass.

Although not initially part of the genome research agenda, there is a current understanding that more effort is needed on the translation process. ‘[N]avigating a course from the base pairs of the human genome sequence to the bedside of patients’ provides a simultaneously compelling opportunity and significant challenge for the next decade [10]. Barriers to this challenge include the underfunding and lack of infrastructure required to conduct the research particularly in T3 and T4 phases of the translation process, which are necessary for moving evidence-based guidelines into practice and then evaluating their health outcomes [11]. A more multidisci-
plinary agenda that reaches beyond the traditional clinical trials framework for evidence has been suggested [12]. Expertise within fields such as communication science are underutilized and may offer innovative approaches for ways to educate health professionals to convey information that changes with regularity. Other behavioral and social sciences can contribute by assessing the impacts of genetic information on individuals, families, and communities.

Translating gene discoveries into clinical and public health applications requires attention to both equity and evidence of utility, and in a complementary fashion, the work toward achieving those accelerates the translation process. Two groups within the health care community with key roles in this process are point-of-care health care providers who deliver genetic services and public health practitioners who implement genomic policies.

**Genetic Medicine and Public Health Genomics: A Case of Sibling Rivalry?**

It would be ideal if there was a shared agenda between these groups to pursue best practices. This is not an impossible goal, but there is a tradition of clinical and public health professionals to exist in separate silos – genetics resides in the world of medicine, and public health genomics exists in the domain of public health.

Each has responded to genomic discoveries differently. Genetic medicine embraced the potential for important applications of genomics in health care and disease prevention. Advances in genetic testing and pharmacogenomics offer hope, with personalized medicine being a link between genomic discovery and medical practice [13]. In contrast, genomic applications that can benefit public health are fewer in number (e.g. family health history, newborn screening), and the impact is usually described in modest terms. Given their distinct histories, it is to be expected that the 2 disciplines function out of differing paradigms. As Khoury [14] describes, the mission of public health, with its population perspective, is seemingly at odds with the ‘one person at a time’ vision of genomic medicine.

These 2 missions are accompanied by different social mandates and thus occupy different places in society. What they have in common is that both disciplines emerged out of a series of historic events and grew to achieve the status of a social institution. There are a number of institutions that surround us as part of the social structure, for example, education, religion, and economics. If one moves down a hierarchy of structures, the health care system exists and makes sense within those other larger institutions and dominant social values. We can place medicine and public health another level down, and they coexist in the way of siblings; like siblings they come with very different personalities.

As a way to compare them, visualize medical hospitals with their dramatic architecture, conspicuous signs out front, and sophisticated ads on television. Public health buildings are harder to find and often situated in poor parts of the city. Any ads that you might see are likely educational messages on the side of a bus. As a second point of comparison, there are the schools – medical and nursing schools are very straightforward – what you see is what you get. They are based on very clear missions and students end up with recognizable credentials. Schools of public health are wide ranging with a broad reach. Students receive some common instruction on population health but very quickly head off into sub fields that range from biostatistics to maternal and child health. At graduation there is no symbolic nursing pin or stethoscope to be bestowed; indeed, graduates from public health programs typically have to explain their degrees.

The previous is an outsider view of public health, but to get at an insider view requires identifying shared values that serve to define its members. What is the paradigm that defines the field, which can then be used to promote the implementation of genomics?

It is easier to think about the subcultures within public health than it is to consider public health as a single system. The provider in a public health clinic who cares for individual patients, the public health practitioner who focuses on health of the population, and the health researcher who has the long view in mind have differing expectations of what genomics can contribute. However, considering both outsider and insider views, one of the key features of public health as a social institution is its ability to rise up and become visible when called upon. Public health has an almost subterranean presence, going unnoticed day to day, but emerging full force when presented with a challenge.

This feature leads to a paradigmatic skill and shared value within the public health workforce as being master of the ‘teachable moment’. After a dramatic food borne disease outbreak, we all learn about food safety and surveillance. Flu season teaches about vaccines and government programs. Parents become educated about state newborn screening programs when they get a positive result. Most, however, don’t know the test was done, which highlights another interesting feature of the field: some
practitioners focus on population-wide issues, and others deal more with a ‘need to know’ audience.

The diversity of public health targets of intervention, and the workforce itself, is a strength – it has a wide societal reach – but a disadvantage when trying to identify the genomic knowledge and skills required in the tool kits of practitioners. In a way, we are talking about anticipation: comprehension must be in place ahead of time to take advantage of teachable moments when they appear. An example is direct-to-consumer genetic testing, which when marketed challenged health professionals to respond to a new demand for information and guidance. One question for the field, then, is how can education not only keep up with rapid change, but also anticipate social implications or unanticipated consequences?

**Keeping Pace with Emerging Knowledge and Changing Attitudes**

To move the genomic agenda forward, there needs to be a convergence of minds among stakeholders. These key players include enthusiastic health professionals, forward-thinking public health practitioners, and a supportive and well-informed public. The hard work required to address the complex issues of equity, utility, and translation provides rich opportunities for these 3 groups to collaborate.

Workforce readiness has received considerable attention, and the current consensus is that the present day approaches to health professional education do not prepare providers for the new era of genomic medicine [15]. Attempting to comprehend and synthesize the continual onslaught of new scientific and technologic discoveries has been compared to drinking from a fire hose. This is then complicated by the slew of complex social, behavioral, cultural, and ethical issues that appear in the wake of the discoveries. Attitudes toward these do not remain static, but change in response to social forces. Thus, general awareness is not enough; professionals need skills to understand the issues on a deeper level, so that when social attitudes shift they can proactively provide appropriate responses.

More has been written on the educational and training needs of point of care health providers [16, 17] and consumers [18, 19] than of public health professionals [20]. One reason is the difficulty of mapping out where genomics might enter the field. Public health professionals wear many hats as they fulfill their social mandate to assess the needs of populations, determine the burden of disease, and assure that appropriate services are available to individuals, families, and communities. There are those who work in Federal or state governments, in academia, in professional, community or lay organizations, and in the private sector. Identifying a role for genomics also depends on the public health focus, be it environmental, health, prevention, maternal and child health, epidemiology, or public health marketing.

Similar to clinical practitioners, many public health providers in the field today received their formal education before genomics became a critical aspect of health sciences. The majority of training programs include neither genetics nor genomics in their curricula, they lack any content on how to apply genomic innovations in health promotion or early detection for common conditions, nor do they require course offerings in these topics for accreditation purposes.

Genomics is not new to the world of public health. Most state health departments administer newborn genetic screening plus other genetic disease prevention programs, and several states employ genetics coordinators. There are positive impacts of some genomic applications. For example, cancer genetics is helping to identify high risk individuals and their families with implications for prevention, screening and treatment. However, the lack of strong evidence for concrete health benefits for common disease with a population impact provides a barrier to genetic education and training. Some within the public health community view genomics as a low-yield investment. Local issues, national and international pandemics, and social and environmental causes of morbidity and mortality are seen as more important priorities, particularly in the context of limited public health funding.

**Genetics Education and Training Recommendations**

The Secretary’s Advisory Committee on Genetics, Health and Society (SACGHS) recently devoted 3 years to studying health professional workforce issues and consumer needs to address the integration of genetic and genomic technologies into health care and public health. As one of its final activities before its charter expired in 2011, SACGHS presented the Department of Health and Human Services (HHS) Secretary Sebelius with the report, ‘Genetics Education and Training’ [21], which can be accessed online (http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS_education_report_2011.pdf). Table 1 lists the SACGHS recommendations.
The process was informed by acknowledgement within the Committee that people move seamlessly between clinical, public health, and public sectors as they seek information or care. Rather than treating health care providers, public health professionals, and consumers or patients as separate and distinct entities, effort was made to identify educational mechanisms that facilitate and enhance synergy among these 3 communities. Data were collected using multiple methods: (1) review of relevant published and unpublished literature, (2) surveys of selected health professional organizations and individuals, and (3) interviews with experts in genomics education and advocacy outreach.
The Committee found an increase in genomics education and training efforts in the private and public sectors, and a number of strategies to incorporate genomics into health care have emerged. However, these efforts have not kept pace with the emerging understanding of the human genome and the rapid evolution of genetic technologies. Accreditation, certification, and licensure requirements are not sufficiently linked to education and training efforts. Genetic tests are becoming more widely available, and with this, patients and consumers are increasingly using genetic information in their health decisions. The need for genetic literacy is only going to increase in the coming years.

These and other findings led SACGHS to recommend to the Secretary of HHS that a series of actions be taken to assure a ready workforce and educated consumers. Some of the specific recommendations listed in Table 1 reflect familiar solutions, but with suggestions for new approaches. For example, development of professional education models would benefit from including new partners at the table so that private and public interests as well as academic, industry, and consumer perspectives are used to identify best practices that incorporate up-to-date content that is easily accessible. Because of the diversity of the public health workforce, it is difficult to target educational efforts that are relevant across groups. A systematic evaluation of its composition with identification of current job responsibilities related to genetics and genomics is a first step. Future priorities and their impact must also be considered, one such example is the promise of affordable genomic analysis. Consumer and community needs are also addressed in the report with actions recommended to prevent genomics from widening the disparity gap. The Committee responded to consumer advocates with a specific recommendation for HHS to develop and sustain an Internet portal that provides trustworthy information for consumers.

**Conclusion**

An educated professional workforce and an informed population cannot alone guarantee that genomics actually improves public health, but in collaboration, these groups are in a unique position to be advocates for the 3 elements of achieving health equity, increasing evaluation efforts to measure utility, and accelerating translation of discoveries to clinical and public health interventions. Within the paradigm of public health, the new discipline of public health genomics is poised and ready to contribute to these goals. The traditional place of public health within the communities it serves and its history of addressing health disparities provide a framework for public health genomics to assure equitable distribution of appropriate genomic technologies and services. Similarly, the clinical validity of genetic information is highly dependent on population characteristics (i.e., prevalence of the genetic variant, strength of its association with disease, interactions with other risk factors), and thus the skills and tools practitioners are very applicable.

The road to translation has many steps, each one building on its predecessor. One thing all phases in the translation process have in common is the need for effective communication. Health education is no longer tightly controlled by a select group of acknowledged experts who speak with one voice. With the democratization of information, consumers and patients have a landscape of media options available, including social media. A person can get facts from a professional or governmental website, figure out how to interpret that information with an online support group or by watching Oprah, and then create something new from these experiences on his or her own blog, which then enters the media as another source of information for the next group of seekers. With its long-standing role in health education and growing expertise in health marketing, the field of public health stands ready to be part of that conversation.

The talents required of individuals in the public health workforce are diverse and must remain relevant in the context of the new normal that is our collective reality. Rather than viewing this as a daunting challenge, a strong case can be made that public health – as a discipline, a philosophy, a paradigm – is well situated to take the lead in advancing the genomics agenda. Although typically hidden from public view, it quietly assures basic health, keeps an eye on health trends, works as part of the communities it serves, and continually builds on its ability to respond quickly to emergencies and new discoveries. Public health genomics is at the center of this picture helping to realize the promise of genomics to improve population health and assure the benefits reach the least among us.

**Disclosure Statement**

The author was a member of the SACGHS and led its work on the Genetics Education and Training Report. Any views expressed in this article are the author’s and do not necessarily represent those of the Committee.
References
