Conference on Implications for Genomics and Public Health

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The revolution in human genomics may be the defining medical achievement of the 21st century [1]. The completion of the Human Genome Project and ongoing advancements in genetic research, technologies, tests and therapies promise to enhance personal health outcomes. Beyond individual benefits (and incumbent risks) in the development and proliferation of genetic technologies and knowledge are potential populational uses that may improve public health outcomes. Incorporating genomics into public health is complicated and uncertain. There is no national plan for the application of genetic science into public health practice and research. On October 7–8, 2004, the Institute of Medicine (IOM) of the National Academy of Sciences, with support of the US Centers for Disease Control and Prevention (CDC) Office for Genomics, convened a stellar group of scholars, geneticists, public health practitioners and others to examine the implications for genomics and the public's health, including:

- major scientific and policy issues
- support for (and challenges to) the translation of genetic research into population health benefits
- integration of genomic information into strategies for promoting health and preventing disease

Highlights

The conference, chaired by Lawrence O Gostin (Georgetown University Law Center, DC, USA), featured strong presentations from premier experts representing a diverse range of disciplines

(e.g., internal medicine, biochemistry, genetic counseling, public health science and practice, biotechnology, informatics, law, ethics, economics, philosophy, psychology, psychiatry, and sociology). Keynote addresses by Gilbert Omenn (University of Michigan Medical School, MI, USA) on 'Genomics and the Public's Health: A Vision for the Future' and Alexandra Shields (Georgetown University, DC, USA) on 'Stratification, Justice, and Opportunity', provided structure and context for each day's sessions. Omenn examined the breadth of new medical data and knowledge flowing from the genetics revolution, concluding that scientific advances in genetics, proteomics, comparative genomics, ecogenetics, toxicogenomics, bioinformatics and computational biology have the potential to improve public health outcomes. Shields explored the predictable potential for genetic knowledge and advancements to further stratify communities, with arising injustices and fundamental unfairness. Together, they captured the quintessential theme running through virtually all conference discussions: that genomics offers tremendous benefits for the population's health, but also significant risks and challenges. While this theme is routinely perpetuated in debates on the clinical and research applications of human genomics, this conference looked well beyond individualistic concerns to assess the benefits and risks to families, vulnerable groups, regional populations, nations, and global communities.

Geneticists, including Dr Aravinda Chakravarti (Johns Hopkins School of Medicine, MD, USA), Dr Alfred Berg (University of Washington School of Medicine, WA, USA), and Dr David Romain (UCLA School of Medicine, CA, USA), cautiously emphasized the significant medical findings in human genomics and their interconnectivity with other medical fields and pharmaceuticals. Collectively, they noted that these findings lead to as many questions as answers. For example, virtually all health conditions have genetic components, yet very few of these conditions are purely, or even mostly, the result of genetic flaws.

How environmental factors interact with genetic features so as to contribute to diseases or other conditions in human populations was examined by Dr David Eaton (University of Washington School of Public Health and Community Medicine, WA, USA). Genetic researchers know that environmental factors are often key, but causality is uncertain. Public health practitioners and researchers may be further relied upon to help 'discover' these factors, and employ tools of public health to benefit at-risk groups.

Dr William Foege (Emory University Rollins School of Public Health, GA, USA), former CDC Director and a virtual legend in public health, examined how to effect changes in the population through genetic, environmental and behavioral contexts. "Genomics will expand the opportunities and widen the disparities," noted Dr Foege. With such "unbelievable opportunities," he continued, there can be "unbelievable inequities." Foege's admonition was clear: without ethical allocations of resources, equitable sharing of knowledge, and societal commitments to global benefits, the public health promise of human

Box 1. Key lessons.

Genetic science

- Scientific and technological advances in genetics, proteomics and related sciences, and bioinformatics have the potential to improve public health outcomes.
- Multifactor components underlie virtually all genetic conditions. Proving causality of genomics to the onset of disease or other conditions is complicated.
- Predicting the functional effect of various genetic sequences is critical, but again complicated.
- Additional research is essential to further identify and validate genetic variants.

Genetics and public health

- Existing public health approaches to genetic diseases (e.g., populational screening and universal diagnosis) can be inappropriate or inefficient.
- The framework for genetic risk assessments for population health research requires risk identification, characterization, and reduction.
- Community-based participatory research improves societal knowledge and awareness.
- Progress is being made to identify and prevent gene–environment interactions with correlating benefits.
- Continued research, funding and education will enhance the ability of public health authorities to incorporate genetics into public health.

Genetics, information and behaviors

- Primary care is critical to delivering genetic services to individuals and the population.
- The use of evidence-based methods to substantiate the use of genetic tests is compromised.
- Cost-effectiveness analysis involving an assessment of opportunity costs helps determine whether a genetic intervention is over- or under-utilized.
- Individual behaviors are key factors to improving public health, including improvements through genetic means.
- Linked, multi-part interventions promote positive behavioral changes.

Public health infrastructure

- Assuring the conditions in which people can be healthy is the overarching objective for public health.
- Genetics has relevance to all essential services and functions of the public health system.
- The heterogeneous public health system is not well organized to accomplish goals in genomics and public health.
- Capacity for public health genomics is tied to the competencies of the members of the public health workforce and academia.
- Financing genetic tests and services within populations is subject to healthcare insurer mindsets, and challenged by limitations of existing genetic tests (i.e., accuracy, costs, and utility).

Ethical, legal and social issues

- Public attitudes concerning genetic research are affected by public confidence, perceptions of utility, moral beliefs, and terminology.
- Individuals have significant concerns of bodily and health information privacy and discrimination [2].
- Protection of disease-specific groups and other vulnerable groups are not significantly addressed through legal or ethical norms.
- The Health Insurance Portability and Accountability Act (HIPAA) Privacy Rule, which applies to identifiable genetic data, allows public health authorities to collect such data for public health purposes [3].
- Additional issues in genetic privacy, such as duty to warn, are increasingly being addressed through litigation.
- Laws can facilitate (and complicate) access rights of providers and patients to genetic test results.
- Social stratification invariably leads to discrimination – the objective is to avoid undesirable genetic discrimination.

genomics will be lost for whole populations for generations.

Additional discussions on day two of the conference turned to existing and future efforts to integrate genomics into public health practice. Presentations by Dr J Michael McGinnis (The Robert Wood Johnson Foundation, NJ, USA) Dr Kristine Gebbie (Columbia University School of Nursing, NY, USA), Dr Bartha Knoppers (University of Montreal Center for Public Law Research, QC, Canada) and others examined challenges to further integrate genomics in the existing US and

global public health systems. These challenges include:

- lack of strong public health infrastructure at the international, national and regional levels
- chronic, insufficient funding
- competing priorities in public health
- inadequate capacity of the system to incorporate genomics into practice
- divergent and unprotected data biobanking
- public misunderstanding and apprehension on the role of genomics in the population

The legal environment supporting population-based genomics, addressed by Dr Ellen Wright Clayton (Vanderbilt University, TN, USA) does not fully protect individuals from the potential risks related to research abuses, health information privacy, discrimination, and social stratification.

Collectively, the presentations and perspectives produced a series of key lessons that pervade the collective disciplines and justify the use of tools and interventions to protect the public's health. Organized within five major

categories, or themes, of the conference, these lessons are listed in **Box 1**.

Outlook

Public health officials are already refining their approach to advancing the collective health of the population through genomics. Genetic advances allow them to assess more effectively the complex interactions between environment, behavior, and genes, and to target appropriate populations for public health interventions. Integrating genomics into public health is essential because improving individual and communal health through genetic advancements is synergistic. Clinical genetic interventions and research provide key data and outcomes that collectively contribute to healthier populations. Conversely, the tools of public health practice (e.g., surveillance, screening, testing, and education) [4] and communal research contribute to measuring and assessing the benefits to individuals and populations. The interconnectivity of genomics and the environment underscores this synergy. Individualized genomic markers or anomalies are like clues of a larger puzzle. Only through the coupling of these discoveries with environmental factors (often analyzed through the public health system) can significant, population-based advancements be made. If the mission of human genomics is to truly go beyond improving the health status of a relatively small class of individuals who may have access to these technologies, reliance on public health systems and tools is needed. In short, significant genetic benefits for individuals and

populations are only possible through the organized, informed and committed integration of genomics and public health.

Though neatly summarized, this goal is far from realized. The charge to the IOM from conference presenters and participants is to continue to explore key challenges and issues at the intersection of genomics and public health. Among issues requiring additional exploration are the following:

- unraveling the advances in genetic science and research to identify clearer objectives for public health
- enhanced application of these advances to public health methodology and practice
- assessment of the infrastructural improvements that are essential to integrating genetics into public health
- funding and development of genetic, medical and public health research to support and measure improvements in public health outcomes
- melding social and behavioral research and methods into public health genetics
- bridging healthcare and public health practitioners (and others) within the intersectoral public health system
- developing techniques for integrating genetics into public health practice that overcome challenges of limited funding, technology, and knowledge
- assuring access to public health genetics in ways that are equitable and sensitive to existing health disparities
- building public trust for public health genetic data collections through attention to culturally relevant factors and complex legal and ethical issues

- developing effective public education on public health genetics through specific objectives, targeted audiences, and multiple channels
- assuring that the public health workforce and its partners are capable of using genomics in real practice settings
- assessing fiscal realities suggesting that insurance underwriting of existing genetic testing, pharmaceuticals and services is limited
- reforming the legal framework to ensure greater access to genetic tests, encourage individuals and groups [5] to use genetic information, and delineate the conditions for third parties to responsibly use identifiable genetic data

With greater resources, additional study, and attention to these and other issues, integration of genomics and public health may reach a highly desired end: 'healthier populations'.

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