

Explore the Emerging Role of Public Health in Integrating Genomics in Surveillance, Outbreak Investigations, and Control and Prevention of Infectious Diseases

WORKING GROUP DESCRIPTION

Background

According to the 2005 Institute of Medicine workshop report on the implications of genomics for public health, “public health genomics” can be defined as “an emerging field that assesses the impact of genes and their interaction with behavior, diet and the environment on the population’s health.” The priorities for this field are to:

1. Accumulate data on the relationships between genetic traits and diseases across populations
2. Use this information to develop strategies to promote health and prevent disease in populations
3. Target and evaluate population-based interventions

The “public health system”, which includes federal agencies such as the Centers for Disease Control and Prevention, state health departments, and academic public health institutions, is beginning to work closely with basic scientists, professional organizations, consumer groups, and the private sector to “translate” advances in genomics into actions to prevent and control infectious diseases at the population level (Centers for Disease Control and Prevention, n.d.). Increasingly, genetic information from pathogens, the human hosts, as well as vectors will be used to understand the pathogenicity, natural history, and genetic susceptibility to infectious agents. These new types of data could have profound influences on how the public health system conducts its surveillance functions, acute outbreak investigations (Lingappa and Lindegren, 2003), and community-level programs for targeting interventions, such as vaccines and medications. A major challenge is how to apply this information on the population level to

affect reduction of the burden of infectious diseases in communities. Current public health education has not fully integrated genomics into its basic competences and core curricula. The practicing public health workforce is not adequately prepared to meet the genomics challenge (Institute of Medicine, 2003; Shortell et al., 2004).

The Problem

What should public health systems do to prepare and respond to the emergence of genomic tools in infectious diseases, in terms of surveillance, outbreak investigations, developing and deploying new interventions (e.g., vaccines), and in its efforts to control of infectious diseases, including bioterrorism events, at the population level?

1. Consider how public health systems should incorporate genomics into acute public health investigations such as outbreak response? What should be the current priorities? Because host genomic factors are involved in determining who will be sick from infectious agents, should public health systems routinely collect such information in their investigations? Should they develop biologic specimen repositories involving pathology tissues, human DNA, etc., to explore the host response to infectious agents and gene and protein expression profiles?

2. Consider how public health systems should integrate genomics into surveillance functions for infectious disease occurrence and tracking in the population. While it may be easier to consider pathogen genomics in surveillance, is there a role for routine collection of human genetic information in such data collection? What tools are needed to make epidemiologic surveillance efforts more in real time?

3. Consider the ethical, legal, and social implications of integrating genomics into public health surveillance and response (e.g., privacy and confidentiality, informed consent) and provide recommendations for action and policy change.

4. Consider the role of genomics in developing and evaluating community interventions for the control of infectious diseases. Such programs include administration of vaccines and working with communities and providers to implement control and prevention measures. Consider the social factors that also play a role in who gets sick from infectious agents (e.g., poor nutrition because of low socioeconomic status could influence one's immunity). What

should public health systems do to interact with and educate the public and the provider communities in genomics?

5. Consider the traditional public health data collection categories (e.g., race and ethnicity). Recent articles have underscored one of the consequences of the mapping of the human genome by calling into question our traditional notions of race. Consider how we should categorize individuals given that traditional notions of race and ethnicity are being challenged.

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WORKING GROUP SUMMARY

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Summary

Back in 1918 it could take weeks to travel from one country to another, and yet the Spanish flu pandemic claimed more than 20 million lives as it spread around the globe. Today one can travel halfway around the world in a matter of hours and disease can spread farther and faster than ever before. Fast-paced international travel and trade have brought the world closer together. This new intimacy has brought infectious disease to the forefront of global public health issues. Thus, infectious disease control is now a shared burden—as a growing threat in developed nations, and a clear and present danger in developing nations—and it is therefore a perfect place for a new science like genomics to play an important role. The need to develop new approaches to fight infectious disease was evident in the sentiments of scientists, engineers, and medical researchers gathered in Irvine, California, at the Third Annual National Academies Keck *Futures Initiative* Conference.

Although the developed world focuses much of its medical research on the diagnosis and treatment of chronic diseases, the developing world is constantly compromised by infectious disease. In these countries, people still suffer from a lack of basic care, such as unclean drinking water, malnutrition, and poor sanitation.

Genomics is a discovery science whose specific applications have yet to be completely realized. While it is difficult to predict exactly how much benefit will result from investing in this kind of research, the time is ripe for public health to begin to explore its potential contribution to mitigating illness and death from infectious diseases. Thanks to the Human Genome Project and enthusiastic news coverage, genomics has received a lot of public attention. But celebrity can result in mistaking genomics for the answer to just about everything in health

care, and proponents risk overselling its potential. Aware of these dangers, the 12 members of the group discussed the future of genomics in the battle against infectious disease.

The group agreed that it is essential to set priorities with respect to how best to proceed in applying innovative science in the fight against infectious disease. While innovative science is essential to advance the knowledge base for public health, resource limitation is an omnipresent barrier to improving public health. The investment in genomic science has to be balanced against opportunities for implementation of programs that use inexpensive intervention. For example, a simple bed net to protect against malaria-infected mosquitoes may be just as effective, and far cheaper, than high-tech treatments based in genomic science. Investment in genomic solutions must therefore be viewed through the long-term lens of the opportunity cost—that is, what will we not be able to afford to do if we spend resources on genomics, and what benefits are likely to accrue in the future.

To help understand the etiology and pathogenesis of a disease, determining the genetic sequence of a pathogen might be a higher priority than sequencing the human hosts, because sequencing the human genome costs are enormous and may yield less information about the infectious disease. While resistance and susceptibility factors in the human genome will no doubt prove important in the future, the price of such discovery has to be balanced against more pressing public health needs. In addition, diseases carrying the highest burdens should be given the highest priority for such research, so that the public benefit to scientific discoveries is possible. Time and resources should be allotted appropriately so that these diseases, namely AIDS, malaria, and tuberculosis, are studied first. Resources should also be given to apply genomics to further research and development of new vaccines and medications because vaccines have historically topped the list of groundbreaking improvements made to public health. Finally, a focus should also be placed on identifying and characterizing specific disease-causing organisms in order to break the chain of disease transmission. If genomics can facilitate this activity, then the expenditure of resources will be justified.

It is clear that genomics fits into the circle of public health. It is important, and not too early, to invest in genomics research—a balanced public health portfolio should include allocating and utilizing resources in discovery science. Analysis of large and diverse kinds of data will be crucial. For instance, host genomic research may lead to a better understanding of susceptibility to infections and risk for disease, as well as the beneficial and adverse effects of

drugs and vaccines. The group supported the idea of a bioresource—a place where quality-controlled information could be collected, analyzed, and applied to diagnose, treat, and prevent disease.

Determining exactly what goes into the bioresource was one of the first matters to tackle. In addition to information that is traditionally collected (such as epidemiologic and demographic data), biosamples, ecological information, and genotypic information on pathogens and hosts should also be compiled. The locations of collection and storage sites must be geographically distributed to ensure that participating countries and organizations are engaged and share ownership and responsibility in maintaining the bioresource—a model of data and information gathering that contrasts with the more “colonial” model of data reposing only in the first world. Designers of the bioresource must place a strong emphasis on standardizing the information collected and enforcing quality control. This will ensure that researchers and healthcare professionals who use the bioresource for diagnoses are working with accurate, standardized datasets.

The group also agreed that the data that come out of the bioresource are as important as the resources devoted to it. Therefore, the resource must be managed as an intelligent system that learns and adapts, continually improving its ability to predict and diagnose health problems as more information is collected in the system. As the study of genomics matures and more discoveries are made, the bioresource would be built to continually adjust to these new insights with new applications. Finally, and key to its success, the bioresource’s output of information has to be almost universally accessible: easy to use by anyone from anywhere.

The benefits of such a bioresource could be far-reaching, not only as a research tool but also as a device to improve public health. Programs for collecting samples and information for single diseases exist: preexisting HIV and malaria projects act as good starting points from which to develop a larger resource that covers multiple diseases. An excellent model is the Global AIDS Program, which partners with communities, scientists, and public officials to prioritize health concerns and direct prevention programs appropriately. However, the Global AIDS Program funding is specifically allocated for prevention programs, and not for research, which would be a crucial additional benefit of the proposed bioresource.

At the same time, it is important to remember that genomics can be successful only if the scientific community takes the time and energy to teach the public how it will improve their

health, because of the inherent need for collaboration between the two groups. The group unanimously agreed that education, in the form of a dialog among all parties, is essential to promoting the use of genomics as a public health tool, especially when scientists need the cooperation and understanding of the public to collect genetic information in the form of blood or saliva samples. The public can in turn teach the scientific community what health issues are of greatest concern to them and offer insight into environmental and social aspects of pathogenicity. Both sides have something to learn from each other, and the process will help everyone build trust in the science and one another.

The public's fears that new technologies may be misused or create inequalities can be put at ease only by taking seriously, and then frankly addressing, their concerns. The public may be opposed to a bioresource because of its potential to be used inappropriately and possible negative implications from collecting genetic information from infected individuals. Discrimination has historically been a complication of communicable disease outbreaks. Characterization of lepers as unclean is but one example of the stigmatization resulting from infectious disease. Genetic identification gives the public cause to worry about another form of discrimination. We hope that by educating both healthcare workers and the public, we can prevent these problems.

The task of winning over the global public is never easy. With the social and economic disparities that already exist between the developing and the developed nations, it is difficult to make public health programs attractive to those who are uncertain of their place in the definition of "public" health. The bioresource could potentially serve as a tool to help engage all participating and interested parties in the collection, storage, analysis, and dissemination of the information. However, it will likely be a challenge to articulate the message so as to compel a strong commitment to investing in this as a shared public health global resource.

The International Human Genome and HapMap project exemplify the inherent challenges of equality in such ambitious global health initiatives. The principle behind the project is to make all information freely available to any scientist in the world. While this open access model appears wholly equitable, many scientists and the public can't use the open access model for human genome sequence and HapMap data because they have neither the training, equipment, nor the funding to make use of the available information. Without a commitment to provide public education and open access to equipment, resources such as the human genome sequence

and HapMap (or the proposed bioresource) will have only limited impact, particularly for disenfranchised groups and those in traditionally underserved communities.

In addition to solving the problems of inequality and global accessibility of the resource's assets, there are many ethical concerns that come with a system as encompassing as the group's proposed bioresource. When donating genomic material to a biobank like the bioresource, participants are typically expected to provide consent for the use of such materials in subsequent research. In exchange, investigators pledge to protect confidentiality and assure contributors that their genetic information will remain secure. Safety is of utmost importance with any repository of infectious materials, and steps must be taken to protect the resource from any harmful misuse of its samples. However, individuals will also have compelling interests in knowing the health status of themselves and their families, especially in the cases of treatable diseases.

With genomics still in its infancy, now is the perfect time to concentrate on the challenges and promises of this new science in winning the battle against common infectious diseases in their threat to human life and the public health.