
The Integration of Genomics into Public Health Research, Policy and Practice in the United States

Laura M. Beskow^{a,b} Muin J. Khoury^a Timothy G. Baker^a
James F. Thrasher^{a,b}

^aOffice of Genetics and Disease Prevention, Centers for Disease Control and Prevention, Atlanta, Ga.,

^bSchool of Public Health, University of North Carolina at Chapel Hill, Chapel Hill, N.C., USA

Key Words

Genetics/genomics · Public health · Epidemiology · Assessment · Policy development · Assurance

Abstract

Objectives: To examine the opportunities for and responsibilities of the public health community in bridging the gap between gene discovery and the application of genetic information to improve health and prevent disease. **Methods:** We developed genetics-related definitions for the core functions and essential services of public health. We combined these definitions with a visual model to create one possible 'blueprint' for integrating genomics into public health activities. **Results:** The proposed blueprint and accompanying examples illustrate the important role for genomics throughout public health research, policy and practice. Further refinement and implementation of this blueprint represents an ambitious public health leadership agenda. **Conclusions:** Opportunities for immediate action include strategic planning for the integration of genomics across programs, developing genomics competencies among health professionals, enhancing surveillance and epidemiologic capacity to aid evidence-based policy making,

building partnerships and seeking input from stakeholders and incorporating information about genomics into health communications.

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Introduction

During the past century, achievements in public health led to dramatic improvements in the health and life expectancy of people in the US and around the world [1]. Immunization programs and better sanitation practices resulted in the eradication or reduction of many infectious diseases and safer food and water supplies. Advances in occupational safety considerably decreased the number of work-related injuries, illnesses and deaths. In the past 30 years, identification of behavioral risk factors, such as smoking, inactivity and poor dietary habits, gave rise to educational interventions and a decline in death rates from certain chronic diseases.

Perhaps because of these accomplishments, the determinants of disease and disability – whether natural or human made – are often perceived as originating outside the body. Although it has long been recognized that disease generally results from a constellation of host- and

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Laura M. Beskow
c/o Office of Genetics and Disease Prevention, Centers for Disease Control and Prevention
4770 Buford Highway NE, Mailstop K-28
Atlanta, GA 30341-3724 (USA)
Tel. +1 770 488 3235, Fax +1 770 488 3236, E-Mail laura_beskow@unc.edu

environment-specific factors, scientific and technologic limits have concentrated attention on the environment. Exogenous influences will continue to be vital for public health, but focusing solely on these influences may lead to diminishing rates of return compared to the triumphs of the past. To continue making significant strides, we must strengthen the effectiveness of public health interventions by more fully incorporating knowledge of internal, host-specific factors and their interactions with environmental exposures.

Rapid advances in human genetics and accompanying technologies are making this expanded approach to public health research, policy and practice increasingly possible. Until recently, the field of genetics was largely confined to the realm of rare disorders caused by mutations in single genes. Even so, the public health community included genetics components in some of its work, experiencing noteworthy success in birth defects prevention [2–5], screening of newborns for inborn errors of metabolism [6, 7] and development of genetic services capacity [8, 9]. Today, the mounting accomplishments of the Human Genome Project demand that we rethink the role of genomics in every condition of public health interest. Microarray technology, for instance, and dense maps of common human DNA sequence variations called single-nucleotide polymorphisms will be a boon for genome-wide association studies of complex disorders such as cancer, heart disease and diabetes [10]. This information explosion is expected to bring about nothing short of a revolution in medicine and public health [11, 12]. Dr. Francis Collins, director of the National Human Genome Research Institute, depicts a potential outcome of this revolution in his description of a hypothetical clinical encounter in 2010 [13]. John, a 23-year-old man, consults with his doctor in selecting from a battery of genetic tests that will provide information about his personal relative and lifetime risks for a number of common diseases:

Confronted with the reality of his own genetic data, [John] arrives at that crucial ‘teachable moment’ when a lifelong change in health-related behavior, focused on reducing specific risks, is possible. And there is much to offer. By 2010, the field of pharmacogenomics has blossomed, and a prophylactic drug regimen based on the knowledge of John’s personal genetic data can be precisely prescribed to reduce his cholesterol level and the risk of coronary artery disease to normal levels. His risk of colon cancer can be addressed by beginning a program of annual colonoscopy at the age of 45, which in his situation is a very cost-effective way to avoid colon cancer. His substantial risk of contracting lung cancer provides the key motivation for him to join a support group of persons at genetically high risk for serious complications of smoking, and he successfully kicks the habit [13].

In the excitement about this kind of vision for genetically based, individualized prevention, however, it is easy to overlook the immense gap between the scientific products of the Human Genome Project and the ability to use genetic information to benefit health. Information about genes and DNA sequences must be translated into knowledge about genetic susceptibility to disease and the interactions between these susceptibilities and modifiable risk factors. We must also formulate policies that will promote the safety, accessibility and quality of genetic tests and services and develop effective programs for targeting interventions to people at increased risk. Bridging this gap – the ‘Grand Canyon’ between advances in human genetics and the application of genetic information to improve health and prevent disease – requires a wide range of activities drawing upon all of the core functions and essential services of public health.

How then should the public health community approach the task of translating the already occurring deluge of genetic discoveries into public health action? How can public health agencies prepare their workforce and their constituencies to ensure that information about gene-environment interactions is used appropriately? The prototypical public health paradigm in genetics has been newborn screening and we can learn much from these experiences. However, the ‘new’ genetics calls for even greater partnership and coordination between clinical medicine and public health activities. While much genetic testing and screening may one day take place as Dr. Collins describes – in a clinician’s office on an individual basis – public health researchers must analyze genetic information on a population level in order to inform health policy and program development. The public health community must work with public and private health care sectors to ensure that genetic tests are valid, available and accessible – especially in underserved populations – and to assure that individuals in the population have access to proven interventions. Public health professionals also have a crucial role in educating other health professionals and stakeholders and in evaluating the impact and cost effectiveness of integrating genomics into health promotion and disease prevention programs.

To highlight the requirements of the new genetics, we have developed definitions for the role of genomics in each of the public health functions and services. We propose these definitions here together with a visual model, thus creating one possible ‘blueprint’ for integrating genomics into the complete range of public health activities. We include examples of current genetics-related activities in the US within each of the essential services to stimulate

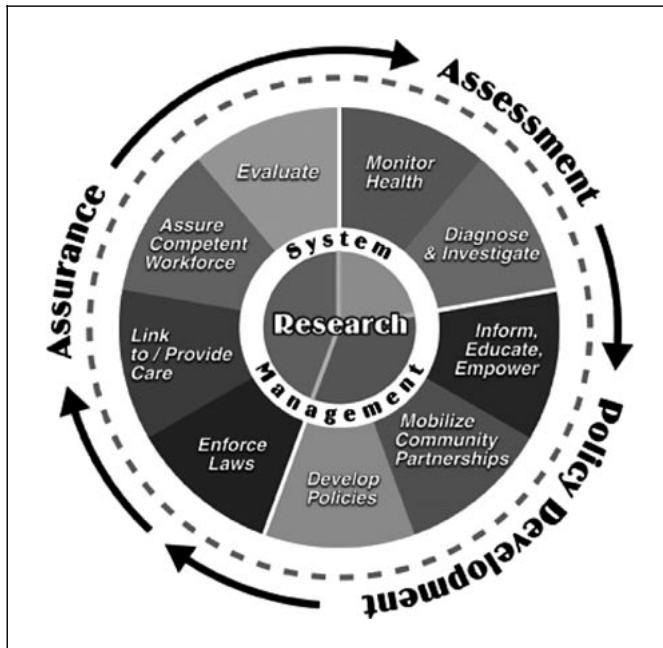


Fig. 1. The integration of genomics into public health. This model has been adapted from that of the Public Health Functions Team [21].

discussion regarding how genomics should be incorporated throughout public health research, policy and practice.

Defining Key Concepts

This blueprint uses several key terms. ‘Human genome epidemiology’ is the study of the role of genetic factors and their interaction with environmental factors in the occurrence of disease in human populations [14, 15]. ‘Stakeholders’ are all groups of people with an interest in the use of genetic information, such as the general public, patients, support and advocacy groups, health professionals, scientists, policy makers and pharmaceutical, biotechnology and insurance industry personnel. ‘Public health infrastructure’ consists of the resources needed to deliver the essential public health services to every community [16]. ‘Gene variant’ refers to both heritable mutations and polymorphisms. ‘Environmental factors’ encompasses all exogenous factors – chemical, physical, infectious, nutritional, social and behavioral. Finally, ‘prevention’ means the use of environmental interventions to reduce the risk of disease among people susceptible because of their genetic makeup.

Blueprint for Integrating Genomics into Public Health

In its landmark report *The Future of Public Health*, the Institute of Medicine (IOM) defined three core functions of public health: assessment, policy development and assurance [17]. To operationalize these core functions, several groups developed more specific descriptions of public health processes, including a list of 10 essential services [18]. We use these functions and services as a foundation for our blueprint, both as a way to demonstrate the merger of genomics into existing public health activities and to emphasize that genetics is not an isolated specialty requiring a separate framework. This approach is in accordance with previous commentary by a Genetics Working Group at the Centers for Disease Control and Prevention (CDC), which used the IOM paradigm to examine the continuum from genetic technology to public health practice [19], and with CDC’s agency-wide strategic plan for genetics activities [20].

The blueprint is proposed as a nonprescriptive tool to promote dialogue and to assist public health professionals at federal, state and local levels to effectively and systematically integrate genomics into public health research, policy and practice. It may also be helpful for educating other key stakeholders, such as policy makers, about the use of genomics to improve health and prevent disease. It is intended to encompass both single-gene disorders and complex diseases involving multiple genetic and environmental factors. It is based on the US health system and would need to be modified for use in other contexts.

The blueprint comprises the visual model depicted in figure 1, together with the genetics-related definitions proposed in table 1. The visual model is based upon that created by the Public Health Functions Team [21], which we adapted as follows. The traditional list of 10 essential public health services includes ‘research’. In this broad sense, we define public health research involving genomics to mean: *a systematic investigation designed to develop or contribute to generalizable knowledge of the impact of human genetic variation on health and disease*. The visual representation created by the Public Health Functions Team places research in the center, as the hub of a ‘wheel’ of public health services. This illustrates how research provides the scientific underpinnings that enable each of the other services. To highlight the array of research activities needed to integrate genomics into public health, we segmented ‘research’ into three areas of inquiry corresponding to the three core functions as shown in figure 1 and described in table 1.

Table 1. Proposed genetics-related definitions

<i>Assessment</i>	<p><i>Core function</i> The regular systematic collection, assembly, analysis and dissemination of information, including human genome epidemiologic information, on the health of the community.</p> <p><i>Related essential services</i></p> <ul style="list-style-type: none">• Epidemiologic and laboratory research: quantifying the impact of gene variants on human health and identifying and quantifying the impact on human health of environmental risk factors that interact with gene variants.• Monitoring health: monitoring health status, including genetic factors, to identify health problems within the community.• Diagnosing and investigating: investigating the distribution of genetic and modifiable risk factors within the community to determine their contribution to identified health problems and to improve health outcomes.
<i>Policy Development</i>	<p><i>Core function</i> The formulation of standards and guidelines, in collaboration with stakeholders, which promote the appropriate use of genetic information and the effectiveness, accessibility and quality of genetic tests and services.</p> <p><i>Related essential services</i></p> <ul style="list-style-type: none">• Policy and communications research: identifying and analyzing the economic, social, ethical and political implications of advances in human genetics, including the information and communications needs of stakeholders.• Informing, educating, empowering: facilitating communication and education about the integration of genomics into health promotion and disease prevention programs.• Mobilizing partnerships: fostering collaboration between public and private agencies and constituent groups to promote effective and efficient communication and policy making about genomics.• Developing policies: establishing standards and guidelines for when and how genetic information should be applied to promote health and prevent disease.
<i>Assurance</i>	<p><i>Core function</i> Assuring constituents that genetic information is used appropriately and that genetic tests and services meet agreed-upon goals for effectiveness, accessibility and quality.</p> <p><i>Related essential services</i></p> <ul style="list-style-type: none">• Health services research: identifying and analyzing the factors that influence the impact of genetic information and the delivery, utilization and quality of genetic tests and services.• Enforcing laws: promoting the enforcement of policies and standards enacted to ensure the appropriate use of genetic information and the effectiveness, accessibility and quality of genetic tests and services.• Linking to/providing care: ensuring the availability and accessibility of genetic tests and services and associated interventions to improve health and prevent disease.• Assuring a competent workforce: ensuring that present and future health professionals have training and skills in the appropriate use of genetic information to promote health and prevent disease.• Evaluating: evaluating the impact of genetic information and the effectiveness, accessibility and quality of genetic tests and services.
<i>System Management</i>	Building and maintaining the capacity of the public health infrastructure to integrate genomics into public health research and practice.

As portrayed in figure 1, performance of public health functions and services is neither linear nor discrete. Assessment activities provide the knowledge base for policy development concerning genomics and for assuring the

proper implementation of programs and services that involve genomic components. Policy development activities help identify gaps in scientific knowledge and form the foundation for assuring the effectiveness, accessibility

Table 2. Epidemiologic and laboratory research in genomics

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- Topics addressed in human genome epidemiology include [14]:
- Prevalence of gene variants in different populations.
 - Magnitude of disease risk associated with gene variants in different populations (relative and absolute risks).
 - Contribution of gene variants to the occurrence of disease in different populations (attributable risks).
 - Magnitude of disease risk associated with gene-gene and gene-environment interactions in different populations.
 - Validity (sensitivity, specificity, predictive value) and clinical utility of genetic tests in different populations.
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and quality of programs and services. The assurance function, in addition to ensuring that genomics is properly integrated into health-related services, also supplies evaluative information for continuing efforts in assessment and policy development.

Examples of Current Genomics Activities in Public Health

Below, we provide brief descriptions and examples from the US to demonstrate practical applications of the genetics-related definitions. These examples are not intended to be comprehensive or even representative of all current or potential genomic activities in public health, but to stimulate further discussion and development of the blueprint for integrating genomics into public health research, policy and practice.

Epidemiologic and Laboratory Research

Gene discovery is not enough; population-based research is essential for translating such discoveries into opportunities for treating and preventing disease. Population-based studies are needed to quantify the impact of gene variants on the risk of disease and to quantify the effect of modifiable factors that interact with gene variants (table 2), forming the scientific basis for developing sound policies and effective interventions.

Example: Researchers are using large-scale epidemiologic studies such as the Atherosclerosis Risk in Communities Study [22], the Framingham Heart Study [23] and the National Health and Nutrition Examination Survey [24] to measure associations between gene variants, environmental factors (e.g. smoking, physical activity, diet) and the risk of cardiovascular disease.

Monitoring Health

Public health professionals use vital records, census data, hospital discharge data and a variety of other sources to gather health-related information about populations. Incorporating knowledge and awareness of the genetic contribution to health problems will enable refined decision making about resource allocation and provide a basis for prioritizing and targeting public health program objectives.

Example: Hereditary hemochromatosis is a common genetic disorder characterized by excessive iron absorption and accumulation, which can lead to illnesses such as cirrhosis, diabetes, heart disease and arthritis [25, 26]. Enhancing surveillance for hemochromatosis through population surveys, provider surveys and population-based registries would help measure the magnitude of the problem, identify research needs, improve understanding of the natural history of the disease, identify changes in screening practices and evaluate the effect of prevention and control measures [27].

Diagnosing and Investigating

Once health problems have been identified in a population, further diagnosis and investigation may be warranted to improve health outcomes. Careful identification of risk factors, including host-specific factors, is a first step toward gaining a thorough understanding of the biologic mechanisms of disease and establishing new avenues for prevention and treatment.

Example: Infectious disease investigations are being expanded to include host genetic factors that influence susceptibility to disease or the severity of disease and that affect responsiveness to vaccines and therapies [28]. Researchers have already identified gene-disease associations for viral, bacterial, parasitic and fungal infections such as HIV [29], hepatitis [30], tuberculosis [31], cholera [32] and malaria [33], providing critical clues to continuing public health efforts to control these diseases.

Policy and Communications Research

Some potential uses of health information raise concerns about the fairness of the use of such information, privacy and confidentiality, individual and group harms and health service delivery issues, among others [34]. Public health research to identify and analyze the implications of these types of issues, together with scientific data, provides the foundation for developing effective policies about the appropriate use of genetic and other health information.

Example: Factor V Leiden is the most common genetic risk factor for venous thrombosis, a leading cause of inpa-

tient and maternal death [35, 36]. Policies concerning population screening for Factor V Leiden, for example before prescribing oral contraceptives or during pregnancy, should be guided not only by clinical and epidemiologic data on risk-to-benefit ratio, but also by an examination of possible personal and social morbidity, overall economic costs and public attitudes and opinions [37].

Informing, Educating, Empowering

Another key to developing effective policies and minimizing the possibility of harm is to empower all stakeholders to make informed decisions about the uses of genetic information with realistic expectations about the risks and benefits. The public health community has a major role to play in raising the level of general genomic literacy, developing targeted messages about the uses of genetic information in disease prevention and coordinating communication strategies with stakeholder groups.

Example: The National Human Genome Research Institute's 'Consumer Day 2000' [38] is one example of how the general public is being empowered with information and education about advances in human genetics. Another is the 'Consumer Voices Network', a program of the Genetic Alliance to foster an informed and articulate consumer voice about genomics issues, as well as to represent consumer perspectives nationally with the news media and health care policy makers [39].

Mobilizing Partnerships

Encouraging new collaborations among partners with diverse perspectives – as well as incorporating genomics into existing collaborations – will help improve the quality of genetics-related policies and reduce duplication of effort.

Example: Asthma, one of our nation's most common and costly diseases, in all likelihood results from a genetic predisposition triggered by environmental allergens, pollutants and infectious agents [40, 41]. The emerging role of genomics in diagnosing, treating and preventing asthma should be shaped by mobilizing government, professional and consumer organizations, such as CDC [42], the Health Resources and Services Administration [8], the National Heart, Lung and Blood Institute [43], the Asthma and Allergy Foundation of America [44], the Allergy and Asthma Network – Mothers of Asthmatics [45], the American Lung Association [46], the American College of Allergy, Asthma, and Immunology [47], the Association of State and Territorial Health Officials [48] and legislative and industry leaders.

Developing Policies

According to the IOM, policy development 'is the means by which problem identification, technical knowledge of possible solutions, and societal values join to set a course of action' [17]. Thus, creating policy regarding when and how genetic information should be applied requires a thorough assessment of all aspects of genetic testing, including medical, epidemiologic and economic data, personal and societal benefits and risks and laboratory and infrastructure concerns.

Example: In 1997, CDC convened a workshop to discuss the clinical, social and economic outcomes of newborn screening for cystic fibrosis and to make recommendations to assist medical professionals, public health officials and state policy makers in evaluating these issues [49]. This statement is one of a number concerning genetic testing for cystic fibrosis, including those from the National Institutes of Health [50] and the American Society of Human Genetics [51].

Health Services Research

Assurances to the public that undergoing genetic testing is safe and beneficial rely on health services research. Questions include: How do the characteristics of different health care systems (e.g. managed care) influence the provision of genetic tests and subsequent clinical or preventive services? Is it cost effective to tailor interventions based on genetic information? What factors affect compliance with proposed intervention strategies?

Example: Colorectal cancer is the second leading cause of death from cancer in the US; however, when it is detected at an early stage, the 5-year survival rate approaches 90% [52]. People who have a family history of colorectal cancer, an indicator of possible genetic involvement, often develop the disease at a younger age and thus may benefit from regular screening earlier than otherwise recommended [53]. Health services researchers are investigating whether educating patients directly about the importance of knowing one's family history of disease results in better compliance with screening guidelines than does educating physicians [54].

Enforcing Laws

Public health policies and programs must be responsive to federal, state and local legislation as well as case law on matters such as health information privacy, confidentiality, duty to warn and informed consent. Public health professionals may also take part in advocating for new legislative actions that will enable people to benefit from genetic information, such as laws to prevent genetic

discrimination and requiring third party payment for genetic testing and clinical or preventive services where appropriate.

Example: President Clinton signed an Executive Order banning discrimination in federal employment based on genetic information and endorsed legislation that would extend these protections to the private sector and to individuals purchasing health insurance (H.R. 2457) [55]. At the state level, the National Conference of State Legislatures has compiled a table of statutory law concerning genomics issues, many of which are relevant to public health, such as employment, insurance, laboratories/genetic services/DNA data and privacy [56].

Linking to/Providing Care

Public health professionals have an important role in assuring the availability of and access to health care, sometimes by directly providing it and at other times by assuring others provide it. To avoid exacerbating existing health disparities – and possibly to help ameliorate them – this role should include assuring access to high-quality genetic testing programs and management/intervention services, especially for the uninsured and underserved.

Example: Untreated maternal phenylketonuria increases the risk of mental retardation, microcephaly, low birth weight and congenital heart disease in offspring, although strict dietary control before and during pregnancy reduces these risks [57]. Public health professionals are working to assure that women of childbearing age who have phenylketonuria have access to dietary formula and low-protein food products, either through state metabolic programs or mandated insurance coverage [58], and are provided with culturally appropriate community resources and support [59].

Assuring a Competent Workforce

Because most health professionals were trained before these advances in genomics, few have the education or experience necessary to participate in this rapidly emerging field. Present and future health professionals must develop the knowledge base, skills and attitudes needed to effectively integrate advances in genomics into their work [60].

Example: CDC is building on national efforts to develop the public health workforce [61, 62] by convening experts to identify the genomic competencies that will soon be required to perform everyday public health functions. These genomic competencies will be used to create core and specialized training curricula and relevant performance standards. Other examples of assuring a compe-

tent workforce include the pioneering work by the University of Washington and the University of Michigan in developing genetics curricula within schools of public health [63, 64].

Evaluating

Systematic evaluation is critical for improving, accounting for and prioritizing public health actions involving genomics. Public health must evaluate in different populations (1) the impact of using genetic information on morbidity, disability and mortality associated with disease, (2) the magnitude and determinants of the utilization of genetic tests and services and (3) the quality of genetic tests and clinical and preventive services relative to established standards and guidelines.

Example: A landmark study indicating that penicillin prophylaxis reduced the incidence of serious infection in young children with sickle cell disease led to the widespread adoption of newborn screening programs for sickle cell disease [65]. Lower death rates among children who have sickle cell disease identified by newborn screening is one measure of the effectiveness of prevention [66], although linking vital records and laboratory, hospital and clinic databases would allow a more comprehensive evaluation of other short- and long-term outcomes of newborn screening [67], such as complication rates and actual utilization of early medical interventions.

System Management

System management is neither a core function nor an essential service of public health. However, by including it in its visual model, the Public Health Functions Team acknowledged the vital role of the public health infrastructure as both a support for and conduit between research and practice.

Example: The final report of a federal Task Force on Genetic Testing called upon government agencies, particularly the National Institutes of Health and CDC, to support collaborative efforts to facilitate the collection of data on the safety and effectiveness of new genetic tests [68]. In response, a Health and Human Services Interagency Working Group on Genetic Testing was formed and has proposed a public-private partnership framework for the collection and dissemination of data on the clinical validity and clinical utility of genetic tests [54]. The Secretary's Advisory Committee on Genetic Testing endorsed testing the feasibility of this approach [69], and pilot studies on cystic fibrosis and hemochromatosis are under way. At the state level, a 1999 Health Resources and Services Administration initiative funded states to develop genet-

ics plans, setting a framework for a genetic service infrastructure and partnerships among state public health programs, primary care providers, the genetics community and service consumers [8].

Conclusions

Rapid advances in human genetics and accompanying technologies are making it possible to explore the full constellation of factors that affect human health and disease – external influences as well as internal, host-specific factors. Understanding the impact of environmental exposures on people who carry specific gene variants offers the possibility of more effective public health interventions targeted at those who are most susceptible to particular diseases. These range from diseases affecting the health of infants and children to adult chronic diseases to disorders stemming from exposure to infectious agents or environmental hazards. Our challenge is to translate gene discoveries into opportunities to improve health and prevent disease in a way that maximizes the benefits of using genetic information, minimizes the risks and conserves health care resources.

We have proposed here one possible blueprint for integrating genomics into the full spectrum of core functions and essential services of public health in the US. This blueprint demonstrates the important role for genomics throughout public health research, policy and practice – not as a separate specialty but as a fundamental component of existing disease prevention and health promotion programs. Further refinement and implementation of this blueprint and others like it represents an ambitious public health leadership agenda. Recommendations for immediate action include the following:

(1) An urgent priority for state public health agencies is to *develop a sound strategic plan* that supports the integration of genomics across its programs. Such plans should be shaped by input from every branch of the organization, including maternal and child health, environmental health, chronic disease, laboratory services and infectious disease.

(2) A key component of the successful assimilation of genomics into public health will be to *train the workforce*. Strong partnerships between schools of public health, academic genetics centers, professional organizations, industry and government agencies will ensure the development of genomic competencies and skills among current and future public health professionals.

(3) The public health community will need to *enhance surveillance and epidemiologic capacity* to collect and analyze the information flowing from community-based assessments of the impact of genetic variation on the burden of various diseases and from the evaluation of genetic tests and services. These data will allow health care providers and policy makers to make evidence-based decisions about the appropriate use of genetic information to improve health and prevent disease.

(4) Public health professionals must *build partnerships* with and seek continuous input from stakeholders such as community groups and professional organizations. This can be accomplished in many ways (e.g. advisory committees, task forces), and the best avenues for communication may be different for different diseases.

(5) Finally, it will be crucial for public health agencies to *communicate about genomic issues* to policy makers, health professionals and the general public. As in other areas of public health, communication messages must be developed for a variety of audiences to educate and empower them regarding the role of genetic information in disease prevention and health promotion.

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