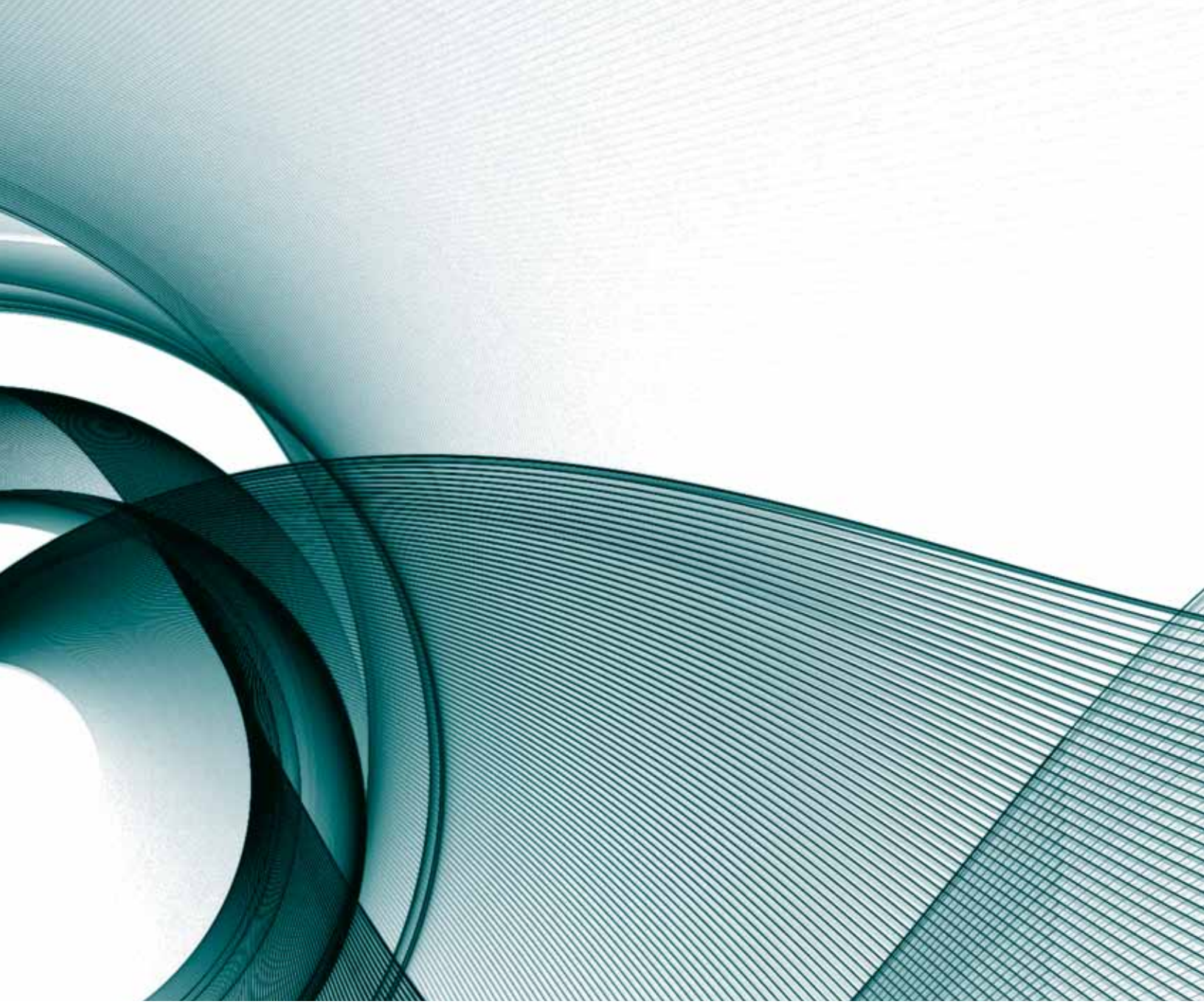

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The 2010 State Public Health Genomics Resource Guide

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Introduction

The rapidly evolving field of genomics offers new opportunities to improve individual and public health. In recent years, researchers have achieved important milestones related to genomics, such as identifying genes that play a role in breast cancer, diabetes and other diseases. Breakthroughs in genomics provide great promise for improving health outcomes for the population, which is the central concern of public health. With strong leadership, discoveries in genomics can be increasingly translated into disease detection, prevention and treatment initiatives that improve public health.

Translating genomic science into public health practice is a challenge for public health leaders. As new genomics breakthroughs expand what we know about our current and future health, health officials and policymakers are facing a plethora of new and evolving ethical, legal and social policy questions.

- What is the appropriate role for policymakers and public health officials related to the growing market of direct-to-consumer genetic tests? As tests become increasingly available to consumers (sometimes without counseling from a qualified healthcare provider), policymakers and health officials are trying to strike a balance between consumer protection (e.g., ensuring that patient genetic data is confidential) and empowerment.
- What are the ethical, legal and societal issues that surround storing biological specimens, such as newborn screening dried blood spots, indefinitely for genetic research? How do state agencies and policy leaders balance the potential public health benefits with privacy and uncertainty about how the samples will be used in the future?
- What ethical principles guide state genomics policies? What role should community engagement play in determining these policies?

Despite the challenges, public health leaders, practitioners and their partners in the public and private sectors are integrating genomics into public health practice. Some common approaches described in this report include:

- **Educating the Public and Health Care Providers about the Importance of Family Health History to Individual and Public Health.** Family history is a risk factor for many common diseases. Individuals who are aware of their family health history can make informed healthcare and lifestyle choices to reduce their disease risk. Integrating family history into environmental health, chronic disease and other areas can help address health disparities and improve public health.
- **Engaging the Community in Planning and Prioritizing to Ensure That State Genomics Activities Reflect Community Priorities and Values.** Integrating community feedback into state genomic needs assessment and planning activities guides policies, programs and funding. Several public health agencies collaborate with diverse populations to gather information about public perceptions and needs and connect people with genomics resources and services.
- **Collaborating with Partners to Maximize Limited Resources.** Partnerships are essential for all the programs outlined in this guide. State health departments are collaborating with other state agencies, schools of public health, provider organizations, policy experts and policymakers to achieve specific goals. These include: informing the public about family health history, educating medical providers about integrating genomics into practice, and ensuring an adequate primary care and genetics workforce. For more information on partnerships, see the final section in this report, "Charting a Path in Your State."

"Translating the knowledge we are gaining from gene discoveries into practical clinical and public health applications will be critical for realizing the potential of personalized health care and improving the health of the nation."

– Muin J. Khoury, MD, PhD, Director, CDC Office of Public Health Genomics

About This Guide

In 2003, ASTHO published Genomics: A Guide for Public Health, which provided health officials with practical tools for integrating genomics into their policies and practices. Over the past seven years, much has happened on the genomics front, including major technology and research breakthroughs, landmark federal legislation and significant policy and program changes at the state level. These changes in policy, funding and research prompted ASTHO to develop *The 2010 State Public Health Genomics Resource Guide*.

This 2nd edition highlights innovative approaches and challenges by states to integrate genomics into their public health programs, with the goal of helping state public health leaders and program managers identify solutions for addressing their state's unique challenges. Included are weblinks to toolkits, presentations and research reports that may be useful resources for states that are considering a similar approach.

ASTHO staff developed the Genomics Resource Guide, with assistance and guidance from a workgroup of state genetic coordinators, CDC staff and state public health managers. The workgroup provided feedback on the 2003 publication and made recommendations for the 2nd edition. Workgroup members also helped ASTHO identify states with innovative approaches. Several members provided detailed information about their own programs to ASTHO staff.

Process and Methodology. At the outset, ASTHO staff asked the workgroup to provide input on the original guide to assess strengths and weaknesses. The workgroup suggested key topics for the 2010 edition. The workgroup said that the state examples in the original guide were extremely effective because the primary audience, public health department managers, want to see what other states are doing and how they are doing it.

As a result, the ASTHO team developed a framework for The 2010 State Public Health Genomics Resource Guide that features state examples and provides tools and resources that can be used in other states. In addition to Internet research, ASTHO gathered information about state programs through telephone interviews with public health genomics program managers. ASTHO staff used a standardized interview guide to gather information about program funding, staffing, activities and outcomes. The interviewees and the state health official in each of the states reviewed their state profiles for accuracy.

How ASTHO Selected States. ASTHO conducted Internet research and solicited input from the workgroup to identify states with innovative programs. Although not formally defined here, innovative programs achieve results in new ways, with new or different funding approaches and/or partners. In many cases, these states are doing more with less by taking advantage of strong public and private partnerships. With guidance from the workgroup, ASTHO narrowed down the field of state examples to reflect differences in geography, funding and core public health function. ASTHO acknowledges that the states included in this guide are not comprehensive and there are other states that could be included in this guide. For that reason, ASTHO will maintain and update this guide on its website to include new genomics trends and examples.

How The Guide Is Organized

The 2010 State Public Health Genomics Resource Guide is divided into three major sections:

I **Overview of Public Health Genomics Today.**
The first section provides background on public health genomics, including what it is, why it is important to public health, and how the field of genomics has changed.

II **Promising Practices in Public Health Genomics.**
This section profiles states that are taking steps to integrate genomics into their public health systems and includes tools and resources. It also describes strategies that help states fulfill their three core public health functions of assessment, assurance and policy development.

III **Charting a Path in Your State.**
The final section provides information and resources to help states assess their current work in genomics and identify options for developing their programs.

1 Section One

"Genomics has a role in 9 of the 10 leading causes of death in the United States, for example, heart disease, cancer, diabetes. All human beings are 99.9 percent identical in their genetic makeup. Differences in the remaining 0.1 percent hold important clues about the causes of diseases. Having a better understanding of the interactions between genes and the environment is helping us find better ways to improve health and prevent disease."

-- National Human Genome Research Institute

Overview Of Public Health Genomics Today

What Is Genomics?

Each species has its own unique genome. Think of the human genome as a book. Genes are inherited pieces of DNA, passed from parent to child. Individual genes tell part of the story, like a chapter in a book, but only a complete collection of genes, a genome, comprises the entire book.

Table 1. Common Terms Defined

Term	Definition
Genetics	"A term that refers to the study of genes and their role in inheritance – the way certain traits or conditions are passed down from one generation to another. Genetics involves scientific studies of single genes and their effects…Genetics determines much (but not all) of a person's appearance and health status, but environmental differences also play a part. Examples of single gene disorders that would be considered 'genetics' include cystic fibrosis and PKU (phenylketonria.)" (1)
Genomics	"A relatively new term that describes the study of all of a person's genes including interactions of those genes with each other and the person's environment. Genomics involves the scientific study of complex diseases such as heart disease, asthma, diabetes and cancer because they are caused more by a combination of genetic and environmental factors. Genomics is offering new possibilities for therapies and treatment of some diseases, as well as new diagnostic methods. The major tools and methods related to genomics studies are bioinformatics, genetic analysis, measurement of gene expression, and determination of gene function." (2)
Public Health Genomics	"A multidisciplinary field focused on the effective and responsible translation of human genome-based information and applications into health care practices to improve population health. It uses population data on genetic variation and gene-environment interactions to develop evidence-based tools for improving health and preventing disease." (3)

Sources: (1) and (2) National Institutes of Health, National Human Genome Research Institute, Frequently Asked Questions about Genetic and Genomic Science. (3) Centers for Disease Control and Prevention, About Us: Public Health Genomics Program Review.

Table 2. What's in a Name? Genetics vs. Genomics

Genetics	Genomics
Study of the inheritance of individual genes	Study of all genes in a person or organism
Diseases primarily caused by a single gene	Traits/diseases influenced by multiple genes and environmental factors
Rare disorders (e.g., Huntington's Disease, Fragile X Syndrome)	Common diseases, such as cancer, diabetes and heart disease
Public health activities include newborn screening, prenatal testing and carrier screening	Public health impact in chronic disease, infectious disease, environmental health, epidemiology and other areas

Source: CDC, "Genetics, Genomics and Public Health: An Overview," 2009

In this report, we use the term "genomics" to encompass all of the related scientific discoveries and the health and social implications, which include:

- The development of new predictive tests, preventive measures, and treatment for a wide range of diseases.
- The privacy and confidentiality, discrimination, and informed consent concerns that accompany disease prevention, tests and treatment.

Genomics And Disease

Scientists' understanding of genes and disease has grown from understanding the role that genes play in rare, single gene disorders (e.g., sickle cell anemia) to the current awareness of their role in a wide range of common and chronic diseases. In fact, every individual has thousands of genetic variations that could influence disease risk. Genetic variations have been identified that play a role in:

- Chronic diseases (e.g., cancer, cardiovascular disease, Alzheimer's disease)
- Environmental diseases (e.g., asthma)
- Occupational diseases (e.g., bladder cancer)
- Infectious diseases (e.g., HIV/AIDS)

While all genes are inherited, all genetic mutations are not. Some mutations are inherited in families, while others occur in an individual randomly or as a result of environmental factors, such as drugs and infections. Most common diseases have multi-factorial causes, with a complex combination of genetic and environmental factors at work, including modifiable risk factors. As shown below in Figure 1, the relative contribution of genetic and environment factors varies by condition.

Some genetic variations—defined as differences in gene frequencies between people or among populations—put individuals at increased risk for developing certain diseases, but they do not assure that those diseases will occur. Others, including fully-penetrant conditions such as Huntington's Disease, cause the disease to occur.

Figure 1. Contribution of Genetic and Environmental Factors by Condition

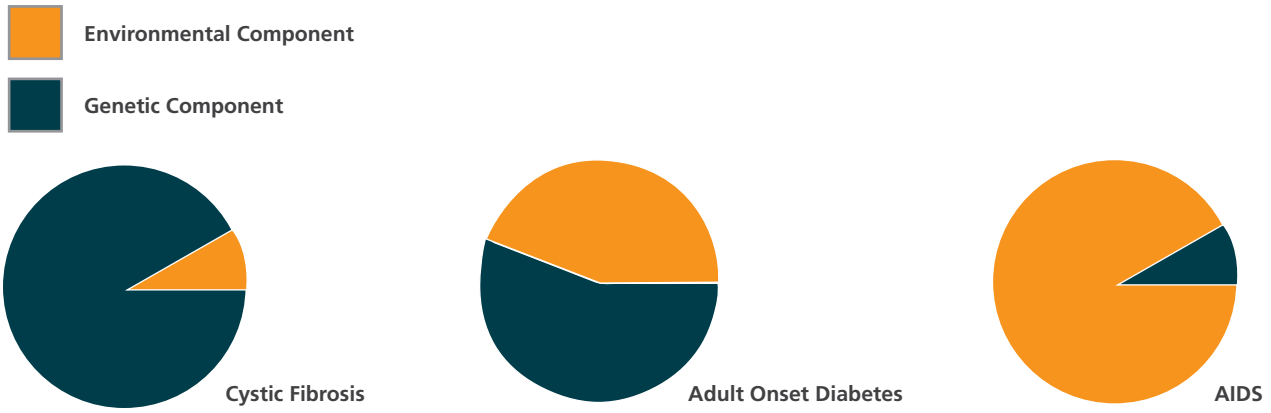
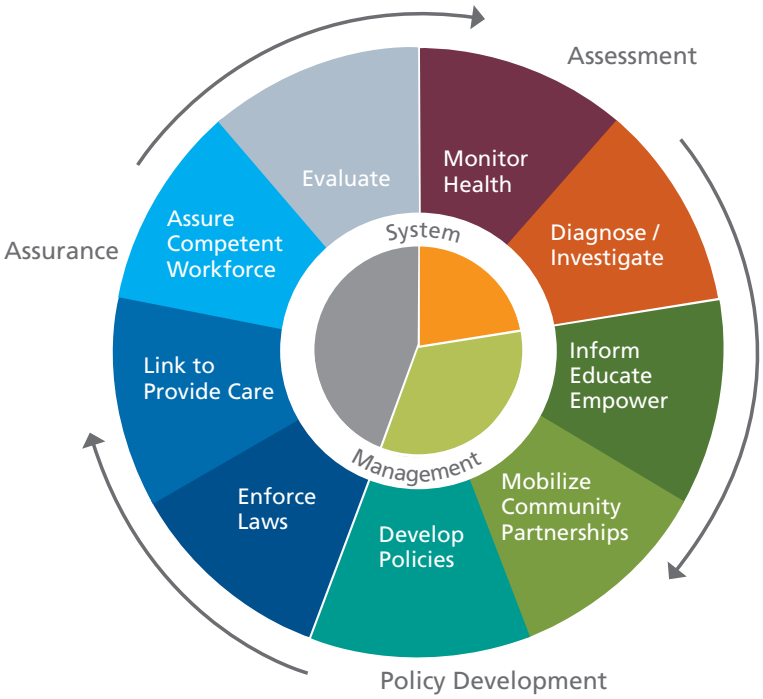


Figure 2. Core Public Health Functions and Essential Services



Source: CDC, Genetics, Genomics and Public Health: An Overview, 2009.

Why Genomics Matters to Public Health

State and local public health officials are challenged to set standards for genomics within the core public health functions of assessment, policy development and assurance, shown in Figure 2. These core functions are defined as follows:

- **Assessment:** The regular systematic collection, assembly, analysis and dissemination of information, including genetic epidemiologic information, on the health of the community.
- **Policy Development:** The formulation of standards and guidelines in collaboration with stakeholders that promote the appropriate use of genomic information and the effectiveness, accessibility and quality of genetic tests and services.
- **Assurance:** Assuring that genomic information is used appropriately and that genetic tests and services meet agreed-upon goals for effectiveness, accessibility and quality. States fulfill these core functions by conducting various essential public health services (listed at left).

Most states do not have the resources to perform all 10 services, and instead focus on delivering the highest priority ones. One state might address genetics workforce gaps while another examines the role of family history and other risk factors for sudden cardiac death in young people. As shown in the next section, states rely heavily on partnerships with academia, health care providers and organizations, policy experts, private foundations and public agencies to deliver these services.

Ten Essential Services of Public Health

1. Monitor health status to identify community health problems.
2. Diagnose and investigate health problems and health hazards in the community.
3. Inform, educate and empower people about health issues.
4. Mobilize community partnerships to identify and solve health problems.
5. Develop policies and plans that support individual and community health efforts.
6. Enforce laws and regulations that protect health and ensure safety.
7. Link people to needed personal health services and assure the provision of health care when otherwise unattainable.
8. Assure a competent public health and personal healthcare workforce.
9. Evaluate effectiveness, accessibility and quality of personal and population-based health services.
10. Research for new insights and innovative solutions to health problems.

Source: American Public Health Association.

The Changing Genomics Landscape

The genomics landscape is vastly different today than it was when ASTHO published the first edition in 2003. Some of the notable changes in recent years are summarized next.

Research and Technology. By 2003, the Human Genome Project had achieved all of its initial goals, including its landmark achievement, sequencing the human genome. According to the National Human Genome Research Institute (NHGRI), "The international effort to sequence the 3 billion DNA letters in the human genome is considered by many to be one of the most ambitious scientific undertakings of all time, even compared to splitting the atom or going to the moon." This accomplishment paved the way for subsequent research and discovery to "combat disease and improve human health." To that end, the NHGRI has supported public and private research that pursues:

- New tools that identify hereditary contributions to common diseases, such as diabetes, heart disease and mental illness.
- New methods for the early detection of disease.
- New technologies that can sequence the entire genome of any person for less than $1,000.
- Wider access to tools and technologies of "chemical genomics" to improve the understanding of biological pathways and accelerate drug discovery.

Researchers have achieved many of these goals, and they continue to discover new tools and technologies that promise to transform how health care is delivered. According to the NHGRI, "Genome-based research is already enabling medical researchers to develop more effective diagnostic tools, to better understand the health needs of people based on their individual genetic make-ups, and to design new treatments for disease."

Public Policy. Although advances in genomics and genetic medicine offer great promise to public health, they also present serious consumer protection challenges, in part due to the predictive nature of genetic information. As a result, states and the federal government have passed legislation to protect patient privacy and prohibit discrimination based on predictive genetic and family history information.

- The Health Insurance Portability and Accountability Act of 1996 prohibited group health plans from using genetic information to establish rules for eligibility, treating genetic information as a pre-existing condition, and using genetic information to set premium contributions.
- In 2000, President Clinton signed an Executive Order that prohibited federal departments and agencies from using genetic information in hiring or promotion activities.
- The Genetic Information Nondiscrimination Act of 2008, or GINA, prohibits health plans and employers from discriminating against individuals based on their genetic information. The landmark federal legislation offers several other protections, such as prohibiting health insurers from requiring genetic testing and requiring employers to treat employee's genetic information like a confidential medical record.
- Prior to GINA, many states already had laws that restricted the use of genetic information; however, the laws vary considerably from state to state. According to the National Conference of State Legislatures, 34 states and the District of Columbia prohibit employer discrimination based on genetic information and 44 states and the District prohibit health insurers from using genetic information to determine eligibility.

States also are examining the need for policies that allow consumers to decide if they want to participate in genetic research. Oregon's Genetic Privacy Law, for example, requires health care providers to inform patients about their rights and allow them to opt-out of anonymous or coded genetic research. Privacy laws reflect concerns about patient privacy; however, lawmakers grapple with how to implement these policies in a way that does not hinder valuable genetic research.

For more information on GINA and Genetic Privacy, see the list of resources and links in Section II. The National Human Genome Research Institute has a GINA fact sheet at **http://www.genome.gov/10002328**.

In addition to these policy developments, federal and state policymakers and public health officials also are passing or considering laws and policies that address a range of other topics, including direct-to-consumer genetic tests, repositories of DNA samples (bio-banks) and workforce competency.

Toward Evidence-Based Practice. The new Healthy People 2020 Genomics Topic Area and Objectives that were recently approved by the Healthy People 2020 Federal Interagency Workgroup reflect the increasing scientific evidence supporting the health benefits of using genetic tests and family health history to guide interventions. The two new objectives focus on implementing evidence-based recommendations from the U.S. Preventive Services Task Force for hereditary breast and ovarian cancer (BRCA) and the Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group for hereditary colorectal cancer (Lynch syndrome). The potential for improving health is great by putting these recommendations into practice, and public health agencies can make important contributions by assuring that policy, provider and public education, and surveillance activities are informed by the current state of scientific evidence for these and other genomic applications.

The movement toward evidence-based practice is reflected in federal funding priorities. CDC's Office of Public Health Genomics (OPHG) has been an important funding source for a small number of states. From 2003 to 2008, OPHG funded four state health departments (Michigan, Minnesota, Oregon and Utah) to establish genomics programs to help integrate evidence-based genomics knowledge, tools (e.g., family history assessments), and surveillance findings into state and local chronic disease prevention and health promotion strategies and activities. Some federally-funded chronic disease programs, such as CDC's WISEWOMAN[i] program requires participating states to integrate family health history risk assessments and educational activities into their program activities.

In 2008, the OPHG funding priorities shifted from capacity-building initiatives to specific projects that translate genomics research findings and information into education, surveillance and policy interventions. These projects fall under the CDC's translation initiative known as the Genomic Applications in Practice and Prevention Network (GAPPNet). This initiative involves researchers, practitioners, policy makers, educators and other professionals who are sponsoring and evaluating research findings, and disseminating high quality information on candidate genomic applications. GAPPNet was formed in 2009 by the CDC, the National Cancer Institute's Division of Cancer Control and Population Sciences and other stakeholders. The key components of GAPPNet are summarized below.

For more information about CDC's genomics activities, visit **www.cdc.gov/genomics/**. For more information about CDC funding for genomics activities, visit **www.cdc.gov/genomics/about/funding/index.htm**

- **Knowledge Synthesis.** Researchers involved in GAPPNet synthesize information on the validity, utility and impact of genomic applications (such as genetic tests or family history) and disseminate information to researchers, policymakers, providers and consumers. Several tools are available that ensure a systematic approach for reviewing research and developing recommendations. For example, the U.S. Preventive Services Task Force Procedures Manual outlines methods for developing reviews and recommendations for clinical preventive services.
- **Evidence-Based Recommendations.** Researchers use the synthesized information to develop evidence-based recommendations about the use of genomic applications. These recommendations help policymakers, healthcare providers and consumers make informed decisions about genetic tests and technologies, family health history and other genomic applications. The EGAPP Working Group was formed in 2005 to develop a systematic process for assessing the wide-ranging evidence about genetic tests for clinical practice. The EGAPP Working Group is an independent, multi-disciplinary panel of experts that prioritizes tests, reviews evidence, identifies information gaps and provides guidance on the appropriate use of tests.

- **Translation Research.** Translation research involves the following: using knowledge to develop a health application (such as a new genetic test); assessing the value of these applications; moving evidence-based recommendations into health practice; and evaluating the health outcomes for each application. Several federal agencies are funding investigators to conduct translation research, including the National Cancer Institute at the National Institutes of Health and the CDC's National Office of Public Health Genomics.
- **Translation Programs.** GAPPNet-funded programs help to translate genomic knowledge and applications into public health and clinical practice programs. State health agencies in Michigan and Oregon have received CDC funding to translate research into education, surveillance and policy interventions for chronic diseases. The Michigan Department of Community Health received funding to promote cancer genomics best practices and decrease morbidity and mortality related to hereditary cancers in the state. The Oregon Genomics Surveillance Program received funding to "develop, implement, and evaluate a surveillance program to monitor awareness, knowledge, and use among health care providers and the public of cancer-related genomic tests and family history in Oregon."

This shift in federal funding priorities has therefore impacted states that had relied on federal funds to support their genomics programs; however, it provided new funding opportunities for state health agencies that focus on translating research into health practice.

(1) Centers for Disease Control and Prevention. *10 Years of Public Health Genomics at CDC*. December 2007. Available at **http://www.cdc.gov/genomics/about/reports/2007/index.htm.**
(2) U.S. Department of Energy Office of Science, Human Genome Program. "International Consortium Completes Human Genome Project." Available at **http://www.ornl.gov/sci/techresources/Human_Genome/project/50yr/press4_2003.shtml. Accessed 06-09.**
(3) Ibid.
(i) The CDC's Division of Heart Disease and Stroke Prevention administers the WISEWOMAN program, which provides eligible women (low-income women between the ages of 40 and 64 who are under-insured or uninsured) with chronic disease risk factor screening, lifestyle intervention, and referral services to prevent cardiovascular disease. CDC funds 21 WISEWOMAN programs in states and tribal organizations.

2

Section Two

Promising Practices

State public health agencies have adopted numerous approaches for integrating genomics into their public health infrastructure. This section profiles states that have adopted strategies to fulfill their core public health functions of assessment, policy development and assurance. These states demonstrate that genomics is a valuable tool for achieving the core functions and essential services, listed in Section One. Some examples of state actions include:

- Collecting and analyzing data about a public health problem, such as sudden cardiac death of the young, and using the data to develop policies and procedures to prevent and manage health care conditions.
- Informing and educating the public and health care providers about the importance of family health history in preventive and primary care.
- Developing and enforcing laws that ensure privacy and prohibit discrimination based on genetic information.
- Ensuring that the public has access to genetic services and providers.

The state examples that follow in Table 3 illustrate what states do, how they do it, who they work with and what problems they encountered along the way. Included are in-depth state profiles and summaries. The state examples included in this chapter are listed in Table 4. It should be noted that while a state example falls under one public health core function, the strategy may likely encompass activities that fall under another area. The core public health functions merely provide a framework for presenting state strategies.

Table 3. Summary of States Profiled in Section II

Public Health Function	Action Area	Success Stories
Assessment	Improving Data About Genetic Services and Inherited Conditions	Public Health Genomics Surveillance in **Oregon**
		Examining Sudden Cardiac Death of the Young in **Michigan**
Policy Development	Integrating Genomics into Public Health Programs	**Michigan's** Healthy Homes University: Using Family Health History to Achieve Healthy Homes
	Building Public Health Infrastructure and Capacity to Address Genomics	**Hawaii** Integrates Community Needs into Planning and Programs
		Developing Public Health Genomics Capacity in **Connecticut**
	Achieving Public Genomic Literacy	Using Family Health History to Increase Genomic Literacy in **Utah**
Policy Development/ Assurance	Addressing Emerging Public Policy Issues	• Three-State Collaboration to Inform the Public about Direct-to-Consumer marketing for BRCA Genetic Testing • Examining Feasibility of a Statewide Bio-Bank in **Connecticut** • Protecting Patient Privacy and Discrimination in **Oregon**
Assurance	Ensuring Access to Genomic Resources and Services	**Hawaii's** Community Genetics Program Addresses Workforce Gaps
		Addressing Access Challenges in **Illinois**
	Addressing Workforce Issues to Meet Demand for Genetic Services	Meeting Demand for Genetic Services in Various States (e.g., **Connecticut, Illinois, Michigan, Oregon** and **Washington**)

How This Section Is Organized

The remainder of this chapter provides an overview of each of the action areas, as well as one or more state profiles or examples. The chapter is organized as follows.

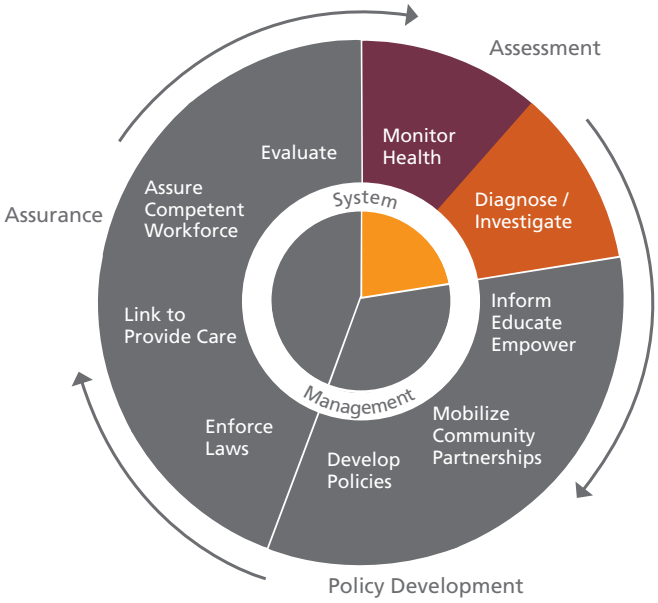
- **Core Public Health Function.** The three core public health functions include assessment, assurance and policy development. Included under each core public health function are concrete state examples that address the public health function. For example, the assurance section contains examples relating to ensuring access to genomic services and addressing workforce issues.
- **Action Area.** Each core public health function contains one or more key action areas. For example, in the area of policy development, states are taking action to integrate genomics into public health, build public health infrastructure and achieve public genomic literacy. The action area sections include background on why the area is important, common challenges and an overview of state strategies.
- **State Profiles and Success Stories.** State profiles include detailed information about a state strategy, including their blueprint for action, what they learned (labeled "take-away messages," and links for further information on the program and links to resources and tools, such as family health history toolkits, brochures and provider handbooks.

Public Health Function: Assessment

Assessment Defined: The regular systematic collection, assembly, analysis and dissemination of information, including genetic epidemiologic information, on the health of the community.

States perform various activities related to the assessment function, including: identifying public health problems, identifying available resources, evaluating their effectiveness, and presenting findings to decision makers. For example, states utilize population-based surveillance systems and registries to obtain data about birth defects and disease prevalence, as well as to gather information about public understanding and perceptions of genomics. As described later in this section, the Michigan Department of Community Health collects and analyzes data from various sources to examine the role of family history and other risk factors for sudden cardiac death in young people. It uses these data to recommend health care system changes and family-based interventions to prevent premature death.

This section on assessment strategies profiles state actions that focus on improving data about genetic services and inherited conditions.



Source: CDC, Genetics, Genomics and Public Health: An Overview, 2009.

Action Area

Improving Data about Genetic Services, Inherited Conditions and Birth Defects

"Adding questions to public health surveillance systems demonstrates a concrete method of integrating genomics into public health programs."

— Oregon Department of Human Services

Surveillance Defined: "The regular systematic collection, assembly, analysis and dissemination of information, including genetic epidemiologic information on the health of the community."

Source: CDC presentation, Genetics, Genomics and Public Health: An Overview, 2009.

Assessing the population's health is a core public health function. According to *Genetics and Public Health in the 21st Century*, "The practice of public health begins with effective surveillance of physical characteristics, diseases, behavior and environmental conditions that significantly affect a population's well-being." Public health agencies collect and analyze a wide range of data about the population's health, from information about the prevalence of birth defects to assessing the public's awareness of genetic tests.

States integrate genomics into surveillance systems, population-based surveys and disease registries to gather important information about how genetic factors interact with environmental, behavioral and other factors to impact the public's health. Examples include utilizing existing cancer registries to determine the relationship between family history of breast cancer and development of the disease, or evaluating clinical records to assess health care provider practices related to collecting and using family health history. Another common approach for states includes adding genomics questions on existing population-based surveys, such as the Behavioral Risk Factor Surveillance System (BRFSS), to gather additional information about genetic conditions and awareness and use of family health history or genetic tests. The BRFSS and other commonly used data sources are described at left. These data provide critical information that helps public health leaders identify unmet needs, monitor trends and develop targeted public health interventions to improve public health. For example Oregon's Genetics Program, described later in this section, collects information about family history and health care provider practices to monitor health status and improve public health practices.

Commonly Used Data Sources

States have numerous existing surveillance systems that can be used to gather important information about genetic conditions, health behaviors and environmental factors.

- **Behavioral Risk Factor Surveillance System:** State-based system of health surveys that gather information about health risk behaviors, clinical practices, and health care access and use. Several states include questions about public understanding and experience with genomics issues, including family health history and genetic testing.
- **Disease Registries:** State-based **systems** to collect information about individuals diagnosed with specific conditions, such as cancer, diabetes and birth defects. Disease registries may already collect important information about family disease history.
- **Pregnancy Risk Assessment Monitoring System (PRAMS):** CDC and state health department surveillance that collects population-based data on maternal attitudes and experiences related to pregnancy. The Oregon Genetics Program uses PRAMS to monitor health care provider practices related to topics such as folic acid consumption and awareness and prenatal tests. Michigan uses PRAMS to gather information about folic acid and newborn screening.
- **Youth Risk Behavior Surveillance System (YRBSS):** CDC and state health department surveillance that monitors health risk behaviors, such as sexual activity and alcohol, drug and tobacco use, and the prevalence of obesity and asthma among children and young adults.
- **Vital Statistics:** Systems for recording births and deaths, available through state vital records offices, as well as through national sources, such as the National Center for Health Statistics.

Challenges

- **Public Concerns about Surveillance Risks.** The general public, and specific population segments affected by the surveillance activity, may have concerns about potential economic, employment, social and other risks associated with sharing genetic and other health information. Examples of state strategies for addressing this challenge include public information and education about the use of genetic information, policies that ensure anonymity and/or patient privacy, and policies that allow consumer to "opt-out."
- **Resource Demands.** Developing policies and educating the public and providers about surveillance systems demands staff time and resources. States rely on partnerships with internal and/or external stakeholders to address their highest-priority genomics data needs. These partnerships are especially important in states that do not have a genomics staff.

Strategies to Improve Surveillance and Monitoring

Public health agencies have adopted various strategies to collect important information about family health history and inherited conditions. As shown in Table 4, state strategies vary, but some of the more common approaches focus on maximizing readily available data, as well as integrating genomics-related questions into existing surveillance systems. In some cases, states design new surveys and procedures to fill in the gaps. Oregon, as described below, adopts all of these strategies.

Up Close: Public Health Genomics Surveillance in Oregon

The Oregon Genetics Program uses multiple data sources to collect information about family history, health care provider practices, genomics, genetics, and chronic disease. According to the program website, "By collecting and analyzing population-based data, our program monitors health trends and uses the data for program improvement and evaluation." Some examples of Oregon's surveillance activities include:

- The Oregon General Knowledge Survey includes various questions about the public's understanding of genetics and family history, their collection of family history, where they obtain genetics and family history information (e.g., television, Internet, health care provider, etc.) and topics of interest.
- The Behavioral Risk Factor Surveillance System includes questions about genomics topics, including family history of disease, health care provider practices, patient behaviors and direct-to-consumer marketing of genetic tests.
- The genetics program staff examined family history collection and use in all 26 of its federally qualified health centers and developed a family history template that incorporated family history research and clinician recommendations.

For more information on Oregon's surveillance activities, see the genetics program website at **http://www.oregon.gov/DHS/ph/genetics/surveillance.shtml**

Table 4. Examples of State Surveillance Approaches

Objective	Activities and Tactics
Maximize Existing Data	• Evaluate and analyze genomics-related information already collected through surveys and registries. Some states have identified valuable, but under-utilized, genomics-related data that has already been collected, but perhaps not analyzed. For example, Utah's Breast and Cervical Cancer Program had been collecting, but not analyzing, family health history information for over 20 years. In 2006, the public health genomics program staff analyzed the cancer database to examine the relationship between family history and the risk of developing breast cancer, and found a positive family history in 46 percent of breast cancer cases. • Evaluate and analyze genomics-related information that is already collected for clinical purposes including medical records reviews, and client enrollment forms for cancer programs. Several states, including Michigan, Minnesota and Utah have analyzed existing clinical data on family health history available through doctor's office chart reviews, electronic medical records, local public health encounters and other sources. Michigan's WISEWOMAN program, asks enrollees about family history of heart disease and stroke, as well as smoking history and medical history. This family history information is integrated into counseling and follow-up. • Gather data from multiple sources to advance public health leaders' understanding of genetics, environment and disease. For example, Michigan's Sudden Cardiac Death of the Young project uses various data sources to increase understanding of the problem and guide development of education and prevention policies to prevent premature death.
Integrate Genomics into Existing Systems	• Identify existing surveillance systems that could be expanded to include genomics-related questions. Several states, including Connecticut, Michigan, Minnesota, Oregon, Utah, Washington and Wisconsin, have included questions about family history of chronic disease, knowledge of genetic tests, perceived risks and other topics in their Behavioral Risk Factor Surveillance System.
Develop New Systems to Address Data Gaps	• Identify gaps in surveillance and develop new surveillance systems that provide essential data for public health planning. • The Oregon General Knowledge Survey, described on the previous page, assesses public understanding of genetics and identifies knowledge gaps.
Improve the Quality of Existing Data	• Develop systems to improve the accuracy and effectiveness of genomics-related surveillance systems.

Success Story: Michigan at a Glance

Mobilizing Partners to Examine Sudden Cardiac Death of the Young in Michigan

Agency	Michigan Department of Community Health (MDCH)
Program Location	MDCH, Division of Genomics, Perinatal Health and Chronic Disease Epidemiology
Funding	In 2005-2008, used CDC cooperative agreement to fund surveillance of death records, autopsy results, medical records review, next of kin interviews, and investigation of circumstance around individual deaths with expert panel review to suggest actions to prevent sudden cardiac death of the young in future cases. Currently there are no CDC funds available for this project.
Staff	Currently, the adult genetics and genomics coordinator devotes about one-third of her time to this project. Other staff, including the chief medical executive, vital records staff member, cardiovascular public health consultant, and epidemiologist provide some time to the project (i.e., 10 percent or less).
Sustainability	State newborn screening funds are used to support the genomic coordinator's staff time, and the project is moving ahead because of commitment from work group members, an "invested" chief medical executive and MDCH staff who are rolling out the 2008 report recommendations.
State Plan	http://www.michigan.gov/documents/MIgeneticsplanandassessment__118168_7.pdf
Program Website	http://www.michigan.gov/mdch/0,1607,7-132-2942_4911_4916-85137--,00.html

"The best way to address any public health program is to engage everyone around a core issue. When you find the right core issue, the egos walk out of the room. The key is finding the public health issue and engaging the right people around it."

– Debra Duquette, Adult Genetics and Genomics Coordinator

Sudden Cardiac Death of the Young (SCDY) claims the lives of more than 300 Michigan children and young adults between the ages of 1 and 39 years annually, and many of these deaths could be prevented through screening, detection and treatment. To address this important problem, health department staff mobilized a motivated group of internal and external experts to investigate and reduce the burden of sudden cardiac death of the young (SCDY) in Michigan.

The genomics coordinator oversees the surveillance project and coordinates the various work groups and project tasks, which includes convening a multi-disciplinary group of experts to analyze mortality data, educate the public and providers about risk factors and develop prevention and screening policies. In 2008, with funding and support from the MDCH Heart Disease and Stroke Prevention Unit, MDCH published the report, Too Young to Die: Impact of Sudden Cardiac Death of the Young in Michigan, which identified several action steps aimed at improving pre-participation sports screening, provider education and public awareness, and emergency response protocols. As a result of this report, one work group reviewed sports participation forms from other states and made recommendations to the Michigan High School Athletic Association to revise Michigan sports forms based on best practices and professional recommendations from the American Heart Association and the 2004 consensus guidelines from multiple organizations (including the American Academy of Family Physicians, American Academy of Pediatrics, America College of Sports Medicine, and others). Although funds are not available for current activities, stakeholders from the health department, health plans, academic centers, hospitals, health systems, private industry, surviving family members, and foundations are implementing these action steps.

Program Goal

"Reduce the burden of SCD of the young in Michigan by identifying public health and medical system changes as well as family-based interventions that might be undertaken to increase awareness of opportunities for prevention, as well as appropriate screening and treatment for relatives potentially at risk."

Source: Michigan Department of Community Health

For More on State Genomics Surveillance Activities:

1. The University of Washington Center for Genomics and Public Health compiled genomics and family history questions from several states and federal population-based surveys from 1999-2006. This includes the Behavioral Risk Factor Surveillance System, the National Health and Nutrition Examination Survey and others. **http://depts.washington.edu/cgph/pdf/Compiled_Genomics_Questions_BRFSS.pdf**
2. Four states' experiences with using the BRFSS to gather genomics information is summarized in a one-page poster at **http://egov.oregon.gov/DHS/ph/genetics/docs/poster11x17_large.pdf**
3. Two states' experiences with reviewing doctor's office chart reviews to examine medical practices related to family health history is summarized at **http://www.cdc.gov/genomics/about/reports/2006/sect3.htm**

Table 5. Methods for Achieving SCDY Objectives in Michigan

Select SCDY Objectives	Activities and Tactics
• Implement and refine a process to collect and review demographic and medical data regarding Sudden Cardiac Death of the Young (SCDY). Establish a SCDY review system. • Identify public health and medical system changes. • Develop consensus-based recommendations to guide public health prevention efforts for reducing SCDY in Michigan. • Inform families of potential risk factors related to heritable causes of SCDY. Identify unmet needs for education, support and medical and genetic resources. • Increase awareness of opportunities for prevention, appropriate screening and treatment for relatives potentially at risk.	• Interviewed medical experts to gather information about practices and procedures following unexpected deaths. • Presented to the Cardiovascular Health Advisory Committee meeting in 2005 and confirmed stakeholder interest in developing a mortality review system. • Hosted a SCDY symposium in 2005 to review causes of SCDY and assess the need for a state surveillance system and awareness campaign. • Analyzed existing mortality records through death certificate data. • Asked family history questions on the 2007 Behavioral Risk Factor Survey to identify how many Michigan families are affected by SCDY. • Partnered with Michigan State University to develop definitions, protocols and data collection instruments for SCDY investigation. • Assured that all information is confidential and used for public health purposes only. • Identified recent SCDY cases through death certificates to thoroughly investigate through review of autopsy report and medical records, and conducting next of kin interviews. • The Expert Reviewer Panel met to review recent SCDY cases and suggest possible factors that could prevent SCDY in future cases. • In 2008, the MDCH published the Too Young to Die report and convened a Call to Action Meeting to identify action steps and assign responsibilities to experts and stakeholders. • Work groups are currently addressing action steps in five major areas: pre-participation sports screening; provider education and public awareness of SCDY risk factors; public awareness of cardiac symptoms and CPR training; emergency response protocols; and medical examiner protocols.

Take Away Messages

- Find the right issue to bring a right people together.
- Use multiple avenues to gather information.
- Partnerships make it possible to do more with less.

Informational Resources

Too Young to Die: Impact of Sudden Cardiac Death of the Young in Michigan 1998-2008 Final Report
http://www.michigan.gov/documents/mdch/SCDYReportfinalJan09_269478_7.pdf

MDCH Burden of Cardiovascular Disease Website, with reports and fact sheets
www.michigan.gov/cvh

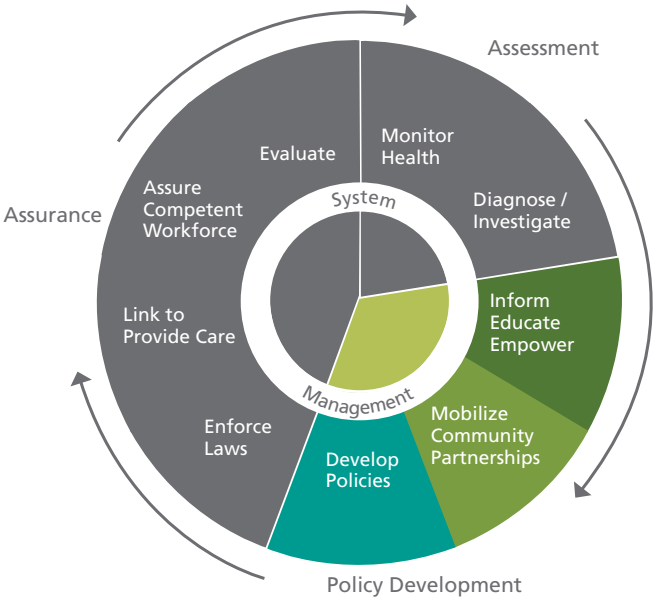
Public Health Function: Policy Development

Policy Development Defined: The formulation of standards and guidelines in collaboration with stakeholders that promote the appropriate use of genomic information and the effectiveness, accessibility and quality of genetic tests and services.

Public health leaders are challenged to develop balanced policies that reflect the dual goals of improving public health and protecting individuals. Policy development activities include formulating standards and guidelines that promote the appropriate use of genomic information, as well as ensuring that genetic services and technologies are effective and accessible. In many cases, the policy development activities rely on productive partnerships among public and private stakeholders. For example, state health agencies develop policies and processes for parental informed consent for newborn screening, as well as fact sheets to inform consumers about a wide range of genomics topics, such as knowing when to seek genetic counseling and testing.

This section on policy development profiles state actions in Michigan, Hawaii, Connecticut and Utah that focus on the following:

- Integrating Genomics into Public Health Programs
- Building Public Health Infrastructure and Capacity to Address Genomics
- Achieving Public Genomic Literacy
- Addressing Emerging Public Policy Issues



Source: CDC, Genetics, Genomics and Public Health: An Overview, 2009.

Action Area

Integrating Genomics into Public Health Programs and Practice

States are integrating genomics in public health programs, for example, working with other programs to build knowledge and partnerships, as well as embed genomics tools and services into practices of other programs.

Public health activities traditionally associated with genetics have included newborn screening, reproductive health, and clinical genetic services. In recent years, state genomics programs have built relationships with a growing number of programs, including chronic disease, infectious disease, environmental health, epidemiology, health disparities and others. In many cases, genomics staff members have focused on integrating family health history tools into cancer, diabetes, cardiovascular disease or asthma programs to identify people at risk and educate them about reducing their disease risk.

Although there are other areas for collaboration, this focus on family health history has garnered many early successes since it is an inexpensive, relatively simple, and effective method for integrating genomics into public health programs and practices.

Challenges

- **Staying Up to Speed.** Public health leaders and staff may not understand genomics or its implications for their work. For many, their public health training may have pre-dated recent advances in genomics. Educating health department colleagues and sustaining support requires a productive and ongoing relationship, which is especially challenging in states without a genomics expert or team. States in this circumstance might consider partnering with genetics professionals in state to provide continuing education, seminars, etc.

Why Integration Matters:

Integrating genomics/family health history into public health practice offers a number of potential benefits for state and local health officials as they set priorities, make funding proposals, guide and motivate staff, and develop communication strategies. Integrating genomics/family health history offers:

1. Creates more targeted, cost-effective public health programs with improved outcomes.
2. Serves as a unifying force for public health agencies.
3. Boosts the impact of risk reduction campaigns and messages.
4. Conveys the urgency of public health initiatives to policymakers, providing evidence for the need for increased funding and thoughtful legislation in the many areas that are impacted by genomics.
5. Sparks collaboration at the state and community levels with important new constituencies. Increased partnerships offer the potential to share resources, broker influence, expand credibility, and gain entry to communities and organizations.
6. Maintains relevancy of public health at a time of great potential change in medical treatment toward personalized health care.

Challenges (continued)

- **Lack of Leadership Support.** Many state programs struggle to begin or maintain genomics work because agency leaders do not support staff time and expenses related to genomics. Obtaining support from agency leaders requires that they understand public health genomics and see how it fits within the public health infrastructure. They also want to know the impact on public health outcomes.
- **Lack of Dedicated Funding.** Genomics is not always an agency priority and must compete for funding with many other efforts, including mandated programs. It is difficult to maintain and expand genomics programs when many states are cutting budgets or committing resources to more immediate needs. As a result, states pursue funding from new sources to supplement or replace state funding. Without funds, states often rely heavily on the contribution of a genomics champion who advocates for genomics and identifies opportunities when possible.
- **Lack of Evidence Demonstrating Public Health Outcomes.** Public health officials often lack substantive data about the public health impact of genomics activities. As a result, other public health program staff and agency leadership may not understand the added value of genomics in disease prevention and improving public health. Whenever possible, identifying public health and cost effectiveness data is therefore a key component of any genomics activity.

Public Health Programs and Practice

- Public Health Programs: Strategies aimed at working with other health programs to build knowledge and awareness of genomics, and identify areas for collaboration. For example, the Michigan Department of Community Health worked with another program to integrate family health history into a healthy homes outreach program.
- Public Health Practice: Embedding genomics tools and services into public health activities. As an example, some public health programs work with public health providers to use family health history forms and protocols in their medical practices.

Strategies To Integrate Genomics Into Public Health

Integrating genomics into policy and practice happens in many ways, but the basic components typically include internal genomics expertise, relationship-building and ongoing consultation and collaboration among program staff. The following table summarizes activities and practices that states have adopted (or considered adopting) to facilitate genomics integration.[1]

Table 6. Examples of State Activities that Integrate Genomics into Public Health

Objectives	Activities and Tactics
Promote Integration of Genomics into Public Health Infrastructure	**State public health agencies:** • Provide resources and funding to support genomics staff members who are charged with implementing state plans, staying "current" on genomics and public health, and coordinating internal and external partnerships and projects. • Consider program location in the organization. To avoid creating a separate program "silo," the Connecticut Department of Health Genomics Office is in the Department's Planning Branch and the Michigan Department of Community Health's Genomics and Genetic Disorders Section is in the Bureau of Epidemiology, rather than in a program area such as newborn screening or chronic disease. This facilitates integration into multiple program areas and department projects. **State Genomics Programs:** • Develop and maintain internal relationships by providing education, expertise and ongoing consultation. This promotes sustainability by integrating genomics into other program grants and activities, such as cancer control plans, WISEWOMAN, WIC counseling or Healthy Homes University programs. • Conduct environmental scans with consideration of genomic advances and readiness of other public health programs to identify opportunities to strengthen public health activities, such as the Michigan Department of Community Health's tobacco genomics workgroup and gene-environment workgroup. • Establish staff buy-in by focusing on integrating genomics into chronic disease programs. Oregon, for example, accomplished this by using a Stages of Change model to assess chronic disease program readiness to integrate genetic tools and information. • Integrate family health history into population surveillance systems. By piggybacking on existing data collection efforts, states are gathering key information about genomics without having to develop new surveillance systems. For an example of this, see Michigan's approach to integrating genomics into an existing surveillance system later in this section. • Develop external partnerships to share human and financial resources, and maximize results. Every state struggles with limited resource. Partnering with external experts help states leverage departmental resources and expand knowledge and expertise to advance projects.
Promote Integration into Health Care System	• Inform health care providers about genomic science and provide realistic and effective methods for integrating into health care practice. State health departments educate providers in a variety of ways, through websites, online manuals, online curricula for nurses and other professionals, workshops and continuing education opportunities. • Develop partnerships with genetics, primary care providers and other health professional organizations. Several state programs partner with national and state provider organizations to utilize their expertise, disseminate information and roll-out practice guidelines and tools. • Strengthen provider ability to provide basic genetic services to patients throughout the lifespan. For example, the Illinois Department of Public Health developed a family health history screening tool for local health departments and genetic clinics to identify individuals who need additional services. States also are informing practitioners and patients about genetic tests.

1. Most of the activities are being implemented by state health departments; however, others have been identified by state health departments, through their state genomics plans or other planning documents, as a recommended path for achieving integration. These are included here to provide a broad menu of options for states that wish to consider alternatives for integrating genomics into their public health infrastructure.

Success Story: Michigan at a Glance

Healthy Homes University: Integrating Genomics into Environmental Health in Michigan

Agency/Program Name	Michigan Department of Community Health, Bureau of Epidemiology, Healthy Homes Section
Program Location	The Healthy Homes University program is located in the Michigan Department of Community Health (MDCH) Healthy Homes Section (HHS), in the Bureau of Epidemiology. The MDCH Genomics and Genetic Disorders Section (GGDS) provide staff time to integrate family health history into program activities.
Funding	The U.S. Department of Housing and Urban Development (HUD) awarded approximately $1 million to the HHS to fund the Healthy Homes University (HHU) program from 2005 to 2008, and $875,000 to fund the program from 2008-2011 (HUD funds do not cover GGDS staff time).
Staff	In the first phase, two GGDS staff members worked part-time on the HHU project (10 percent or less) and that portion of their salary was covered by CDC cooperative agreement funds. In the second phase, the genomics coordinator's time on the project (approximately 10 percent or less) is covered by state funds.
Sustainability	HUD funding sustains the HHU program, while provision of genomics expertise has been incorporated into the genomics coordinator's ongoing role.
State Plan	http://www.michigan.gov/documents/MIgeneticsplanandassessment__118168_7.pdf
Program Website	http://www.michigan.gov/genomics

Issue In Brief

In 1999, Congress established the U.S. Department of Housing and Urban Development (HUD) Healthy Homes Initiative to "develop and implement a program of research and demonstration projects that would address multiple housing-related problems affecting the health of children." Since that time, HUD has awarded grants to state and local agencies, non-profits and academic institutions to address housing-related health problems.

Michigan's Healthy Homes University, which began in 2005, illustrates how family health history can be a powerful tool for addressing complex environmental health problems, such as childhood asthma and allergies. The HHU program provides education as well as a range of targeted intervention products and services (e.g., HEPA vacuums, furnace filters, carpet removal, pillow and mattress covers) to eligible homes. Eligible families have low to moderate incomes and a child under the age of 18 with asthma. The Healthy Homes Section in Michigan's Department of Community Health administers the program, and the Genomics and Genetic Disorders Section lends staff time to integrate family health history information into the program's activities.

According to Debra Duquette, Genomics Coordinator, integrating family history into the program not only improved health outcomes, it helped to open doors and build trust among participants who may have otherwise been wary of the "state com[ing] into their home." It "could have been all about the house," but instead, the focus on family health history and health builds trust and "creates a different dialogue." What's more, the program seems to be working. Between 2005 and 2008, the program reported a 50 percent decrease in self-reported asthma symptoms and 70 percent reduction in emergency department and hospital visits for the primary child identified with asthma in each home.

"Many [states] have had these state Healthy Homes Programs, and many have faltered because they couldn't recruit households. It's hard to have the 'State' come into your home. But when you include family history, it opens up the lines of communication with people. The state isn't coming to talk about your home and its cleanliness; they're coming to educate the family and reduce asthma symptoms."

– Debra Duquette, Adult Genetics and Genomics Coordinator

Table 7. Blueprint For Integrating Genomics Into The Healthy Homes University Program

Select HHU Objectives and Desired Outcomes	Select Activities and Interventions
• Apply principles of gene-environment interactions and family history knowledge in an actual public health project. • Show the public health impact of collecting information on family history of asthma. • Promote positive change in family knowledge, attitudes and behaviors regarding asthma triggers and injury reduction. • Reduce frequency of asthma and injury emergency care events and school absence.	• The Healthy Homes Section partnered with the Genomics and Genetic Disorders Section to integrate family history questions into the baseline survey of families. These questions gather information about family members with asthma. By collecting family history for all family members, and not just the participating child, the program can more accurately capture program impact, since the interventions are typically affecting more than one individual in each home. • Develop educational materials to inform families about asthma. The genomics staff collaborated with the asthma program to develop family health history fact cards that HHU program staff members distribute to participating families. • Educate participating families about asthma and healthy homes. HHU staff members visited homes a minimum of four times during the six-month enrollment period to collect baseline information about the family and the house, provide asthma and injury control education, install asthma or injury reduction products or services, and evaluate knowledge, attitudes and family behaviors. • Enroll and work with families to reduce the effects of environmental factors on children's health. Between 2005 and 2008, 301 homes enrolled in the program for a 6-month period. Between 2008 and 2011, the HHU program plans to enroll 250 homes, with 50 of those homes enrolling for a 12-month period. Program staff will collect family history information for all enrolling families. • Document health outcomes. Between 2005 and 2008, the program reported a 50 percent decrease in self-reported symptoms and 70 percent reduction in emergency department and hospital visits for the primary child identified with asthma in each home.

Take-Away Message

- Internal and external partnerships are critical.
- Family health history is an integral tool for public health initiatives.
- Genomics/family health history provides an innovative way to expand and document the reach of an environmental project.

Informational Resources on the Healthy Homes Program

Healthy Homes University Program Application
http://www.mich.gov/documents/HHU_Application_147414_7.pdf

HUD Healthy Homes Project Abstracts by Region
http://www.hud.gov/offices/lead/hhi/hhabstracts.cfm

Tools You Can Use

Asthma Family History Fact Card
http://www.migeneticsconnection.org/factcards.shtml

Action Area

Building Public Health Infrastructure and Capacity to Address Genomics

"Developing a state genomics infrastructure requires invested leadership, genomics expertise and accessible genetics data."

– Illinois State Genetic Services Plan, 2007

The public health genomics infrastructure refers to the capacity of the public health agency to address current and emerging issues, as well as the public health system at large to integrate genomics advances into public health practice.

Public health agencies vary considerably along the infrastructure continuum. Most states lack genomics resources and capacity, and some have well-developed, sustainable programs that have formed critical linkages with internal and external partners. Although funding is a critical ingredient for these states, they also benefit from other factors, such as support from senior management and other programs, as well as a champion who identifies windows of opportunity with other program areas and helps to move genetics activities forward in the state. "Without a doubt," according to the 2007 report Genomics and Public Health Practice, "the most often cited factor contributing to success was the creativity, perseverance, and commitment of program staff."

ASTHO identified several common building blocks of state public health genomics infrastructure, including:

- **Personnel:** Dedicated staff with a genetics and public health background, and resources;
- **Funding:** Sustainability through short and long-term funds;
- **Organizational Chart:** A "genetics home" where current genetics activities originate;
- **State Plan:** A genetics state plan and/or needs assessment that guides the state's activities and policies;
- **Education:** Centralized information clearinghouse (e.g., a program website) with basic genetics information, a directory of resources, and technical assistance;
- **Data:** Genetics data collected through registries or other databases;
- **Marketing:** Marketing genetics resources to the public, providers, legislators and public health leaders;
- **Communication:** Coordination and communication among various partners, such as state agencies, laboratories, and providers in community health centers, health departments and private practice;
- **Partners:** Internal partnerships and expert advisory committees/councils to provide input on policies, research and program direction.

Public Health Infrastructure Defined:
"Local public health infrastructure includes the systems, competencies, frameworks, relationships, and resources that enable public health agencies to perform their core functions and essential services."

Source: National Association of County and City Health Officials

Challenges

- **Lack of Dedicated Funding.** One interviewee said, "Without a doubt, funding was the biggest factor" in her state's dismantling of the public health genomics programs and activities. Without funding, genomics staff members are transferred to other areas, making it difficult to advance genomics in the department.
- **Lack of Leadership and Staff Support.** Building and maintaining an effective public health infrastructure requires two things that many managers find in short supply, staff time for genomics staff and other program areas to collaborate, and resources to support salaries, services and program activities.

Strategies To Build Public Health Genomics Infrastructure

Developing state public health genomics infrastructure takes place in many ways as shown in Table 8, often beginning with assessing current capacity (e.g., through needs assessments or key informant interviews) and expanding from there. Securing funding is a critical step, as is developing staff expertise, informational and data resources.

Table 8. Examples of State Infrastructure Development Activities

Objectives	Activities and Tactics
Assess Current Infrastructure	• Identify where genetics resides in the state, which could be the newborn screening program, a planning division, or a separate program. • Identify staff knowledge of genomics and/or public health. Assess opportunities for informing and educating agency staff about genomics through meetings, brown bag lunches and internal working groups. • Examine current funding and support for genomics, including grants (e.g., maternal and child health block grants), and state support for staff time and program activities. • Assess scope of current clinical genetics services, identifying service providers, scope of services and locations. A needs assessment or report may contain further data about the quality, availability, and affordability of services. • Several states have conducted needs assessments to identify unmet needs and prioritize funds and resources to address those needs.
Increase State Genetics Capacity	• Identify program staffing and resource needs. • Develop a genomics plan with goals, objectives and activities. These should show how the genomics activities can contribute to public health interventions that are designed to prevent disease and improve health. • Develop a plan to ensure short and long term funding for program staff and activities. Consider including genomics activities into other program grants (e.g., including family history in a cancer or cardiovascular disease grant), identifying private sources (e.g., convening hospitals and third party payers to address workforce inadequacies). • Consider where genetics staff and resources should reside, and the value of centralizing information and human resources into a state genetics program. • Develop and maintain an accessible website which contains genetics information, resources and technical assistance. • Market the genetics program and its resources to the general public, health care providers, and key public health and other state policy leaders. • Develop partnerships with genetic service providers, including genetic counselors and clinical geneticists, to build knowledge and capacity.
Ensure that the State Genetics Program Content Reflects National, State and Community Priorities*	• Convene an expert advisory body that might include key agency staff, external policy and medical experts, consumer representatives and genetics and other health care providers to guide program direction and provide input into policies. • Create opportunities for public dialogue and input to guide program activities and decisions. • Monitor genetics and health policy issues and incorporate them into genetics program planning and activities. • Develop an ongoing evaluation process for the state genetics program and the strategic plan.
Enhance Genetics Content and Competency of Other Public Health Programs and Services*	• Create internal partnerships to identify opportunities for incorporating genetics into existing programs and services. • Develop educational opportunities in genetics for state and local public health program staff. • Assure that genetics program staff and genetics professionals participate in work groups or activities that address public health concerns.

Up Close: Hawaii Integrates Community Needs into Genetics Planning and Programs

In 2000 Hawaii received funds from the U.S. Health Resources and Services Administration (HRSA) to expand and update a state genetics needs assessment and plan. Community involvement has been a key feature of Hawaii's state planning process. To understand the most pressing needs surrounding genetics in the state, the Hawaii Department of Health surveyed practitioners (e.g., public health nurses, physicians and genetics providers) and public health administrators and conducted focus groups with a broad range of the general public. This input became the blueprint for the state genetics activities.

According to Sylvia Au, State Genetics Coordinator in the Genetics Office, the state plan helps her prioritize program activities, develop policies and obtain funding. For example, the needs assessment process revealed strong support for expanding the newborn screening panel. They developed a pilot program for expanded newborn screening and evaluated family satisfaction with the process. This feedback provided critical information that shaped state newborn screening policies.

In addition, consumers and teachers said that there was a need to develop a genetics curriculum and advocate for its inclusion in the local schools. As a result, the Genetics Office staff, which benefited from having a former high school biology teacher on staff, developed a resource kit for teachers, and provided lesson plans and tests that were aligned with state education standards. Hawaii moved forward with its genetics agenda because the health department hired staff to implement the plan. "Without a dedicated state genetics coordinator," Au asked, "who is the one who is going to carry the torch or even use the state plan?" Many states tap their newborn screening coordinator to take on genetics, on top of their newborn screening duties. Hawaii benefited from a state health department decision to fund a permanent state genetics coordinator with formal training in genetics. In addition, the office is able to "do a lot of the extras," Au said, because it uses federal funds to support additional staff members, including four full-time genetic counselors and a project assistant. The office relies on partnerships with academic institutions, third party-payers and community members to achieve the state's genetics goals.

Partnering with community members is an important and ongoing process in Hawaii. Au explained that staff members work closely with Native Hawaiian and Asian/Pacific Islander communities to develop responsive policies and programs, and ensure ethics in research. "We need to be a leader in how policies are developed to protect the public and allow them to take advantage of (genetic) services."

Success Story: Connecticut at a Glance

Developing Public Health Genomics Capacity

Agency	Connecticut Department of Public Health – Genomics Office (DPH-GO)
Program Location	The DPH-GO is located in the Department of Public Health Planning Branch.
Funding	The department funds the genomics office staff salaries, but there is no operational budget for the genomics office, and it does not receive federal funding. The office sometimes receives one-time funding from other program areas to complete special projects, and other program areas have included genomics activities in their federally-funded projects.
Staff	Two part-time staff, including a genomicscoordinator and genomics epidemiologist
Sustainability	Staff is critical to sustain genomics activities, stay current on science and policy, and ensure that genomics has a presence in public health policy and practice. With limited staff and funds, partnerships are a critical tool for sustaining genomics activities.

Issue In Brief

In 2002, Connecticut received federal planning grant funds to assess the state's genomic needs and develop a Connecticut Genomics Action Plan. This planning process identified the need for a permanent genomics office to weave genomics through the public health department activities and in the state. When the multi-year planning grant funds were gone, the momentum among key staff to implement the plan was not. "We didn't let (the Plan) die on the shelf," said Beverly Burke, Connecticut's Genomics Office Coordinator. Instead, the department's high-level planning branch oversaw the plan and coordinated departmental efforts, which focused on forging active internal and external relationships to achieve the state's public health genomics goals. In 2008, the Connecticut Department of Public Health Commissioner established the Genomics Office, which was intentionally kept under the Department's Planning Branch to promote genomics throughout the department's programs, and not just within one program area, such as newborn screening or chronic disease. Keeping genomics at a high level, combined with formal internal and external partnerships, are key components of Connecticut's strategy. As a result, according to Burke and Genomics Epidemiologist Joan Foland, genomics is not another "silo" in public health. Instead, genomics in Connecticut is formalized into the public health infrastructure and integrated throughout the department.

Getting Genomics on the Radar (and Keeping It There)

Getting other public health programs to think about genomics requires the Connecticut Genomics Office staff to build relationships, educate and inform, and consult on projects. Building relationships and keeping people engaged is a critical step in the integration process. Coordinator Beverly Burke explained that a mix of formal and informal meetings and conversations helped them "build bridges" among health programs. The Genomics Office staff found that one-on-one time with staff from other program areas, while time-consuming, has been an effective way to open doors with other program staff. This was important in the beginning, and remains critical once genomics "crosses the threshold" into other program areas, Burke said. Once it is part of other program activities, the tasks shift from educating and obtaining buy-in to helping colleagues integrate genomics into their public health activities by including genomics and hereditary issues into the asthma program's seven-year plan, for example. There is also a time and place for formal training sessions and meetings. Case in point: a DPH intern developed a brief presentation on family health history and presented it to various programs during their regularly-scheduled staff meeting. The presentation provided basic information on family health history, as well as specific information tailored to the program area. At the end, the staff completed a survey and indicated their interest in doing more with family history. Based on this feedback, the Genomics Office staff pulled together a group of interested staff to further develop family health history information. The end result was widely-used family history tools that integrate genomics and chronic disease in the form of a workbook, pocket guide and poster.

Download Copies at:

Family Health History Pocket Guide at http://www.ct.gov/dph/lib/dph/genomics/fhh_guide.pdf

Family Health History Workbook at http://www.ct.gov/dph/lib/dph/genomics/fhh_wkbk.pdf

Family Health History Poster at http://www.ct.gov/dph/lib/dph/genomics/fhh_poster.pdf

Table 9. Blueprint for Developing Genomics Capacity in Connecticut

Goal	Activities that Help the Genomics Office Achieve Goals
Integrate Genomics into DPH Programs	• Establish and formalize relationships with other program areas through the internal Council of Genomics (COG), which convenes 15 members on a monthly basis to discuss, advise on, and participate in Genomics Office activities. The COG membership is cross-cutting, and includes staff from areas such as chronic disease, laboratory services, research and development and health surveillance. • Establish a broad-based internal liaison group, known as the Gene Team. This group meets twice a year and consists of about 35 members from various DPH programs. Members are "genomics ambassadors" to their programs.
Foster Collaboration within DPH and with Local, Regional, and National Partners to Further Integrate Genomics into Public Health Programs	• Establish and formalize relationships with external experts. This is achieved through the Expert Genomics Advisory Panel, which convenes experts from medical, insurance, academia, law, bioethics, genetic counseling and consumer advocacy communities. Their charge is to provide independent guidance and advice to DPH on clinical, scientific, legal, educational, ethical, and social issues related to public health genomics. • Partner with other groups to achieve results. Among its partners are other Connecticut state agencies, academic institutions, the New England Regional Genetics Group, the CDC National Office of Public Health Genomics, the Genetic Alliance and others.
Contribute Effectively to Genomics Policy Development	• Engage external and internal partners to review genomics issues (e.g., direct-to-consumer genetic testing) and develop genomics policies. The Genomics Office sought the external panel's input on marketing of direct-to-consumer (DTC) testing. The Genomics Office staff will integrate this feedback into educational materials for consumers on DTC testing. • The Expert Genomics Advisory Panel identified a need to integrate genetic testing for hereditary colorectal cancer screening into colorectal cancer screening program documents. • Use existing surveillance tools, like the BRFSS survey, to assess the state population's awareness and needs pertaining to genetic testing and family health history.

Take-Away Message / Why It Works

- Partnerships are essential.
- Program location matters.
- Agency buy-in is critical.

Informational Resources

Connecticut Genomics Action Plan, 2007 Update
http://www.ct.gov/dph/lib/dph/state_health_planning/dphplans/genomics_action_plan_2007_update.pdf

Tools You Can Use

Family Health History Pocket Guide, Workbook and Poster
http://www.ct.gov/dph/cwp/view.asp?a=3134&q=387814

Web-based Information Resource on Colorectal Cancer, Including Information on Genetic Testing
http://www.ct.gov/dph/cwp/view.asp?a=3134&q=387814

Action Area

Achieving Public Genomic Literacy

"There is an important role for public health in providing accurate information and a balanced view of genetic technology."

– Michigan Department of Community Health, Genetics State Plan and Needs Assessment

Family health history and genetic tests are powerful tools for assessing a person's disease risk, but in order to realize the benefits of these tools and avoid potential harm, people need to understand genomics and how these tools impact their health. While most people and healthcare providers know that family health history provides important health information, many of them may not know what information to gather and how they can use it to assess their disease risk and determine which screenings are right for them. To fill the gap, states are adopting various strategies to educate and empower the public to learn about their family history and use that information to improve their health. Improving public understanding of genomics is important for several reasons.

- As the field of genomics grows, the ethical, legal and social implications of genomics are growing increasingly complex. Policymakers and state officials need to understand these implications in order to develop policies and laws.
- Without an adequate understanding of genomics, consumers are less likely to understand why tests or services are important.
- Informed individuals have the tools and knowledge to be proactive about their health and open a dialogue with their health care provider about lifestyle and other changes that reduce their risk for developing heritable conditions and improve their overall health.
- Consumers have direct access to an increasing number of genetic tests and medical providers are not always involved to explain test results and limitations. Consumers should have adequate knowledge of what they are being tested for, what the test results mean for them and where they can go for more information.

Up Close: Why are States Focusing on Family Health History?

In 2009, the National Institutes of Health convened a multi-disciplinary panel of providers and public officials at the Family History State-of-the-Science Conference. According to the panel, "An individual's family history has the potential to capture information about shared factors that contribute to that individual's risk for developing common diseases, such as diabetes, stroke, cancer, and heart disease. Family history is also used routinely in many other ways, including its well-defined use in determining who might benefit from genetic testing and its use in the interpretation of genetic test results."

The CDC's Office of Public Health Genomics stated that it "has been and will continue to be a core component of clinical and public health practice." Individuals who understand their family health history can use that information to make informed healthcare and lifestyle choices that reduce their disease risk.

States recognize the importance of family health history in public health practice. As a result, many states are focusing on educating the public and the public health workforce about how to collect and analyze family history information. Many states focus on family health history initiatives because there are tools and resources available. The federal government, through the Surgeon General's Family Health History Initiative and other initiatives, and states and localities are using family health history to inform the public about genomics and improve public health. According to the University of Washington's Center for Genomics and Public Health, "Because family history is an important risk factor for most diseases of public health importance, it is an appealing tool that is currently available. In addition, collecting family history information is accessible and inexpensive."

However, although it is an appealing tool for states, the State-of-the-Science panel pointed to a lack of evidence demonstrating the effectiveness of family history. In addition, the panel identified other barriers that affect health care providers, including lack of provider compensation for obtaining and interpreting family history, and lack of tools and technology to analyze and interpret the data.

Challenges

- **Lack of Dedicated Funding.** Lack of funding impedes genomics education and family health history initiatives. Partnering with other programs (e.g., to include family health history in a chronic disease consumer fact sheet) provides opportunities to do more with less.
- **Human Resource Requirements.** Maintaining departmental expertise in the rapidly changing field of genomics requires resources for training and staff development.
- **Targeted Communication:** Reaching individuals who lack Internet access, transportation, health coverage, or a medical home is a challenge. Developing effective and clear public health genomics communication strategies and reaching intended recipients requires culturally-sensitive public health messages that are delivered in multiple ways.

Strategies To Improve Public Genomic Literacy

States are adopting varied approaches to improving public knowledge and awareness of how genomics fits into their health and lifestyle decisions.

Table 10. Examples of State Activities to Improve Public Understanding of Genomics Issues

Objectives	Activities and Tactics
Assess Public Awareness	• Survey residents to assess their understanding of genomics and related topics. For example, several states (Connecticut, Michigan, Oregon and Utah) asked questions on the Behavioral Risk Factor Surveillance System questionnaires to assess public understanding of genetic tests and personal genome scans.
Inform Public about Available Genetic Services and Resources	• Develop information about how to access services in the state, including consumer-oriented statewide directories of genetic providers, and information about paying for services. Several states have developed centralized sources of information on genomics, service providers, specific diseases, newborn screening and state policies. • The Michigan Genetics Connection provides information for the general public, health care providers, patients and families, and teachers and students. • Oregon's Genetics Program provides information about privacy policies as well as opt-out forms for consumers who do not want their genetic specimens to be used in anonymous genetic research.
Improve Public Understanding of Basic Genetic Concepts	• Develop programs to improve understanding among children enrolled in public schools. Several states provide genomics curriculum and lesson plans to teachers, and teacher training. • The Utah Department of Health worked with the Genetic Science Learning Center to develop teacher, student and parent materials on heredity, inheritance, and family health history. These materials are available in English, Spanish, and Tongan, and are aligned with national and state standards. • The Genetics Education Committee of Oklahoma developed a genetics toolkit to help science teachers incorporate genetics in their classroom. • Wisconsin received funds from the National Human Genome Research Institute to support genetic counselor participation in Wisconsin classrooms on national DNA Day. • The Illinois Department of Public Health invited 9th to 12th graders to participate in the 4th Annual National DNA Day Essay Contest.
Disseminate Information through Multiple Channels	• Distribute publications and resources to public libraries, and offer workshops and public lecture series in targeted locations (e.g., senior centers and community health centers). • The Connecticut Department of Public Health distributed a print copy of *Understanding Genetics: A New England Guide for Patients and Public Health Professionals* to each public library in the state. • The New York State Department of Health Wadsworth Center holds free public lecture series each year to educate the public about genetics and other biomedical topics. • Utilize other sources for information, including the National Coordinating Center for the Genetics and Newborn Screening Regional and Collaborative Groups, the Genetic Alliance and the National Coalition for Health Professional Education in Genetics.

Success Story: Utah at a Glance

Using Family Health History to Increase Genomic Literacy in Utah

Agency/Program Name	Utah Department of Health (UDOH)
Program Location	The Chronic Disease Genomics Program (CDGP) was located in the UDOH Bureau of Health Promotion, Division of Community and Family Health Services.
Funding	CDC funding from 2003-2008 supported the UDOH CDGP, which dissolved in 2008 because of lack of funding. Funds of less than $200,000 per year from 2003-2008 covered all staff costs, materials and operating expenses.
Staff	One full-time health educator, a half-time epidemiologist, and a half-time program manager
Sustainability	The CDGP dissolved in 2008. An external genetics advisory committee currently advises the health department on genomics issues, but many of the genomics activities have ended without dedicated staff. The family health history materials are currently available, as well as consultation on a limited basis. Utah's family health history materials continue to be heavily used in Utah and other states.
State Plan	Utah Genomics Plan 2006-2010 available at http://www.health.utah.gov/genomics/Genomics%20plan%20final_Sept2008.pdf
Program Website	http://www.health.utah.gov/genomics/

Issue In Brief

Utah is recognized as a leader in developing widely-used, culturally-appropriate tools to educate the public and public health practitioners about genomics and health. Recognizing that genomics is complex and intimidating, Utah focused on building interest and awareness through family health history, an effective platform for promoting collaboration among the public health department, providers and the members of the public, as well as a proven tool for preventing disease and promoting health. According to Jenny Johnson, who was the CDGP health educator, people "get" family history. "It resonates with the public because it connects them with their past," and, she said, it fits within a practitioner's activities because many already use family history in their daily activities. The Chronic Disease Genomics Program developed a balanced family history initiative. Staff dedicated their time to the development, dissemination and evaluation of materials. The end result, the program produced high-quality, culturally-appropriate family health history materials and used various channels, such as media, senior centers, libraries, elementary and high school science and health classes, to reach specific populations. Although the chronic disease genomics program no longer exists, its activities and tools continue to be a model for other states that want to integrate genomics into their public health department activities and public health practice.

"Family health history is a simple and cost-effective tool for assessing an individual's or population's risk for disease and guiding tailored interventions that may increase behavior change."

– Utah Department of Health, CDGP Success Stories 2003-2008

Table 11. Blueprint for Increasing Genetic Literacy in Utah

Goal	Activities that Help the CDGP Achieve Goals
Develop Community and Public Health Leadership in Genomics and Chronic Disease	• Developed internal partnerships to integrate genomics into various program areas (e.g., asthma, diabetes, cancer, and Women, Infants and Children (WIC)). The WIC program integrated family history in the counseling process by tailoring nutritional information based on the client's risk factor. In addition, the cancer program integrated family history into the WISEWOMAN program grant. • Forged external partnerships to conduct research, develop materials and disseminate information. Partners included academic institutions, provider groups (e.g., genetic counselors), media agencies, the Family History Library, private companies and community-based organizations (such as the National Tongan American Society). • Awarded community mini-grants to community agencies and universities to implement projects relating to family history.
Develop Family History Interventions	• Developed an Electronic Health Family Tree (eHFT) in 2005. • Developed a family health history toolkit, available in English, Spanish and for seniors, and other materials (e.g., family reunion packet and Senior Center packet) to help families talk about, collect and share their family health history. • Developed a targeted dissemination strategy for family history toolkits, including a media campaign using radio, TV and print media. The public obtained toolkits through the CDGP website, classes at the Family History Library (FHL) and from the UDOH resource line. • Partnered with the Genetic Science Learning Center to develop a high-school health history curriculum and adapt existing 5th grade genetics modules on family health history for biology and health students and their families. • Partnered with Hispanic/Latino and Pacific Islander advisory committees to adapt classroom curricula and develop culturally and linguistically-appropriate take-home activities. The 5th grade curriculum is available in English, Spanish and Tongan, and the high school curriculum is available in English and Spanish.
Assess Family History Interventions	• The State of Utah Employee Wellness Program in 2005 and 2006, and high-school students in 2007 pilot-tested the electronic Health Family Tree Program. • High-school teachers and more than 500 students evaluated the High School Family Health History Curriculum. The findings, information is easy to understand, teacher guides are effective, the reading level is appropriate. An evaluation of the Spanish curriculum in 2008 found that materials were culturally-appropriate and the activities were engaging and easy to understand. • Fifth grade teachers and students gave high ratings for cultural appropriateness, student engagement and learning outcomes.

Take-Away Message

- Sustaining genomics requires staff.
- Finding the right skill set is critical.

Informational Resources

UDOH CDGP Success Stories
http://health.utah.gov/genomics/successstoriesbook_webonly.pdf

Tools You Can Use

Health Family Tree Tool
http://health.utah.gov/genomics/familyhistory/fhrp.html

Family Health History Toolkit
http://health.utah.gov/genomics/familyhistory/toolkit.html

High School and 5th Grade Curriculum
http://learn.genetics.utah.edu

Action Area

Addressing Emerging Public Policy Issues

"The presumption of modern science, including medical genetics, has always been that knowledge is fundamentally good for human beings, and that the more we know about ourselves the better we will be able to live the kind of lives we want to live. Yet the truth of this supposition remains in doubt as we lift the lid of the Pandora's box of our genomic inheritance."

–The President's Council on Bioethics, 2008

Public health leaders and policymakers are increasingly challenged to address the emerging ethical, legal and social implications related to new genetic technologies and services. As new genetic tests are available, health policy leaders are taking steps to protect consumer privacy, prevent discrimination based on genetic information and ensure that the public has sufficient knowledge to make well-informed decisions. Despite the challenges, however, genetic testing also offers potentially significant individual and public health benefits. As a result, health leaders are challenged to strike a balance between protecting patients and product regulation on the one hand, and fostering genomic research and translation on the other. Some examples of other emerging and complex policy issues include:

- **Increasing Availability of Direct-to-Consumer Genetic Tests.** Consumers have greater access to genetic tests and genomic profiles that provide information about their disease risk. While these tests may provide valuable information, they raise several issues for policymakers and health officials, such as whether the tests are effective and reliable, and how to ensure that consumers understand test results.
- **Expansion of Newborn Screening Programs and Genetic Bio-Banks.** In recent years, technology advances have expanded the number of conditions that can be screened and identified. Although expanded newborn screening capacity offers public health benefits, it also raises ethical questions about what conditions to test and under what circumstances, as well as concerns about access to, and availability of services for an increasing number of children identified through the newborn screening process. Moreover, states have different policies regarding the storage and use of residual newborn screening dried blood spots and other genetic samples. As genetic screening and research have evolved, researchers and other stakeholders have pushed to store these samples in centralized locations, known as bio-banks, for current and future clinical care and health research. However, retaining samples for research presents a number of dilemmas for health leaders, such as determining whether and how parents should be notified if their child's sample will be used for research, or if researchers identify that a child is at-risk.
- **Safeguarding Patient Privacy and Preventing Discrimination.** As more and more genetic information is obtained, stored, often electronically in large databases, and used for research, many worry about the potential uses and misuses of this information. State and federal policymakers have passed legislation, including the federal Genetic Information Nondiscrimination Act of 2008, to protect consumers from employer or insurer discrimination based on genetic information.

These are not the only public policy "hot topics," but they are among the most pressing issues facing states today. They are likely to grow in importance as new technologies move from research to marketplace. Because these issues and challenges are so fluid, this section does not provide a comprehensive overview of all public policy issues, but rather, it summarizes some of the highest-priority issues affecting states today, and provides resources to help health policy leaders anticipate changes and develop sound policies for the future.

Direct To Consumer Genetic Tests

The field of genetic testing is rapidly evolving. In 2009, tests were available for more than 1,800 diseases, up from just 100 in 1993. Genetic tests cover a broad spectrum, including traditional diagnostic and newborn screening tests, as well as an emerging field of tests and genomic profiles that predict a person's risk for developing various chronic diseases.

Although most tests are made available through a patient's health care provider, including diagnostic and prenatal testing, an increasing number are directly available to consumers. For example, consumers can go online to one of many direct-to-consumer (DTC) genetic testing and genomic profiling companies and purchase a test to find out if they are predisposed to certain cancers, diabetes, cardiovascular disease and a long list of other diseases. However, there is not a uniform system to ensure that these tests are clinically valid and therefore some tests may not provide valid and reliable information.

Policy Issues. These products have the theoretical potential to provide information to help consumers make lifestyle changes and improve their health. Consumers who learn that they have a higher-than-normal risk for cardiovascular disease, for example, can modify their exercise and dietary habits and possibly reduce their disease risk. Some say that direct access to genetic tests and personal health information democratizes genomics by promoting access to genomic tools and information. In addition, expanding the genetics research database, made possible when companies and researchers obtain large numbers of genetic samples, enhances research and knowledge about the interaction between genes and disease, which offers significant public health benefits.

Table 12. Types of Genetic Tests

Test	Uses for Test
Newborn Screening	To identify genetic disorders that can be treated early in life. All states test infants for phenylketonuria and congenital hypothyroidism and most states also test for other genetic disorders.(1)
Diagnostic Testing	To identify or rule out a specific genetic or chromosomal condition. In many cases, genetic testing is used to confirm a diagnosis when a particular condition is suspected based on physical signs and symptoms. Diagnostic testing can be performed before birth or at any time during a person's life, but is not available for all genes or all genetic conditions.(2)
Carrier Testing	To identify people who carry one copy of a gene mutation that, when present in two copies, causes a genetic disorder. This type of testing is offered to individuals who have a family history of a genetic disorder and to people in certain ethnic groups with an increased risk of specific genetic conditions. If both parents are tested, the test can provide information about a couple's risk of having a child with a genetic condition.(3)
Pre-implantation Testing	To detect genetic changes in oocytes, or embryos that were created using assisted reproductive techniques such as in-vitro fertilization.(4)
Predictive and Presymptomatic Testing	To detect gene mutations associated with disorders that appear after birth, often later in life. Predictive testing can identify mutations that increase a person's risk of developing disorders with a genetic basis, such as certain types of cancer. Presymptomatic testing can determine whether a person will develop a genetic disorder…before any signs or symptoms appear.(6)
Forensic Testing	Uses DNA sequences to identify an individual for legal purposes.(7)
Prognostic Testing	Used to predict disease severity/outcome based on genetic profiling of disease cells. Prognostic testing is primarily used in oncology to alter treatment strategies.(8)
Pharmacogenomic Testing	Used to identify genetically determined response to specific pharmaceutical agents. This testing is used to predict drug effectiveness and side effects.(9)

Source: (1-7) U.S. National Library of Medicine, Genetics Home Reference, http://ghr.nlm.nih.gov/handbook/testing/uses and (8-9) CDC, "Genetics, Genomics and Public Health: An Overview," 2009.

However, direct access to genetic tests raises many public policy concerns, including concerns about regulatory oversight, potentially misleading practices, lack of provider involvement and questionable accuracy and utility. Each of these concerns is discussed below.

Lack of Oversight. Although there is currently federal oversight of genetic tests and laboratories, it is limited. Critics warn that the current regulatory framework does not assure that tests are accurate and valid, nor does it monitor the claims made by DTC companies. According to the Genetics and Public Policy Center, "there is no uniform or comprehensive system to assess the analytic and clinical validity of tests before they are offered to patients, and there are no laboratory standards that specifically address molecular or biochemical genetic testing or require laboratories to enroll in proficiency testing programs that assess their ability to perform the tests correctly."

According to a 2008 report by the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS), there are oversight gaps in five main areas:

- Regulations governing clinical laboratory quality;
- Oversight of the clinical validity of genetic tests (defined on the following page);
- Transparency of genetic testing;
- Level of current knowledge about clinical usefulness of genetic tests;
- Meeting the educational needs of health care professionals, the public health community, patients and consumers.

The SACGHS makes several recommendations to fill these gaps, such as requiring the Food and Drug Administration to address all laboratory tests and funding for a lead agency to develop a web-based registry for laboratory tests.

Federal Oversight of DTC Tests

- The Food and Drug Administration (FDA) regulates the safety and effectiveness of medical devices, but it currently only regulates a small portion of direct-to-consumer tests. The FDA does not regulate genetic tests that are manufactured "in-house" in laboratories, but only those that are manufactured and sold to other companies that perform testing. According to a 2006 Federal Trade Commission fact sheet, " …while most other home-use medical tests undergo FDA review to provide a reasonable assurance of their safety and effectiveness, no at-home genetic tests have been reviewed by the FDA, and the FDA has not evaluated the accuracy of their claims."

- Federal legislation, known as the Clinical Laboratory Improvement Amendments of 1988 (CLIA) requires certification for all clinical laboratories (including genetic testing labs), and addresses a range of issues, including personnel qualifications and quality control. In addition, the law requires labs to conduct proficiency testing, but the federal government relies on DTC test manufacturers to conduct the testing. In other words, there is not federal oversight to ensure that genetic test results are accurate or valid.

State Oversight of DTC Tests

- State regulation of genetic testing varies. Many states rely on CLIA requirements to regulate genetic testing laboratories, but New York and Washington operate their own laboratory certification programs. State policies related to direct-to-consumer genetic testing companies also vary. According to a 2007 state survey by the Genetics and Public Policy Center at Johns Hopkins, 25 states and the District of Columbia permit tests without restrictions, 13 states prohibit them (and require that an authorized provider orders the test) and 12 states allow certain direct-to-consumer tests, such as pregnancy, glucose and cholesterol.

Potentially Misleading Practices. Critics argue that some companies make false or misleading claims about their tests, or fail to adequately educate consumers about test reliability and validity. This has the potential to mislead consumers and health care providers alike, who may not understand the limitations of a specific product.

Lack of Provider Involvement. The American College of Medical Genetics recommends that genetic testing should be provided only through "an appropriately qualified health care professional," who orders and interprets genetic tests. Although many DTC companies encourage consumers to share their results with their health care providers, and some even provide information to help them analyze and interpret test results, this is not a requirement. Removing health care providers from the genetic testing equation is potentially harmful to consumers, who may not understand what the test can (and can not) determine, or how to interpret test results. This could lead them to make serious and permanent decisions to address their perceived risk of developing a disease.

Questionable Accuracy and Utility. Before tests are ready for sale, critics argue that certain standards should be met. A 2008 article in the New England Journal of Medicine suggests that direct to consumer genome services and gene profiles raise questions about their analytical and clinical validity and clinical utility (defined below). "Measures of utility address the question at the heart of the clinical application of a test: If a patient is found to be at risk for a disease, what can be done about it?" At the moment, there is no conclusive answer to this question.

When Genetic Tests Are Ready for Prime Time

- **Analytic Validity**: Ability of the test to accurately and reliably measure the genetic markers it is intended to measure. Does the test produce the right answer?
- **Clinical Validity:** Ability of the genetic test to accurately and reliably diagnose or predict the condition of interest. Does the test relate to a person's health risk of developing disease?
- **Clinical Utility:** Likelihood that using the genetic test will significantly improve health-related outcomes.

Source: CDC, Genetics, Genomics and Public Health: An Overview, presentation, 2009.

"There are very few observational studies and almost no clinical trials that demonstrate the risks and benefits associated with screening for individual gene variants, let alone testing for many hundreds of thousands of variants."

Policy Responses. Federal and state health policy leaders are developing policies to address many concerns associated with direct-to-consumer testing.

- California law requires that a state licensed physician be involved, consumer counseling is available, and proof that tests are valid and conducted at certified laboratories.
- New York law requires that authorized health care providers order tests, and therefore direct access is not permitted. The State of New York certifies all labs that do business in the state, including labs located outside of New York, through its Clinical Laboratory Evaluation Program (CLEP) which certifies laboratory directors, issues laboratory permits and conducts proficiency testing.

States that have sought to end the sale of DTC tests have encountered challenges by companies that argue that they are not selling medical information or genetic tests and therefore should not be regulated as such.

In response to concerns about lack of public awareness and knowledge about genetic tests, several state health agencies have surveyed the public about their understanding and use of DTC tests (i.e., by including questions on existing behavioral surveys), and many states provide information to consumers and health care providers to help them understand genetic tests. Other policy responses include strengthened oversight of company disclosure and patient information materials to ensure that they disclose information about the reliability of test results and provide adequate information to help consumers and their health care providers interpret test results.

In addition, federal and state policymakers have taken numerous steps to address concerns over privacy and discrimination by preventing employer and insurer discrimination based on genetic information.

Up Close: Three-State Public Health Collaboration to Educate the Public and Providers About Cancer Genetic Testing

Preparing for a large-scale DTC marketing campaign for BRCA1 and BRCA2 testing brought together three state health agencies to provide public and provider education and a consistent public health message. (Mutations in BRCA1 and BRCA2 genes are linked to hereditary breast and ovarian cancer.) Health department staff in Connecticut, Massachusetts and New York used this campaign as a window of opportunity to educate the public and providers about genetic testing and available genetic resources and providers. According to a summary of the project, "The public health community must be prepared with accurate, up to date and unbiased information for providers, the general public, and those truly at high risk. Additionally, health care clinicians need to be aware of the genetics resources in their community, such as genetic counseling services, so that their patients can make well-informed decisions."

To market a consistent public health message and to educate the public and providers, the health agencies sent materials to over 20,000 providers in three states. Some professional organizations (e.g., the Connecticut Medical Society) included some of these materials on their websites or newsletters.

For More Information on Genetic Testing

FAQ about Genetic Testing	National Human Genome Research Institute (NHGRI), http://www.genome.gov/19516567
What is Direct-to-Consumer Genetic Testing	Genetics Home Reference, http://ghr.nlm.nih.gov/handbook/testing/directtoconsumer
Promoting Safe and Effective Testing in the United States	NHGRI, http://www.genome.gov/10002393#EXECUTIVE
Issue Brief: Who Regulates Genetic Tests?	Genetics and Public Policy Center (GPPC), http://www.dnapolicy.org/policy.issue.php?action=detail&issuebrief_id=10
Survey of Direct-to-Consumer Testing Statutes and Regulations	GPPC, http://www.dnapolicy.org/resources/DTCStateLawChart.pdf
Preparing for a Direct to Consumer Marketing Campaign for BRCA-1 and BRCA-2 Testing: A Three State Public Health Collaboration	Connecticut Department of Public Health, Massachusetts Department of Public Health, New York State Department of Health http://www.cdc.gov/genomics/events/file/print/10year/48_DTC_BRCA.pdf

Newborn Screening and Genetic "Biobanks"

Advances in newborn screening have been fueled by new technologies and growing interest in retaining newborn screening samples for current and future research. According to a 2008 report by The President's Council on Bioethics, "Over the next years and decades, anticipated developments in the technology and the practice of medicine are likely to alter the landscape of newborn screening entirely, ushering in a potentially vast increase in the kinds and amounts of genetic data that can be routinely collected upon the birth of a child." Although these advances offer significant research and public health opportunities, they also present myriad ethical, social and legal challenges for public health leaders. In response, states are adopting varied strategies to inform the public and health care providers about newborn screening policies, develop guidelines for storing and sharing genetic samples, and ensure access to genetic services for children who have been identified through the newborn screening process. Some states rely upon uniform standards developed.

State screening programs began in the 1960s to test for a single, serious disorder known as phenylketonuria. Over the years, new screening technologies have made it possible to test newborn blood spots for an increasing number of genetic and metabolic disorders, and currently some states screen for up to 30 or more of these conditions. The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) was established in 2003 to advise the Secretary of the U.S. Department of Health and Human Services on newborn screening tests, technologies and policies, and many states rely on their recommendations to guide state newborn screening policy. In addition to screening, many states provide a range of other services, including follow-up diagnostic, treatment and counseling services for children identified through newborn screening, and provider and consumer education.

As newborn screening programs capture more genetic information, there is increasing interest in collecting and retaining dried blood spots for current and future research. These centralized repositories of genetic samples, known as biobanks, DNA warehouses, and other names, are important, because large databases allow researchers to analyze the relationships between genes and disease. Moreover, retaining this information provides significant public health benefits, such as identifying individuals who might benefit from new treatments. Critics argue that these systems could jeopardize patient privacy, lead to discrimination by employers and insurers, and could conflict with individuals' principles and religious beliefs. A recent court case in Texas illustrates the complex legal issues surrounding newborn screening programs and procedures. Although there is now a law permitting

the Texas Department of State Health Services to retain genetic material (unless parents "opt out" and fill out a form specifying that the sample should be destroyed), the state did not have such a process in 2002 when it began retaining blood samples. The lawsuit alleged, among other things, that the practice of retaining samples indefinitely and without parental consent violated fundamental privacy rights and violated medical research standards of obtaining informed consent. As a result of a settlement between the Texas Civil Rights Project and the Texas Department of State Health Services, the state was required in 2010 to destroy newborn blood samples collected over the past seven years.

Policy Issues and Responses. There are numerous challenging policy questions related to newborn screening and bio-banking. Table 13 summarizes these challenges, and provides policy options and resources for addressing them.

Biobanking Defined: A place that collects, stores, processes and distributes biological samples and the data associated with those materials. Biobanks may be used for clinical care or health research. In the United States, research biobanks are governed by ethical principles for human subject research established by federal guidelines.

Source: Michigan BioTrust for Health

Table 13. Examples of State Newborn Screening and Biobanking Activities

Policy Challenges	Options and Resources
Technological advances in newborn screening make it possible to screen for an increasing number of conditions.	• States have developed varied approaches for identifying the list of conditions for newborn screening. Some state laws specify the panel of disorders screened during NBS, while others adopt the recommendations of another agency or organization (e.g., the Recommended Uniform Screening Panel of the ACHDNC). • Other options for determining a state's newborn screening panel include reviewing other state newborn screening programs, and utilizing frameworks for evaluating new technologies and tests. States typically seek to incorporate community, provider and policy expertise into decision-making process.
A growing number of childhood conditions are identified through newborn screening, demanding more from the newborn screening infrastructure.	• States and regional collaboratives are connecting children in remote locations to metabolic and genetic specialists through telemedicine, interstate licensure, and favorable reimbursements. For example, the Hawaii Community Genetics initiative, described later in this section, promotes access through telehealth visits with a pediatric geneticist. • Several state agencies and professional organizations offer genetic training to primary care providers and medical students. Online fact sheets and resource guides inform providers about screening, referrals and communicating with patients. • The federal Newborn Screening Saves Lives Act of 2007 establishes grants to provide education and outreach on newborn screening and follow-up care. • States are developing short- and long-term follow-up procedures for children conditions identified through newborn screening process. • Regional collaborative projects track patients long-term to capture clinical history and progress. Participating providers enter patient information into centralized database. These records are accessible to all of the patient's providers. • Several states require coverage of medically necessary foods and treatment of disorders identified through NBS. • States are supporting provider education projects (e.g., online information clearinghouses and provider training) to ensure primary care providers have genetic knowledge. Other policy options include adopting strategies to expand the number of clinical geneticists and genetic counselors.
Need for parental notification and education.	Newborn screening and storage of samples requires parental notification and processes for educating and empowering families to opt-in or out of various stages of the newborn screening process. Examples of state options include: • Developing informed consent policies and determining requirements for anonymous and identifiable samples. • Determining policies for families that do not want their child's sample stored, including developing informational materials, opt-out or opt-in forms. • Developing parental notification policies, including establishing policies for communicating test findings to individuals and groups.

Table 13. Examples of State Newborn Screening and Biobanking Activities (continued)

Policy Challenges	Options and Resources
Developing large-scale databases of genetic information necessitates health agencies to develop research standards and protocols.	Storing, researching and sharing genetic samples requires policy leaders to develop standards to ensure patient privacy, set standards for participating research organizations, and determine how long samples will be stored and for what purposes. Examples of state options include: • Requiring researchers to obtain approval from an ethics or review board before obtaining blood spots and other genetic samples. • Determining time period for storage. Are samples stored indefinitely or discarded after initial testing? • Encouraging community input into storage and research standards. The Michigan Biotrust for Health, for example, informs and solicits input from the public through a state website and online survey, which is also an educational tool. • Developing research protocols for sharing results with parents. • Developing safeguards to protect patient privacy and confidentiality, which is a challenge with sharing of records among multiple research organization via electronic file-sharing. • Developing policies to ensure that researchers meet privacy, ethics and research standards. • Examining potential commercial use of genetic samples and information.
Maintaining newborn screening during public health emergencies is a critical public health function and challenge.	• Some states and regional projects are addressing the need for improved medical records, which is essential for displaced patients with genetic or metabolic disorders. These records "follow" them if they move, allowing new providers to understand their medical history and medications. • State health agencies are partnering with other states to maintain essential services. In the aftermath of Hurricane Katrina, the Louisiana laboratory transferred responsibilities to the Iowa Public Health Laboratory. In 2007, Maryland lawmakers passed House Bill 344, authorizing the Maryland public health laboratory to enter into a mutual aid agreement with another state's public health lab during an emergency.
Addressing ethical dilemmas associated with newborn screening.	Expanding newborn screening creates several ethical dilemmas, such as testing for conditions that do not currently have an available treatment. Examples of state options include: • Engaging broad participation by the public, providers, policy and genetics experts to develop ethical framework for genetic testing. Develop ethical principles that help define newborn screening panels. The Connecticut Department of Health conducted a feasibility study for a statewide bio-bank to study pre-term births and birth defects. The office of genomics followed the study with a half-day symposium on bio-banks, allowing for public discussion. • Michigan retains newborn blood spots indefinitely, a departure from their prior policy of discarding samples. The state is educating parents about this policy through community engagement, focus groups with the public and an online survey which is also an educational tool.

Up Close: Connecticut Examines Feasibility of a Statewide Bio-bank to Study Preterm Births and Birth Defects

Premature births in Connecticut are a serious public health problem. Premature births disproportionately occur among non-Hispanic Black/African American women, and come at great economic and social cost to the state. According to a 2007 study, "the total cost of hospitalizations in Connecticut for all premature births was over $85 million in 2004." In response, the Connecticut Department of Public Health convened a panel of experts to examine the feasibility of a statewide population-based bio-bank of donated human tissue and information to help study preterm births and birth defects. The reason: a statewide bio-bank offers significant public health benefits, including the potential to analyze and understand the genetic and biological determinants of preterm births and birth defects. Although small, private bio-banks already operate in the state, they do not provide as many research opportunities due to the small sample size.

The expert panel discussed four possible models of bio-banks, and evaluated these against five specific criteria:

1. Level of increased funding to the state from external sources;
2. Level of increased research potential;
3. Affordability and sustainability;
4. Degree to which ethical, legal and social issues are addressed; and
5. Level of positive perception.

According to the 2007 Feasibility Study, "The Panel members felt that a bio-bank in Connecticut would increase funding to the state from federal and private sources, would increase research potential, and could be accomplished to minimize ELSI (i.e., ethical, legal or social) issues." However, the study found that more information was needed about the public perception of a large-scale bio-bank, specifically from potential donors, the general public and policymakers. As a result, the Panel recommended that lawmakers mandate a comprehensive statewide feasibility study to address cost, oversight and funding, prevalence of birth defects and other issues.

In 2008, the Connecticut Department of Public Health's Office of Genomics followed up the feasibility study with a half-day symposium, "Bio-banks: The Promise and Public Health Challenges," to inform and engage the public about bio-banks in the state.

For More Information on Newborn Screening and Biobank Issues

National Newborn Screening Status Report, July 2009	National Newborn Screening and Genetics Resource Center (NNSGRC), http://genes-r-us.uthscsa.edu/nbsdisorders.pdf
Resources and recommendations related to newborn screen tests, technologies and guidelines.	Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC), http://www.hrsa.gov/heritabledisorderscommittee/
Using Genomic Data-bases to Study Complex Diseases	Genetics and Public Policy Center, http://www.dnapolicy.org/policy.issue.php?action=detail&issuebrief_id=38
U.S. Newborn Screening Programs, State Map, August 2007	NNSGRC, http://genes-r-us.uthscsa.edu/resources/consumer/statemap.htm
The Changing Moral Focus of Newborn Screening: An Ethical Analysis by the President's Council on Bioethics, 2008	President's Council on Bioethics, http://www.bioethics.gov/reports/newborn_screening/index.html
The Newborn Screening Saves Lives Act of 2007	The Library of Congress, http://thomas.loc.gov/cgi-bin/bdquery/z?d110:SN00634:
Expanding Newborn Screening: Process, Policies and Priorities	The United States Preventive Services Task Force, http://www.ahrq.gov/CLINIC/uspstf08/methods/newbscr.htm
Genetics Legislation database	National Conference of State Legislatures (NCSL), http://www.ncsl.org/default.aspx?tabid=14408
Newborn Genetic and Metabolic Disease Screening Laws	NCSL, http://www.ncsl.org/default.aspx?tabid=14416
Insurance Coverage of Medically Necessary Foods	NCSL, http://www.ncsl.org/default.aspx?tabid=13865\
Feasibility Study for a Statewide Bio-bank to Study Preterm Births and Birth Defects	Bio-bank Feasibility Expert Panel, Connecticut Department of Public Health, http://www.ct.gov/dph/lib/dph/BIOBANK_Final_Report_111407.pdf

Protecting Patient Privacy and Preventing Discrimination

Although genomic advances hold promise for improving public health, many consumers and health leaders are concerned about how this information will be used and managed. Dr. Francis Collins, Director of the National Human Genome Research Institute, said in congressional testimony that, "The misuse of genetic information has the potential to be a very serious problem, both in terms of people's access to employment and health insurance and the continued ability to undertake important genetic research."

Policy Issues

Participating in genomic research and gathering information about one's own genetics, through testing or family history, provides enormous opportunity to improve individual and public health. However, as more of this sensitive information is gathered, many worry that the risks associated with misuse of this information could effect a person's employment and health and life insurance options. Although genetic discrimination is not widespread, the public's concerns about potential misuses of this information are significant.

Policy Responses

Federal Policies. Congress passed, and President Bush signed, the Genetic Information Nondiscrimination Act of 2008, or GINA, that prohibits health plans from requiring genetic testing. It also prohibits health plans from using predictive genetic information for underwriting purposes. The law protects employees from discrimination by their employers based on genetic information, and requires employers to treat any employee's genetic information as a confidential medical record. Questions remain about implementation and there is some uncertainty about how these protections will impact consumers in practice.

Prior to GINA, other federal legislation protected consumers from discrimination based on their genetic information. On February 8, 2000, President Clinton signed an executive order prohibiting every federal department and agency from using genetic information in hiring or promotion activities. Before that, the Health Insurance Portability and Accountability Act of 1996 prohibited a group health plan from using genetic information to establish rules for eligibility or continued eligibility. The act provides that genetic information shall not be treated as a preexisting condition in the absence of the diagnosis of the condition related to such information. It also prohibited a group health plan or issuer of a group health plan from using genetic information in setting a premium contribution.

State Policies. The majority of states already have laws that restrict the use of genetic information by employers and insurance companies. Lawmakers across the country continue to strengthen protections. According to the National Conference of State Legislatures, 34 states and the District of Colombia prohibit employers from discriminating based on genetic information and 44 states and the District prohibit health insurers from basing eligibility on genetic information.

Up Close: Oregon's Genetic Privacy Policy and Health Agency Response

In 1995, the Oregon legislature enacted comprehensive genetic privacy legislation to protect individuals from employment and insurance discrimination based on genetic test results. The law broadly prohibits disclosure of genetic information, and contains several provisions related to patient privacy, including a recent requirement that healthcare providers and health systems give patients an opportunity to opt-out of anonymous or coded genetic research. Anonymous research is defined in statute as "scientific or medical genetic research conducted in such a manner that any DNA sample or genetic information used in the research is unidentified."

Policymakers have revisited the law several times over the years, as shown in Table 14. The creation of a Genetic Research Advisory Committee in 1999, changed to the Advisory Committee on Genetic Privacy and Research in 2001, formalized a process for revisiting and revising state law. The Committee is required to report every two years to the Oregon Legislature on the use and disclosure of genetic information and make recommendations for changing the law, if needed. In addition to monitoring the state policy environment, the Committee also addresses the implications of federal privacy laws, including the Health Insurance Portability and Accountability Act of 1996 and the Genetic Information Nondiscrimination Act of 2008, on the state's existing requirements. Other Committee tasks include advising the Oregon Department of Human Services on administrative rules (e.g., informed consent policies), creating opportunities for public education, and obtaining public input on genetic privacy and research issues.

Oregon's experience illustrates the fluid nature of genomics policy development, as well as the ongoing role for state health officials. Implementing the 2005 opt-out requirement proved challenging for stakeholders, especially hospitals, providers and laboratories who were required to develop new procedures and invest resources to comply with the requirements. The dynamic nature of Oregon's privacy legislation impacts other stakeholders, including state health agency staff, who are responsible for informing the public about privacy protections, overseeing institutional review boards that review genetic research, and developing and implementing policies (e.g., informed consent processes).

Additionally, the Legislature has never appropriated any funding for any of these activities. The legislative process demonstrates that enhancing the public good while providing privacy protections is a complex and ongoing process, best achieved by allowing for changes based on stakeholder feedback and objective research and analysis.

Table 14. Oregon Legislative Timeline

1995	• Legislature enacted the Comprehensive Genetic Privacy Act to protect the privacy of genetic samples and protect individuals from employment and insurance discrimination based on test results. • The law created privacy protections for obtaining, retaining and disclosing genetic material. • The law defined genetic information as the "property" of the individual. • As directed in statute, in 1996 the Oregon Health Division adopted administrative rules for consent forms for genetic testing.
1997	• In response to charges that the property clause was having negative effects on research, the Legislature amended the statute to exempt anonymous research from the privacy act because anonymous research could not result in discrimination.
1999	• The Legislature created a Genetic Research Advisory Committee to "study the use and disclosure of genetic information and…develop a legal framework that defines the rights of individuals whose DNA samples and genetic information are collected, stored, analyzed and disclosed."
2000	• The Advisory Committee recommended that the Legislature replace the controversial property provision with a confidentiality clause. • The Advisory Committee published the report, Assuring Genetic Privacy in Oregon, which made recommendations for the remedy of violations, family issues, informed consent, property and continued oversight.
2001	• The Legislature deleted the property provision and specified that genetic information and DNA samples are private and that individuals and their families have a right to protection of that privacy. • The law contained several other changes, such as requiring researchers to obtain informed consent except when the person's identity is anonymous or encrypted. It also created penalties for intentional violations.
2003	• The Legislature added some provisions to the statute, including new definitions (e.g., anonymous research) and new standards for regulating coded research.
2005	• The Advisory Committee made several recommendations in their 2005 report to the Legislature, including: - Exempt routine disclosures of genetic information by providers and health insurers from special protections. - Prohibit use of family members' medical history for health insurance and employment decisions. - Prohibit employers and health insurers from using information concerning whether a person sought genetic counseling. - Modify informed consent requirements for research. • The Legislature modified informed consent requirements for research under certain limited conditions and required health care providers and health systems to inform patients about their right to "opt-out" of anonymous or coded IRB-approved research.
2007	• The Legislature amended the statute to align the Oregon statute with federal HIPAA requirements.

Source: Oregon Department of Human Services, Genetics Program, History of Oregon's Genetic Privacy Law, http://www.oregon.gov/DHS/ph/genetics/docs/HistoryofOrego021408.pdf.

Informational Resources on Oregon's Privacy Laws

History of Oregon's Genetic Privacy Law
http://www.oregon.gov/DHS/ph/genetics/docs/HistoryofOrego021408.pdf.

2009 Report to the Oregon Legislature from the Advisory Committee on Genetic Privacy and Research
http://www.oregon.gov/DHS/ph/genetics/docs/ACGPR2009LegReport.pdf

Oregon Genetic Privacy Laws
http://www.oregon.gov/DHS/ph/genetics/docs/Oregon_Genetic_Privacy_Statutes_07.pdf

For More Information on Privacy and Anti-Discrimination Policies

The Genetic Information Nondiscrimination Act of 2008: Information for Researchers and Health Care Professionals	U.S. Department of Health and Human Services, http://www.genome.gov/Pages/PolicyEthics/GeneticDiscrimination/GINAInfoDoc.pdf
GINA Fact Sheet	National Human Genome Research Institute, http://www.genome.gov/10002328
Genetic Privacy Laws	National Conference of State Legislatures (NCSL) http://www.ncsl.org/IssuesResearch/Health/GeneticPrivacyLaws/tabid/14287/Default.aspx
Genetic and Health Insurance Anti-Discrimination Laws	NCSL,http://www.ncsl.org/default.aspx?tabid=14374
GINA Legislation	Human Genome Project Information, http://www.ornl.gov/sci/techresources/Human_Genome/publicat/GINAMay2008.pdf
Legal Issues Related to Discrimination and Privacy	Congressional Research Service, http://assets.opencrs.com/rpts/RL30006_20080310.pdf
Genetic Privacy and Discrimination	Genetics and Public Policy Center (GPPC),http://www.dnapolicy.org/policy.privacy.php
Issue Brief: The Genetic Information and Nondiscrimination Act	GPPC, http://www.dnapolicy.org/policy.issue.php?action=detail&issuebrief_id=37
Oregon Genetics Program Online Resources, including: genetic testing guidelines, genetic research opt-out information, and genetic privacy and research information.	Oregon Department of Human Services, Genetics Program,http://www.oregon.gov/DHS/ph/genetics/

Taking A Step Back: Common Policy Considerations For Genomics Issues

Although the public policy issues related to genomics cover a wide spectrum, policymakers and health officials approach these issues by addressing several common policy considerations.

Underlying Principles about Genetic Exceptionalism. There is debate about the need to establish separate policies for genetic information. Some argue that genetic information is no different than other medical information and should be covered in existing legislation, such as the federal Health Insurance Portability and Accountability Act. However, many policymakers argue that genetic information is special because, as Oregon legislation said, genetic tests have the "unique ability…to predict a person's future health." Sorting through these underlying assumptions is an important but challenging process for state policymakers and health officials.

Accessibility and Availability. As new technologies are available, health policy leaders examine their effect on access to genetic services and whether they improve access to services or create access disparities. Health leaders are identifying these barriers (e.g., maldistribution of services, lack of insurance, or lack of provider and public understanding of genetics) and considering policies to mitigate them. Some examples of questions that states are addressing include:

- How should the state develop its list of conditions under the newborn screening program? Some states defer to recommendations from other organizations, and other states sort through the information using frameworks.
- Do children identified through newborn screening have access to medically necessary follow-up services and specialty care? Is there an adequate network of providers and specialists to treat the growing number of children identified as having genetic conditions?
- What factors prevent people from receiving care and services, such as lack of insurance, lack of transportation, lack of information? Can public policy effectively and efficiently reduce these barriers?
- Do new technologies and services, such as direct-to-consumer genetic tests, enhance or thwart access to genetic services? What are the public health benefits and risks?

- Is the workforce adequately prepared to integrate new products and technologies? Does the public health workforce possess the needed competencies to translate genomics research into practice? Are primary care providers prepared to interpret and incorporate genetic tests into their patient care plan?

Effectiveness and Quality. State health leaders need evidence of effectiveness for a specific test or technology in order to ensure that it has the capacity to improve public health outcomes. As a result, they are examining the appropriate role for public health agencies, and developing standards and policies to ensure that products and services are effective, reliable and valid. Moreover, some health leaders are taking steps to ensure that genetic test manufacturers clearly communicate results, and provide adequate information to help consumers and their providers interpret those results.

Ethical, Legal and Social Implications. In order to protect the public from premature and inappropriate use of genetic information, health leaders are developing information, education, policy and legislation. Genomic advances involve often-controversial ethical issues. Health leaders must examine multiple perspectives, such as the ethical principles related to genetic testing and newborn screening, and legal protections for individuals and groups from genetic discrimination.

Standards and Guidelines. Federal, state and local health officials are being asked to set standards for the role of genomics within the scope of the core public health functions (assessment, policy development and assurance). The challenge can be staggering. For example, genomic advances are transforming the newborn screening process and new technologies are making it possible to identify an increasing number of genetic conditions. Although there is capacity to test for more conditions, many states are grappling with the process of developing a list of conditions that meet ethical, social, research and public health standards. Just because technology is available, should it be adopted? What are the social, ethical and public health costs and benefits of expanding newborn screening? According to a 2008 report by the Agency for Healthcare Research and Quality, "Screening recommendations for public health programs should be transparent, unbiased, evidence-based, and attentive to important social values, especially if they will affect every child born in the United States." This can be a tall order for policymakers and health officials, since it requires extensive genetics and policy expertise and data and community engagement.

As a result, states are examining the optimal process for developing policies and identifying the appropriate stakeholders to help develop standards and considering the following questions:

- What role should community engagement play in new initiatives, such as genetic "bio-banks" or genetic privacy laws?
- As new technologies are available, what role do state officials play in ensuring that these services and tests are effective and valid?
- Should states regulate direct-to-consumer tests and manufacturers? If so, how should this occur in an age of Internet commerce?
- Should new parents be able to decline or opt-out of having their child's dried blood spot retained? If so, what are the most effective ways to communicate newborn screening policies?

Conceptual Framework for Analyzing Newborn Screening Expansion

1. What is the ultimate goal, and how does the intervention achieve those ends?
2. How strong is the evidence that the intervention can improve patient outcomes?
3. How strong is the evidence that the intervention will work in the setting specific to the policymaker?
4. What constitutes "good enough" evidence for a policy decision?
5. What other considerations are relevant to policy decisions?

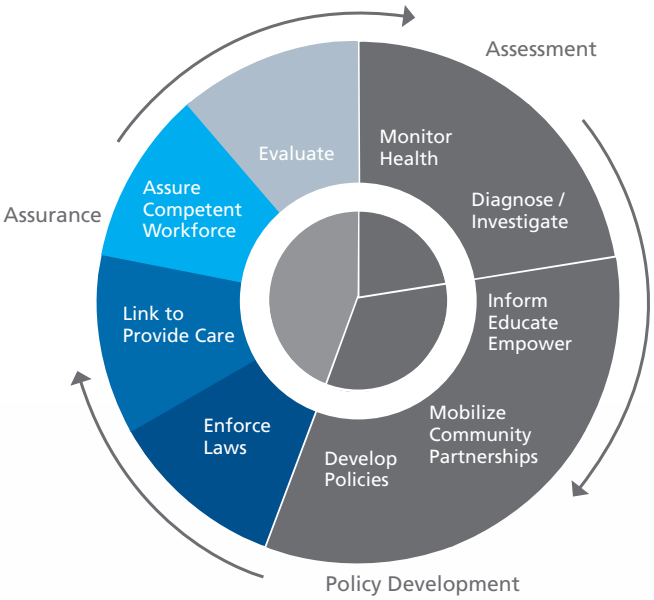
Source: David Atkins et al., Making Policy When the Evidence is in Dispute, Health Affairs, 2005.

Public Health Function: Assurance

Assurance Defined: Assuring that genomic information is used appropriately and that genetic tests and services meet agreed upon goals for effectiveness, accessibility and quality.

States conduct various assurance activities, ranging from developing genomics training and educational opportunities for healthcare providers and public health practitioners to providing or funding services in clinics, hospitals and health departments. Some state agencies, such as the Hawaii Genetics Office, monitor genetics research to ensure that it meets the public's expectations about the use of information, as well as the process for communicating findings to participants and their families.

This section on assurance strategies profiles state actions in Hawaii, Illinois and other states that focus on ensuring access to genetic services and addressing workforce issues to meet current and future demand for genetic services.



Source: CDC, Genetics, Genomics and Public Health: An Overview, 2009.

Action Area

Ensuring Access to Genomic Resources and Services

"Advances in genetic science, particularly through the Human Genome Project, have generated a wealth of information that is crucial for the prevention, early diagnosis and treatment of genetics-based diseases and conditions. Unfortunately, … communities in Illinois with a high concentration of racial and ethnic minorities are not benefiting from these scientific advances due to their limited knowledge of genetics, lack of understanding of the potential for improving health, and the lack of linguistic and culturally appropriate genetics services."

– University of Illinois at Chicago Midwest Latino Health Research, Training & Policy Center

Access to genomic services and technologies depends on several factors. Services must be available and affordable. Individuals need to understand what the services entail, why they are important and how they relate to their health.

Reducing Access Barriers. Many of the same factors that create disparities in the health care system at large are also problems that impede access to genetic services. States have taken steps to address some of these barriers. For example, some states have addressed cost barriers by mandating coverage for certain genetic tests (e.g., colorectal cancer screening for high-risk individuals) or mandating coverage for medically-necessary foods and formula for children with inborn errors of metabolism.

Improving the Availability and Quality of Services and Providers. In many communities, the genetics infrastructure, which includes providers, genetic clinics, public health departments, health centers, universities and laboratories, is ill-equipped to meet current and future demand for services. The problem can relate to quantity (e.g. not enough providers or sites, lack of capacity among current providers to meet demand for services) or other workforce characteristics, such as lack of provider knowledge about genomics. States have taken several steps to inform the public about existing services. Several states include directories of genetic services in their states on their websites. For example, the Oregon Genetics Program website contains contact information for genetic service providers, newborn hearing screening and newborn blood test screening. In addition, state genetics networks, such as the Mountain States Genetics Network, provide directories of services and providers.

Access Barriers Include:

- **Economic:** Lack of insurance, transportation.
- **Geographic:** Long distances to available services, lack of clinics/providers serving a community.
- **Language:** Lack of interpreters, culturally-competent providers and staff.
- **Educational:** Lack of knowledge about genetics and genetic services.
- **Cultural:** Some individuals have cultural or spiritual objections to genetic services, such as prenatal screening.

Challenges

- **Lack of Dedicated Funding.** Addressing access barriers can be expensive. Costs include paying for genetic, translator and transportation services, or providing financial incentives for providers to pursue genetic specialties. Many states do not have the funds to support projects and programs that are not mandated.
- **Lack of Leadership Support.** Garnering support from policymakers, state health officials, and state agencies can be a challenge, especially when these initiatives are competing with other programs for limited resources. As a result, demonstrating public health impact and return on investment is critical for leaders who make data-driven decisions.
- **Complexity.** Addressing complex problems for which there is no simple, inexpensive, or short-term solution, such as shortages and mal-distribution of providers, is a daunting challenge for policymakers, health officials and other stakeholders. Public and private partnerships enable states to develop solutions with multiple stakeholders and share the time and cost of implementing the solutions.
- **Lack of Reimbursement for Genetic Services.** A lack of appropriate billing and diagnosis codes for genetic services and testing is a challenge because providers often do not receive adequate reimbursement for their services. Some states are developing policies that recognize genetics providers (e.g., licensure for genetics counselors) and reimburse these providers for genetics services.

Strategies to Improve Access to Genomic Services

Because access to services depends on multiple factors almost every state genomics strategy is also an access strategy.

Table 15. Examples of State Activities to Improve Access to Genetic Services

Objective	Activities and Tactics
Ensure Availability of Genetic Services and Resources	• Support local health departments and genetic clinics that provide information, services and referrals. • Participate in statewide and regional collaborative projects that share services and providers through tele-health and sharing of human, financial and technology resources. • Promote professional development for rural providers and encourage opportunities for them to collaborate with genetic service providers. • Consider licensure for genetic counselors. According to the National Conference of State Legislatures, six states have laws that require a license to practice genetic counseling.
Decrease Cultural, Economic and Other Access Barriers	• Disseminate culturally-sensitive information about genetic services to target populations. • Address concerns over privacy and discrimination. The federal Genetic Information Nondiscrimination Act of 2008 addresses privacy and discrimination concerns, and several states prohibit genetic discrimination in employment, health insurance and other types of insurance. • Provide transportation services or reimbursement for individuals who lack transportation. • Address financial barriers that impede or delay access for insured individuals. Some states require insurers to cover certain procedures for adults based on positive genetic testing or family history.
Improve Public Awareness of Basic Genomics	• Develop educational initiatives, such as K-12 educational programs to introduce genomics and family history. • Implement educational and informational projects, such as awareness days, public service advertisements, community workshops and fact sheets. • Create informational websites to disseminate information about a range of genomics topics, including genetic testing, family history, privacy and anti-discrimination.
Educate Providers about Genomics	• Promote opportunities for health care providers and other staff to learn about cultural values and perceptions that relate to genetic services. • Train "front-line" providers, such as physicians, physician assistants and nurses, about genomics, and provide them with knowledge and tools to incorporate into practice. • Provide online information and curricula for public health providers including manuals, family history tools, and information about specific genetic conditions. • Offer or require genetics courses in graduate medical education and
Expand Access to Newborn Screening Services	• Develop and distribute culturally-sensitive information about newborn screening services to providers and families. • Provide information about state policies regarding privacy and storage of genetic information, and procedures for participating or opting-out of genomics research.
Address Workforce Needs and Gaps in Services	• Develop strategies and incentives (e.g., loan forgiveness programs) to attract more students to the field of public health genomics, as well as the number of physicians who specialize in genetics. • Develop strategies (e.g., telemedicine, state policies to allow for interstate licensure) to connect genetic service providers to underserved populations. • Develop a triage system that trains public health nurses to recognize when genetic services are needed.

Up Close: Federal-State Partnerships to Improve Access to Services

The CDC's Division of Heart Disease and Stroke Prevention administers the WISEWOMAN program, which provides eligible women (low-income women between the ages of 40 and 64 who are under-insured or uninsured) with chronic disease risk factor screening, lifestyle intervention, and referral services to prevent cardiovascular disease. CDC funds 21 WISEWOMAN programs in states and tribal organizations. The CDC requires all participating states to include family health history questions in their WISEWOMAN activities.

The Minnesota WISEWOMAN program, called Sage Plus. The Department of Health Cancer Screening and Heart Health Program administers the program and trains participating clinics to screen patients and provide education and lifestyle counseling. According to the program website, "[t]he mission of Sage Plus is to provide women with knowledge, skills, and opportunities to improve their diet, physical activity, and other life habits to delay or control cardiovascular and other chronic diseases."

Specific activities include:

- Individual risk reporting, which includes an individualized heart health profile and report that summarizes blood pressure, total cholesterol, glucose and other factors
- Lifestyle assessment which examines physical activity, smoking and dietary habits.
- Education about chronic disease risk factors and healthy behaviors through fact sheets and other materials.
- Lifestyle change counseling between the patient and a lifestyle counselor.
- Action planning between the lifestyle counselor and patient. The patient signs a "Lifestyle Change Contract" which contains concrete steps to reach their goals.
- Self-monitoring by patients.
- Follow-up between lifestyle counselors and patients to monitor progress, modify goals or set new goals.

Up Close: Hawaii's Community Genetics Program Addresses Workforce Gaps

Hawaii's needs assessment reflects input from providers, public health administrators and consumers about the most pressing needs surrounding genetics services in Hawaii. Many stakeholders, for example, reported that Hawaii's lack of pediatric geneticists was a top concern, and therefore, for several years, Hawaii did not have any pediatric geneticists, partly because they do not generate significant reimbursements for hospitals. As a result, hospitals did not recruit them.

To fill this gap, a diverse group of stakeholders came together to form Hawaii Community Genetics, a collaborative effort of the Department of Health, two of Hawaii's main medical centers, the University of Hawaii John A. Burns School of Medicine, and the Hawaii Medical Services Association (Hawaii's major third party payer). According to Au, State Genetics Coordinator, it works because "everyone contributes time and resources." The resources include funding, clinic space, support staff, genetic counselors, metabolic nutritionist, as well as dedicated staff to help with reimbursement policies.

By bringing together these stakeholders, Hawaii has recruited one pediatric geneticist who provides in-person outreach clinics, as well as tele-health visits, allowing the geneticist to see patients in diverse geographic locations and cut down on travel expenses. Today there is a nine-month waiting period for in-person services. Au explained that Hawaii is in the process of recruiting one or two more pediatric geneticists. Partnerships in this model are critical. Having the third-party payer at the table helps because it facilitates tele-health visits through favorable reimbursement policies and eliminates the need for pre-authorization for certain genetic tests. Au believes this model could work in states that do not have any geneticists. In her opinion, the key element to replicate this model is "a leader who can help get people to work together."

Success Story: Illinois at a Glance

Addressing Access Challenges on Many Fronts in Illinois

Agency	Genetics and Newborn Screening Program (GNSP)
Program Location	The GNSP is located in the Illinois Department of Public Health (IDPH) Office of Health Promotion.
Funding	Using newborn screening funds, the Genetics Program administered $2.9 million in 2009 in grants to genetic clinics, health departments for clinical services and State Genetics Plan grantees, which address five key areas identified in the state plan.
Staff	The Program has 13 staff members, including one Genetics Program Administrator and two nurses. One nurse focuses on newborn screening, the other on managing grants.
Sustainability	Reliable funding source in newborn screening funds.

Issue In Brief

The Genetics and Newborn Screening Program promotes access to genomic services by addressing multiple access obstacles including geographic, economic, language and cultural and educational barriers. Importantly, the Program promotes access to services through a statewide network of local public health agencies that provide information, services and genetic referrals for families. "Through this system," said Claudia Nash, Genetics Program Administrator, "no family is denied services."

The program does not stop there. Ensuring access to services depends on a number of other factors, including affordable services, public awareness, and culturally-competent providers. To that end, the program provides several grants to organizations that address reducing access barriers; public education; professional education; finance and reimbursement; and ethical, legal and social issues related to genomics, also known as ELSI. One grantee, the Midwest Latino Health Research, Training and Policy Center, offers genetics training to clinic and health department staff who interact directly with clients, including interpreters, genetic clinic front desk staff, and community health workers. At the front lines of patient care, these workers are an important conduit to genetic services to community members. In addition, the community health worker training program educates about delivering family history awareness sessions to community members. The Midwest Latino Center partners with other organizations and community stakeholders to improve awareness of genomics through events, media campaigns, a bilingual genetics website, and with the Chicago Center for Jewish Genetic Disorders to promote family history awareness through DNA Day.

" … [A]ccess to genetic services is not equally available and utilized by all segments of the population, particularly in underserved rural, low-income and minority communities. Understanding and reducing potential barriers to access to genetic services is key in providing equal opportunities for disease prevention and health promotion."

— 2007 Illinois State Genetic Services Plan

Table 16. Blueprint for Improving Access to Genetic Services in Illinois

Access Goals	Activities and Tactics
Promote High Quality, Comprehensive and Accessible Genetic Services for all Illinois Residents	• The Illinois Department of Public Health (IDPH) supports a statewide network of clinical genetic centers through a clinical genetics grant program. The IDPH provides grants to 18 clinical genetic centers which provide outreach services at 28 satellite clinics across the state. • The IDPH provided grants to 38 local health departments in 2009 to assure follow-up, referral, tracking and educational services to infants with a confirmed diagnosis; identify individuals and families who may benefit from genetic counseling and/or genetic services; refer all appropriate clients; and provide consumer and professional education. Some of these health departments sub-contract to smaller health departments to provide genetic screening, referral, and follow-up services. • The IDPH addresses cost access barriers by assisting families with transportation costs through grant funding to local public health agencies.
Promote Integration of Genomics into Illinois' Health Care Delivery System Through Provider Education and Training	• For more than 20 years, the IDPH has offered a one-week medical genetics training course for local health department and hospital nurses. • Through a partnership with the Chicago Center for Jewish Genetic Disorders and the UIC Midwest Latino Center, the IDPH funded the development of tool kits for IDPH Genetic Grantees and College Wellness Centers, including newborn screening and family health history fact sheets. • The UIC Midwest Latino Health Research, Training and Policy Center (an IDPH grantee) conducted the following activities: - Developed one-day training to educate medical interpreters in genetics. - Developed training program to educate front desk/support staff on providing culturally competent service. - Conducted a family history awareness pilot training program to train community health workers on how to use the U.S. Surgeon General's Family History Portrait. As of June, 2009, these trained CHWs had conducted six community workshops aimed at educating families and referring those at risk to available services and programs. • Northwestern University, an IDPH grantee, conducted various activities to improve provider awareness of genomics, including: - Partnered with health professional organizations (e.g., the Illinois Chapter of the American Academy of Pediatrics) to secure genetic speakers for physicians at grand rounds and other Illinois education programs for health providers. - Conducted and evaluated a genomics continuing medical education program track for clinical physician assistants at the 2008 Illinois Academy of Physician Assistants annual conference. - Identified and distributed Spanish language genetic fact sheets at local health fairs. - Conducted a genomics education needs assessment in Illinois' chronic disease prevention and control programs to identify education needs and programs to offer.
Improve Genomic Awareness and Literacy Among Illinois Residents	• The Midwest Latino Center developed a bilingual English/Spanish genetics website (**www.easylearngenetics.net**) to educate the public on genetics topics. The website's target audience are people with limited genetic literacy and ethnic minority populations such as Hispanics/Latinos in Illinois. • The Chicago Center for Jewish Genetic Disorder and the Midwest Latino Center developed and distributed brochures, flyers and posters to promote family health history and newborn screening, as well as 2009 DNA Day materials to organizations, clinics and the general public through health fairs and community outreach activities. • The 2009 DNA Day mass media campaign included four live call-in shows on a Chicago cable access channel. Genetic counselors conducted TV interviews in English and Spanish. Spanish language newspapers published press releases about the newborn screening program. • Researchers at the University of Chicago, an IDPH grantee, surveyed parents and pediatricians to ascertain their understanding of newborn screening, Cystic Fibrosis and Sickle Cell Disease.
Address Genetic Services Financing and Reimbursement Issues that Impact Genetic Service Providers	• Northwestern University, an IDPH grantee focusing on finance and reimbursement issues, evaluated billing practices of Illinois genetic service providers and reimbursement policies of public and private insurance providers to determine gaps in genetic services that were provided, billed and reimbursed.

Take-Away Message

- Complex problems demand comprehensive solutions.
- Creative communication strategies are critical.

Informational Resources

IDPH Genetics Website
http://www.idph.state.il.us/HealthWellness/genetics_prog.htm

Bilingual Genetics Website
http://easylearngenetics.net/illinois/illinois.html

2007 State Genetic Services Plan
http://idph.state.i.us/HealthWellness/GeneticServicesPlan.pdf

Action Area

Addressing Workforce Issues to Meet Demand for Genetic Services

"Increasingly, public health workers will be asked to incorporate genetic information into public health practice. An understanding of genetics as it pertains to each public health discipline will be essential to develop programs that benefit the public."

– CDC FAQ's About Competencies

Competencies Defined: "Competencies are cross-cutting because they are for all workers in public health and are also specific to certain disciplines within public health. (They) are a guide for public health workforce development including curriculum and the content of public health education and training programs."

Source: CDC, FAQ's About Workforce Competencies, **http://www.cdc.gov/genomics/translation/competencies/faqcomps.htm**

A competent workforce, according to the CDC, "possesses the skills, knowledge and attitudes necessary for the effective practice of public health." The challenge to building competency is made more complex by the broadness of the public health genomics workforce, which is comprised of state and local health agency employees, public health providers and support staff, laboratory workers, and others.

State public health agencies address workforce competency in two major ways:

- Genomics competency among public health administrators and agency employees. Does the agency have a qualified expert to help guide activities; inform and educate staff and external stakeholders, and identify opportunities for collaboration? Does chronic disease program staff understand the role that genomic tools, such as genetic testing and family history, play in program activities? Do agency staff members have a sufficient knowledge of genomics to ensure that they are applying knowledge to benefit public health and take steps to protect the public?
- Genomics competency among the public health workforce. Is the primary care public health workforce prepared to interpret genetic test results, make referrals when needed, and apply genomic tools into their practices?

Up Close: Genomic Competencies for Public Health Professionals and Administrators/Leaders

The 2001 document, *Genomics Workforce Competencies* was developed by an interdisciplinary team of individuals representing local, state and federal public health programs. According to the CDC, these competencies were developed "as a tool for public health programs and schools of public health to incorporate genomics into existing competencies and program training goals."

A public health professional within his/her professional field and program is able to:

1. Apply the basic public health sciences, including behavioral and social sciences, biostatistics, epidemiology, informatics, and environmental health to genomic issues, studies and genetic testing, using the genomic vocabulary to attain the goal of disease prevention;
2. Identify ethical and medical limitations to genetic testing, including uses that do not benefit the individual;
3. Maintain up-to-date knowledge of the development of genetic advances and learn the uses of them to achieve public health goals related to his/her field;
4. Identify the role of cultural, social, behavioral, environmental and genetic factors in development of disease, disease prevention, and health promoting behaviors and their impact on medical service organization and delivery of services to maximize wellness;
5. Participate in strategic policy planning and development related to genetic testing or genomic programs;
6. Collaborate with existing and emerging health agencies and organizations, academic, research, private and commercial enterprises to identify and solve genomic-related problems;
7. Participate in the evaluation of program effectiveness, accessibility, cost benefit, cost effectiveness, quality of personal, and population-based genomic services in public health;
8. Develop protocols to ensure informed consent and human subject protection in research.

A public health leader/administrator as appropriate to a specific agency or program is able to:

- Communicate the role of genomics in public health to policy makers, community members and staff;
- Develop a clear understanding of the perspectives of various community stakeholders that may apply genetic information beyond the individual and/or family;
- Identify the political, legal, social, ethical and economic issues associated with integrating genomics into public health;
- Effectively integrate genomic issues into policies and programs;
- Assure that current science and research are used in planning and delivery of genomic services;
- Include genomic competencies in staffing plans to ensure adequate capacity building;
- Assure that all workers develop appropriate genomic competencies and can appropriately apply genomic knowledge and tools within the parameters of their professional duties;
- Manage genomic program fiscal and human resources and strategies for developing budget priorities and proposals for funding from external sources;
- Promote a genomics oriented policy agenda that effectively addresses the appropriate use of genetic tests, effective service delivery, and adequate funding streams.

Source: CDC, Genomics Competencies, **http://www.cdc.gov/genomics/translation/competencies/index.htm**

Challenges

- Lack of Dedicated Funding. Cost is a major barrier for public health agency internal efforts (e.g., informing and educating departmental staff), as well as external activities (e.g. educating health care providers).
- Lack of Leadership Support. Genomics staff members may struggle to maintain leadership support and encouragement of genomics training opportunities and partnerships.
- Competing Public Policy Priorities. Health care provider shortage is a widespread problem. The field of medical genetics and genetic counseling is just one of many specialties suffering. Policymakers who develop and fund workforce development activities must make difficult decisions about distributing limited resources among wide-ranging fields. Presenting data about the genetics workforce and how shortages impact public health in affected areas is key.

Strategies to Improve Workforce Competencies

State public health leaders adopt a wide range of strategies to bolster the public health workforce. Some state public health departments support an agency genomics expert and/or team to build knowledge within the agency and coordinate internal and external activities. Several state and local health departments provide, coordinate or advertise workforce training and education opportunities through summer institutes, online courses, provider manuals and fact sheets.

Improving Competency in the Public Health Agency. Many of the state public health agencies that ASTHO interviewed for this publication have at least one full-time staff member who is responsible for coordinating genomics activities. According to the 2007 report, Genomics and Public Health Practice: Lessons from State Pilot Projects, these individuals are critical drivers of success. "Without a doubt," the report found, "the most often cited factor contributing to success was the creativity, perseverance and commitment of program staff." Without such a person, states struggle to maintain momentum for genomics integration activities.
State public health agencies hire individuals with public health and/or genetics expertise and training to perform a wide range of functions, including:

- Provide leadership, direction, management, planning and coordination for genetic/genomics program activities;
- Coordinate genetics/genomics activities in the division and agency;
- Communicate and coordinate with internal and external stakeholders (e.g., academic institutions, third party payers, public health providers, policy experts, policymakers, national organizations) to advise about genomics issues, implement and evaluate genomics activities, and monitor legislation, research and technology advances;
- Perform needs assessments, program evaluations and/or cost effectiveness activities;
- Implement the state genetics plan, strategic plan or other planning document;
- Manage newborn screening programs;
- Manage grants with federal agencies, local service providers and other funding partners; and,
- Perform program evaluation for genetic/genomic services, education and other program activities.

These individuals develop genomics knowledge within the agency through education (e.g., presentations at staff meetings), fundraising and partnerships with internal and external stakeholders. In addition, these staff members perform other core public health functions, including informing others about genomics through websites, reports and testimony for legislators, and educational activities (e.g. DNA Days, lecture series and provider opportunities).
According to the Wang and Watts report, "Staff members of public health genomics departments tended to identify with one of the two disciplinary backgrounds: public health or genetics, but rarely both." ASTHO reviewed state genetics/genomics job descriptions for this report and found examples of states that are trying to recruit staff members with both backgrounds by requiring a public health background (i.e., a master's of public health degree or equivalent) with some coursework in human genetics.

Having a public health background offers several advantages. People with this background use public health language and tools to communicate with other program areas, and they have a broad understanding of how public health programs operate. Several public health agency managers told ASTHO that fluency in public health is critical for integrating genomics throughout the agency. Moreover, states that assigned genetics duties to their newborn screening managers often lost out on important opportunities to apply genomics broadly within the department. "They need to speak the public health language," one person told us during interviews for this report. Individuals with training in genetics also bring important strengths and skills, such as an ability to understand and interpret genetic science, and work with health care providers to translate research into practice.

Some states contract with consultants to provide specific functions, such as developing guidelines for health care providers, conducting research, and marketing and outreach. Others rely on maximizing agency expertise and resources. For example, Connecticut's Internal Council of Genomics has representation from chronic disease, environmental health, infectious disease, newborn screening and other programs. Engaging broad and cross-cutting staff participation is enables the agency to stretch limited staff and financial resources.

Improving Competency among the Public Health Provider Workforce. State public health agencies perform a wide range of activities designed to improve provider awareness and use of genomics tools and resources.

- The Illinois Department of Public Health (IDPH) offers a one-week medical genetics training course for local health department and hospital nurses. In addition, an IDPH grantee provides training programs to medical interpreters, front desk and support staff on genetics and providing culturally competent service.
- The Oregon Genetics Program offers education and training opportunities for health care providers, including a summer seminar series that focuses on different genetics/genomics topics, such as colon cancer and family history.
- The University of Washington Center for Genomics and Public Health sponsors the Summer Institute in Public Health Genomics. The Institute features faculty from the CDC, state health agencies, private sector and academic institutions. The Institute provides an introduction to public health genomics for individuals "interested in obtaining knowledge and skills for integrating genomics principles and applications into health practice and policy." In 2009, the curriculum consisted of modules that focus on a variety of topics including translating public health genomics into practice, genetic testing, genomics and health literacy, and bio-banks and bio-trusts.
- A free, online workshop, "Incorporating Genetics and Genomics Into Your Curriculum," is available to nurse educators through the TRAIN (TrainingFinder Real-time Affiliate Integrated Network) program, a national learning resource for public health professionals. Users can register for workshop on the Connecticut Department of Public Health website and receive four continuing education units from the National League for Nursing. The workshop covers chromosomal disorders, modes of inheritance, family history development, and integrating genetics in nursing curricula. The project is a partnership between the St. Vincent's College, the Connecticut Department of Health, the New England Genetics Collaborative, and the U.S. Health Resources and Services Administration.
- The Genetic Alliance and the New England Public Health Genetics Education Collaborative, with health department representatives from Connecticut, Rhode Island, Massachusetts, Vermont, New Hampshire and Maine, published *Understanding Genetics: A New England Guide for Patients and Public Health Professionals* in 2007. The publication contains information for providers (e.g., indications for referrals, information on ethical, legal and social issues, as well as fact sheets and other information for consumers.

For More on Workforce Competency:

- CDC Genomic Workforce Competencies provides list of competencies for different types of public health professionals.
- National Center for Cultural Competencies at Georgetown University Center for Child and Human Development includes organizational self-assessments, definitions, tools and policies for achieving cultural competence.
- March of Dimes "Genetics and Your Practice" provides information for providers about integrating genetics into their practices, with information about genetic testing, family history and referrals for genetic services.
- Core Competencies in Genetics for Health Professionals outlines health care professional competencies for taking and interpreting family health history. (National Coalition for Health Professional Education in Genetics)
- Genetic Counselor Competencies specifies competencies for genetic counselors. (American Board of Genetic Counseling)
- *Essentials of Genetic and Genomic Nursing: Competencies and Curricula Guidelines and Outcome Indicators* defines genetic and genomic competencies for registered nurses.
- Secretary's Advisory Committee on Genetics, Health and Society Resolution on Genetics Education and Training of Health Professionals.
- "Genetics in Clinical Practice: A Team Approach" is a "virtual practicum" for healthcare providers (and students) who see patients with genetic disorders. Participants learn through patient simulations in which the learner's decisions and knowledge about clinical genetics affect the patient's health outcomes. Users have access to a Learning Resource Room that provides lectures, interviews and online resources.

1 Oregon Department of Human Services. "Using BRFSS to Collect Genomics Data: State Findings." Available at **http://egov.oregon.gov/DHS/ph/genetics/docs/poster11x17_large.pdf**. Accessed in 07-09.

2 The WISEWOMAN program is described in greater detail later in this section under access initiatives.

3 Michigan Department of Community Health. "Genetics Through the Life Cycle: Improving Health and Preventing Disease." Available at **http://www.michigan.gov/documents/MIgenetics planandassessment__118168_7.pdf**. Accessed in 07-09.

4 Rick Weiss, "Genetic Testing Gets Personal," Washington Post, March 25, 2008, **www.washingtonpost.com/wp-dyn/content/article/2008/03/24/AR2008032402750.html**.

5 Secretary's Advisory Committee on Genetics, Health and Society. "U.S. System of Oversight of Genetic Testing: A Response to the Charge of the Secretary of Health and Human Services." April 2008. Available at **http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS_oversight_report.pdf**.

6 Gail Javitt, "Too Much Information," transcript from an online conference, washingtonpost.com, June 13, 2006, **www.washingtonpost.com/wp-dyn/content/discussion/2006/06/12/DI2006061200946.html.**

7 New England Journal of Medicine. "Letting the Genome Out of the Bottle, Will We Get Our Wish?" January 10, 2008. Available at **http://content.nejm.org/cgi/content/full/358/2/105.**

8 Martin S. "Changes for the Louisiana Public Health Laboratory System." Louisiana Morbidity Report. 17: 3-5. Available at **http://www.dhh. louisiana.gov/offices/publications/pubs-205/worker_health.pdf.**

9 Chapter 74 of the Public Health Code is available at **http://mlis.state.md.us/2007RS/chapters_noln/Ch_74_hb0344T.pdf.**

10 Connecticut Department of Public Health. Feasibility Study for a Statewide Biobank to Study Preterm Births and Birth Defects. November 2007. Available at **http://www.ct.gov/dph/lib/dph/BIOBANK_Final_Report_111407.pdf**.

11 Michigan Department of Community Health. "Michigan BioTrust for Health." Available at **https://sso.state.mi.us/prod-bsltpublic/blankslatepublic/viewslate.aspx. Accessed 06-2009**.

12 Virginia A. Moyer, Ned Calonge, Steven M. Teutsch, and Jeffrey R. Botkin, on behalf of the United States Preventive Services Task Force. "Expanding Newborn Screening: Process, Policy, and Priorities," Hastings Center Report 38, no. 3 (2008):32-39. Available at **www.thehastingscenter.org.**

13 Centers for Disease Control and Prevention. "Genomics Workforce Competencies." Available at **http://www.cdc.gov/genomics/translation/competencies/faqcomps.htm**. Accessed 08-22-09.

3 Section Three

Charting A Path For Genomics In Your State

Genomics, which many public health leaders may think of as a new program or expense, must compete with a vast array of mandated programs and services, not to mention emerging public health issues that demand resources and attention. Between July 2008 and December 2009, state public health departments lost over 3,600 workers through layoffs and attrition."

Given these circumstances, the notion of increasing attention, much less funding or staff time, to a non-mandated program or area may seem unrealistic at best. For that reason, any discussion of next steps must be grounded in realistic and inexpensive options that offer concrete benefits. The most viable options typically share a few common characteristics: they bring together key stakeholders who share resources and expertise; they offer important benefits to stakeholders; and they address a specific and pressing public health problem. In addition, making the case to public health leaders demands that the activities are tied to the agency's core functions and mission.

The field of genomics is advancing rapidly. As a result, public health agencies often find themselves reacting to emerging hot topics, such as direct to consumer marketing of genetic tests. Having a plan in place is needed for states to take a proactive approach in addressing these innovations.

The remainder of this resource guide focuses on how states can overcome barriers and set goals related to genomics and public health. A state's next steps will vary based on a number of factors including: funding, organizational support, political will, genomics experience and training among staff, and the state's track record for collaboration.

Making the Case to State Public Health Leaders

There are compelling and important reasons for supporting state public health genomics.

1. Genomics is a tool for achieving population health goals from infancy through adulthood. By understanding how genes are affected by one's environment and behavior, we can develop better interventions to prevent, detect and treat disease.

2. State public health departments play an important role by providing unbiased education and information about genetics/genomics and available testing for the general public. Genomics can strengthen the impact of risk reduction campaigns and messages.

3. Rapid advances in genetic services and testing call for oversight and assurance, one of the core public health functions.

4. Public health officials can use their expertise and credibility to inform policymakers on the numerous ethical, legal and social issues in genomics.

5. Genomics enhances surveillance opportunities at state health agencies. Some states are identifying important connections between family health history and disease, and developing public health interventions to reduce disease risk among certain populations.

6. The cross-cutting nature of genomics represents an opportunity for aligning goals and objectives from various program areas. Stakeholders from academic institutions, government, foundations, consumer groups and provider groups can forge partnerships to create and maintain genomics initiatives, and public health agencies can coordinate and oversee these joint initiatives.

Step 1: Obtaining Agency Support And Buy-In

Convincing public health leaders and policymakers about the need for integrating genomics into public health activities is often an uphill battle, partly because genomics is a relatively new and separate field of science. One health department employee said, "it's intimidating and that turns people off." To be sure, the field of genomics is complex and rapidly evolving; however, it is also creating unprecedented opportunities for public health officials that are practical, immediate and valuable.

Examples of Questions to Ask:

- Who performs genomic/genetic functions in the department? Is there a genetics/genomics coordinator?
- Did the agency complete a needs assessment or state genetics plan? If so, is it being used currently?
- What are the current genetic/genomic activities and services? Is there a centralized website?
- What genomics data is currently being gathered? Are there registries that already gather family history or other genomic information?
- What are the existing and potential funding sources for genomics?
- Who are internal and external partners? What are they addressing?
- What are staff perceptions about gaps in services, strengths, weaknesses, opportunities and threats related to genomics?
- What hot topics need to be addressed?

For More Information:

- The National Newborn Screening and Genetics Resource Center (NNSGRC) has a link to state genetics plans at **http://genes-r-us.uthscsa.edu/resources/genetics/geneticsplan.htm**.
- The NNSGRC also maintains links to state genetics or newborn screening programs at **http://genes-r-us.uthscsa.edu/resources/genetics/state.htm**.
- The National Coordinating Center for the Genetics and Newborn Screening Regional Collaborative Groups maintains links to seven regional collaboratives at **http://www.nccrcg.org/disclaimer.asp?loc=http://www.nccrcg.org/regional.asp.**

Step 2: Taking Stock

Understanding where genetics and genomics currently reside in the public health agency is key. Traditionally, genetics has been associated with newborn screening programs, which are usually administered by the maternal and child health or laboratory division in the public health agency. Key informant interviews and informal conversations may be effective methods for gathering information about who "does" genomics, where they are located, how they are funded, and with whom they collaborate. It is important to identify whether a genetics or genomics state plan or needs assessment exists, and whether it is currently being used. Many states completed needs assessments but lost momentum when funding disappeared. Although it may now be several years old, the document may be a focal point for staff to discuss next steps and identify activities or gaps that can be realistically addressed.

Step 3: Charting A Course

Many states have a common framework, such as a needs assessment, state plan, or strategic plan for addressing genomics and making decisions about the future. Although not all states actively apply them. The plan provides a starting point for stakeholders to revisit needs and identify new opportunities that may have emerged since they completed their plan.

A needs assessment helps the health agency and its partners identify the need for integrating genomics into public health and set relevant priorities. Table 17 summarizes why and how states have conducted needs assessments.

Examples of Needs Assessments and State Plans

Several states' Needs Assessments and/or State Plans are available online. Some examples include:

- Illinois State Genetic Services Plan at http://www.idph.state.il.us/HealthWellness/GeneticServicesPlan.pdf
- Colorado State Genetics Services Plan at http://www.cdphe.state.co.us/ps/genetics/geneticsplan.pdf
- Michigan Needs Assessment and State Plan at http://www.michigan.gov/documents/MIgeneticsplanandassessment__118168_7.pdf
- Oregon's Assessment Methods and Findings at http://www.oregon.gov/DHS/ph/genetics/docs/needs02.pdf
- Oregon's Strategic Plan for Genetics and Public Health at http://www.oregon.gov/DHS/ph/genetics/docs/orplan02.pdf
- Utah Genomics Plan at http://health.utah.gov/genomics/Genomics%20plan%20final_Sept2008.pdf
- Utah Asthma Genomics Work Plan at http://health.utah.gov/genomics/pages/projects/asthmaconference/Workplan%20with%20notes%20-%20FINAL.pdf

Table 17. Reasons for Conducting a Needs Assessments and State Examples

Reasons	Examples
Bring together a diverse group of stakeholders (e.g., community members, providers, health department officials, policymakers, third party payers, etc.) to identify and prioritize needs.	• Community involvement was a cornerstone of Hawaii's needs assessment process. It included health care providers, public health administrators and community members.
Gather baseline data about the current and future level and use of services.	• A genomics needs assessment could investigate the number of children with inherited disorders, where they receive services, and the availability and accessibility of those services. • The Michigan needs assessment summarizes the current supply and distribution of hospitals and clinics, as well as the supply of geneticists and genetic counselors relative to the population.
Assess the capacity of the health agency to support expected growth in the genomics field.	• An objective of the Illinois Genetic Services Plan is to develop a public health infrastructure to support genomics activities. This includes raising awareness among policymakers and agency leaders. • The Michigan needs assessment summarizes the agency's current organizational and administrative structure of genetics, including staffing and funding sources at the state and local level.

The specific work processes and products included in a needs assessment vary by state, but they typically share common characteristics, including:

- Stakeholder input, with representation from consumers, public health programs, health care providers, third party insurers, advocacy groups, researchers and others. Needs assessments typically gather information about public attitudes and beliefs, as well as provider perceptions and practices.
- State population demographics.
- Availability and utilization of existing genetics programs and resources, including direct and indirect genetic services, laboratory services, etc.
- Projected needs for services and genetic resources.
- Conclusions and recommendations, which often include consensus-based goals and objectives, as well as strategies for achieving these goals.
- Data collection using different methods, such as telephone interviews, focus groups, key informant interviews, and analysis of state data sources (e.g., registries, databases, etc.).

The needs assessment often drives the state genetic services plan, which outlines how the state will address its needs. Some states focus their plans on one or more diseases. Utah's Asthma Genomics Workplan, for example, contains strategies for addressing several key issues related to asthma and genetics.

What if a needs assessment isn't realistic in my state?

If this is not an option, consider less formal ways to convene a core group of staff members to pursue one or two pressing action items. States can still move ahead without a formal plan. Connecting with interested colleagues who already see the value of using genomics tools in their activities is an effective strategy. Look for things that can be done easily without a lot of resources. Working with colleagues in the chronic disease program to integrate family health history into consumer guides and fact sheets, for example, may be a useful and feasible step. Some states dovetail efforts with emerging issues, such as news coverage of direct-to-consumer genetic tests, to maximize impact. Refer to another state's plan if it helps you narrow down the field of issues and develop an action plan.

Step 4: Taking Action

The states highlighted in this publication suggest that certain factors are essential building blocks for genomic activities. Every state must chart their own course based on political, agency, fiscal and other factors, but the experiences highlighted in the previous section demonstrate the importance of developing productive partnerships to move forward with genomics activities. This section highlights strategies and resources that states have used to implement genomics strategies.

Keys To Success

States and local public health departments have used diverse strategies to address the challenges of integrating genomics into their programs. In their 2007 survey of state genomics programs, Grace Wang and Carolyn Watts identified several keys to success.

1. **Passionate People.** "Without a doubt," the report stated, "the most often cited factor contributing to success was the creativity, perseverance, and commitment of program staff." While most champions reside in the health departments, some states have external champions from universities, health care providers and faculty that help to move projects along and keep a focus on genomics.

2. **Knowing When to Lead and When to Follow.** Programs fulfilled both a leadership role with genomics in their department, as well as a supportive role, in which genomics staff would provide support and guidance to other program areas. "Creating these secondary opportunities was crucial," the Wang and Watts report found,"both to make the best use of limited resources but more importantly to assure the continuation of genomics activities."

3. **Forge Collaborations with Early Adopters.** Program staff worked with colleagues in other areas to "weave genomics into their work" through other program grants or by developing genomics content for inclusion in other programs' education and communication materials.

4. **Build on Existing Data Systems.** Several state genomics program officials have worked with other programs and agencies to include genomics questions or data collection into existing surveillance systems, such as the Behavioral Risk Factor Surveillance System. These approaches utilize existing systems and create opportunities for data collection and linkages between program areas.

5. **Explicit Partnerships.** State genomics programs can not accomplish their goals alone. Instead, they rely on numerous "productive partnerships" with other state programs and agencies, advisory committees, academia, federal programs, and other state genomics programs. "With limited budgets and limited personnel, genomics programs relied on the assistance of other agencies and organizations to extend their work."

6. **Organizational Factors.** States with a genetics plan or needs assessment have an advantage because these provide guidance for state activities. In addition, ASTHO's research found that state decisions about where to locate genomics functions within the health agency were critical factors to program success. For example, one state official said that having genomics at a high level within the organization facilitated integration with various program areas.

7. **Maximizing Opportunity.** The Wang and Watts report found that programs benefited from maximizing "windows of opportunity," taking advantage of rising public interest in genomics topics can be an effective way to educate and inform consumers and policymakers. Several states have developed DNA Days, or other public information campaigns aimed at improving awareness and genetic literacy.

Partnering For Results

Smart collaborations leverage the state role and resources with individuals and organizations that contribute different skill sets and resources. In addition, partnerships improve access to new constituencies, such as policymakers, community-based organizations and providers.

State health agencies typically take several common steps when building and maintaining relationships.

1. **Identify potential partners at the local, state and national level that can help to achieve specific goals and address unmet genomics needs.** The field of public health is full of potential partners. State health agencies need to understand their options and be strategic, in pursuing them. Effective partnerships will provide states with the needed knowledge, skills or training to address high-priority needs while maximizing their limited resources.

2. **Develop a strategy for building and maintaining relationships.**

3. **Identify clear goals and track progress towards achieving them.** A hallmark of successful partnerships is the focus on an achievable and measurable goal. It is important to track milestones, celebrate successes and change courses if needed.

There are numerous types of collaborations, including partnerships within state agencies, with other states' agencies, with federal or local governments, with community-based organizations, and with private and other stakeholders.

Inter- and Intra-Agency Partnerships. Many state genomics programs partner with other health programs or other agencies to complete specific projects, such as data collection, policy development, and fundraising. This promotes sustainability by integrating genomics into other program grants and activities, such as Cancer Control Plans, Women, Infants and Children (WIC) counseling or other programs. Several state genomic programs develop and maintain internal relationships by providing education, expertise and ongoing consultation. Some other examples of inter-agency partnerships include:

- In Michigan, the genomics program collaborates with the Healthy Homes Section to integrate family health history into the Healthy Homes University program.
- Utah and other states consult with chronic disease programs to weave family health history into other program activities, including grants, and public education.
- Several states (including Michigan, Minnesota, Oregon and Utah) integrate family health history into the Behavioral Risk Factor Surveillance System. By piggybacking on existing data collection efforts, states are gathering key information about genomics without having to develop new surveillance systems.
- Connecticut establishes and formalizes relationships with other program areas through the internal Council of Genomics (COG), which convenes 15 members on a monthly basis to discuss genomics office activities. The COG membership is cross-cutting, and includes staff from areas such as chronic disease, laboratory services, research and development and health surveillance.
- The Connecticut Department of Health also establishes a broad-based internal liaison group, known as the Gene Team. This group meets twice a year and consists of about 35 members from various DPH programs. Members are "genomics ambassadors" to their programs.

State-to-State Partnerships. There are numerous examples of state health agencies that collaborate with agencies in other states to share resources and ideas, and address common issues and problems. Some of these are ad hoc partnerships driven by a specific issue or challenge facing states, such as the collaboration among Connecticut, Massachusetts and New York to address public information needs regarding a large-scale direct-to-consumer marketing campaign.

Regional collaboratives are an effective mechanism for bringing neighboring states together to address issues and share information. The Genetic Services Branch in the U.S. Health Resources and Services Administration supports seven regional genetics and newborn screening collaborative groups (see Table 18).

Table 18. U.S. Regional Genetics and Newborn Screening Collaboratives

Name	Participating States	Website
New England Genetics Collaborative	Connecticut, Massachusetts, Maine, New Hampshire, Rhode Island, and Vermont	http://www.negenetics.org/
New York-Mid-Atlantic Consortium for Genetics and Newborn Screening Services	Delaware, District of Columbia, Maryland, New Jersey, New York, Pennsylvania, Virginia and West Virginia	http://www.wadsworth.org/newborn/nymac/
Southeast Newborn Screening and Genetics Collaborative	Alabama, Florida, Georgia, Louisiana, Mississippi, North Carolina, Puerto Rico, South Carolina, Tennessee, and the Virgin Islands	http://southeastgenetics.org/
Region 4 Genetics Collaborative	Illinois, Indiana, Kentucky, Michigan, Minnesota, Ohio and Wisconsin	http://region4genetics.org/
Heartland Regional Genetics and Newborn Screening Collaborative	Arkansas, Iowa, Kansas, Missouri, North Dakota, Nebraska, Oklahoma, and South Dakota	http://www.heartlandcollaborative.org/
Mountain States Genetics Regional Collaborative Center	Arizona, Colorado, Montana, New Mexico, Nevada, Texas, Utah, and Wyoming	http://www.msgrcc.org/
Western States Genetic Services Collaborative	Alaska, California, Guam, Hawaii, Idaho, Oregon, and Washington	http://www.westernstatesgenetics.org/

Federal-State and Other Public Partnerships. Several states partner with federal, state and local agencies. For example, the CDC's Office of Public Health Genomics (OPHG) funds state initiatives and provides an online information clearinghouse of research, policies, funding, training opportunities, resources, and other emerging issues. The Office of Public Health Genomics, with other stakeholders, formed the Genomics Applications in Practice and Prevention Network (GAPPNet) in 2009 to, among other things: convene individuals and groups conducting genomics translation research; synthesize and evaluates research findings; and develop and disseminate validated genomic knowledge and applications for use in medicine and public health.

In addition, the OPHG supports two Centers for Genomics and Public Health at the University of Washington and University of Michigan. These Centers provide information, technical assistance and partnership opportunities for state health agencies and other stakeholders.

Community-Based Partnerships. Recognizing the importance of educating and engaging the public, many states have partnered with community groups and specific populations to gather input and deliver information and services. Some examples include:

- Utah partnered with Hispanic/Latino and Pacific Islander advisory committees to adapt classroom curricula and develop culturally and linguistically-appropriate take-home activities about family health history. The 5th grade curriculum is available in English, Spanish and Tongan. The high school curriculum is available in English and Spanish.
- Utah awarded community mini-grants to community agencies and universities to implement family history projects.
- The Hawaii needs assessment reflects input from providers, public health administrators and consumers about the most pressing needs surrounding genetics services in Hawaii.
- The Midwest Latino Center, which receives funding from the Illinois Department of Public Health, partners with other organizations and community stakeholders to improve awareness of genomics through events, media campaigns and a bilingual genetics website.
- Through a partnership with the Chicago Center for Jewish Genetic Disorders, the Illinois Department of Public Health funded the development of toolkits for IDPH Genetic Grantees and College Wellness Centers, including newborn screening and family health history fact sheets.

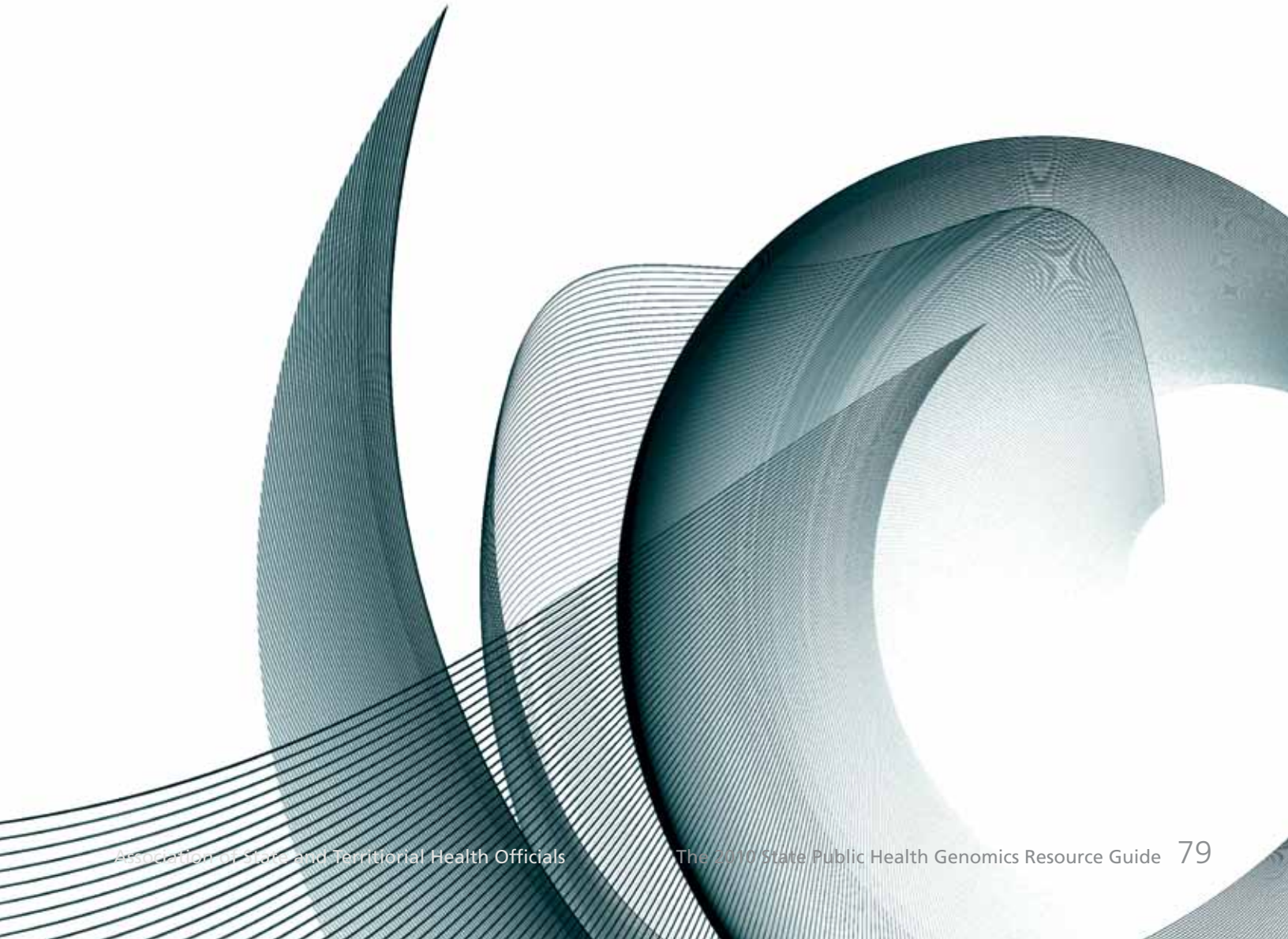
Public-Private and Academic Partnerships.
Several state agencies collaborate with academic institutions or form advisory committees with representation from the public and private sectors in order to integrate genomics into their programs and conduct outreach. Some examples include:

- Utah forged external partnerships to conduct research, develop materials and disseminate information. Partners included academic institutions, provider groups (e.g. genetic counselors), media agencies, the Family History Library, private companies and community-based organizations (e.g. National Tongan American Society).
- Utah partnered with the Genetic Science Learning Center to develop a high-school health history curriculum and adapt existing 5th grade genetics modules on family health history for biology and health students and their families.
- The Hawaii Genetics Program assembled a diverse group of stakeholders, including the Department of Health, the Hawaii Medical Services Association (Hawaii's major third party payer), two of Hawaii's main medical centers and the University of Hawaii School of Medicine to address a pressing workforce problem.
- The Illinois Department of Public Health partnered with health professional organizations (e.g., the Illinois Chapter of the Academy of Pediatrics) to secure genetic speakers for physicians at grand rounds and other Illinois education programs for health providers. The IDPH also conducted and evaluated a genomics continuing medical education program track for clinical physician assistants at the 2008 Illinois Academy of Physician Assistants annual conference.
- Connecticut established and formalized relationships with external experts through its Expert Genomics Advisory Panel, which convenes medical, insurance, academia, law, bioethics, genetic counseling and consumer advocacy communities. Their charge is to provide independent guidance and advice to DPH on clinical, scientific, legal, educational, ethical, and social issues related to public health genomics.
- The Connecticut Department of Public Health convened a panel of experts to examine the feasibility of a statewide population-based biobank of donated human tissue and information that would study preterm births and birth defects.
- The Oregon Genetics Program partners with the Oregon Health and Science University (OHSU) to offer workforce training and education on genomics-related topics. In addition, an OHSU MD/Masters of Public Health intern worked with the genetics program to examine family history collection at the state's federally qualified health centers and develop a recommended template for collecting this patient information.
- Recognizing the policy issues related to storing dried blood spots left over from newborn screening, the Michigan Department of Community Health is seeking public input to help develop policies for a formal biobank, known as the Michigan BioTrust for Health. The website provides information about newborn screening, dried blood spots, biobanking, privacy and potential research uses. The website also includes a survey that allows the public to provide input on the issues.

Conclusion

As your agency contemplates its next steps, think about the wide range of state strategies profiled in this guide and determine whether any of these options makes sense in your own state. Charting a course for the future is undoubtedly a challenge. However, the states in this guide illustrate that progress can happen even in uncertain times.

The field of genomics is advancing rapidly, providing challenges and opportunities alike to state and local public health agencies. Public health leaders will be called upon to develop policies that help agencies protect the public and improve the population's health. As they have always been, public health staff will have to be creative and efficient as they develop approaches that bring diverse stakeholders together to solve complex public health problems.



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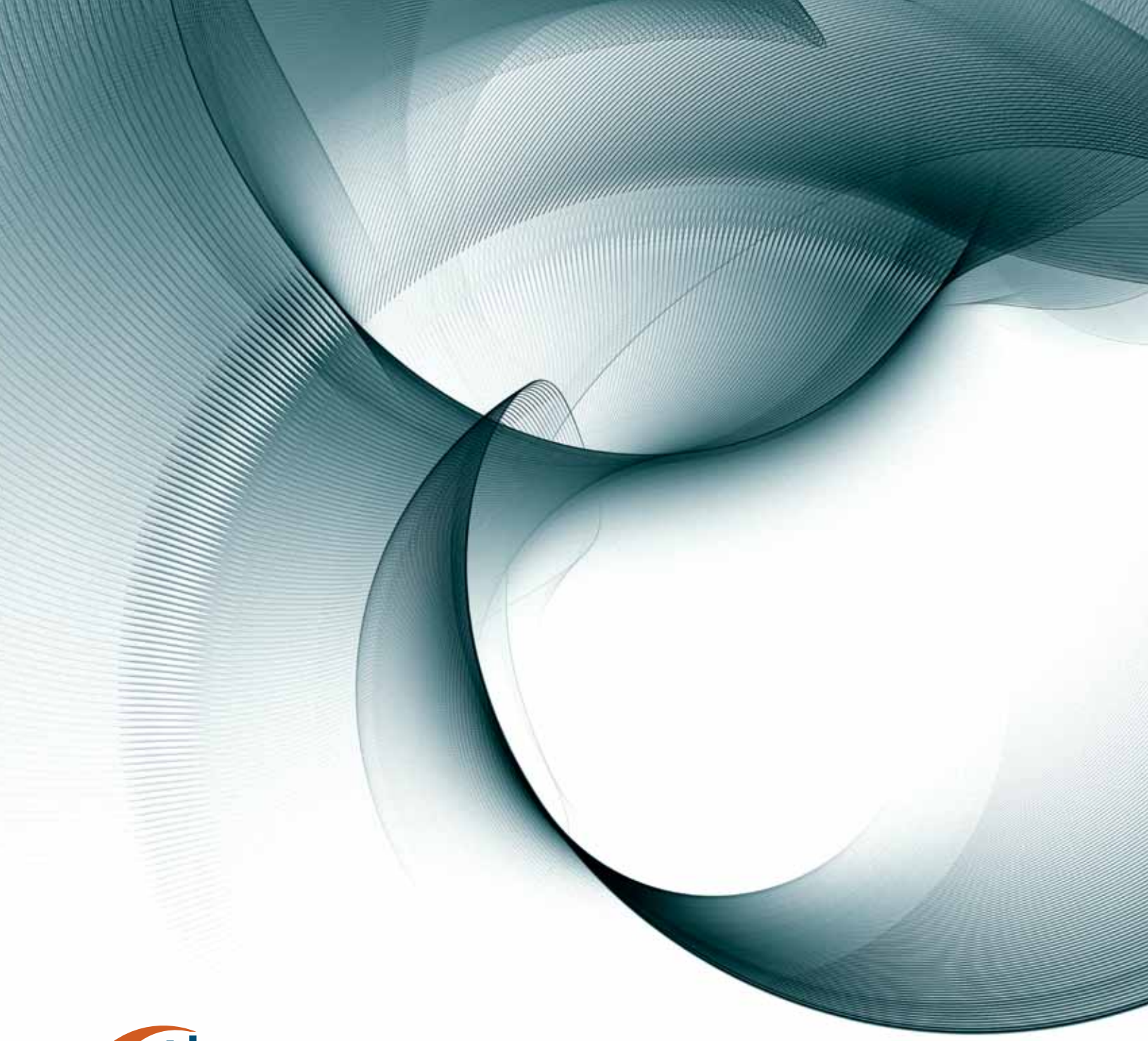

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