



PARTNERSHIP FOR PREVENTION

WASHINGTON, DC



Harnessing Genetics To Prevent Disease & Improve Health

A State Policy Guide

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introduction

For policymakers it is not a question of *if*, but *when* and *how* the advances resulting from the Human Genome Project and other genetic research will be integrated into society, medicine, and public health. State policymakers have the opportunity *now* to:

- Protect consumers.
- Monitor the implications of genetics for health, social, and environmental policy goals.
- Assure genetic advances will be tapped not only to treat medical conditions, but also to prevent disease and improve health before people become ill.

On a daily basis, research generates information about the influence of genes and the genome on human health. This research should spawn new ways to safeguard and improve the health status of individuals and population groups. The great potential of the genomic era is the development of interventions to prevent or better manage costly, common chronic diseases such as cancers, heart disease, and diabetes. Medical interventions could include drugs and preventive measures that are tailored to a person's genetic profile. Beyond the clinical setting, genetic advances should yield new public health strategies to assess health risks, target disease prevention efforts, and eliminate harmful environmental exposures.

This promise can be realized if state policymakers actively monitor genetic advances and carefully consider the implications for health, social, and environmental policy goals. This report will help **governors**, **state legislators**, and other **policymakers** to establish the legal protections, research systems, genetic services, market incentives, and educational programs necessary to harness genetics for improving the health of citizens. **Grantmakers**, both private and public, can contribute by convening diverse stakeholders and informing policy discussions.

DEFINITIONS FOR KEY TERMS

Definitions for key terms — as used in this report — are provided below.

Genetics — the scientific study of genes and their roles in health and disease, physiology, and evolution of human development.¹

Genomics — the science and study of the sequence and functioning of the human genome (i.e., all the genetic material or the complete inheritance of a particular organism).¹

Environment — defined broadly as factors that influence health, including air, water, food, infectious agents, chemicals, and many other substances. Exposures in the human environment are determined in part by behavioral choices, such as tobacco use, alcohol use, diet, degree of physical activity, and sites for recreation and work.² Interactions between genes and the environment are a key health factor.

For definitions of other terms used in this report, see "Glossary" in Appendix A, page 27.



This symbol marks information about the role of grantmakers.

OVERVIEW OF THE HUMAN GENOME PROJECT

The ultimate goal of the Human Genome Project (HGP) is to develop effective approaches for disease prevention, diagnosis, and treatment through a better understanding of the contribution of genes to the development and functioning of the human body.²³ In particular, the HGP will expand knowledge and improve understanding about the interactions among genes and between genes and environmental influences.²⁴ The project is designed to:

- Locate and map all human genes (draft published in 2001^{25,26} and completed in 2003 for the 50th anniversary celebration of the publication of the double-helix structure of DNA^{27,28}).
- Discover the entire DNA sequence of the human genome.
- Store this information in databases accessible to researchers and the public and create resources for data analysis.
- Map and sequence the genomes of other living organisms.
- Study the impact that genetic information and technologies may have on society, including ethical and legal ramifications.

In the fall of 2002, NIH launched an additional program to develop a map of groups of genes (haplotypes) that will link variations in the genome to differences in health and increased risk for common diseases. The “HapMap” could be complete by fall 2005.

By building on the existing foundation of health sciences, the advances resulting from the HGP will lead to new ways to prevent, detect, and manage infectious and chronic diseases.

CDC OFFICE OF GENOMICS AND DISEASE PREVENTION

At the Centers for Disease Control and Prevention (CDC), the Office of Genomics and Disease Prevention (OGDP) works to integrate advances from the Human Genome Project into all aspects of public health research, policy, and programs. The office convenes experts to advise on genetic policy, translates science for public health uses, distributes information, and trains and educates the public health workforce. The OGDP website provides news and information about human genome advances and the likely impact on public health and the prevention of disease. (www.cdc.gov/genomics)

About this Guide



This guide focuses on policy issues related to the harnessing of genetics for disease prevention and health improvement. While directed primarily to state legislators and executive branch officials, it should also be useful to other state officials responsible for incorporating genetics into health policy and practice. In addition, a special section identifies roles for private grantmaking organizations and public funding agencies to participate in the policy development and implementation process.

In brief, the guide:

- 1) Summarizes the top policy issues related to the use of genetic and genomic advances to prevent disease and premature death.
- 2) Offers policy guidance for reaping the benefits of genetic discoveries to advance individual and population health while guarding against the premature use of technologies with untested benefits.
- 3) **Identifies specific policy actions necessary to integrate genetics and associated genetic technologies into broad health policies and programs.** These actions are intended to:
 - Strengthen privacy regulations to safeguard personal genetic information.
 - Assure access to proven genetic services and technologies to improve health in high-risk populations and reduce health disparities.
 - Increase support for applied research and health tracking systems to enhance knowledge of the relationship between genetics, environmental factors, and chronic disease prevention.
 - Address gaps in health care financing that could be a barrier to using genetic advances for disease prevention among at-risk populations.
 - Expand public and professional education to broaden knowledge and understanding of genetics and its potential impact on health and ensure that information about genetic services is readily available.
- 4) Provides helpful information and resources (see appendices).

By focusing on disease prevention and health improvement, this information complements the detailed policy reports regarding other genetic services and technologies available from the National Conference of State Legislatures (NCSL), Association of State and Territorial Health Officials, and others.³⁻²²

Disease prevention and health improvement, as used in this guide, refer to activities that help people avoid illness, injury, and premature death; enhance health; and improve quality of life. Other sources of information should be sought regarding genetics policy related to family law, cloning, stem cell research, genetic selection, or forensic DNA. NCSL’s genetic technology reports cover some of these topics.

Preventing Disease Using Advances in Genetics: Top Policy Issues

Likely Benefits

The Human Genome Project and genetic research will yield powerful information about the causes of common diseases and methods to improve health. For example, genetic advances may expand opportunities for disease prevention through services or practices that:

- Improve disease risk assessment.
- Make earlier, more accurate diagnoses.
- Enhance medical care and public health practice.

Health professionals will increasingly use genetic tests and family histories to assess risk for disease in individual patients, families, and populations. Once clinicians and public health professionals identify increased risk, they can recommend preventive measures such as:

- Increased frequency of preventive health services (e.g., cancer screening, lipid tests) for persons with a specific genetic variation.
- Immunizations of population groups that are particularly susceptible to an infectious disease, perhaps with genetically improved vaccines.
- Customized treatments based on an individual's genes.
- Removal of harmful exposures at home, school, work, and public spaces to reduce the odds of gene-environment interactions that contribute to disease.

To maximize the benefit of genetic advances for disease prevention and health improvement, policymakers will want to integrate genetics into programs and policies that seek to protect individuals and reduce behavioral, environmental, and other health risks.²⁹⁻³¹

Potential Risks

Concerns about inappropriate uses of genetic information may impede the application of genetic discoveries.^{32,33} Specifically, individuals who could benefit from genetic-related services may be reluctant to access such care because of the potential for privacy breaches, stigmatization, and discrimination.

Without laws that protect the confidentiality of personal health information, consumers may be:

- Unwilling to use genetic services, which could weaken consumer demand and subsequently inhibit the development of new genetic services and technologies.
- Less likely to participate in genetic research, which needs adequate numbers of participants to evaluate and assess promising services and technologies.^{18, 33}
- Reluctant to share family history or information from genetic tests, which may diminish health professionals' ability to help patients.

APPLYING GENETICS TO PREVENT CANCERS

Over 1.25 million new cancer cases were diagnosed in 2002, and approximately 550,000 cancer deaths occurred during that year. A small proportion of three cancers — colorectal, breast, and prostate — are known to result from inherited characteristics. Tobacco use, sun exposure, toxins, physical inactivity, and heavy alcohol use also can cause cancer.

Genetics research holds great promise for understanding, treating, and even preventing the many forms of cancer. Genetic tests are now available for some genes associated with increased risk for breast, colorectal, and ovarian cancers.

The Centers for Disease Control and Prevention (CDC) is developing tools to assess the benefits and risks of existing genetic tests for cancers by examining availability, quality, and usefulness. The results of these assessments will inform future recommendations about cancer-gene testing in the general population.

In the future, state health agencies could provide guidance to health systems about criteria to help health professionals make decisions about referring patients for genetic counseling. Websites underwritten by state and private funds might provide consumers with information about genetic tests, including clinical utility and validity.^{44,45}

Conversely, policies that overly protect individual interests may hinder the attainment of public or societal goals,^{6,34} such as those outlined below.

- Public health agencies may be unable to respond to changes in community health if laws prevent the use of personal genetic information in health tracking systems, such as cancer registries or surveillance systems for birth defects or infectious diseases.
- Employers and regulators may be unable to identify and remove low-level, harmful occupational exposures without the ability to monitor gene-environment interactions.
- Insurers may be less willing to offer affordable insurance in the individual or small employer market if high-risk individuals are able to choose generous coverage while keeping information about their high risk from the insurer (sometimes called *adverse selection*). This scenario may lead to unnecessarily high premiums if, in order to meet obligations to policyholders, insurers raise premiums because they cannot adequately assess risk in the underwriting process.

In brief, genetics policy must find **an acceptable balance between providing strong protection for individuals' interests while enabling society to benefit from the genomic era.**

Genes, Health Behaviors, and the Environment

Policymakers must guard against the perception that genes alone determine present or future health status.³¹ Only a very small segment of a state's disease burden can be traced to single-gene disorders (e.g., inherited diseases such as phenylketonuria (PKU) and Huntington's disease). In contrast, **the majority of diseases** — including infectious diseases and chronic diseases — **result from gene-gene or gene-environment interactions.**

Environmental factors that affect health encompass everything from the food, water, and drugs people consume to the air they breathe and their levels of physical exercise. Toxins and pathogens in the external environment have a profound and cumulative effect upon human health.³⁶ In fact, long before the mapping of the human genome began, researchers identified tobacco use, unhealthy diet, physical inactivity, excessive alcohol use, infections, and trauma as the underlying causes of preventable death and illness.³⁷ Research has also connected many health disparities to environmental exposures and socio-economic factors rather than genetic inheritance.

Genetics should eventually enable health professionals to target disease prevention/health improvement interventions to people who will benefit most while avoiding adverse effects of testing and treatments in individuals with less risk. However, attaining these benefits requires additional study of how environmental factors influence the development of diseases with a genetic component. Because of the complexity of sorting out gene-gene and gene-environment interactions, researchers may need years or decades to develop applications from genetic sciences for preventing or treating many health conditions.^{20,31}

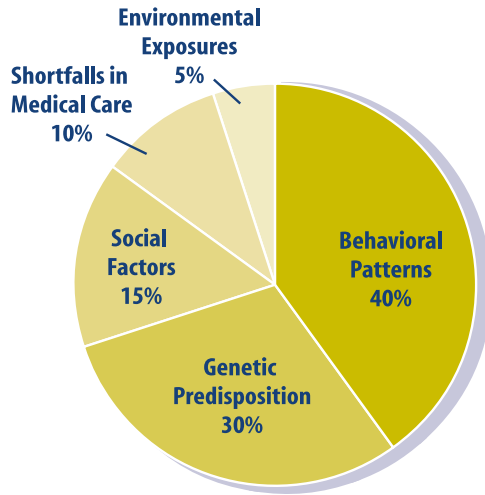
In the short-term, **policymakers already have the means to significantly curtail premature death and avoidable disability by investing in interventions recommended by the Task Force on Community Preventive Services** (www.thecommunityguide.org). Currently, many of these high-impact disease prevention opportunities have yet to be fully exploited.³⁸⁻⁴⁰ In the long-term, the integration of genetics into health policy and practice will maximize the benefits of the genome era for disease prevention, especially when combined with use of other proven preventive services.

“The more science learns about genes, the more we will learn about the importance of the environment.”⁴¹

FIGURE 1

Leading Causes of Premature Deaths

A leading public health expert compiled multiple studies to estimate percentages of premature deaths attributable to various factors.



Source: Health Affairs 2002;21(2):78-9342

WHAT IS PREVENTION?

Prevention encompasses a host of activities to help people avoid illness, injury, and premature death. Some preventive measures result from **health, social, and economic policies**. Examples are laws that seek to deter drunk driving; requirements to fortify foods with needed vitamins and minerals; regulations to improve air and water quality; and employment policies to prevent worksite injuries or reduce toxic exposures.

Other measures are **clinical preventive services**, such as immunizations, the use of cholesterol-lowering drugs, and periodic health screenings. These services protect against diseases or help detect diseases at an early stage when treatment can be most effective and less expensive than for late-stage diseases.

A third type is **community-based interventions**. Health education campaigns to encourage people to switch to low-fat milk, wear seat belts, or quit smoking are preventive measures. Other community-based interventions are programs that help to reduce violence and abuse, track infectious diseases like HIV/AIDS, or improve employee health and productivity with worksite-based services.³⁵

FIGURE 2

U.S. Deaths by Disease or Disorder

Chronic and Infectious Diseases (2000)

Diseases of the Heart	710,760
Malignant Neoplasms (cancers)	553,091
Diabetes Mellitus	69,301
HIV	14,478
Pneumonia	3,548

Genetic Diseases

Sickle Cell Anemia (1999)	498
Cystic Fibrosis (2001)	409

This chart illustrates that common chronic and infectious diseases are a greater source of mortality than genetic diseases. Although relatively uncommon, deaths related to genetic disease often occur at a young age, creating substantial losses for families and communities.

Sources: National Vital Statistics Report Vol. 50, No. 16., Sept. 16, 2002. Cystic Fibrosis Foundation Patient Registry: Annual Data Report 2001. National Heart Lung and Blood Institute Morbidity and Mortality Chartbook 2002.¹⁰⁰

Rationale for State Policy Intervention and Oversight

HOW DOES HIPAA AFFECT STATE GENETICS POLICY?

The federal Health Insurance Portability and Accountability Act (HIPAA) preamble specifies that protected health information includes genetic information. HIPAA sets a floor in protecting the privacy of personal health information; thus, states may establish laws that are more stringent than the HIPAA privacy provisions (but not less stringent). Further, HIPAA does *not* preempt state laws governing:

- The collection and analysis of public health and vital statistics, which often are based on personal health information.
- Health plan regulation and monitoring, which may require review of individual medical records.
- Standards for health research that protect the privacy of personal health information while enabling health researchers access to medical records for studies that meet specific requirements.

HIPAA regulations:

- Forbid discrimination on the basis of an individual's health status or his/her medical and genetic information.
- Prohibit group health plans and health insurance issuers from:
 - denying coverage based on the presence of a genetic marker without the diagnosis of illness associated with that genetic information; and
 - altering eligibility requirements or premiums for individuals in a group on the basis of their genetic status.

The regulatory provisions above would preempt state laws that are contrary, unless states obtain authorization from the Secretary of Health and Human Services.

The full range of practical applications from the Human Genome Project will not emerge for many years, perhaps decades.⁴³ Nonetheless, the first wave of genetic tests for chronic diseases is entering the commercial market. For this reason, **public policies need to be put in place in the near term** that protect the privacy and confidentiality of genetic information, ensure appropriate use of genetic services and technologies, and encourage the integration of genetics in health, environmental, and social policy.

Figure 3 and the maps in Figures 4-7 show that many states have initiated policies to capture benefits and minimize problems that could emerge from genetic advances. In particular, states have actively sought to **prevent misuse of personal genetic information by enacting privacy and confidentiality standards**. These standards often give consumers greater protection than provided in the federal Health Insurance Portability and Accountability Act (HIPAA) (see the “How Does HIPAA Affect State Genetics Policy?” sidebar). As genetic services and technologies become widespread, privacy policies warrant regular review to minimize unintended consequences such as incomplete medical treatment or adverse selection in health insurance.⁶ Policymakers may need to review privacy policies regarding access to individual genetic information that could be useful to family members.

An important first step for states is to begin **integrating genetic advances into programs and policies focused on preventing common and costly chronic diseases** such as cancers, heart disease, asthma, and diabetes.⁴⁴ New genetic findings also can be incorporated into state infectious disease control and environmental health programs.

Second-stage steps would address more challenging policy issues. Few states are debating how they might **assure that beneficial genetic services and technologies are available, affordable, and accessible**. The free market will supply a wide range of these services and technologies, but state policymakers will need to decide how to fill gaps where the commercial sector lacks sufficient incentives. Expanding access to genetic services could build on current public policies and efforts that address market gaps, such as:

- Programs to screen for preventable disorders.
- Financing to provide HIV/AIDS treatment or prenatal care to the uninsured.
- Loans to attract health professionals to medically underserved areas.
- Toll-free quitlines to help state residents curb tobacco use.
- Laws that require health plans to cover preventive services such as mammography (note: these mandates do not apply to self-insured plans).

Policymakers will need to deliberate how best to address gaps in health insurance coverage, promote the provision of genetic services in medically underserved areas, and minimize socio-cultural barriers that may impede access to genetic services and technologies.

A third timely action would be to review, and revise as needed, state licensing examinations to include competencies in genetics. The goal would be to **assure that consumers receive high-quality care and accurate information from health professionals**. State policymakers could also review the process — or establish one if needed — that **assures a systematic evaluation of genetic tests** before including them for payment in state-financed health programs.

Figure 3 highlights how some states are integrating genetics into health, social, and environmental policy realms to meet the demands of a rapidly expanding science.

FIGURE 3**Examples of Current State Genetics Policies and Programs**

POLICY REALM	PROGRAMS AND POLICIES ³⁻⁵
Insurance	Majority of states prohibit insurers from requiring genetic testing or using genetic information to deny health insurance (see Figure 4).
Employment	Many states prohibit genetic discrimination in hiring and retaining employees (see Figure 5).
Research	<p>Many state-supported institutions conduct genetic discovery studies, usually with federal grants. Some universities are conducting genetics policy research.</p> <p>At least one state (MI) is investing in life science research (which includes genetics) as a part of an <i>economic development strategy</i> to attract and retain life sciences industries to the state.</p>
Training	State-supported universities are beginning to incorporate genetics competencies into health professional training.
Public & Professional Education	<p>With support from CDC Office of Genomics and Disease Prevention, state chronic disease programs are developing plans to educate health professionals about the use of genetics to strengthen disease prevention.</p> <p>Some states are expanding their public school (K-12) genetics curriculum.</p>
Consumer Protection	<p>State regulations require informed consent for specified medical procedures and for participation in research.</p> <p>States also have laws assuring the privacy of personal health and genetic information (see Figure 7).</p>
Civil Rights	Some states include genetics as a protected class in anti-discrimination laws.
Public Health Services	<p>All states offer newborn screening for genetic and other abnormalities, and many require it. Most states provide pre- and post-test counseling.</p> <p>State laboratories analyze newborn and other genetic tests.</p> <p>State health agencies are incorporating genetic and genomic science into chronic disease prevention and surveillance programs.^{44,45}</p>
Licensure and Accreditation	State laws establish procedures for licensing health professionals and health care organizations. State governments select members of boards that determine criteria, such as competencies in genetics, for licensure and accreditation.
Environmental Protection	One state (NY) uses molecular epidemiology and tracks biomarkers to probe gene-environment interactions in the development of health conditions, including those associated with the clean up of the World Trade Center. ⁴⁶
Other	States have updated family, criminal, and civil law to incorporate advances in genetics. ⁴⁴

APPLYING GENETICS TO PREVENT OVERWEIGHT/OBESITY

Among U.S. adults, over 60% are overweight or obese. Health behaviors and gene-environment interactions contribute to this multifactorial chronic condition. Excess weight is much more than a cosmetic issue: almost 80% of obese adults have diabetes, high blood pressure, heart disease, high blood cholesterol levels, and/or osteoarthritis.

Current research indicates that multiple genes appear to influence the intake of foods and the expenditure of energy. Large, long-term studies are needed to better understand overweight/obesity and to translate genetic research into treatment and prevention strategies.

In the future, state health agencies could conduct public education campaigns with the message that even persons with genetic risks for obesity can achieve and maintain a healthy weight.^{44,45,67}

Grantmakers' Role in Independent Policy Guidance



Grantmakers in the fields of health and medicine can help translate genetic research and technologies into sound public policy. Historically, these organizations have been particularly **effective at convening diverse groups of stakeholders to analyze and recommend policy change**. The diversity and expertise of their boards of directors, staff, and grantees enable grantmakers to **provide leadership and input that spans the public and private sectors**. By convening and providing leadership, grantmakers can help stakeholders and experts advise on technical issues or build consensus about complex genetics policies.

Many grantmakers are viewed as trustworthy sources of health-related information.¹⁸ By **funding independent policy analyses**, grantmakers can help clarify issues related to genetics and **build support for policy goals**.

Private grantmaking organizations and public funding agencies can **support the state genetics policy process** in additional ways.

- Disseminate genetic and genomic research findings in formats tailored for special populations (e.g., policymakers, clinicians).
- Conduct or sponsor public education campaigns that are culturally and linguistically appropriate.
- Conduct or sponsor community-wide discussions or conferences regarding the implications of genetic research for special populations and general audiences.
- Assist state policymakers with identifying needed policy changes, analyzing policy alternatives, and evaluating outcomes.
- Fund research and demonstration projects to translate genetic developments into disease prevention services, especially for diverse populations where disparities in health outcomes may be substantial.
- Sponsor interdisciplinary training in genetics for health professionals, especially on topics – such as family histories – for which commercial sponsors are few or nonexistent.
- Facilitate dialogues between health researchers and the media and sponsor training programs for reporters to ensure consumers receive accurate, useful updates on genetic developments with balanced coverage of potential harms and benefits.

According to chair Kathryn Malvern, the Zeta Phi Beta National Education Foundation's educational forums let minority communities "know that there is another side to the genetics equation – that not all [diseases] are genetic [in origin], that it has to do with genetics or the environment, or genetics and the environment. Minority and underserved communities should be concerned about genetic screening, in part, because research shows that people of color are more likely to be exposed to hazardous agents in their neighborhoods and work places."⁴⁷

The States' Role in Balancing Opportunities and Risks Associated with Genetics

Genetic advances feed hopes for new ways to extend life and improve health while also arousing fears about the privacy of health information and potential for discrimination. Some of these fears may stem from misuses of genetic information in the 20th century that violated now-accepted principles of bioethics (see the “Historic Misuses of Genetics” sidebar on page 10).

To harvest the benefits of genetics research, balance individual rights and societal needs,^{48, 49} and discourage misuse of genetic information, state leaders can:

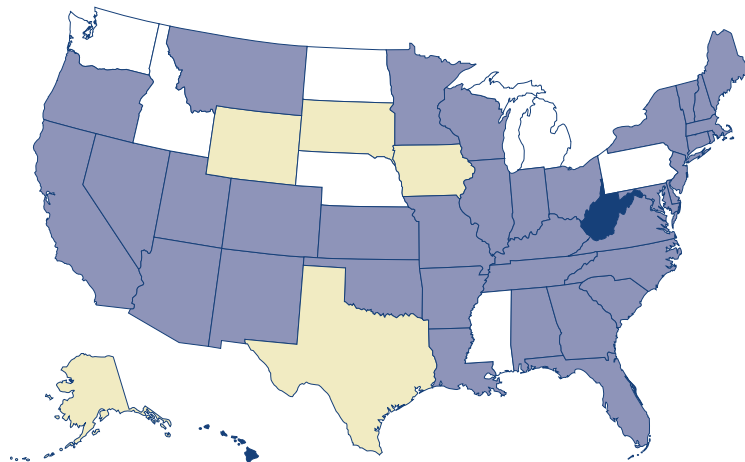
- **Increase their understanding of genetics** by learning more about the relationship between genes, environmental factors, and health in order to make science-based decisions.
- **Involve an array of stakeholders, especially consumers, in policy development.** Broad stakeholder involvement reduces the odds for unintended, negative consequences resulting from policy decisions. Advisory groups of stakeholders and experts can monitor the short- and long-term effects associated with genetic policy; this information can inform future policy changes.
- **Assign responsibility** (such as to the state health agency or an advisory group) **for establishing criteria and processes to distinguish beneficial genetic tests from tests lacking documented utility.** State policymakers can use the criteria — such as test validity and the availability of treatment — to evaluate benefits, risks, and costs. (See policy principle 2, page 15 and “Population Screening Using Genetic Tests,” page 14.)
- **Provide opportunities for the public to increase knowledge and broaden understanding of genetics through educational programs and community-based discussions.** State institutions and agencies can offer formal instruction and sponsor educational campaigns for a variety of audiences.

“The learning curve for legislators and the public is great...but people need to know what genetic testing means and what the options are,” says Washington Senator Rosa Franklin, a former nurse, speaking about the importance of the findings from the Human Genome Project.⁷

FIGURE 4

State Genetic Laws Assuring Nondiscrimination in Health Insurance

KEY:  No state laws  State law(s) applies to all insurers
 Applies to group policies only  Applies to individual policies only



Source: NCSL Genetic Technologies Project⁴⁹

Note: As of August 7, 2002, these states have laws that prohibit insurers from instituting eligibility rules based on genetic information.

HISTORIC MISUSES OF GENETICS

Current health and medical ethics have been shaped by both prior misuse of science and unfounded “scientific” beliefs that harmed vulnerable populations. Memories of these wrongdoings remain strong and influence policymaking today.

Eugenics was one of those misuses. In the late 19th to the mid-20th centuries, the eugenics movement promoted genetic selection to encourage reproduction among people with “good” genes while seeking to restrict births in certain immigrant and racial groups and persons deemed “unfit.” Initially, eugenics had the cover of science, but scientists later discovered that many of the “undesirable” traits targeted by the movement were due to environmental influences, not genetic inheritance. Beyond its scientific flaws, eugenics violated basic human rights.

Many groups are wary about genetic testing because of a history of public institutions not protecting the health and safety of all people and not involving the public in policy or program planning. For example, in the latter decades of the 20th century, sickle cell carrier screening was conducted without sufficient education and involvement of communities of color. Screening resulted in discrimination and stigmatization as well as widespread confusion within and outside targeted populations about the disease versus the harmless sickle cell trait. As a result, some people fear future applications of genetics will disadvantage demographic groups that share — or are believed to share — common genetic traits.

Only in the 1970s did the concept of *protection of human subjects* become codified in federal regulations. These regulations established the practice of informed consent and the use of research review boards to discourage unethical practices.

There is still the risk of misusing genetics and further marginalizing population groups. Policymakers, researchers, and health professionals can turn to ethics experts in universities for guidance. Schools of public health can assist by involving diverse communities in research planning and policy debates — another essential way to address fears and avert misuses.² Grantmakers can



play a role by funding university ethics centers, community-based research and policy debates, and educational programs.

For additional information on eugenics, see the Cold Spring Harbor Laboratory/National Human Genome Research Institute website (www.eugenicsarchive.org/eugenics/).

- **Monitor potentially beneficial and harmful uses of genetic information,** especially by accessing national resources such as the CDC Office of Genomics and Disease Prevention. Documentation of actual uses of genetic information will enable policymakers to disentangle myths and fears from reality.³³ This information should be used to evaluate proposed or existing health policies.

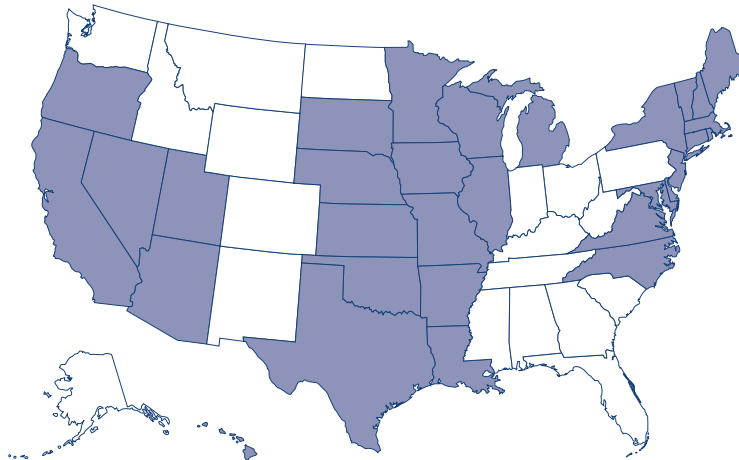
- › The media has reported cases of loss of employment and insurance coverage based on genetic test results, but the actual extent of and risk for such events are poorly documented and should be a component of a state’s policy research agenda.
- › Insurers are concerned about individuals purchasing generous insurance coverage because they know they have, or are at risk for, a genetic condition, while those who test negative for severe conditions minimize their insurance coverage. Known as *adverse selection* or *reverse discrimination*, such use of genetic information can undermine insurers’ attempts to spread risk over the entire insured population and thereby maintain affordable rates.^{3,50} Working with state insurance commissioners, policymakers should seek evidence whether adverse selection is occurring or, conversely, whether insurers are inflating premiums on the assumption of adverse selection.
- › Biotechnology industry and consumer groups may advocate for coverage of new genetic services and technologies in state-financed health programs and state requirements for health plans. To substantiate purported benefits, policymakers will want data from randomized, controlled clinical trials that document the utility and limitations of the proposed service or test.



To help policymakers make informed decisions and balance individual rights with societal needs, grantmakers can sponsor educational briefings at the state capitol that enable policymakers to interact with experts in genetics, population health, clinical care, law, and ethics. They also can convene stakeholders to make policy recommendations and fund independent analyses of policy proposals.

FIGURE 5

State Genetic Laws Assuring Nondiscrimination in Employment



Source: NCSL Genetic Technologies Project⁵²

Note: As of February 3, 2003, the shaded states have laws that proscribe genetic discrimination in employment or dismissal and/or employment terms, conditions, or privileges.

Policy Foundation: Integration or Exceptionalism

Before formulating new genetics policies, state policymakers and their advisors will need to decide on a philosophical perspective. Policy strategies will differ greatly based on whether the decision is for integration, exceptionalism, or a blend of the two perspectives.

Exceptionalism views genetic information as distinct from — and more sensitive than — other personal health information; moreover, exceptionalism considers genetic information as so closely associated with personal, family, and group identity that its misuse could affect life opportunities in severe and singular ways. Thus, genetic issues are considered so exceptional that they merit distinctive policy treatment. Policies based on exceptionalism give genetic information special status and protection, distinct from other health information, especially in privacy and anti-discrimination laws. In a similar way, many state laws treat information about HIV status differently from information about other infectious diseases (e.g., pneumonia).

In contrast, the **integration perspective views genetic information as so integral to health that it cannot be separated from other health information.** This perspective promotes the integration of genetics into existing health policies by strengthening laws addressing privacy and confidentiality, scientific research, insurance and employment regulation, public health programs, and medical practice. For example, Minnesota and Washington include genetic information in existing privacy statutes, an approach that is primarily one of integration. (The federal HIPAA (see page 6) also applies an integration approach.)

A few states have adopted a **blended perspective** (sometimes called *limited exceptionalism*) that provides special treatment only for some types or uses of genetic information.⁵ For more discussion of exceptionalism and integration, see the NCSL publications *Genetics Policy and Law* and *Genetics Policy Report: Privacy*.^{5,6}

The Case for Integration

In the past, political realities may have necessitated genetics policies based on exceptionalism.⁴ However, laws recently enacted in Michigan, Nebraska, South Dakota, and Washington signal a switch to an integration or blended approach.

This report encourages integration of genetics into health, environment, and social policy and programs — rather than separation — for several reasons.

First, a growing body of knowledge supports the premise that all health conditions (with the exception of trauma) have a genetic basis.¹ Thus, genetic information is so integral to health and medicine that laws separating genetics from other personal data may not succeed.^{30, 55}

Second, because the most common, and costly, diseases result from gene-environment interactions, **genetic advances are likely to extend and expand — not supplant — current practices in medicine, public health, environmental protection, and other disciplines.**

Third, the sensitivity of genetic information depends on how much it conveys about a person's current or future health. Some genetic variations may be associated with greater health risks than others. Developing effective policy to cover this wide range of sensitivity is difficult especially if an exceptionalism approach is used.

MICHIGAN AND MINNESOTA APPLY THE INTEGRATION APPROACH

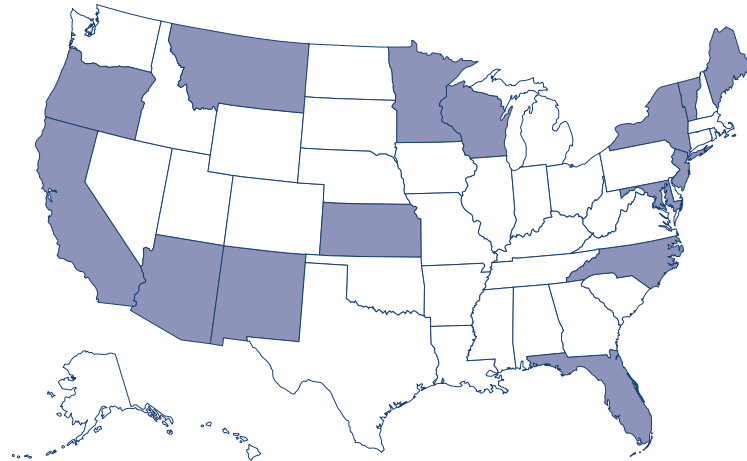
At a time when many state policies were based on exceptionalism, the Michigan Governor's Commission on Genetic Privacy and Progress adopted an integration perspective and recommended that genetic issues be dealt with in the context of overall medical care values and principles.⁶⁰

Currently, Michigan law reflects a blended approach. Administering a genetic test requires written informed consent from patients, whereas other medical tests do not. Genetic information, including test results, is no more or less private and confidential than any other medical or health information.⁶¹ In the area of employment, Michigan law specifically prohibits companies from discriminating against applicants or employees because of genetic information that is unrelated to job duties.

A future-oriented Minnesota law, in effect since 1983, does not distinguish between genetic and other health information for employment decisions. The law enables employers to use information that is job-related but bars access to all non-job-related medical information. By not specifying *genetic information*, the law avoids the problem of defining this term. It also permits employers to provide workers with essential protections (e.g., monitoring the health effects of environmental exposures, assessing candidates' medical fitness for job responsibilities, and implementing programs to reduce occupational injuries and disability).

FIGURE 6

State Genetic Laws Assuring Nondiscrimination in Life Insurance



Source: NCSL Genetic Technologies Project⁴³
Note: As of October 14, 2002, the shaded states have laws that limit how life insurers may use genetic information.

SUGGESTIONS FOR DEFINING GENETIC INFORMATION

For the term *genetic information*, the American College of Medical Genetics recommends definitions be sufficiently:

- Broad to accommodate single-gene disorders and the contribution of multiple genes to common, complex diseases.
- Flexible to avoid becoming rapidly outdated by new developments.
- Narrow and clear to avoid confusion in application of statutory protection in the current system of health delivery.

The college further advises against arbitrary distinctions that unfairly exclude some individuals from protections afforded others.¹³

An integration approach may avoid the problem of defining genetic information if current definitions of health information apply to genetic information.

Fourth, integration offers the best opportunity to advance the science and practice of disease prevention and health improvement.^{8, 16, 44, 55-59} For individuals, integration should improve disease diagnoses or risk assessments for future disease. For families and society, integration should enable public health agencies to track and modify behavioral, environmental, and socioeconomic factors associated with common and preventable health conditions.^{2, 56}

Finally, the difficulties associated with using the exceptionalism approach are multiple and serious.

- It is difficult to develop a precise legal definition of *genetic information* that distinguishes it from other health information and does not obstruct legitimate uses (e.g., for medical treatment, test results commonly used in the insurance underwriting process, or occupational safety policies that protect all workers).^{3, 4, 6, 55}
- Exceptionalism may inadvertently provide greater legal protection to those with genetic disorders than to those with equally serious diseases attributable to other causes.
- Exceptionalism may inadvertently stigmatize people with genetic diseases by setting them apart as a distinct group.
- By restricting uses of genetic information, exceptionalism could delay timely use of beneficial genetic services and technologies.

In recognizing that all health conditions have a genetic component, the integration approach sidesteps these problems.

Examples of How Integration Would Work

Integration will allow private health systems and public health agencies to use genetics and genomics to prevent disease and improve health by:

- Strengthening chronic disease programs (e.g., provide consumers with nondirective information about genetic tests relevant to specific chronic diseases).
- Using genetic and other health research to guide the allocation of resources to programs most likely to benefit groups who have high risk of disease.

- Expanding laboratory capacity for analyzing new genetic tests and expeditious reporting of results to health professionals and patients.
- Updating health-tracking systems to capture genetic information that can be used for health planning.
- Assuring that all genetic tests and services include informed consent and are accompanied by culturally proficient patient education and genetic counseling.
- Requiring licensed or certified health professionals to receive training in applications of genetics for disease prevention and health improvement.

Genetics can be integrated into other state (and local) government activities.

Education (K-12) – Add questions to standardized tests to assess students’ understanding of genetics. Train health and science educators to incorporate genetics into curricula.

Occupational health/safety – Advise employers about ways to lawfully monitor and protect employees who have genetic predispositions for diseases associated with worksite exposures.

Environmental health – Reduce common environmental exposures known to have harmful gene-environment interactions.

Consumer protection – Ensure current laws protect consumers from misleading marketing and fraudulent practices related to genetic services.

Insurance – Assure that affordable insurance is available for those with genetic disorders and other serious health conditions and that reverse discrimination or adverse selection (see pages 4 and 10) are minimized.



Grantmakers can support the integration of genetics into health, environmental, and social policies. For example, they can sponsor:

- Efforts to develop and disseminate educational materials for diverse populations groups.
- Independent assessments of employers’ use of genetic information to advance occupational health.
- Educational seminars for health professionals, social service professionals, as well as policymakers.
- Projects to identify and promote protocols that protect the privacy and confidentiality of personal health information – including genetic data.

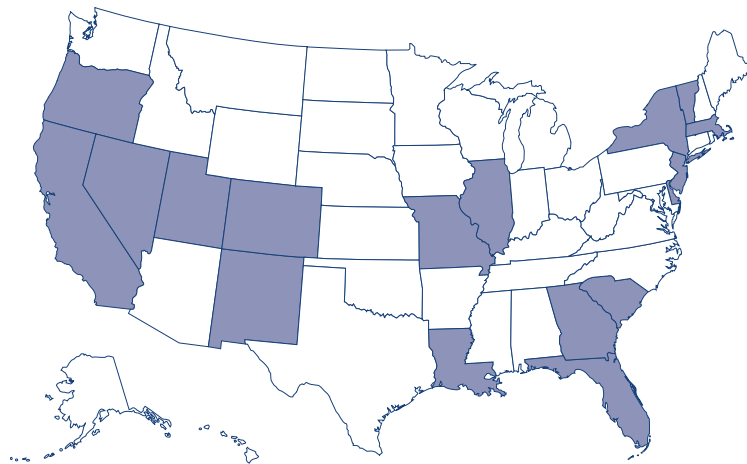
EXPERTS IDENTIFY DIFFICULTIES WITH EXCEPTIONALISM

Following extensive testimony and long deliberation, the Taskforce on Genetics and Insurance — a subcommittee of the Human Genome Project Advisory Committee on Ethics, Legal, and Social Issues — determined that “treating genetic information differently [from other health and medical data] is conceptually confused, practically infeasible, and ethically indefensible.”

In terms of health care coverage, the task force concluded that genetic information did not differ substantially from other kinds of health-related information.⁶²

FIGURE 7

State Laws Imposing Penalties for Violations of Genetic Privacy



Source: NCSL Genetic Technologies Project⁶⁴

Note: As of April 15, 2002, the shaded states have laws that impose specific penalties for violations of genetic privacy.

Population Screening Using Genetic Tests

DEFINITION OF GENETIC TESTS, SCREENING, AND STANDARDS

Genetic tests analyze human DNA, RNA, chromosomes, proteins, and certain metabolites to detect the presence or absence of inherited or acquired genes or genetic markers. Such tests are frequently conducted on blood samples, but other body fluids or tissues may also be used. Types of genetics tests include diagnostic, predictive, pharmacogenetic, newborn screening, carrier, prenatal, and forensic identification (i.e., a DNA “fingerprint”). Genetic tests should meet the same standards as any other medical test. Randomized, controlled clinical trials provide the best data on test harms and benefits.

Genetic screening involves the use of a test to evaluate asymptomatic groups of individuals or populations for specific genes or genetic markers.

Before deciding to use genetic tests for population screening, many health professionals seek information about the reliability, utility, and validity of a test.

Reliability is a test’s ability to repeatedly produce the same result.

Clinical utility refers to the usefulness of the test and value of the information to the person being tested for seeking effective treatment or a preventive strategy. The utility of a given test will change as technology and treatments change.

Analytical validity is an indicator of how well a test measures the property or characteristic it is intended to measure. Does it do what its makers claim it does?

Clinical validity refers to the accuracy of the test in diagnosing or predicting the presence or absence of risk for a health condition. Does the test successfully detect disease or predisposition for disease?

Specificity, sensitivity, and positive predictive value are additional criteria used to assess the benefits and harms of genetic tests.²³

Population screening involves testing large numbers of people for the presence of a disease, disability, or genetic variation. Such screening should be done only with sufficient evidence that positive health outcomes will benefit those tested.

Since the late 1960s states have supported population-based newborn screening programs. These programs enable early detection and treatment for costly and often devastating genetic diseases that appear early in life. A substantial body of research informed the development of these screening programs. The effectiveness of these programs in reducing the burden of PKU and other health conditions in newborns is a direct result of the availability of diagnostic tests plus treatments that are effective, safe, relatively inexpensive, and acceptable to most parents.

Future population screening programs will be more complex than ones for newborn screening. Many genetic tests in development or now entering the market are for health conditions, such as cancers, that usually appear in adulthood when testing is most appropriate. While genetic testing for chronic diseases may assist in risk assessment, environmental exposures — which often have a greater influence than genes on the development of diseases⁶⁸ — also must be taken into account.

Public health experts recommend policymakers follow the principles below when considering whether genetic tests should be available on a population-wide basis.^{11, 69, 70}

- The disease or health condition is an important public health problem.
- Scientific data document the occurrence of the genetic trait and the burden of disease associated with that trait.
- The natural history of the disease is understood.
- Research establishes the safety, effectiveness, and predictive value of the test.
- The target population (i.e., specific demographic groups) finds the screening to be acceptable.
- Screening, counseling, follow-up, and effective interventions are available, accessible, and safe.
- Safeguards assure informed consent and voluntary testing, protect the privacy of individuals who are tested, and deter stigmatization and discrimination.
- The benefits of screening outweigh the harms, and the economic costs are in balance with the benefits.

Currently, over 900 genetic tests are available, but genetic counseling is often the only intervention available for persons found to have a genetic trait.^{71, 72} As a result, it may be many years before effective population screening using genetic tests, other than those for newborns, can be put into wide use to improve the health of population groups.

The policy implications of screening for and preventing diseases based on genetic tests and information are complex. They will require careful study, consultation, and ongoing policy review. (For tips on this process, see “Genetics Policy Development Framework,” pages 18 and 31.)

Policy Principles for Disease Prevention in the Genomic Era

Whether following an integration, exceptionalism, or blended approach, state policymakers can use the genetics policy principles offered below. The principles are inter-related, providing a balance between individual and societal interests while realizing the goal of tapping genetic and genomic advances for disease prevention and health improvement. Policymakers can tailor the principles for their states or use them as a checklist during policy formation. (Appendix D, “Actions for Policymakers,” illustrates how these principles can aid policy development.)

1. Utilize genetic advances to reduce the burden of disease and decrease disparities in health outcomes.

As genetic research on diseases and potential treatments accelerates, public policy and scarce public dollars should be directed to reduce the prevalence of those diseases — such as cancers, heart disease, diabetes, asthma — that place a heavy burden on individuals and society (see Figure 2, page 5). To do so, policymakers will need information to assess:

- Who is in greatest need.
- What help should be given.
- What approach will provide the greatest public good.

Integrating genetics and genomics into state chronic disease prevention programs — already being initiated in several states — is one strategy likely to yield beneficial results at a reasonable cost.

2. Limit publicly funded programs to genetic services and technologies that scientific evidence shows to be *safe, reliable, and beneficial*.

Criteria for investing public resources should include the utility, validity, and reliability of tests (see “Genetic Tests” sidebar on page 14) as well as the availability of effective treatments or preventive measures. Scientific data, analyzed by public health and medical experts, should be used to compare the costs, benefits, and harms of alternative options to protect and improve health. (For example, medical family histories may provide as much or greater insight into future health risks than a series of genetic tests.⁷³) Analyses using these criteria can help policymakers, taxpayers, and interest groups understand the potential impact of incorporating genetics into health policy and practice. (For information about federal regulation of genetic tests, see Appendix B.)

3. Safeguard individuals and communities from potential harm by protecting health privacy and assuring the use of informed consent and the provision of quality counseling.

Preventing the inappropriate use of genetic or any other personal health information will help avert stigmatization and discrimination. Existing laws and regulations may already provide sufficient privacy protections; if not, policy revisions may be needed.

Requiring informed consent and allowing for informed refusal (see Appendix E) helps assure the privacy and confidentiality of personal genetic information and promotes personal autonomy in genetic testing. Informed consent is not required for most newborn screening tests.

APPLYING GENETICS TO PREVENT ASTHMA

About 15 million people in the United States, many of them children, suffer from asthma. Each year asthma accounts for about 1.5 million emergency room visits, 350,000 hospitalizations, and over 5,000 deaths.

This complicated disease results from the interaction of genetic, environmental, and behavioral factors. Genetics research should yield insights into the onset of the disease, the biological processes that trigger asthma attacks, and opportunities to prevent or control asthma. For example, studies of genes and environmental triggers could identify the need for environmental policies that further reduce air pollutants.

In the future, state health agencies could promote the use of clinical guidelines that integrate knowledge about gene-environment interactions associated with asthma onset and asthma attacks.^{44, 45, 63, 64}

GRANTMAKERS ENGAGE MINORITY COMMUNITIES IN GENETICS

In 1998, the Zeta Phi Beta Sorority, through its National Education Foundation, began building awareness of the Human Genome Project among minority communities. The foundation held conferences around the country to provide a forum for minority communities to learn about genetics and develop policy recommendations.

With support from U.S. Department of Energy and other federal and private funders, the foundation planned four large conferences. One conference brought together Pennsylvania legislators and community representatives to develop genetics policy recommendations for the Pennsylvania Black Caucus. In collaboration with the Consumer Health Foundation and Howard University, another conference facilitated a dialogue between minority communities and expert presenters. Participants discussed ownership of genetic information, ways to discourage genetic discrimination, confidentiality of medical records, minority involvement in clinical trials, and outreach to encourage minority students to pursue studies in genetics and related fields. Zeta Phi Beta compiled recommendations into a report to inform national legislation.

To expand its programmatic reach, the foundation awarded mini-grants to Zeta chapters for one-day informational forums in local minority communities throughout the country. Future endeavors may continue to make the case for minority involvement in genetic policymaking and research.^{47,75}

Further, genetic education and counseling by trained health professionals can help individual patients and families make informed choices by assuring they understand the benefits and risks of genetic testing and the implications of test results. (The American Board of Medical Genetics and the American Board of Genetic Counseling certify genetic counselors.) Educational materials and counseling interventions must be linguistically appropriate and culturally proficient for diverse populations.

4. Reduce health disparities by directing publicly financed genetic services to populations most at risk or those with the greatest need.

Not all individuals and communities benefit equally from medical and public health services, including advances in genetics. It is well documented that specific population groups in the United States persistently bear a disproportionate share of preventable health conditions and have a shorter lifespan than other population groups.⁷⁴ These disparities could be further exacerbated if disadvantaged communities have limited access to genetic services and technologies.

Because health status influences educational and occupational achievements, public policy should be used to reduce health disparities. One option is to assure that, to the extent possible, essential genetic services are available and affordable to everyone whether through health insurance coverage or via publicly funded programs if private insurance is unavailable or inaccessible.

It is essential to consult with proposed beneficiaries when developing policies or programs to increase access to genetic services and technologies. Also, efforts to reduce disparities should uphold the right of individuals to make fully informed, independent decisions concerning genetic services that are free of coercion.

5. Require community consultation in the planning and design of: a) public health programs that incorporate genetics; and b) publicly financed genetic research.

To assure that policies or research do not further disadvantage vulnerable populations, states should require consultation with diverse stakeholders, including those whom the policy will affect and research subjects. Community consultation enhances policy development and research by:

- Educating individuals and communities about genetics and their health.
- Assuring that informed consent and other protocols are culturally and linguistically sensitive.
- Promoting community debate and deliberation about genetic issues.
- Using the target population's perspective to inform the design and evaluation of a policy or research project.
- Cultivating community leadership on genetic issues.



Likewise, grantmakers can require grantees to involve disadvantaged communities in their programs and research. They can sponsor workshops to help program planners and researchers more effectively involve diverse communities, invest in programs to educate the public about genetics, and encourage public involvement in policymaking and research.

6. Promote the collection and analysis of scientific data to protect public health and safety and prevent disease.

Public policies and resources are often necessary to support applied research that translates science into disease prevention practice.⁷⁶ Public health experts expect it could be years before sufficient data accumulate to determine the cost-effectiveness of new genetic services and technologies. In the interim, states should support studies to identify which policies, programs, and services are likely to protect and improve the public's health at a reasonable cost.

Policy support is needed for environmental and public health researchers to gather sufficient data to: 1) clarify the relationships between genes, environmental factors, and health; and 2) develop interventions to effectively prevent or treat health conditions that have a genetic component. This research will require large population studies, investigative trials, prevention effectiveness studies, and applied practice investigations.

Balanced policies are needed that protect the privacy of individuals while supporting the information needs of qualified researchers to conduct these studies.⁷⁷ Information contained in individual medical records is also vital to many public health functions such as monitoring community health, conducting epidemiologic studies, and evaluating publicly financed programs.

7. Guard against premature commercialization while assuring that the potential health gains from commercial development of genetic services and technologies are realized.

Many tests for genetic disorders are still in the investigational stage; even fewer therapies or preventive measures based on genetic research are currently available. Although the full market potential may take years to realize, the commercial promotion of genetic tests, services, and other technologies is underway even as legal, ethical, social, and economic policy issues are debated.

This may lead to business and public expectations quickly getting ahead of policy responses and could result in inappropriate use of genetic services and excess costs. Policymakers must be prepared to resist pressure for quick action in order to review scientific data about the harms, benefits, and costs of new genetic services and technologies. At the same time, the initiation of appropriate policies and programs can ensure that beneficial genetic advances are obtainable by all persons needing these services.

WHAT IS PREVENTION RESEARCH?

Prevention research answers three basic questions.

- A. What are the risk factors associated with illness, disability, and injury (such as sedentary lifestyles, exposure to environmental toxins, and substance abuse)?
- B. How common are these risk factors and the ill health they produce?
- C. What are the best ways to address these factors and thereby promote health?

By answering these questions, prevention research serves as the critical link between biomedical research (which focuses on human physiology or disease treatment) and public policies that encourage wellness and reduce the need for sick care. Among many achievements, prevention research has:

- Developed vaccines and vaccination policies that eradicated the ordinary transmission of smallpox.⁷⁸
- Made the case for lead-free gasoline policies, which has reduced the level of lead in children's blood by 80%.⁷⁹
- Substantially lessened the threat of death from cervical cancer by improving the quality of the Pap test and supporting its widespread use in routine care.⁷⁸

While federal agencies underwrite most prevention research, states can assure that health researchers have access to data for large and long-term population studies. For example, public agencies need genetic and environmental data to identify ways to reduce birth defects. Health researchers must use individual health records to assess the potential benefits and harms of genetic testing.

Genetics Policy Development Framework

A policy framework is a tool for policymakers to use in navigating complex issues and competing interests. As public and private health systems look to capitalize on genetics, a policy framework can help policymakers:

- Become familiar with research findings and applications for medical and public health practices that are complex and new.
- Examine the ethical, legal, social, and economic implications of policy options.
- Balance the interests of individuals with those of their families, other groups with similar genetic risks or diseases, and the public at large.
- Obtain timely expert and stakeholder input at all stages to maximize positive outcomes and reduce the odds of unintended consequences.
- Articulate desired outcomes for new policies and programs.

The sample policy framework in Figure 8 initiates a continuous process to obtain public input, establish principles and goals, review and update policy, and keep abreast of advances in genetics and the policy environment.

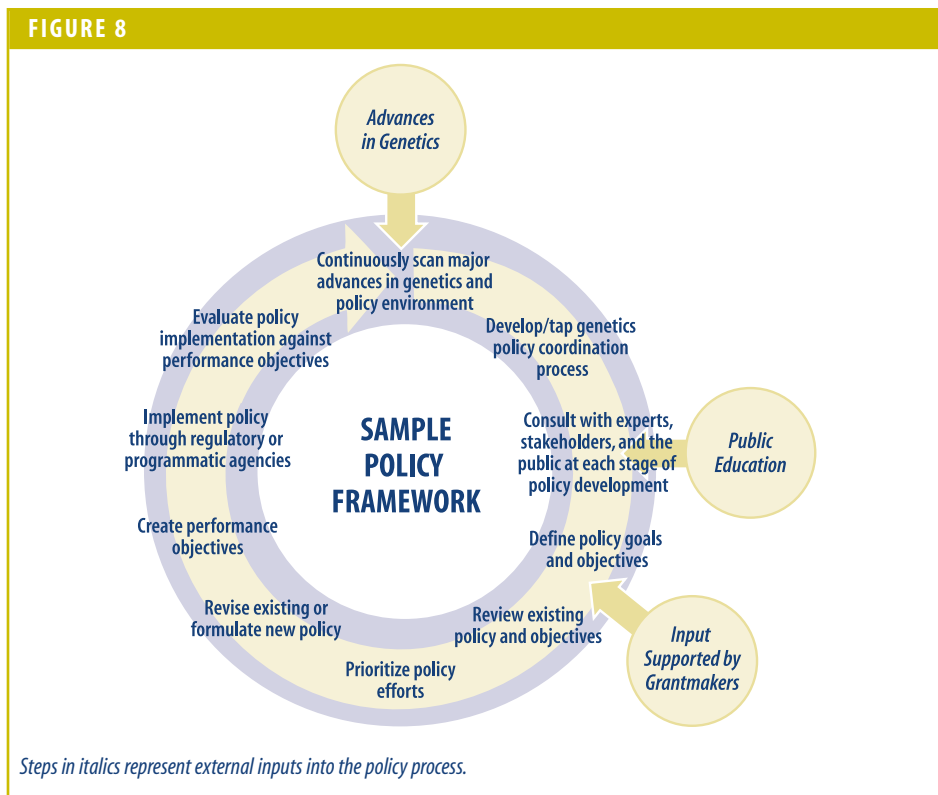
APPLYING GENETICS TO PREVENT HEART DISEASE AND STROKE

Heart disease and stroke are the first and third leading cause of death in the United States. Health professionals have known for many years, based on family history and health research, that genes, environment, and health behaviors contribute to cardiovascular disease (CVD).

The genetic variants associated with CVD are numerous and complex. As genetic factors are identified and genetic tests for CVD risk become available, it may be possible to develop personalized treatment plans. Researchers are also developing family history assessments that health professionals and patients can use to assess risk.

In the future, state health agencies could provide information to health professionals and consumers about genetic factors in heart disease and stroke. They also might promote the use of family history assessments in clinical practice.^{44,45,65}

FIGURE 8



When used to integrate genetics into existing health policy, this framework triggers a continuous scan of science and technology to prompt modifications to current policy, while minimizing the need for frequent formulation of new policy. Scientific evidence of efficacy, effectiveness, and utility can help policymakers and other stakeholders weigh benefits, harms, and costs. The process is iterative with no end-point and includes a mechanism for periodic policy review to accommodate scientific advances, facilitate policy updates, and correct for unintended consequences. See Appendix D for a longer discussion of the elements in this framework.

Checklist of Information Needed

The model framework will help ensure that policy deliberations use available scientific evidence and consider the perspectives of both individual consumers and diverse population groups. The checklist below identifies specific types of information that should be sought.

- ✓ Seek analyses from experts to determine if evidence indicates the policy will have the desired effect.
- ✓ Examine policy recommendations by experts (e.g., recommendations of the Secretary’s Advisory Committee on Genetics, Health and Society; the CDC Office of Genomics and Disease Prevention).
- ✓ Assess the risks associated with the policy including the effects of not implementing the policy or delaying action until sufficient evidence becomes available.
- ✓ Determine the likely outcomes (benefits, harms, and costs) associated with the policy.
- ✓ Investigate what is acceptable to health professionals, consumers, and others who would have to comply with the policy or regulation.
- ✓ For genetic services and technologies, confirm through advisory experts the clinical utility (e.g., how results would aid in prevention, diagnosis, and treatment) and verify the reliability of the data and the processes used to assess utility.
- ✓ Study the fiscal impact of the policy for state and local governments, employers, insurers, taxpayers, and intended policy beneficiaries.
- ✓ Compare to other policy alternatives. A review of similar policy decisions in other states is one way to identify policy alternatives (see Appendix C for other resources).

Use of Standardized Legal, Medical, and Public Health Terminology

To avoid confusion, contradiction, or possible unintended outcomes, terminology and definitions used during policy formation — and in statutes and other policy documents — should be understood by all stakeholders and used consistently. The National Conference of State Legislatures may be able to provide terms and definitions already in use by some states. Other publications discuss definitional issues.^{3, 4, 55} For definitions of terms used in this report, see “Glossary” in Appendix A.

APPLYING GENETICS TO PREVENT DIABETES

Diabetes is increasingly common in the United States, affecting about 16 million people. The prevalence of this dangerous, costly disease is expected to increase over the next few decades, especially due to excessive weight and physical inactivity. As more Americans develop diabetes, their risk for heart disease, kidney failure, blindness, and loss of limb increases.

Genetic studies of type 2 diabetes are especially complex because genes associated with a deficiency in insulin production or response to insulin may appear normal, but function inappropriately. Also, nutrition, physical activity, and weight strongly influence diabetes development and management.

Type 2 diabetes can be prevented and managed without knowing the genetics of the disease. However, genetic research may make pharmacological treatments even more effective. Genetic studies may also help identify susceptible populations or individuals before they become symptomatic, when preventive strategies can have the greatest impact.

Type 2 diabetes disproportionately affects African Americans, Hispanic/Latino Americans, and other racial and ethnic minority groups. In the future, state health agencies could convene groups to review opportunities to use advances in genetics to prevent the onset of diabetes or control the disease within their community.^{45, 66}

NEW YORK INTEGRATES GENETICS INTO PUBLIC HEALTH

The New York State Department of Health is beginning to integrate genetics into many of its programs. These activities build on a strong cornerstone, the Wadsworth Center, which is the first U.S. public health laboratory to license the practice of genetic testing. With a staff of 1,100 and about \$24 million in federal funding, the center also conducts research, sponsors educational opportunities for youth and K-12 instructors, and screens over 250,000 newborns each year.

The center regularly coordinates activities with other department divisions, especially those that are integrating genetics. For example, the division of environmental disease prevention increasingly assesses gene-environment interactions in disease epidemiology. The division of chronic disease and adult health coordinates Internet-based listservs about genetics and chronic diseases. Genetics is also being integrated into the state cancer registry and programs for children with special health care needs.⁴⁶

Recommendations

This report offers guidance to state leaders about genetics policy issues and identifies widely accepted disease prevention principles. This section goes one step further by outlining specific policy recommendations likely to harness genetics to improve the health of state residents.

Integrate Genetics into Existing Disease Prevention/Health Improvement Policies

Problem: An exceptionalism approach may isolate genetics from other state investments in health care and public health, possibly creating redundant, overlapping, or contradictory policies and programs. Exceptionalism can slow the translation of basic research into practice or lead to missed opportunities to advance individual and community health.

Recommendation: An integrated approach is strongly recommended in order to maximize the benefits of genetic and genomic science.^{8, 16, 44, 55-59} For more information, see “The Case for Integration” on page 11.

Establish a Process for Coordinating State Genetics Policy Issues

Problem: Many states lack a coordinated process to achieve a consistent, coherent approach to integrating genetics into policy and programs. In many instances, decisions about integrating genetics into health policy involve complex issues about ethics, costs, benefits, and individual and societal interests. Without coordination, policy decisions in one area, such as medical care, could be at odds with policy in another area, such as research or insurance.

Recommendation: Each state should develop a coordinated approach to genetics policy development. The process should involve expertise in a wide variety of policy issues, have authority to bridge state agency lines, and interact with many stakeholders.

Most states have a genetics coordinator, usually located in the maternal and child health program of the state health agency, who facilitates development and implementation of a state genetics plan. While often focused on newborn services, the coordinator frequently works with multiple stakeholders in planning broadly for genetic services and policy. The activities of this position would continue to provide an essential service and be a starting point for creating the wider functions of a state genetics policy coordination process.

Each state should determine the best approach for this process. At a minimum, the process should include mechanisms to convene state agencies to improve communication and coordination, mediate controversies, and address crosscutting issues such as ethical, legal, and social concerns. The process could also:

- Track and assess genetic advances that may have implications for state policy.
- Monitor the policy environment for unintended consequences.
- Make periodic reports to state policymakers.
- Coordinate programs to prevent or control disease.
- Help identify genetics-related prevention research needs (see recommendation about prevention research on page 25).

To facilitate the coordination process, some states have convened a time-limited advisory panel of experts with staff support from the state health agency. Working groups are another alternative approach to a permanent, funded position. For example, legislatures could create ad hoc teams that bring together advisory groups and staff from various committees to coordinate legislative development and analyses. Persons and agencies involved in the coordination process should be knowledgeable about and representative of the many policy realms presented in Figure 3 (page 7).

Solicit Systematic Advisory Input from an Array of Stakeholders and Experts

Problem: Genetics is a complex policy area with many unknowns, a rapidly expanding knowledge base, and potentially high risks.

Recommendation: State policymakers should formally consult with experts and stakeholders by establishing temporary or permanent advisory panels. Open and extensive dialogue promotes clarification of scientific facts, expression of divergent views, deliberation of benefits and harms, and broad consideration of policy proposals.

Broad stakeholder and expert input are critical to the genetics policy coordination process recommended above. Periodic public hearings in a variety of locations and multiple stakeholder meetings also assure that policy formation takes into account diverse perspectives. For a list of potential advisors, see “Consult with Experts, Stakeholders, and the Public Throughout the Process” on page 31.

Anticipate Emergence of the Genetic Services and Technologies Market

Problems: The private market for genetic services and technologies (e.g., testing, education and counseling, therapies) is quickly emerging. In June 2002, Aetna, Inc., announced it would pay for genetic tests for plan enrollees who are at risk of serious, but treatable diseases. In fall 2002, Myriad Genetics, Inc., began direct-to-consumer marketing of genetic tests for breast and ovarian cancers.

The widening market for genetic services and technologies has multiple policy implications.

- State governments and other employers are straining from double-digit medical inflation. New benefits for genetic services will need to deliver value by improving health or moderating future health care expenses.
- Currently, no one is sure how genetic services will be financed; what will be the demand for and cost of these services; or what services should be offered. Many insurance policies lack specific information about payment for genetic education and counseling, two services essential to high-quality care. Even if these services were covered, gaps in reimbursement and billing systems could impede the delivery of services.⁸⁰
- Because personal health information — including genetic data — has substantial commercial value, there will be great demand and financial incentives offered for access to this information. Policymakers will need to balance profit incentives and public benefits when formulating policy regarding the commercial use of genetic data.⁸¹

Recommendations: Policymakers should require that genetic services financed by the state meet standards for validity, utility, and reliability (see definitions on page 14). Additional criteria should consider estimated costs and potential benefits, the availability of effective prevention or treatment, and provision of education and counseling by qualified health professionals.

Genetic advisory groups or committees can strengthen “the connection between legislators and their health authorities,” according to Tim Baker of the CDC Office of Genomics and Disease Prevention. Advisory groups provide policymakers with a far-reaching picture of what their state is doing to address genetics.

Recognizing the complexity of genetics policy, Oregon Senator Richard Devlin says, “Everything changes so rapidly that the legislature will probably have to revisit the issue every session.”

State policymakers should also prepare for the emerging market by:

- Protecting society from unwanted and potentially harmful genetic information disclosures.³⁰ Policies to protect individual privacy should be robust, yet provide for the information needs of researchers who seek to develop beneficial applications of genetics.
- Authorizing one state agency to collect, investigate, and take appropriate steps regarding reports of discrimination or privacy breaches that involve genetic information.⁸²
- Directing state universities to expand genetics education and training for health professionals and the general public.
- Convening insurers, employers that sponsor coverage, consumer groups, and health professionals to identify and resolve barriers that will impede the timely availability of affordable and beneficial genetic services.

States will need expert and stakeholder input in defining what kinds of genetic services should be billable within standard visits covered by Medicaid or State Child Health Insurance Program (SCHIP). Modifying billing codes may be moot in states that have capitated contracts for Medicaid and SCHIP.

Expand Public Education Programs

Problem: Consumers with little or misleading information cannot make knowledgeable decisions about their health and health care. Educational programs are needed to help the public understand the effects of genetic variation on individual and population health.

Recommendations: Public policy should support well-designed public education programs that increase consumer knowledge. Public education should enable citizens to:²⁰

- Participate effectively in policy debates about genetics and support enactment of sound genetics policy.
- Become informed consumers who can make appropriate personal health decisions.

The public education system (K-12) should integrate genetics into science and health curricula and keep instructional material as up to date as possible. State universities should sponsor short courses or symposia for the general public; universities and community colleges should expand course offerings in genetics for health professionals and health administrators. To minimize costs, many existing educational resources could easily incorporate advances in genetics.



The established communication channels of state health agencies, voluntary health associations, and the media can be used as part of a coordinated public education strategy to introduce genetics and inform audiences about potential uses, benefits, and harms. Policymakers — with assistance from grantmakers — can bring together media representatives with scientists to eliminate communications gaps and encourage balanced media coverage about the risks and benefits of genetics.

Internet websites can integrate genetics into health and disease information available to consumers. Public and private grants to community-based organizations are essential to develop culturally and linguistically appropriate genetics education programs for multi-cultural groups, persons with limited literacy skills, and other vulnerable groups.

Strengthen the Public Health Infrastructure for Genetics

Problem: Many public health agencies lack the personnel and resources necessary to integrate genetics into programs.

Recommendations: To harness genetic advances for disease prevention and health improvement, public health agencies should have resources to train staff and update and expand equipment. State policymakers need to continually inform and educate federal officials about gaps in the capacity of state health agencies to integrate genetics into programs and practice.

For example, because of the potential expansion in genetic tests and screening, state-funded laboratories (and possibly agencies that certify laboratories) may need increased capacity to analyze new and greater numbers of genetic tests. Improved data collection and communications systems for surveillance and epidemiology will enable health agencies to detect, investigate, and respond appropriately to changes in public health.

State policymakers also should assure their public health workforce has access to CDC-supported training and program development assistance. Staff knowledge and skills must be upgraded regularly to keep pace with discoveries in genetics and related fields.²³ Public health staff must be able to translate these advances into health education messages and programs for the general public, especially about common chronic diseases such as diabetes, heart disease, and cancers.

Review Licensing Requirements for Health Professionals, Adding Genetics Competencies as Needed

Problem: Because genetics has long been a medical subspecialty, other health professionals typically receive minimal instruction in the field and lack a full understanding of how to apply genetics effectively. Today's rapid advances in genetic and genomic sciences make it essential that all health professionals begin to develop competencies in genetics. In particular, nurses, health educators, and primary care providers — often the consumers' entry point for medical care — must be able to provide quality genetic information and education to patients.

Genetics proficiency for health professionals may develop slowly unless licensing requirements are updated to include genetics.

Recommendations: First, state policymakers should direct licensing boards to work with health professional organizations to expand opportunities for continuing education in genetics.

Second, governors and state legislators should ensure that all licensing boards have members with genetics expertise or background in related ethical, legal, and social issues. State leaders should ask these boards to develop recommendations concerning:

- Genetics competencies as a certification, licensing, and accreditation requirement.
- The licensure of genetic counselors. Currently, only California and Utah require genetic counselors to be licensed.⁸⁵
- The availability of genetics education programs for practicing health professionals.

Any new licensing requirements should promote comprehensive interdisciplinary training in genetics for health professionals. The University of Washington School of Public Health already provides genetics training across many disciplines, including those outside health and medicine.²

The Centers for Genomics and Public Health, recently funded by CDC at the Universities of Michigan, North Carolina, and Washington, will develop web-based, distance-learning programs for practicing public health professionals (see Appendix C).

WASHINGTON AND OREGON INVEST IN CONSUMER EDUCATION PROGRAMS

This year the Washington State Health Department will roll out an innovative program to build public knowledge about genetics. A series of "front porch" forums, in which neighbors gather on a porch or at a local spot, will stimulate public dialogues about genetics. Newspaper articles reporting on the front porch discussions will carry the issues to the community at large. In time, the department hopes a better public understanding of genetics will improve health education and promotion efforts.

Oregon also plans to invest in public genetics education. In 2001, state policymakers asked the state advisory committee on genetic privacy and research to educate the public about ethical, legal, and scientific issues associated with genetics. The committee is currently researching how to undertake such an effort.⁷

IOM OUTLINES STEPS TO PREPARE PUBLIC HEALTH FOR GENETICS

Looking to the future, the Institute of Medicine (IOM), part of the federally chartered National Academy of Sciences, recently issued calls to action to prepare public health for modern challenges and opportunities. Clearly, the IOM reports agree with the recommendations in this policy guide to integrate genetics into health policy and programs.

In *The Future of the Public's Health*, the IOM calls for public health agencies and other stakeholders — health care systems, employers, media, and academia — to:

- Advance medical and public health practice by supporting not only genetic advances, but also proven behavioral and population interventions to prevent disease, treat illness, and improve health.
- Educate and inform consumers about genetic advances.
- Counter information, products, and practices that may undermine individual or population health.⁸³

In *Who Will Keep the Public Healthy?*, the IOM finds that public health professionals have little or no training in genetics, a fairly new and rapidly changing science. To build professional competencies in genetics, the institute calls for:

- Continuing education for the current public health workforce so it can use genetics as one tool to advance population health.
- Incentives for public health professionals to participate in genetics education and training opportunities.
- Core curricula in public health schools to develop students' ability to apply genetics to basic public health sciences.

Whether for current or future public health professionals, educational programs must build an understanding of ethical, legal, and social issues related to genetics.^{21,84}

Increase the Supply of Diverse, Qualified Health Professionals, Especially Genetic Counselors

Problem: The scarcity of genetic counselors and other health professionals trained to interpret test results and counsel patients impedes the appropriate use of genetic tests for disease prevention and health improvement. These shortages will be especially pronounced in many rural, inner city, and disadvantaged communities.

According to the National Society of Genetic Counselors, the United States has approximately 2,000 to 2,200 genetic counselors. Twenty-six graduate programs in genetic counseling add about 175 counselors each year, and the society expects this number to grow. Other health professional schools are beginning to incorporate genetic counseling into clinical training.⁸⁶

Recommendations: States should offer incentives to increase the supply of qualified genetic counselors and improve the genetic counseling skills of other health professionals.⁸⁷

In addition, state leaders should support universities' efforts to increase the number of ethnic and racial minority students in health professional programs. Scholarships and other assistance also should provide students from medically underserved communities with an incentive to practice in their home communities.

State policymakers should ask federal officials to support expanded genetic counseling training programs. These programs should go well beyond current efforts — largely for primary care providers — to reach more health professionals and provide more in depth instruction. See “Health Resources and Services Administration,” page 29.

Use Existing Prevention Measures Known to Improve Health

Problems: Interventions known to be effective in improving health are readily available, independent of genetic research. However, many of these proven strategies are underutilized,^{74, 88} even while the cost of care rises for cardiovascular disease, cancers, diabetes, and other preventable health conditions.⁸⁹ Implementing these disease prevention strategies would increase aggregate costs, but the return on investment would be healthier citizens. Productivity, quality of life, and independence also could improve.

Recommendation: State leaders should invest in recommendations from the nation's top prevention experts³⁵ while preparing for future developments in genetic services and technologies.

The *U.S. Preventive Services Task Force* (USPSTF) sets the gold standard in its *Guide to Clinical Preventive Services*. Convened by the U.S. Public Health Service, this panel of independent prevention and primary care experts identifies the most effective clinical interventions (vaccines, screening tests, and physician counseling) to help Americans prevent disease and promote health. USPSTF recommendations are posted at www.ahrq.gov/clinic/prevenix.htm. (States should ask federal policymakers to expand the USPSTF agenda to include genetic services and technologies.)

A companion resource, the *Guide to Community Preventive Services* recommends effective strategies to protect the health and safety of a community, such as school programs, public education campaigns, and policy interventions. An independent, non-federal task force convened by the Centers for Disease Control and Prevention develops *The Community Guide*. To access recommendations, go to www.thecommunityguide.org.

Make a Long-term Commitment to Prevention Research Utilizing Genetic Advances

Problems: The average U.S. lifespan would not have increased by 25 years during the 20th century without prevention research, which is the critical link between biomedical research and public health policy and practice.

More prevention studies are needed to link genetic datasets with databases describing disease outcomes, environmental exposures, behavioral risk factors, and use of medical services and technologies. This research cannot occur without sufficient resources to support trained staff and adequate information systems along with policies that allow access by qualified researchers to individual health information.

Recommendations: While much of the funding for prevention research may come from federal agencies, state policymakers and health agencies should collaborate with state universities to develop a genetics research agenda. This joint effort would identify:

- Research priorities needed to fill information gaps about how best to improve the health of state residents.
- Any modifications to state policy necessary to protect individual health privacy rights without obstructing necessary research studies.
- Efforts, if any, needed to increase the supply of prevention research staff and develop sophisticated information systems.



Universities can use the research agenda to prioritize funding requests and research activities. Together, state policymakers and university officials can educate federal policymakers about the need for prevention research funding. They might ask grantmakers to sponsor independent analyses of policy options or studies of special topics, such as cases of genetic discrimination.

Further, states and universities can use the agenda to establish multi-state collaborations that may be necessary to support complex or expensive studies that some research questions require. For example, to assess the benefits, costs, and harms of adding a genetic screening test to Medicaid coverage, universities may need to conduct a randomized clinical trial for several years and with a large study population. Other research may be needed to analyze the costs of requiring insurers to cover genetic tests.

Similarly, epidemiological studies for a serious, but uncommon condition may require the participation of several states in order to produce sufficient data. To examine a high incidence of asthma or lung cancer in a cross-border region, two states might investigate the influence of local environmental exposures on gene expression.

In regards to privacy protections, former Arizona Senator Susan Gerard (now the governor's advisor for health and human service policy) notes "We need to be careful not to make it so difficult to access information that it stifles research."

PENNSYLVANIA INVESTS IN PREVENTION RESEARCH

With some of the state's portion of the national tobacco settlement, the Pennsylvania Department of Health awarded \$23 million in prevention research grants to the University of Pennsylvania, Temple University, and the University of Pittsburgh. These prevention studies will:

- Examine whether online communications between providers and patients can reduce risk of heart disease and stroke.
- Evaluate different interventions to improve hypertension medication compliance among African Americans.
- Identify promising interventions to reduce health disparities among racial and ethnic minority groups.

Later in 2003, the department plans to grant an additional \$59 million in tobacco settlement funds to research institutions in the state.⁹⁰

Conclusions

One hundred years ago no one imagined the impact that electrical energy would have on every aspect of daily life. Similarly, it is impossible today to predict how the science of genetics will alter opportunities to advance individual, family, and community health. State policymakers have already begun to address some of the complex policy issues arising from genetic and genomic sciences. As with other fast-paced scientific and technological advancements, **the intersection between genetics and public policy will continue to require close monitoring and timely action.**

Early in the process of developing or updating public policy to include genetics, policymakers should decide whether such policies are viewed as exceptional (i.e., in a class apart from other laws and regulations governing health and medicine) or should be incorporated into existing policies. Although genetic exceptionalism has been the basis of many recent state laws, health experts believe **the integration of genetics into health, social, and environmental policy is the more efficient and effective strategy.**

Comprehending how genetic advances can be used to promote health and prevent illness is only now beginning. It is clear that many highly publicized possibilities may be decades away from being realized, if ever. To create sound genetics policies and programs, policymakers and those who advise them will want to **follow a coordinated process for policy development that relies on scientific research and ongoing community consultation.**

State leaders should not only establish policies that are flexible enough to adapt to rapid advances in genetics but also make preventing disease and improving health a priority.

Glossary of Terms

*This appendix defines terms used in the report. Definitions of other terms are included (indicated by *) because they may be useful for policymakers. Words used in definitions that are in bold type are defined elsewhere in the glossary.*

***Base pairs/bases:** two complementary, nitrogen-rich components that form the nucleotide units of **DNA**, held together in the shape of a double helix. The bases are adenine, thymine, cytosine, and guanine, often known by their first letters, A, T, C, and G. The order or sequence of sets of these base pairs determines the code for the message from the **cell** nucleus to the cell cytoplasm, where **protein** products are made.

Carrier: a person who has one abnormal **gene** and (usually) one normal gene in the pair inherited from parents. Carrier status is particularly important for a recessive mutated gene. Carriers do not usually develop disease but can pass the mutated gene on to their children. If both parents are carriers of the same disease-causing mutation, the **probability** is one in four that a child will develop the disease or defect.

Carrier Testing: genetic tests can be used to identify individuals who carry disease-causing recessive genes that could be inherited by their children. Carrier testing is designed for people who have no symptoms of the disease, but may be at risk because of family or group history.

Cell: the basic sub-unit of any living thing.

Chromosomes: structures found in the nucleus of a **cell**, which contain the **genes** along lengthy strands of **DNA**. Chromosomes come in pairs, one (part) inherited from each parent. A normal human cell contains 46 chromosomes, 22 pairs of non-sex-determining autosomes, and one pair of sex chromosomes.

Confidentiality: refers to the reasonable expectation of an individual that sensitive information originally disclosed within a confidential relationship (e.g. patient/health professional) will not be redisclosed without explicit consent. Confidentiality, along with secrecy and anonymity, are principles of informational privacy.

Disease Prevention: clinical and community-based activities (such as immunizations, sanitation, tobacco-use restrictions, policies to promote nutrition and physical activity, medicines, and environmental protections) that help people avoid illness, disability, and premature death.

DNA (deoxyribonucleic acid): the substance of heredity, is the molecule of tightly coiled double spiral strands (double helix) of chemical units (see **bases**) that contains the information a **cell** needs to replicate itself and make the **proteins** a cell needs. DNA sequencing determines the exact order of the **base pairs** in a segment of DNA. The strands of DNA are paired, one from each parent, to form **chromosomes**. DNA is uniquely individual and can be used for forensic identification.

Environment: defined broadly to include substances — whether inhaled, ingested, absorbed, or intravenously injected — that affect health, including changes in **genes**. Environmental factors that influence health include air, water, food, infectious agents, chemicals, noise, radiation, and many other exposures. Other environmental factors are affected by behavioral choices, such as tobacco use, alcohol use, diet, and levels of physical activity. Exposures occur in the home, at sites for recreation and work, and in the outdoors. Socio-economic factors represent an additional environmental influence on health.

***Epigenetics:** refers to all modifications in **genes**, other than changes in the **DNA** sequence, that alter a gene's expression. Also, it is the study of changes in gene function (the activation or inactivation of the gene) that occur without changes to the DNA sequence. Epigenetic modifications are also known as "marks."

Expression: the **proteins** that a **gene** instructs a cell to make. Also, the disease, condition, or trait that is the result of a genetic mutation or a gene-gene or gene-environment interaction.

***Familial Diseases:** genetic disorders, such as heart disease and cancers, that are inherited and tend to be common in a family group. Familial diseases can also be non-genetic.

Family History: the recording and analysis of the medical history of a patient and his/her relatives. More in-depth analyses use information collected about disease and age of onset for up to three generations. Sometimes called a "pedigree."

Gene: an informational unit along the sequence of **DNA** containing hereditary information that makes individuals both similar and unique. Genes code for the production of particular **proteins** and are subject to modification (i.e., **mutation**) both randomly during the process of replication and as a result of exposure to environmental agents such as radiation, oxidizing agents, some viruses, etc. In humans, the DNA molecules in the 23 pairs of chromosomes contain a total of about 30,000 to 40,000 genes.

Gene or Genetic Markers: landmarks for a target gene, either detectable traits that are inherited along with the gene or distinctive segments of **DNA** that point the way to a gene.

Gene Mapping: determining the relative positions of genes on a **chromosome**.

Gene Therapy: treating disease by replacing, manipulating, or supplementing nonfunctional genes.

Genetic Counseling: generally a non-directive communications process to inform a patient and his/her family about a genetic disorder or risk of disease and the associated ethical, legal and social issues. Counselors also discuss clinical procedures, treatment, management, and prognosis.

Genetic Information: data about **genes**, gene products, and gene tests. Also, the inherited characteristics that derive from the individual or a family member.

Genetic Testing: examining a sample of blood or other body tissue for biochemical, **chromosomal**, or **genetic markers** that can indicate the presence or absence of disease, be predictive of the risk for future disease, or be used to identify the source of the body tissue.

Genetic Services and Technologies: services include programs designed to educate the public or inform individuals about **genetics** or to counsel patients about risks and benefits of **genetic tests**, genetic diseases, or treatments. Technologies include genetic tests, therapies, or other interventions based on genetics or **genomics**.

Genetics: the scientific study of **genes** and their roles in disease, physiology, and evolution or human development. Genetics studies the qualities or traits transmitted from parents to offspring and those that are modified by the environment.

- *Human genetics* is the science of variation and heredity within the human species.
- *Medical genetics* is the study of hereditary factors and variation in human disease.
- *Clinical genetics* is the discipline whereby comprehensive services are provided to patients, families, and populations suspected of having hereditary disorders.

Genome: all the genetic material in the **chromosomes/DNA**, or complete inheritance, of a particular organism. The genome contains all of the **genetic information** that encodes the form and functions of a specific organism.

Genomics: the science and study of the sequence and functioning of the human **genome** and other living organisms. Also, the growing knowledge and understanding of the function of **genes** and their impact on human health and the use of this information to improve health and prevent disease.

Health Disparities: differences in the incidence, prevalence, mortality, and burden of diseases and other adverse health conditions that exist among population groups.

Health Improvement: (also called “health promotion”) the enhancement of health status and quality of life in individuals and population groups. Also, activities that improve or protect health such as physical education classes, child safety seat distribution and education programs, quit-smoking campaigns, and excise taxes on alcohol.

Human Genome Project: a public/private international research project designed to learn the structure of and understand function of the genomes of humans and other organisms and to share findings with the research community by **mapping DNA** and **sequencing** the genes.

Informed Consent: the process of informing a patient or potential research subject about the purpose, benefits, risks, potential outcomes of a **genetic** test or technology (or other medical procedure), and possible future uses of the data. Informed consent includes a signed document containing the above information and describing the process by which the individual was provided the information. In order to give fully informed consent, the decision must be independent and free of coercion.

Map/Mapping: the process of diagramming or charting **DNA sequences** to show the order in which **genes**, **genetic markers**, or other landmarks are found along the **chromosomes**. A gene map determines the relative positions of genes on a chromosome and the distance between them. A physical map of a species shows the specific physical location of its genes and/or markers on each chromosome. Physical maps are particularly important when searching for disease genes.

Metabolites: something essential for metabolism or a substance produced or used during metabolism (digestion). In drug use, a metabolite usually refers to a substance(s) that remains after metabolism.

Mutations: changes in the **DNA** spelling (the number, arrangement, or molecular **sequence**) of a **gene** that can prevent **proteins** from functioning normally and may lead to specific disorders such as birth defects or chronic diseases. Some mutations are inherited from parents while others are acquired, the result of environmental influences, lifestyle factors, and the aging process. Some mutations are protective, acting to prevent or modify errors in the coding of genes. For instance, genes for sickle cell help protect against malaria.

A common misconception is that once a gene variation or mutation is identified, it is easy to fix it or prevent disease. Another mistaken assumption is that a positive test for a genetic mutation means that one has or is certain to develop a disease. In most cases, the identification of a genetic mutation indicates only an increased risk or **probability** for developing a specific disease. Conversely, some persons without an apparent genetic risk develop similar illnesses.

Newborn Screening: examining blood (or other tissue) samples from an infant to detect disease-related abnormalities or variations in **gene** products.

Pharmacogenetics: the application of genetic science and technology to understand how genetic variations influence responses to drugs and the metabolism of drugs.

***Pharmacogenomics:** the **genome**-wide scan of variation in drug biotransformation and responses to drug action.

Population Health: what society does collectively in both the private and public sector to protect and improve the health and safety of population groups by reducing disease or trauma and preventing premature deaths. Also referred to as “community health.”

Privacy: the right of an individual to control the conditions under which personal information is conveyed to others; the capacity of the individual to determine which information is communicated to whom.

Probability: the possibility or odds that a person with one or more **genetic** factors will at some point in time develop a specific health condition. Determining probability based on genetic information may be influenced by environmental exposures.

***Protein:** a large, complex molecule composed of amino acids that is essential to the structure, function, and regulations of the body. Examples are hormones, enzymes, and antibodies.

Public Health: as used in this report, refers to specifically delineated powers, duties, rights and responsibilities conferred on public health agencies and officials. Others use the term to refer to any program designed to affect the health of large numbers of people or specific population groups and may be sponsored by private organizations or government agencies (see **population health**).

Reliability: the probability that a genetic or other medical test will repeatedly get the same result.

RNA: a chemical similar to **DNA** that transmits genetic code instructing the cell to produce certain **proteins** or perform other cellular activities.

Screening: looking for evidence of a particular disease or risk for future onset of disease. In this report, screening refers specifically to genetic diseases, in people with no symptoms of disease.

***Secondary Uses of DNA:** using **genetic information** stored in databanks for a purpose other than its original use.

Sequence/Sequencing: determining the order in which the letters, or **bases**, occur in a **gene**.

Toxicogenomics: the study of interactions between genes and environmental exposures. See Appendix D, “National Institutes of Health.”

Utility-Clinical: refers to the usefulness of the test and value of the information to the person being tested for seeking effective treatment or a preventive strategy. The utility of a given test will change as technology and treatments change.

Validity-Analytical: an indicator of how well a test measures the property or characteristic it is intended to measure. Does the test do what its manufacturer claims it does?

Validity-Clinical: refers to the accuracy of the test in diagnosing or predicting the presence or absence of risk for a health condition. Does the test successfully detect disease or predisposition for disease?

Additional Glossaries

The CDC Office of Genomics and Disease Prevention provides links to other glossaries. Go to <http://www.cdc.gov/genomics/info/glossary.htm>.

The National Human Genome Research Institute has an extensive glossary. Go to <http://www.genome.gov/glossary.cfm>. Its Spanish-language talking glossary is available at www.genome.gov/sglossary.cfm.

The Federal Role in Genetics

The first section of this appendix describes key federal agencies and some of their policy jurisdictions pertaining to genetics. The second section provides an overview of major federal statutes pertaining to genetics.

Federal Agencies and Committees

Agency for Healthcare Research and Quality (AHRQ) – supports research to improve the outcomes and quality of health care, reduce costs, address patient safety and medical errors, and broaden access to effective services. www.ahrq.gov

Centers for Disease Control and Prevention (CDC) – the federal public health agency for protecting the health and safety of people, providing credible information to enhance health decisions, and promoting health through strong partnerships. www.cdc.gov

In genetics, CDC supports public health efforts through the Office of Genomics and Disease Prevention (OGDP). The office convenes experts to advise genetic policy, translates science for public health uses, distributes information, and trains and educates the public health workforce. OGDP has created the Human Genome Epidemiology Network (HuGE Net), an online database of genetic testing results and other information for researchers. www.cdc.gov/genomics

Centers for Medicare and Medicaid Services (CMS) – administers Medicare, Medicaid, and State Children's Health Insurance Program (SCHIP). www.cms.gov

Food and Drug Administration (FDA) – approves all gene therapies and related drugs. FDA regulates genetic tests assembled and sold as kits, which represent a small portion of all genetic tests. FDA standards apply to the active ingredients used in genetic tests that laboratories develop in-house for clinical laboratory services, but not the tests themselves. www.fda.gov

Health Resources and Services Administration (HRSA) – directs programs that improve the nation's health by expanding access to comprehensive, quality health care particularly in underserved communities.

The Maternal and Child Health Bureau provides funding and resources for many state-based newborn and pediatric genetic services. www.mchb.hrsa.gov

HRSA also funds training programs through the Bureau of Health Professionals (www.bhpr.hrsa.gov), which may be a funding source for genetic counseling training programs. Both bureaus are currently studying national genetics workforce needs.

National Institutes of Health (NIH) – among its many biomedical research institutes are the:

- National Human Genome Research Institute, home of the Human Genome Project, conducts basic research and studies of ethical, legal, and social implications. www.genome.gov
- National Institute of Environmental Health Sciences, home of the Environmental Genome Project, which works to improve understanding of human genetic susceptibility to environmental exposures (www.niehs.nih.gov/envgenom/home.htm). Another division is devoted to the study of toxicogenomics. www.niehs.nih.gov

Office of Human Research Protections (OHRP) – responsible for preventing harm to people participating in biomedical and behavioral research conducted or sponsored by the U.S. Department of Health and Human Services. See also Appendix D, "Resources" section. <http://ohrp.osophs.dhhs.gov>

Secretary's Advisory Committee on Genetic Testing (SACGT) – from 1999 to 2002, the committee advised the Secretary of Health and Human Services about the medical, ethical, legal, and social implications of genetic tests, particularly their safe and effective incorporation into health care. It also made recommendations about enhancing the oversight of genetic tests, among other issues. www4.od.nih.gov/oba/sacgt.htm

Secretary's Advisory Committee on Genetics, Health and Society (SACGHS) – organized in fall 2002, this committee will more broadly consider the impact of genetic technologies on society. www4.od.nih.gov/oba/sacghs.htm

U.S. Department of Health and Human Services (HHS) – department that includes AHRQ, CDC, CMS, FDA, HRSA, NIH, ORHP, and SACGHS. www.hhs.gov

Federal Policy Jurisdiction

Some of the federal laws establishing jurisdiction over genetic services, genetic information, and applications of genetics are described very briefly below. More in-depth summaries and analyses are available.^{3-6, 91, 92}

Americans with Disabilities Act (ADA) – no mention of genetics. Protection from genetic discrimination has yet to be tested in the courts.

Clinical Laboratory Improvements Amendments (CLIA) – provides oversight of clinical laboratories (including state-operated facilities) through a comprehensive evaluation of laboratories' operating environment, personnel, proficiency testing, quality control, and quality assurance. May need to be expanded to cover genetics.

Employee Retirement Income Security Act (ERISA) – prohibits employers from discriminating against employees in firing, promoting, or denying benefits due to any health condition.

Equal Employment Opportunity (EEO) provisions in Title VII of the Civil Rights Act of 1964 – prohibits employment discrimination based on genetic information. Limits employer testing.

Health Insurance Portability and Accountability Act (HIPAA) – establishes minimum protections regarding the privacy of personal health information, including genetic information, and allows states to have more stringent privacy safeguards. Other provisions apply to health insurance coverage and health research. State laws governing the collection and analysis of public health and vital statistics are exempted from the HIPAA privacy provisions. ERISA preempts state laws affecting employers' benefit plans.

Occupational Safety and Health Act (OSHA) – assures, to the extent possible, safe and healthful working conditions for employees. Authorizes the Occupational Safety and Health Administration to establish workplace standards to minimize workers' contact with potentially dangerous exposures. Currently, OSHA provides little guidance on the use of genetic information or tests for occupational safety and health purposes.

Resources

This appendix identifies organizations and initiatives with information to assist state officials with genetic policies and programs.

Association of State and Territorial Health Officials (ASTHO) – in collaboration with many of its affiliates, other health organizations, and CDC – is appraising state policy needs, facilitating policy dialogues, and developing policy options for integrating genetics into public health policy and practice. Two resources for policymakers include the ASTHO *Genomics Impact* electronic newsletter and the *Genetics Briefs* series published with the National Conference of State Legislatures. www.astho.org

CDC-Funded Centers for Excellence in Genomics – among their services, these centers can provide technical assistance to local, state, and regional public health organizations.

- **North Carolina's Center for Genomics and Public Health** provides assistance to the public health community through strategic planning and cost-benefit analysis, especially in the area of cancer genetics. In addition, the center works to build public health professionals' competency in integrating genetics into public health. www.sph.unc.edu/nccgph/index.htm
- **The University of Michigan's Center for Genomics and Public Health** is a partnership between the School of Medicine, the School of Public Health, and the Michigan Department of Community Health. The goal of the program is to further the integration of genetic discoveries into public health practice. Among its many activities, the center will develop web-based, distance-learning programs for practicing public health professionals; explore the ethical, legal, and social issues related to genetics; and address community participation. www.sph.umich.edu/genomics/
- **The University of Washington's Center for Genomics and Public Health** focuses on integrating advances in genetic technology into public health practice and educating students and current public health professionals. Partners include the Northwest Center for Public Health Practice, the Center for Ecogenetics and Environmental Health ELSI Core, Washington State Department of Health Genetic Services Division, and the Northwest Cancer Genetics Network. <http://depts.washington.edu/cgph/>

Coalition of State Genetic Coordinators is an organization of state and territorial genetics coordinators and others who work together to promote core public health functions as they apply to genetics. The coalition sponsors an information service, the National Newborn Screening and Genomics Resource Center. To access the coalition or the resource center, go to: www.stategeneticscoordinators.org.

National Conference of State Legislatures (NCSL) has a Genetic Technologies Project. Since 1999, the project has provided legislators and other policymakers with objective, comprehensive, and scholarly information to facilitate the development of sound genetics-related legislation. As part of this initiative, the project convened a blue ribbon panel to study and report on genetics issues in employment, insurance, privacy concerns, and genetic reproductive technologies. Along with the *Genetics Briefs* series published with ASTHO, the reports from the blue ribbon panel are available at www.ncsl.org.

Task Force on Community Preventive Services, an independent, non-federal task force convened by the Centers for Disease Control and Prevention that recommends effective strategies to protect the health and safety of a community. These recommendations are published in the *Community Guide to Preventive Services*. www.thecommunityguide.org

U.S. Preventive Services Task Force (USPSTF) sets the gold standard or definitive guide to prevention in clinical settings in the *Guide to Clinical Preventive Services*. This panel of independent experts in prevention and primary health care was convened by the U.S. Public Health Service to identify the most effective interventions – vaccines and medicines, screening tests, and physician advice – to help Americans prevent disease and promote health. www.ahrq.gov/clinic/uspstfix.htm

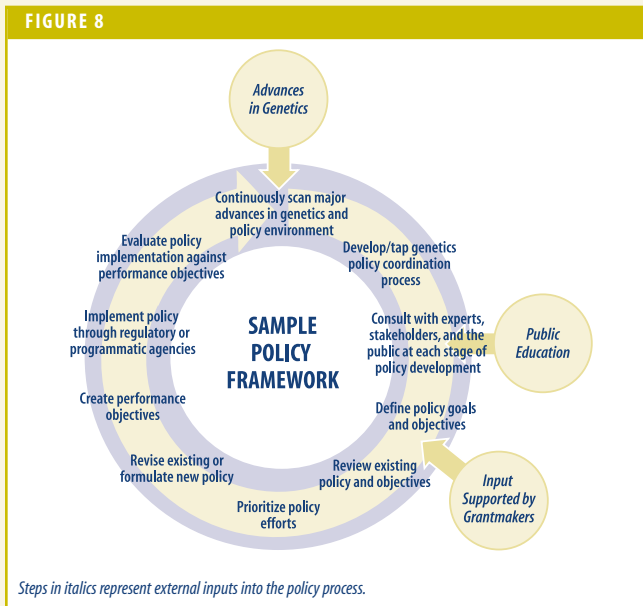
Actions for Policymakers

This appendix provides supplemental information about the sample policy framework described in the Genetics Policy Development Framework section in the main text.

Policy development is “the means by which problem identification, technical knowledge of possible solutions, and societal values join to set a course of action.”⁹³ In order to develop and enact effective genetics policy, state policymakers need to respond consistently to often-competing requests and demands from commercial interests, consumers, the research community, public health, and health care practitioners.

Using the sample policy framework, this appendix describes the steps in greater detail. The information is presented in an iterative form and has no defined endpoint. In practice it may be helpful to revisit prior steps or alter the sequence to reflect a state’s unique policy context. For example, the public consultation process may be enhanced by first providing stakeholders with a basic education in genetics, health policy, and disease prevention. Some steps, especially the policy and science scan, should be continuous.

Suggested Framework for Action by Policymakers



Scan Genetic Discoveries and Policy Environment

As with any complex or potentially controversial deliberation, policymakers will want to **regularly assess major advances in genetics and the current policy environment**. What are potential benefits? What, if any, problems are occurring, and what are potential remedies? Evaluation of existing policies and continuous monitoring as new laws are implemented will help avoid or quickly mitigate unintended consequences.

Develop a Genetic Policy Coordination Process

In order for state policies to keep pace with the rapid advances in genetics, a process for **coordinating information, developing consistent policy, and monitoring policy outcomes** is critical. See recommendation about policy coordination on page 20.

Expand Public Education Programs in Genetics

A knowledgeable public can assist in the policy development process and be supportive of sound genetics policy. Genetic education programs (see recommendation on page 22) can be incorporated in primary, secondary, and university curricula and be integrated into community or media channels.

Consult with Experts, Stakeholders, and the Public Throughout the Process

Policymakers will want to invite a broad array of representatives to advise, debate, and recommend policy options. Consultation with advisory panels is useful at several stages in the policy development process. Whether temporary or permanent, advisory councils provide a venue for gathering and interpreting information needed to develop sound policy recommendations.

Stakeholders with a statutory, economic, or professional interest in the outcome of genetic policies include, but are not limited to:

- Intended policy beneficiaries.
- Employees in group insurance plans, health consumers, and organizations representing diverse communities and institutions.
- Representatives from all levels of government responsible for enacting, implementing, and regulating health policy.
- Insurance companies, health maintenance organizations, and insurance purchasers such as employers and government agencies.
- Public funding agencies and private grantmakers.
- Representatives of the biomedical industry, such as pharmaceutical and biotechnology companies.
- The public, which encompasses individuals who can collectively represent a state’s diverse social groups and geographic regions. Travel subsidies and stipends may be necessary to assure all persons can participate fully.

Experts who can assess the policy implications of research findings and evaluate policy options include, but are not limited to:

- Public health officials from state, local, and federal agencies, including specialists in disease prevention, education, epidemiology, environmental health, and health services.
- Broad representation from the health professions, including, but not limited to, geneticists and faculty from health professional schools.
- Ethicists, both specialists in medical ethics and others with a broad social and philosophical perspective.
- Researchers in genetics, genomics, environmental sciences, and those involved in prevention research.
- Economists who can appraise the impact of genetic discoveries on health costs and other economic effects.
- Actuaries who can evaluate the impact of genetic services and technologies on the cost and availability of health insurance coverage.
- Legal specialists with expertise in privacy and confidentiality issues and matters related to discrimination.
- Educators, from kindergarten to post-graduate levels, to consult on broadening curricula and professional training.



Develop Relationships with Grantmakers

Private foundations and public funding agencies are a valuable source of expertise and financial resources. Grantmakers are well positioned to **convene advisory groups, independently analyze policy options**, and support other aspects of the policy development process (see “Grantmakers’ Role,” page 8).

Define Policy Principles and Objectives

The “Policy Principles” section (page 15) can serve as a starting point for discussion and development of principles and objectives. Broad policy principles — established in consultation with stakeholders, experts, and public representatives — create a foundation for developing more detailed policy objectives. Objectives should address immediate concerns, such as privacy protection, yet be sufficiently broad and flexible to ensure long-range integration of genetics into health policy and practice.

Review Existing Policy

Guided by these principles and objectives, a review of existing health policies and programs can be conducted in many realms, as suggested in Figure 3, page 7. Agencies and advisory bodies with relevant oversight (e.g., the insurance commission or board of health) can take the lead in **analyzing existing policies as they relate to principles and objectives established as part of the policy development process**.

Policymakers might request advisory bodies, or state agencies, to answer specific questions.

- Are there statutory references to genetics? Is the statute text sufficiently broad to include genetics, or should genetics language be added?
- Where are revisions, if any, needed in statutes, regulations, or elsewhere?
- What evidence, if any, exists of a need to provide special protection or treatment for personal genetic information?
- How have other states handled a specific issue?
- Does any federal policy pre-empt state jurisdiction?
- What are the policy positions or recommendations of pertinent professional organizations, the advisory committees associated with the Human Genome Project or the CDC Office of Genomics and Disease Prevention?
- What concepts and terminology should be used in order to promote consistency among different sections of state law and with other states’ and federal policies?
- What equity issues need to be resolved regarding access to genetic services by those in greatest need?
- Does the agency responsible for administering the policy have adequate resources to integrate genetic advances into its procedures and practices?

A critical challenge in the policy review phase is to **distinguish between sound scientific findings and faulty assumptions or questionable research**.

Authoritative institutions and experts can evaluate the sources and soundness of the data on which decisions are to be based.

Prioritize

Because of the complexity and speed of advances in genetics, policymakers will have to **determine which health policy areas to address first**, based on consultation with advisory groups.

Revise Existing or Formulate New Policy

After priorities are identified, **specific policy strategies can be proposed** through open dialogue and debate among advisors and policymakers. In some cases, state policymakers may opt to revise existing laws and regulations; in other cases, formulating new policy may be preferred. The intent of this step is to close gaps and integrate genetics into policies and programs to ultimately advance the policy principles and objectives identified earlier.

When selecting among policy strategies, policymakers may consider the costs, benefits, and effectiveness of the options based on available scientific studies. State data about disease prevention and health improvement needs can help determine whether, for example, a new statewide population screening program is needed or whether the relatively low incidence of the health condition calls for a targeted program of education and testing. Also, diverse and special population groups should be consulted to assess the potential impact of proposed policy and regulations. Broad support from stakeholders, experts, and the public will facilitate adoption and implementation of recommended actions.

Create Performance Objectives

To have a basis for evaluating policy outcomes in the future (see “Evaluation” below), measurable performance objectives should be set. Measurable objectives can be established for public health and medical services, professional and public education programs, and insurance and employment regulations. For example, objectives might be:

- Decrease in preventable health conditions.
- Increase in the number of health professionals who participate in continuing education about genetic testing.
- Increase in the percent of the state insured population with genetic conditions who receive recommended early interventions.
- Quantifiable improvements in public understanding of common genetic concepts.

Before finalizing objectives, a review should assess the logic that links objectives to the policy and a check made of the availability and validity of data sources needed for performance measurement.

Implement Policy

State administering agencies would next implement the policies. They will require sufficient resources to publicize new or revised policy, promulgate regulations, oversee compliance, create systems to capture data relevant to performance objectives, and monitor outcomes of policy implementation.

Evaluate Policy Implementation Against Performance Objectives

Evaluation is an important, and often overlooked, component to any policy process. Pertinent state agencies should create procedures to measure performance objectives (see above) and also solicit qualitative stakeholder feedback. This periodic documentation of outcomes informs policymakers about the extent to which policy objectives are being achieved. Policymakers can then identify any additional revisions to laws or regulations or the need for new policies. Incorporating sunset dates in legislative language or regulatory documents will reinforce the evaluation process.

Informed Consent

This appendix provides only information, not recommendations, about informed consent, including purpose and process.

Informed consent is a comprehensive process, not just a form or piece of paper, which must be signed by a patient or research subject. The process provides the facts, clear alternatives, and anticipated consequences of a test or study that are necessary to make a fully informed, independent decision that is free of coercion. Consent must be voluntary, given only by persons known to be competent,* and based on full, non-directive disclosure. The Secretary's Advisory Committee on Genetic Testing recommended specific informed consent be provided for all genetic tests in clinical and public health practice (with the exception of most newborn screening). Further, the committee recommended practitioners document informed consent for those tests used to predict future risk of disease, as well as for tests advised by a doctor or conducted as part of other hospital tests.⁹⁵

Participants must be accurately and effectively informed (using language that is easily understood and comprehended) about what genetic information is being sought, how it will be used, what will happen to samples when the study is over, and what might be harms and benefits of a test or research. The right of individual to choose to participate or not (*informed refusal*) and to withdraw at any time must be clearly and repeatedly stated. In the case of research studies, the consent process must declare if the donor is to be compensated and disclose proprietary interests of the investigator and institution, the funding source, and any potential commercial use.

The goal is to protect the decision-making autonomy of individuals, prevent harm, and avoid feelings of being cheated, powerless, misled, or betrayed. Informed consent is based on principles of respect for persons, beneficence, and justice. Ethical standards for how consent is achieved should guide disclosure and documentation of the informed consent process. The signed agreement must record the process by which subjects/patients have been informed and describe what measures will be taken to protect subjects from discrimination or stigmatization.

Community Consultation

Medical research on genetic testing and screening interventions affects families and whole communities. Health professionals, boards overseeing research, and community representatives should determine the need for community consultation when data collection could affect many people. Just as individuals must participate in the informed consent process, so must communities be fully informed about tests and studies. The process includes community debate and deliberation to consider the complexities of genetic testing before community decisions are made. The community consultation that precedes community-based research is not optional⁹⁶ and is as important as final community consent.⁹⁷ Persons leading consultations must be cognizant of cultural and language differences. Prior to developing research protocols involving a defined community, it is critical that researchers engage and empower sub-populations to determine through interviews and discussions who can legitimately “speak” for a community.

Additional Protections for Research Subjects

Institutional Review Boards (IRBs) are required for all federally funded human research. Boards are comprised of experts from various disciplines who must review proposals, recommend/require changes, and approve the research. The informed consent process is part of the IRB review. Some states such as Maryland are extending IRB requirements to research funded by the state and private organizations.

Community Advisory Boards (CABs) are formed of representatives from a proposed research community who share a common geography or other identity such as history, symbols, language, or culture. CABs facilitate both individual informed consent (and informed refusal) and community consultation by serving as a two-way communications channel between community and researcher. CABs can assist in recruitment, evaluate risks and benefits, make recommendations to potential participants, monitor the process, identify problems, and disseminate findings. CABs can act as sponsors for research and are an especially important component of community-based prevention research. State and federal funding agencies are calling for more CAB involvement and incorporation into research protocols.⁹⁸

Tissue Trustees are individuals or very small committees responsible for keeping private the identity of research subjects. The trustee functions as a “fire wall” between donor and researcher and communicates between the two. Trustees can set up processes or an infrastructure to balance donor protection with societal benefits resulting from tissue-based research. They can establish ethical guidelines for research and set up a nationwide registry of tissue samples that can be shared.

Certificates of Confidentiality, commonly used in substance abuse research, could be used for genetic research to protect participants’ identities. Current forms of certificates would need to be modified.³⁰

Resources

Office of Human Research Protections, U.S. Department of Health and Human Services, located in the Office of the Secretary of Health and Human Services, ensures the safety and welfare of people who participate in HHS-sponsored research by monitoring research programs; training clinical investigators, members, and staff of institutional review boards; and providing guidance on informed consent. <http://ohrp.osophs.dhhs.gov>

National Bioethics Advisory Commission, created by executive order in 1995, establishes broad principles to govern ethical conduct of research and provides advice and makes recommendations on governmental programs and policies regarding biomedical issues. www.bioethics.gov

Most Americans Want Informed Consent

86% of U.S. adults believe physicians should get informed consent before doing any genetic testing beyond the routine.

93% say written consent must be given for research studies.
(Gallup Poll, 9/00)

* Children cannot provide informed consent. Decisions about genetic testing in children and adolescents must be decided based on the benefits to be derived and the welfare of the child as well as the family and may not be justified unless there are clearly beneficial treatments or effective preventive strategies.⁹⁴

Integrating Genetics into Public Health

This appendix suggests how states might integrate genetics into core public health functions.

In order to protect and advance public health and safety, all states have policies and programs to prevent disease, injury, and disability. In the future, genetics will be incorporated into each of the core public health functions (shown in boldface below) and the associated essential services listed below each function.⁹⁹ Potential applications of genetics are in parentheses.

Assess Health and Hazards

- Monitor health status and hazards to identify and solve community health problems. (Incorporate genetic data in epidemiological studies; track the inappropriate use of genetic information and assess missed opportunities to use genetic services and technologies.)
- Diagnose and investigate health problems and health hazards in the community. (Study diseases resulting from gene-environment interactions.)

Develop Policy

- Inform, educate, and empower people to safeguard their health. (Educate people about the benefits and harms of genetic services and technologies.)
- Mobilize community partnerships to identify and solve health problems. (Include consumers and community representatives in advisory groups with other stakeholders and experts for genetics research or policy review.)
- Develop policies and plans to prevent or mitigate individual and community health problems. (Include social, ethical, and legal considerations when developing genetic policy.)

Provide Assurance

- Enforce laws and regulations that protect health and ensure safety. (Incorporate genetic information into strategies for assuring privacy and confidentiality of personal medical and health information.)
- Link people to needed personal health services and assure the provision of health care when otherwise unavailable. (Provide genetic testing and counseling for high-risk populations that otherwise would lack access.)
- Assure a competent public health and personal health care workforce. (Support training programs for new skills and competencies in genetics.)
- Evaluate the effectiveness, accessibility, and quality of health services in clinical and community settings. (Assure availability of services and competencies in laboratories that analyze genetic tests. Increase availability of individual and group counseling about genetic tests.)
- Conduct prevention research to develop and test interventions to protect population health. (Include genetics and genomics in the research agenda.)

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