



FRAMEWORK FOR PUBLIC HEALTH GENETICS POLICIES AND PRACTICES IN STATE AND LOCAL PUBLIC HEALTH AGENCIES¹

The mission of public health is to “fulfill society’s interest in assuring conditions in which people can be healthy” (Institute of Medicine, 1988). This mission requires state and local public health officials to respond to ever-changing priorities and to ensure that current and future policies and practices are appropriate. It is challenging to achieve and sustain the balance of existing programs with available resources while incorporating new recommendations and technologies. Breakthroughs in human genetics provide great promise for improving the health of the public, but there are significant policy implications and resource needs. It is evident that genetics will become a fundamental component of the policy and practice roles of public health agencies by 2010, making careful consideration of the framework and process for meeting this essential challenge.

Discoveries in genetics are already impacting society’s health in numerous ways. Every day, health professionals and the general public are provided information about exciting discoveries in areas such as cancer, heart disease, and birth defects, creating expectations for better health services. As these expectations evolve, health policymakers will have to determine how and when to make recommendations for incorporating new discoveries into policy and practice and providing adequate financial support. For example, tandem mass spectrometry technology allows newborn screening programs to screen for additional conditions. Currently, states are developing policies to ensure that tests added to newborn screening programs are appropriate.

A larger challenge for state and local public health officials is setting standards for the role of genetics within the broad scope of core public health functions. Performance measures of the efficiency and effectiveness of public health agencies and programs using health outcomes are the gold standards used by health officials to determine priorities. The core functions and essential services of public health are the foundations for these analyses, which use population-based data and proven strategies for considering the relative impact of existing and new interventions and programs. State policymakers depend on these measurements for establishing and sustaining program investments and resource allocation and acquisition. In developing genetics and public health programs, officials will be expected to apply state or local performance measures and outcomes data.

Public health officials may be expected to provide criteria for: 1) using genetic tests to predict the probability of disease and impact of interventions; 2) using genetic screening and services throughout the life span; and 3) preventing inappropriate uses of genetic testing. The ability to measure the impact of these program functions on the prevention of disease will require careful long-range planning. The assessment of the prevalence and incidence of diseases and the appropriate use of genetic testing and screening capabilities will be the responsibility of the state

¹ This document is intended to be a guide for State Health Officials and other health department staff to use for incorporating genetics into health agency programs. The availability of resources and the political climate of the health agency will affect the degree to which each state is able to integrate the framework.

health agency. State policymakers will turn to their State Health Officials to provide guidance concerning the validity and utility of genetic testing and the use of genetic information to improve the public's health without compromising the privacy and economic ability of its citizens.

As genetic tests are developed for particular uses, policies or regulations for oversight and management of laboratory services, clinical services, and genetics services need to be available. The evolving roles and responsibilities of state public health agencies in assuring the incorporation of genetics throughout the public health system, including prevention, education, health promotion, surveillance, and laboratory and clinical services, are outlined in this document. The three core public health functions and the ten essential public health services are used to frame the integration of genetics into public health practices and policies.

Three Core Public Health Functions and Genetics

According to the IOM report *The Future of Public Health*, the goal of public health is to generate an organized community effort to address public concerns about health by applying scientific and technical knowledge (IOM, 1988). While acknowledging that the private sector has a role in promoting health and preventing disease, it is clear that the public sector must provide fundamental building blocks to carry out public health's mission. This sentiment is true for genetics as well. Genetics will offer many opportunities for public and private collaboration, but state health agencies will bear the ultimate responsibility for ensuring that genetics information is integrated into the basic scientific and technical knowledge of public health—the three core public health functions.

Assessment. To improve health, it is important to understand how genetics interacts with other factors. Therefore, it is necessary to regularly collect, analyze, and share information, including genetic information and environmental interactions, related to health conditions, risks, and community resources (Washington State Health Department, 1994). According to the book *Genetics and Public Health in the 21st Century*, surveillance is needed to determine: 1) the population frequency of genetic variants that predispose people to specific diseases, both common and rare; 2) the population frequency of morbidity and mortality associated with such diseases; and 3) the prevalence and effects of environmental factors known to interact with given genotypes in producing disease (Khoury et al., 2000). Establishing criteria for genetic testing recommendations may involve reassessing data using additional vital statistics or other factors. Other factors include the availability of quality genetics resources in the community, the appropriateness of genetics technologies offered to the community, the accessibility of clinical and genetics services, the costs and benefits of using genetics technology, and the community's knowledge of the use of genetics to improve health. This information is necessary for State Health Officials and others responsible for providing health policy guidance to enact policies and programs that are best for their communities.

Policy Development. Sound health policy development requires a combination of scientific guidance and analyses of existing policies, regulations, resources, and strategic priorities. Public health policy aims to improve the health of the community while providing necessary individual protections. Development of good public policies occurs through an informed process that

includes input from a broad-based spectrum of disciplines, professional backgrounds, interest groups, stakeholders, and consumers. Health agency policies underlie priorities for a public health response to identified problems, barriers, and needs such as genetic screening, diagnosis, treatment, and prevention programs. Public health policies also provide members of the public with objective guidance and information to empower them in decision making regarding the use of genetics technologies. Issues such as health insurance discrimination, population screening, and privacy and confidentiality require guidance from State Health Officials to ensure the public's health and minimize potential harm.

Assurance. Public health agencies assure their constituents that services necessary to achieve goals are provided, either by encouraging action by other private or public entities, by requiring such action through regulation, or by providing services directly (IOM, 1988). Agencies may collaborate with other public and private entities and educate public health staff and private health-care workers about the use of genetic information to improve health. Programmatically, the incorporation of up-to-date genetic information in areas such as maternal and child health, occupational health, and disease prevention programs will improve outcomes by providing better prevention information. This information should be available in formats that are appropriate to the target audience in terms of reading level and cultural competence. Enhancement of data systems to include genetic information, with appropriate privacy protections, can be part of ongoing considerations for program improvement. Outcome evaluations that include genetic information will create an opportunity to develop more effective policies and practices. Some health agencies may find it necessary to assure the availability and quality of laboratory and clinical genetics services in their state through licensing and certification activities.

Ten Essential Public Health Services and Genetics

In 1994, the nation's major public health organizations developed and adopted the *Ten Essential Public Health Services* as an enhancement of the core public health functions. The ten essential services are used below to outline the integration of genetics into public health policy and practice, where appropriate, and to identify desired goals.

Monitor health status to identify community health problems. The development and maintenance of a strong health data collection system with the capacity to monitor genetic factors that affect health status and identify health problems within the community is valuable to state public health agencies' efforts to improve the public's health. Population-based data collected through vital statistics systems and ongoing disease surveillance form the basis of monitoring community health status. The inclusion in these databases of genetic information linked to populations and diseases imparts pertinent information for monitoring disease incidence and prevalence. Systems must be capable of capturing clinical and laboratory information within the state generated by public and private services and reporting analyzed data in a useful format. Data collected in these systems could include genetic variants, health status, demographics, interventions, environmental triggers, and safety and efficacy of genetics technologies. The ability of population-based data collection systems to capture associations between genetic and environmental factors and resultant clinical manifestations will expand our understanding of the relationships between these factors and provide new insights into prevention. A first step is to examine existing data sources to identify methods to incorporate

genetics and to assess existing genetic information in surveillance systems, such as the Behavioral Risk Factors Surveillance System and management information systems. Health information systems should collect genetics data as part of overall surveillance and evaluation strategies and be capable of integrating with existing systems.

GOALS:

1. Analyze incidence, mortality, and morbidity data to prevent and reduce the burden of disease and to associate the data with genetic predisposition and environmental triggers.
2. Identify opportunities for including genetic information in existing programs.
3. Develop data collection systems for genetics that can be integrated with existing data systems (e.g., birth defects registries, vital statistics, birth and death certificates, cancer registries, laboratory reporting).
4. Identify genetic information that is currently collected in existing data systems.
5. Identify communities that could benefit from genetic information and interventions.
6. Develop a system for analyzing the validity and utility of genetic tests.

Diagnose and investigate health problems and health hazards in the community. Applied public health research into the causes of health problems, including relevant genetic factors, is key to understanding diseases can be prevented and to reducing their burden in the community. The applications for genetics range from newborn screening to cancer prevention education. State health agencies and environmental agencies, if separate, will need to work together to address environmental factors that may interact with genes to cause negative health outcomes. Genetic information can be used to identify environmental hazards to which individuals may be especially susceptible. This information may be used to reduce avoidable exposures to environmental factors and to modify behaviors to minimize disease. Health agency epidemiologists and social behavioral scientists will need to be capable of incorporating genetic information into their work.

GOALS:

1. Identify genetic risk factors to increase opportunities for early intervention, reduction of disease burden, and primary prevention of disease throughout the life span.
2. Identify environmental elements to which individuals may be particularly susceptible.
3. Develop a health promotion (social marketing) plan that empowers citizens to use genetic information appropriately to reduce their risk of disease.
4. Train personnel to assess genetic factors when investigating environmental health hazards and to create behavior change programs.

Inform, educate, and empower people about health issues. The public and key policymakers need information and education about genetics and its relationship to maintaining good health. Materials used to educate the public should be culturally relevant and made easily available to all populations including underserved populations. Materials are also needed for audiences with low literacy levels and non-English speakers. Social marketing campaigns that include information on the known role of genetics in many diseases empower the public to make better healthcare and lifestyle choices. Individuals that want genetic information about themselves should have the ability to access this information without fear of discrimination to themselves or their families. Educating policymakers and the public about genetics directly impacts the

development of policies that provide necessary protections from the misuse of genetic information.

GOALS:

1. Inform the general public and policymakers about genetics and its impact on health.
2. Provide consistent information through a range of focused health education programs so that informed decisions regarding genetic health issues can be made.
3. Assess community needs for genetic information and services.

Mobilize community partnerships at the state and local levels to identify and solve health problems. The identification of public and private community programs and partners interested in working collaboratively to promote effective and efficient decision making provides for greater understanding about genetics and its contribution to disease prevention and health promotion. Program partnerships with the community provide the basis for broad input on public health issues. Genetic test results have implications not only for the person tested, but also for individuals related to that person. Thus, a single genetic test can have vast implications for a community in which many related individuals reside. To avoid misuse of genetics, community participation in forming genetic policies and practices is necessary. Key community and peer leader members of these partnerships also serve as excellent community informants and can disseminate beneficial genetic information. Partnerships also may focus on securing needed legislation for relevant issues. Partnership members should represent the diversity of the community, be accountable to the community they represent, and have equal levels of participation in decision making.

GOALS:

1. Establish effective communication with community members regarding genetics issues.
2. Establish a committee of accountable community leaders with equal levels of participation in decision making to form genetics policies and practices.
3. Ensure the relevance of genetics policies and programs to the communities they are designed to serve and protect.

Develop policies and practices that support individual and community health efforts. The state health agency is the appropriate body to provide the necessary leadership for the development of public policies and programs that guide the applications genetic information to health promotion and disease prevention. The state health agency must develop and use standards for integrating genetics into public health practices that reflect community values and needs. A strategic planning process can be used to develop a comprehensive plan to incorporate genetics into the activities of the state health agency.

GOALS:

1. Apply population-based genetic information to state policies and programs to improve individual and community health.
2. Develop a strategic plan to guide the integration of genetics into public health practice and policies.

Enforce laws and regulations that protect health and ensure safety. An adequate legislative base and oversight authority for genetic testing and related clinical services is necessary to protect the public from the inappropriate use of genetic information, research, or services. Legislation and regulation regarding genetics should address the effectiveness, accessibility, and

quality of genetic tests and services. Effective legislation establishes guidelines for monitoring compliance and actively enforces statutes and regulations. Issues needing legislative leadership from state health agencies include: prohibitions against insurance discrimination, employment discrimination, and disclosure of genetic/medical information; informed consent requirements; property rights of personal genetic information; and regulation of clinical professions providing genetics services such as counseling and genetic research.

GOALS:

1. Develop legislation, statutes, and regulations that provide for the optimal use of genetic information to improve health, while protecting clients and consumers from the misuse of genetic information.
2. Provide leadership and guidance for public health genetics policies.

Link people to health services, including genetics services, and assure the provision of health care when otherwise unavailable. The availability of appropriate services for preventing and treating disease is fundamental. Where necessary, states may need to establish the capacity for the provision of specific genetics services. By capitalizing on new genetic discoveries, the health agency can provide for more effective and targeted genetics services with greater capacity to improve the public's health. This may include identification of funding sources to provide individual services and to ensure that qualified personnel and facilities are available and accessible to the public. Effective services are community-based and culturally sensitive, and they are able to refer individuals to mainstream health-care providers for genetics services. These services include those aimed at prevention, health education, primary care, and specialty services.

GOALS:

1. Create provisions for high-quality, culturally competent genetics services for those who need or desire them.
2. Ensure that high-quality, clinically valid genetic tests are available.
3. Develop genetic information and services that are culturally competent and effective in improving health.

Assure a public health and personal health care workforce competent in genetics. Current and future health professionals will need training and skills development in the appropriate use of genetic information to promote health and prevent disease. Individuals graduating from schools of public health will need genetics knowledge in order to function up to agency standards and be competitive in the public health workforce. Partnerships with academic institutions may provide mutually beneficial opportunities for educating the public health workforce about genomics. Academia has a vested interest in providing its students with practical experience and connections to employment opportunities. Health agencies need employees with a sound understanding of health promotion, including the role of genetics in health promotion. There will be a growing need for continuing education opportunities for public health professionals in this area. The state health agency may also wish to work with professional organizations to ensure that all health-care providers, especially primary care providers, have continuing education opportunities in genetics and continuing education credit for participation in those programs. Public health genetics competencies have been developed

by the Centers for Disease Control and Prevention in partnership with state public health agency representatives in the following areas, administration, laboratory, environmental health, health education, clinicians, and epidemiology.

GOALS:

1. Create and maintain a public health workforce that is competent in public health genetics.
2. Provide opportunities for the current public health workforce to obtain continuing education in genetics.
3. Create opportunities for continuing education credit for all health professionals in genetics when possible.
4. Prepare current public health students to participate in programs that incorporate genetic information to promote health.

Evaluate effectiveness, accessibility, and quality of personal and population-based health services, including genetics. A system is needed to provide ongoing evaluation of the impact of genetic information and the effectiveness, accessibility, and quality of genetic tests and population-based health services. Quality of services, personnel, cultural competency, and use of surveillance and population-based epidemiological studies are important components of evaluation. Genetic tests will need to be evaluated based on their analytical validity and clinical validity and utility prior to any considerations for population-based genetic testing. The health outcomes of individuals who participated in genetics services should be evaluated to determine the effectiveness of these services in improving health. Ongoing monitoring of the utilization of genetics services also is necessary to develop a comprehensive evaluation of the impact of genetics on public health. Communication and information dissemination will be necessary to provide timely and accurate information to the general public and professionals in order to enhance their basic knowledge about genetics, genetic screening, counseling, and comprehensive services.

GOALS:

1. Assure the availability and accessibility of up-to-date genetics programs, services, tests, and treatments.
2. Conduct outcomes evaluation of available genetics services to determine their effectiveness.
3. Review and evaluate information related to the clinical utility and validity of genetics tests.

Research for new insights and innovative solutions to health problems. There are numerous studies that examine the link between genes and disease and provide insight into reducing the occurrence, morbidity, and mortality of disease. The findings must be analyzed through a public health lens to determine when they should be incorporated into public health practice. The social, economic, and ethical implications of research findings will need special emphasis in determining the benefit of incorporating genetics into public health.

GOALS:

1. Identify and assess genetics research findings to determine the appropriateness of incorporating them into public health practices.

2. Assess the social, economic, and ethical impact of this information in determining its appropriateness for public health.
3. Ensure that genetic information is continually updated and incorporated into the public health infrastructure.

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Appendix A

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(Extracted from http://www.astho.org/templates/display_pub.php?pub_id=453, July 21, 2003)

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Public health officials may be expected to provide criteria for: 1) using genetic tests to predict the probability of disease and impact of interventions; 2) using genetic screening and services throughout the life span; and 3) preventing inappropriate uses of genetic testing. The ability to measure the impact of these program functions on the prevention of disease will require careful long-range planning. The assessment of the prevalence and incidence of diseases and the appropriate use of genetic testing and screening capabilities will be the responsibility of the state health agency. State policymakers will turn to their State Health Officials to provide guidance concerning the validity and utility of genetic testing and the use of genetic information to

improve the public's health without compromising the privacy and economic ability of its citizens.

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Assessment: To improve health, it is important to understand how genetics interacts with other factors. Therefore, it is necessary to regularly collect, analyze, and share information, including genetic information and environmental interactions, related to health conditions, risks, and community resources (Washington State Health Department, 1994). According to the book *Genetics and Public Health in the 21st Century*, surveillance is needed to determine: 1) the population frequency of genetic variants that predispose people to specific diseases, both common and rare; 2) the population frequency of morbidity and mortality associated with such diseases; and 3) the prevalence and effects of environmental factors known to interact with given genotypes in producing disease (Khoury et al., 2000). Establishing criteria for genetic testing recommendations may involve reassessing data using additional vital statistics or other factors. Other factors include the availability of quality genetics resources in the community, the appropriateness of genetics technologies offered to the community, the accessibility of clinical and genetics services, the costs and benefits of using genetics technology, and the community's knowledge of the use of genetics to improve health. This information is necessary for State Health Officials and others responsible for providing health policy guidance to enact policies and programs that are best for their communities.

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health response to identified problems, barriers, and needs such as genetic screening, diagnosis, treatment, and prevention programs. Public health policies also provide members of the public with objective guidance and information to empower them in decision making regarding the use of genetics technologies. Issues such as health insurance discrimination, population screening, and privacy and confidentiality require guidance from State Health Officials to ensure the public's health and minimize potential harm.

Assurance: Public health agencies assure their constituents that services necessary to achieve goals are provided, either by encouraging action by other private or public entities, by requiring such action through regulation, or by providing services directly (IOM, 1988). Agencies may collaborate with other public and private entities and educate public health staff and private health-care workers about the use of genetic information to improve health. Programmatically, the incorporation of up-to-date genetic information in areas such as maternal and child health, occupational health, and disease prevention programs will improve outcomes by providing better prevention information. This information should be available in formats that are appropriate to the target audience in terms of reading level and cultural competence. Enhancement of data systems to include genetic information, with appropriate privacy protections, can be part of ongoing considerations for program improvement. Outcome evaluations that include genetic information will create an opportunity to develop more effective policies and practices. Some health agencies may find it necessary to assure the availability and quality of laboratory and clinical genetics services in their state through licensing and certification activities.

Ten Essential Public Health Services and Genetics

In 1994, the nation's major public health organizations developed and adopted the *Ten Essential Public Health Services* as an enhancement of the core public health functions. The ten essential services are used below to outline the integration of genetics into public health policy and practice, where appropriate, and to identify desired goals.

1. Monitor health status to identify community health problems: The development and maintenance of a strong health data collection system with the capacity to monitor genetic factors that affect health status and identify health problems within the community is valuable to state public health agencies' efforts to improve the public's health. Population-based data collected through vital statistics systems and ongoing disease surveillance form the basis of monitoring community health status. The inclusion in these databases of genetic information linked to populations and diseases imparts pertinent information for monitoring disease incidence and prevalence. Systems must be capable of capturing clinical and laboratory information within the state generated by public and private services and reporting analyzed data in a useful format. Data collected in these systems could include genetic variants, health status, demographics, interventions, environmental triggers, and safety and efficacy of genetics technologies. The ability of population-based data collection systems to capture associations between genetic and environmental factors and resultant clinical manifestations will expand our understanding of the relationships between these factors and provide new insights into prevention. A first step is to examine existing data sources to identify methods to incorporate genetics and to assess existing genetic information in surveillance systems, such as the

Behavioral Risk Factors Surveillance System and management information systems. Health information systems should collect genetics data as part of overall surveillance and evaluation strategies and be capable of integrating with existing systems.

GOALS:

- a. Analyze incidence, mortality, and morbidity data to prevent and reduce the burden of disease and to associate the data with genetic predisposition and environmental triggers.
- b. Identify opportunities for including genetic information in existing programs.
- c. Develop data collection systems for genetics that can be integrated with existing data systems (e.g., birth defects registries, vital statistics, birth and death certificates, cancer registries, laboratory reporting).
- d. Identify genetic information that is currently collected in existing data systems.
- e. Identify communities that could benefit from genetic information and interventions.
- f. Develop a system for analyzing the validity and utility of genetic tests.

2. Diagnose and investigate health problems and health hazards in the community:

Applied public health research into the causes of health problems, including relevant genetic factors, is key to understanding diseases can be prevented and to reducing their burden in the community. The applications for genetics range from newborn screening to cancer prevention education. State health agencies and environmental agencies, if separate, will need to work together to address environmental factors that may interact with genes to cause negative health outcomes. Genetic information can be used to identify environmental hazards to which individuals may be especially susceptible. This information may be used to reduce avoidable exposures to environmental factors and to modify behaviors to minimize disease. Health agency epidemiologists and social behavioral scientists will need to be capable of incorporating genetic information into their work.

GOALS:

- a. Identify genetic risk factors to increase opportunities for early intervention, reduction of disease burden, and primary prevention of disease throughout the life span.
- b. Identify environmental elements to which individuals may be particularly susceptible.
- c. Develop a health promotion (social marketing) plan that empowers citizens to use genetic information appropriately to reduce their risk of disease.
- d. Train personnel to assess genetic factors when investigating environmental health hazards and to create behavior change programs.

3. Inform, educate, and empower people about health issues: The public and key policymakers need information and education about genetics and its relationship to maintaining good health. Materials used to educate the public should be culturally relevant and made easily available to all populations including underserved populations. Materials are also needed for audiences with low literacy levels and non-English speakers. Social marketing campaigns that include information on the known role of genetics in many diseases empower the public to make better healthcare and lifestyle choices. Individuals that want genetic information about themselves should have the ability to access this information without fear of discrimination to themselves or their families. Educating policymakers and the public about genetics directly impacts the development of policies that provide necessary protections from the misuse of genetic information.

GOALS:

- a. Inform the general public and policymakers about genetics and its impact on health.
- b. Provide consistent information through a range of focused health education programs so that informed decisions regarding genetic health issues can be made.
- c. Assess community needs for genetic information and services.

4. Mobilize community partnerships at the state and local levels to identify and solve health problems: The identification of public and private community programs and partners interested in working collaboratively to promote effective and efficient decision making provides for greater understanding about genetics and its contribution to disease prevention and health promotion. Program partnerships with the community provide the basis for broad input on public health issues. Genetic test results have implications not only for the person tested, but also for individuals related to that person. Thus, a single genetic test can have vast implications for a community in which many related individuals reside. To avoid misuse of genetics, community participation in forming genetic policies and practices is necessary. Key community and peer leader members of these partnerships also serve as excellent community informants and can disseminate beneficial genetic information. Partnerships also may focus on securing needed legislation for relevant issues. Partnership members should represent the diversity of the community, be accountable to the community they represent, and have equal levels of participation in decision making.

GOALS:

- a. Establish effective communication with community members regarding genetics issues.
- b. Establish a committee of accountable community leaders with equal levels of participation in decision making to form genetics policies and practices.
- c. Ensure the relevance of genetics policies and programs to the communities they are designed to serve and protect.

5. Develop policies and practices that support individual and community health efforts:

The state health agency is the appropriate body to provide the necessary leadership for the development of public policies and programs that guide the applications genetic information to health promotion and disease prevention. The state health agency must develop and use standards for integrating genetics into public health practices that reflect community values and needs. A strategic planning process can be used to develop a comprehensive plan to incorporate genetics into the activities of the state health agency.

GOALS:

- a. Apply population-based genetic information to state policies and programs to improve individual and community health.
- b. Develop a strategic plan to guide the integration of genetics into public health practice and policies.

6. Enforce laws and regulations that protect health and ensure safety: An adequate legislative base and oversight authority for genetic testing and related clinical services is necessary to protect the public from the inappropriate use of genetic information, research, or

services. Legislation and regulation regarding genetics should address the effectiveness, accessibility, and quality of genetic tests and services. Effective legislation establishes guidelines for monitoring compliance and actively enforces statutes and regulations. Issues needing legislative leadership from state health agencies include: prohibitions against insurance discrimination, employment discrimination, and disclosure of genetic/medical information; informed consent requirements; property rights of personal genetic information; and regulation of clinical professions providing genetics services such as counseling and genetic research.

GOALS:

- a. Develop legislation, statutes, and regulations that provide for the optimal use of genetic information to improve health, while protecting clients and consumers from the misuse of genetic information.
- b. Provide leadership and guidance for public health genetics policies.

7. Link people to health services, including genetics services, and assure the provision of health care when otherwise unavailable: The availability of appropriate services for preventing and treating disease is fundamental. Where necessary, states may need to establish the capacity for the provision of specific genetics services. By capitalizing on new genetic discoveries, the health agency can provide for more effective and targeted genetics services with greater capacity to improve the public's health. This may include identification of funding sources to provide individual services and to ensure that qualified personnel and facilities are available and accessible to the public. Effective services are community-based and culturally sensitive, and they are able to refer individuals to mainstream health-care providers for genetics services. These services include those aimed at prevention, health education, primary care, and specialty services.

GOALS:

- a. Create provisions for high-quality, culturally competent genetics services for those who need or desire them.
- b. Ensure that high-quality, clinically valid genetic tests are available.
- c. Develop genetic information and services that are culturally competent and effective in improving health.

8. Assure a public health and personal health care workforce competent in genetics:

Current and future health professionals will need training and skills development in the appropriate use of genetic information to promote health and prevent disease. Individuals graduating from schools of public health will need genetics knowledge in order to function up to agency standards and be competitive in the public health workforce. Partnerships with academic institutions may provide mutually beneficial opportunities for educating the public health workforce about genomics. Academia has a vested interest in providing its students with practical experience and connections to employment opportunities. Health agencies need employees with a sound understanding of health promotion, including the role of genetics in health promotion. There will be a growing need for continuing education opportunities for public health professionals in this area. The state health agency may also wish to work with professional organizations to ensure that all health-care providers, especially primary care providers, have continuing education opportunities in genetics and continuing education credit for participation in those programs. Public health genetics competencies have been developed

by the Centers for Disease Control and Prevention in partnership with state public health agency representatives in the following areas, administration, laboratory, environmental health, health education, clinicians, and epidemiology.

GOALS:

- a. Create and maintain a public health workforce that is competent in public health genetics.
- b. Provide opportunities for the current public health workforce to obtain continuing education in genetics.
- c. Create opportunities for continuing education credit for all health professionals in genetics when possible.
- d. Prepare current public health students to participate in programs that incorporate genetic information to promote health.

9. Evaluate effectiveness, accessibility, and quality of personal and population-based health services, including genetics: A system is needed to provide ongoing evaluation of the impact of genetic information and the effectiveness, accessibility, and quality of genetic tests and population-based health services. Quality of services, personnel, cultural competency, and use of surveillance and population-based epidemiological studies are important components of evaluation. Genetic tests will need to be evaluated based on their analytical validity and clinical validity and utility prior to any considerations for population-based genetic testing. The health outcomes of individuals who participated in genetics services should be evaluated to determine the effectiveness of these services in improving health. Ongoing monitoring of the utilization of genetics services also is necessary to develop a comprehensive evaluation of the impact of genetics on public health. Communication and information dissemination will be necessary to provide timely and accurate information to the general public and professionals in order to enhance their basic knowledge about genetics, genetic screening, counseling, and comprehensive services.

GOALS:

- a. Assure the availability and accessibility of up-to-date genetics programs, services, tests, and treatments.
- b. Conduct outcomes evaluation of available genetics services to determine their effectiveness.
- c. Review and evaluate information related to the clinical utility and validity of genetics tests.

10. Research for new insights and innovative solutions to health problems: There are numerous studies that examine the link between genes and disease and provide insight into reducing the occurrence, morbidity, and mortality of disease. The findings must be analyzed through a public health lens to determine when they should be incorporated into public health practice. The social, economic, and ethical implications of research findings will need special emphasis in determining the benefit of incorporating genetics into public health.

GOALS:

- a. Identify and assess genetics research findings to determine the appropriateness of incorporating them into public health practices.

- b. Assess the social, economic, and ethical impact of this information in determining its appropriateness for public health.
- c. Ensure that genetic information is continually updated and incorporated into the public health infrastructure.

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APPENDIX B

GENETIC STAKEHOLDER ADVISORY GROUP SUMMARY

To initiate the stakeholder's advisory input process, an introductory session was held in September, 2003, that provided the invited members with background as to the goals of the Genetics Planning Project, and began to identify issues germane to the group's charge. One month following this session, the next in the planned series of genetics advisory meetings was held.

To facilitate a thorough discussion of the potential impact of genomics on public health, a hypothetical public health situation was developed to which the genetics stakeholders' group responded on October 13, 2003. The group addressed directed questions in six major public health areas of concern that included Infrastructure Development, Science, Service, Education, Partnerships, and Policy-making (**Stakeholders' Hypothetical & Stakeholder' Reponses; Appendix**). On November 11, 2003, the group met again to review the responses from the previous meeting and to generalize the responses into a set of recommendations. The meeting discussion points were recorded, and are summarized below.

Infrastructure Development

A small, on-going Genomics Advisory Body should be created that oversees the incorporation of genetics into the DPH infrastructure and that coordinates genomics plans within the state. Group members should be chosen so that efforts in the five general areas identified, and outlined below are coordinated. Members should include representatives from industry (insurance and biotechnology), healthcare, and epidemiology. Scientific experts, bioethicists, educational specialists, and consumer representatives should also sit in the group, and it should be inclusive of diverse ethnic groups. Individuals from within the committee may be called to chair workgroups as needed, in science, service, or education.

In addition to this external advisory body, an internal team of individuals representing diverse units within DPH should be created, call the "Gene Team." This group of individuals should evaluate the ways in which genomics can be incorporated into their programs, and should also evaluate the impact of potential genetics programs on activities within their unit.

A dedicated position, a director of genomics, should be established, and should chair both the Genomics Advisory Body, and the internal Gene Team. It is strongly recommended that the Body's structures and the position of a genomics director be formalized to ensure a stable, enduring integration of genomics into DPH infrastructure. In addition to working with the Genetics Advisory Group and the internal Gene Team, the genetics director should forge a healthy relationship with the existing newborn Genetics Advisory Committee (or GAC), which oversees the newborn screening program within DPH.

In addition, the Genomics Advisory Body should be created to advise the Commissioner on topics related to genetics. This committee will be involved in any activities that ensure the steady incorporation of genetics into DPH activities. The

advisory body will also be involved in making recommendations for the genomics director to, among other duties, chair their group.

General genetics education is a priority, and the most important groups that should be initially targeted for education are healthcare workers, public health workers, legislators, and school age children. One of the first activities overseen by the Genetics Advisory Body should be the development of educational opportunities in genetics to these targeted groups. The Genetics Advisory Body should oversee development of an educational process that is self-sustaining. Additional subsequent populations identified for such educational efforts include the general public, and affected/potentially affected family members, particularly those with diminished or unequal access to genetics information and/or services.

There are many types of genetics programs that could be considered by DPH for implementation, and the steps needed to evaluate and initiate any specific program will require specific methods. The steps outlined below in the areas of science, service, partnerships, and policy should all be considered before initiating any new program in genetics. In addition, educational activities specific to a genetics program under consideration should be considered.

Science

When needed to evaluate potential genetics programs or to develop monitoring protocols for existing programs within the state, an *ad hoc* science workgroup should be created, reportable to the overseeing Genomics Advisory Body. The group should be composed of scientists who can evaluate the scientific merit of a genetic program under consideration, chaired by a representative of the standing Genetics Advisory Body. The workgroup should evaluate the potential impact of any genetics program with methods that include risk/benefit analysis and the potential implications of initiating any program in genetics. The panel should also consider evaluations of a potential program from a variety of sources, including those from national panels and other states.

Having high quality data to monitor any potential program in genetics is important. The nature of the needed data, however, is expected to be specific to any potential genetics program, and should be considered before initiating any genetics program. Birth, death, cancer, and hospital discharge data are expected to play an important role in any surveillance system. The science workgroup should identify data useful for monitoring any specific genetics program, and should identify needed data. Baseline data should be collected before any program in genetics is initiated. It is possible that some types of data may need to be declared “reportable,” and it is also possible that multi-state regional efforts can be used to gather the needed data. Consortia with schools of public health should be considered to collect the information needed to monitor any genetics program.

Service

A variety of services can be envisioned for any genetics program. Examples include offering genetics tests, coordinating short-term or long-term treatment, coordinating genetics counseling, or providing resources or education. The Department may also be

involved in monitoring quality control. Before any program in genetics is initiated, an *ad hoc* service workgroup should be created. The workgroup, chaired by a member of , and reporting to, the overarching Genetics Advisory Body, will identify the type of specific service to be offered by DPH that is appropriate for the genetics program under consideration. The workgroup will assess the ability of the current healthcare workforce to meet the needs of the public, and steps to close those gaps should be performed before any new genetics program is initiated. In addition, the workgroup should evaluate whether services are to be offered by the state DPH or by local departments of health.

If services related *genetic testing* is under consideration, the criteria for testing will depend on the specific genetic test under consideration, and whether or not it will be mandatory. The test could be offered to the general population or to select high-risk populations. Consideration of any genetic test should be sensitive to the perceived needs of the public.

If coordination of genetic testing services is under consideration, then the Department should consider workforce development of individuals sensitive to those cultures expected to be served. Genetics counselors and genetic service providers should be available to serve the needs of the public from varying cultures.

If resources and education related to genetic testing are under consideration, then successful programs such as that used within the HIV division of DPH should be considered as models for any genetics program.

In the current newborn screening program, DPH is involved in testing, tracking, and treatment. This comprehensive set of services may not be appropriate for the general life-span population. It is also generally not considered appropriate for DPH to involve itself directly in long-term treatment.

As services are considered for a potential genetics program, the needs of a diverse community should be considered. Such consideration should include the active participation of insurance companies, and should also include participation by HUSKY and Medicaid providers. If genetics tests are considered for a potential genetics program, reimbursement of genetics counselors should be considered, both for pre-test and post-test counseling.

Education

As mentioned above, genetics education is considered paramount to any genetics program, and general education in genetics should ideally be initiated well in advance of any considered program. Education in genetics should be sustained throughout any genetics state plan. A large variety of groups should be considered for educational opportunities in genetics, and should include public health professionals, schools of public health, schools of law, the judicial system (lawyers, judges, legislators), health care professionals, schools of nursing and medicine, public and private schools, and insurance companies. Educational opportunities should also be offered to the public, consumers, and affected families. Of the groups identified for genetics education, healthcare and public health professionals, legislators, and school-aged children (grades K-12) are considered the highest priority subpopulations. An Education workgroup that reports to the Genomics Advisory Body, should be created that addresses the educational

and curricular needs of the state. It should include representatives from the Department of Education, Department of Higher Education, Schools of Public Health, Medical and Nursing Schools, Universities and Colleges, and other organizations involved in education within the state. The workgroup should be involved in the following activities:

1. Offer internet-based educational programs and distance learning programs; some programs may already exist and could be accessed regionally;
2. Include of a few sentences in existing DPH brochures that discuss the role of genetics in health to target the general population and consumers;
3. Make health and science curricular changes in K-12 that incorporate genetics topics;
4. Develop curricular changes in professional schools (medicine, nursing, public health, and law);

It is important to monitor any trends in public awareness of genetics. On-going polls that monitor public opinion and general knowledge about genetics should be initiated by the Education workgroup, and should be sustained throughout the duration of the state plan. Possible approaches to monitoring public opinion include: telephone polls, questionnaires at seminars, surveys at meetings, mailings to targeted populations. Methods should avoid selection bias, and opinions should be solicited from the lay public. Methods should also be culturally sensitive. Questions in the existing BRFSS survey are one avenue in which the public's awareness of, and attitudes toward, genetics can be assessed. Added questions to other existing, on-going surveys should be considered.

A Speakers' Bureau that can address a variety of audiences should be compiled by the Education workgroup. It should include potential speakers with general and specific knowledge of genetics, and should also include educators at a variety of levels. The Bureau should be involved in the following events:

1. Offer workshops with CME credits and grand rounds in hospitals to target healthcare professionals;
2. Offer talks at high schools that target high school seniors;
3. Offer public-access Genetics 101 workshops and question/answer opportunities at public libraries and other public forums;
4. Access science writers and programmers through the media.

If educational opportunities are needed to address a specific genetics program, the target population should be identified, and experienced, trusted, culturally sensitive teachers should be identified and organized who can educate that target population.

Partnerships

To be effective, a comprehensive genetics plan must involve collaborations with diverse groups within the state, as well as among the Northeastern states. Groups with which DPH should develop partnerships include:

Department of Education, Department of Higher Education, Schools of Public Health, Medical and Nursing Schools, Local Health Departments, Universities and Colleges, National consumer support groups, nonprofit organizations, well-informed legislators, members of the CT Public Health Committee, healthcare providers and insurance

companies, science writers, medical and professional societies, industry groups such as Pfizer, worship organizations, and the Connecticut Public Health Association.

To be efficient, regional programs that break down state-centered programs should be pursued, allowing inter-agency cooperation, and avoiding duplication of efforts. Attempts should be made to pool resources.

Policy-Making

Among the policy issues related to potential programs in genetics, informed consent and discrimination are among the most prominent. The state should promote laws at the federal level that help ensure informed consent and prevent discrimination, such as that recently approved by the US Senate, and that ensure services. These issues should also be addressed by the Genetics Advisory Group. Before initiating any potential program in genetics, the need for legislation and the implications of that legislation should be evaluated. Decisions should be in concert with current information, which should, in turn, integrate information from science and service. Potential legislation should weigh the benefit to the individual *versus* society.

If the issue under consideration includes *informed consent*, the process of informing should include pre-test and post-test evaluation. The potential impact of genetics information on the individual should be evaluated, and the amount of information needed for the public to make informed decisions, should be assessed. For instance, the potential impact of a positive carrier state for cystic fibrosis should be considered relative to that of a drug toxicology screening. Different methods of delivering informed consent should be studied to develop that which is most effective.

Besides regulations needed to stabilize the infrastructure of genetics within DPH, regulation needed to implement any genetics program should be considered. Mandated data needed to monitor a genetics program, and legislation that protects individuals from the consequences of data collection should be considered.

Genetic tests are already regulated by the state, but additional resources may be required to regulate new genetic tests. Among the needed resources is funding, and legislation may be required to enable additional tests.

APPENDIX D PUBLIC HEALTH GENETICS SYMPOSIUM BREAKOUT SESSIONS - SUMMARY

During attendance to an all day symposium on Public Health Genetics, participants, who included professionals throughout the state in genetics, were given the opportunity to respond to questions directed at a public health hypothetical situation (**Symposium Hypothetical; Appendix**). Participants chose one of five breakout sessions, which were chairs by members of the Stakeholders' Advisory Committee and other genetics professionals throughout the state, and discusses a series of directed questions related to one of four areas: Public Policy, Surveillance, Services, or Education. Notes of each session were recorded (**Symposium Responses; Appendix**). At a joint session following the breakout sessions, co-facilitators presented summaries of each breakout, and these highlights are listed below.

Public Policy

Question: How can equal, culturally sensitive access to testing, screening, and genetic counseling be assured?

- Equal, culturally sensitive access can be addressed through legislation, but assurance will require physician, community, state, family support.
- Cultural competencies should be encouraged.

Question: Should the test protocol be regulated?

- Testing and screening protocols can be regulated through state and legal means.
- It is important to maintain protocols that assure testing accuracy.
- Counseling protocols need to be case-specific, considering the genetic structure, disease, and interventions.

Question: How can we ensure confidentiality and privacy of test results, including giving information to family members?

- Laws are in place to hold employers accountable not to discriminate – but legal power may not assure protection.
- Following the current model of newborn screening, results are not given to insurers or employers. Mechanisms are already in place to assure confidentiality – the same should be applied.
- George Annas recommends higher standards for genetics tests than for other diagnostic test results.

Surveillance

- *Define the role of the state: Science versus information dissemination versus resource development.*
- *Prevalence measures: Make use of outside data and utilize newborn blood spots.*
- *Genetic testing: Assure benefit and validity prior to use.*
- Gather data to define high risk target populations at multiple entry points into health care system, such as at birth, entry into school, etc. - include family history.

Services

Question: At what age should people be tested, and how can information be provided to the family to ensure informed consent?

- Prevalence and penetrance of the gene mutation needs to be known to 1) better assess the public health benefit of mass screening or 2) identify those with positive family history subgroup.
- It might be best to target people with positive family history.
- There is a need to know at what age the intervention might have best impact.

Question: Once a test is performed, what type of services might be needed for children, adults and seniors, and should the state monitor the quality of these services?

- Multiple services are needed at multiple points in life - birth, point of independence, marriage/family planning. How will the adult be 're-informed?'
- Caution: The burden on genetic counseling may increase – other family members may want to be tested.*
- Culturally diverse staff are needed.
- The state should monitor quality, and also perform cost-benefit analysis (balancing the effectiveness of intervention with financial implications and increased anxiety).

How can we ensure access to genetic counseling services?

- Nurses are underutilized, and could be used. Peer-educators could also be used.
- Availability of genetic counseling should be increased in non-traditional settings within the community (e.g., church, schools, community centers)

Education

Question: Who needs this information?

The following should receive education:

- The Public, including consumers;
- Health care professionals;
- Legislators;
- Insurers;
- Home care agencies.

Question: How do we deliver this information?

Information could be delivered through:

- Media commercials, using celebrities with genetic conditions, and possibly financed by pharmaceutical companies;
- Family Resource Centers or Parent-Teacher Organization, using consumers such as a parent or a child with the genetic disease.
- Novel Idea: Provide grandparents with the tools to develop their family health history as a gift for their progeny.*
- Health care professionals with CME credits that include genetics as part of the topic. Also, genetics could be integrated into their topics at hand.