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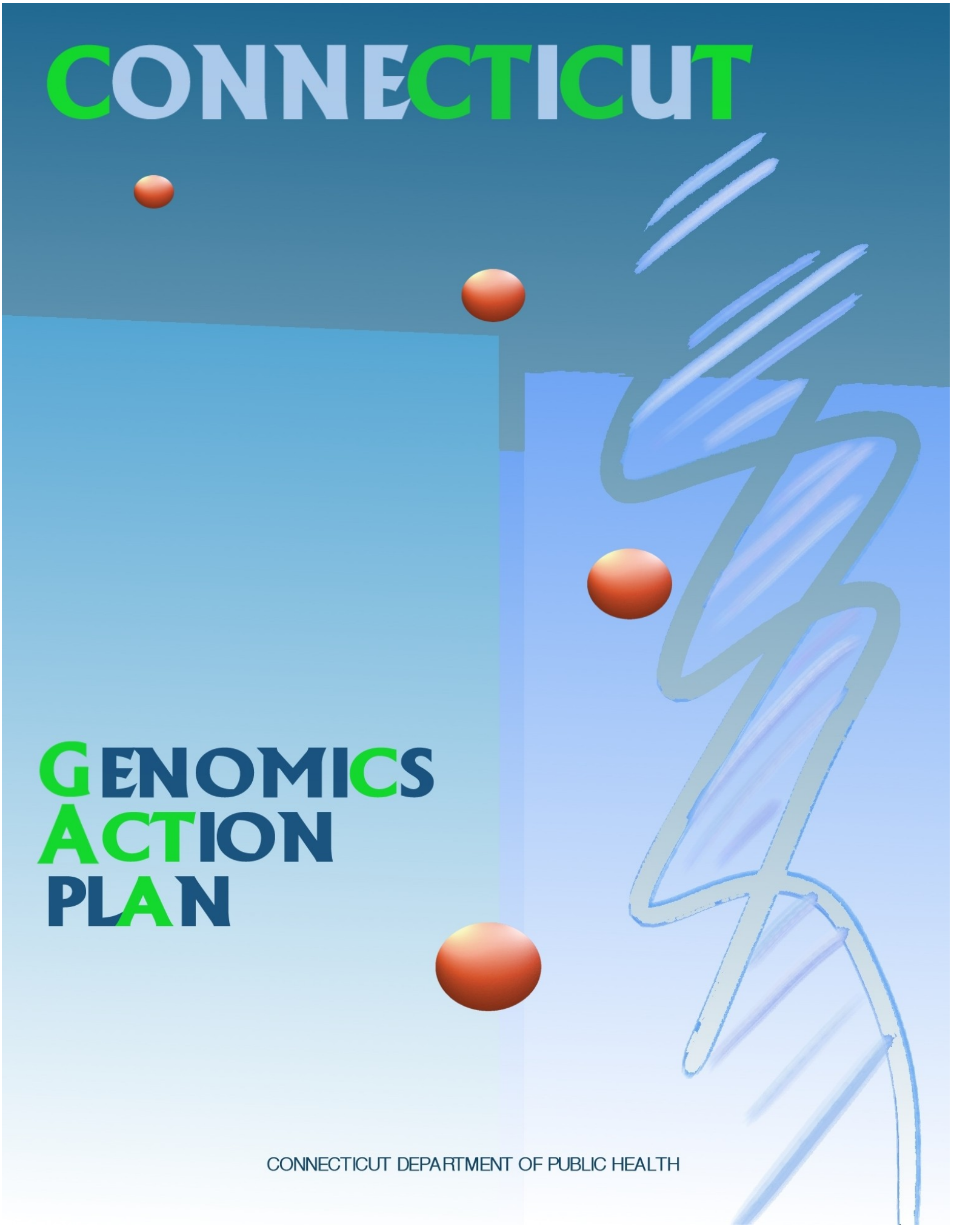

GENOMICS ACTION PLAN

CONNECTICUT DEPARTMENT OF PUBLIC HEALTH

CONNECTICUT GENOMICS ACTION PLAN
July 2005

An electronic version of this Plan is available on the Internet at:

www.dph.state.ct.us

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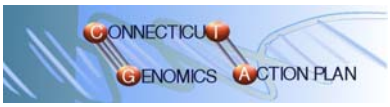


Executive Summary

Rapid technological advances in the field of genetics and the potential impact of these advances on population health make this area a high priority for public health consideration and planning. We now understand that most human disease results from an underlying genetic susceptibility that is modified by environmental factors. From a public health perspective, the term "genomics" is being used to describe the study of how an individual's genes interact with each other and with the environment in order to identify their influence on health and disease. Public health genetics is expanding beyond issues related to birth defects and rare genetic disorders detected through newborn screening. There is increasing evidence of the relationship between genomics and chronic diseases, including asthma, cancer, diabetes, and heart disease. Environmental health is changing as more is learned about individual susceptibility to environmental exposures, such as cigarette smoke and toxic chemicals. The study of HIV/AIDS, SARS, tuberculosis, and other infectious diseases has also been influenced by the advancement of genomics.

Recognizing the increasing role that genomic discoveries will play in disease detection, prevention, and treatment, the Connecticut Department of Public Health (DPH) embarked on a multi-year planning process to assess statewide genetic service needs and to develop a CT Genomics Action Plan ("Plan") to address those needs. Funding for this project was provided by a federal grant from the Maternal and Child Health Bureau (MCHB) of the Health Resources and Services Administration. Implementation of the Plan will help DPH reach national Healthy People 2010 objectives as well as MCHB Title V Block Grant state and core objectives.

The planning process was coordinated by a DPH Genetics Planning Team ("Team") with guidance from an external Genetic Stakeholders Advisory Group. Multiple strategies were employed to inform the process. For example, input was solicited via surveys from general medical practitioners, genetics professionals, and families affected by genetic conditions in Connecticut. Feedback was also obtained during several educational workshops and a symposium in public health genetics. During these sessions, attendees were privileged to receive insight from nationally recognized genetics experts, including Lori Andrews, J.D., Distinguished Professor of Law at Chicago-Kent College of Law and Director of the Institute for Science, Law, and Technology; Wylie Burke, M.D., Ph.D., Chair, Department of Medical History and Ethics, University of Washington School of Medicine; Neil Holtzman, M.D., M.P.H., Director of Genetics and Public Policy, Johns Hopkins University; Muin Khoury, M.D., Ph.D., Director, Office of Genomics and Disease Prevention, Centers for Disease Control and Prevention; Sharon Krag, Ph.D., Professor, Associate Dean of Graduate Education and Research, Johns Hopkins University, Bloomberg School of Public Health; and Bradford Therrell, Jr., Ph.D., Director, National Newborn Screening and Genetics Resource Center and Professor, Department of Pediatrics, University of Texas Health Science Center.

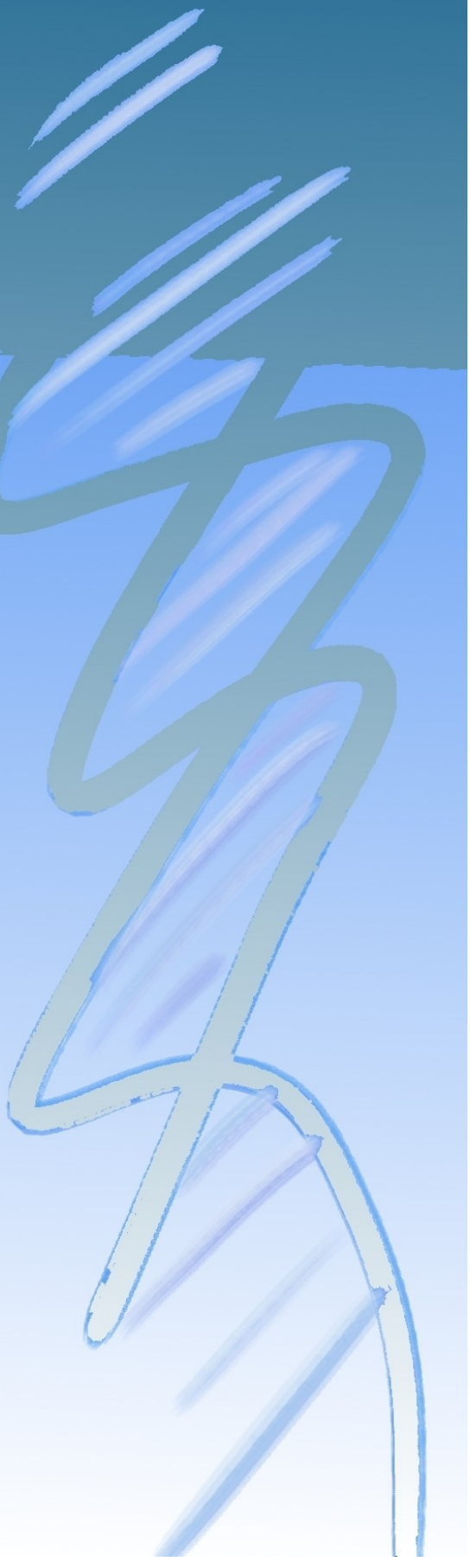
The Plan provides an overview of the history and current directions of genomic science and services in Connecticut. It identifies demographic changes that are likely to affect future genetic service delivery. The Plan assesses public health's role in data integration of children's health information systems, genetic health services availability and accessibility, genetics education, workforce development, and genetic health policy development.

Based on the findings, the CT Genomics Action Plan was developed to guide genetic-integration activities of DPH during the next 5 years. To achieve the goals of the Plan, an adequately funded genomics program with agency-wide visibility is needed to provide leadership and assurance that Connecticut's residents will benefit appropriately from genomic advances.

Key Findings:

- ★ Traditional public health activities associated with genetics have pertained primarily to newborn screening and reproductive health, but genes play a role in the development of disease across the lifespan. There is a need for DPH to continue to broaden its activities to consider the impact of genomic advances on chronic diseases, infectious diseases, environmental health, and epidemiology.
- ★ Ongoing demographic changes in Connecticut will continue to affect the health care needs and delivery of services in the state. Trends, such as the aging population, growing ethnic diversity, childbearing at older ages, and socioeconomic disparities, need to be considered when determining future genomic service needs.
- ★ There is a need to develop a child health information system within DPH to assure that Connecticut's children are receiving comprehensive and coordinated health care, particularly those children and youth identified as having special health care needs.
- ★ There is a need to assure that a competent genetics workforce is available to meet the growing demands of genetic testing in Connecticut.
- ★ Barriers remain for accessing genetic services, including an uneven distribution of trained providers and lack of insurance reimbursement for services.
- ★ There is a need to enhance public understanding of genetics and the impact of genetics on health.
- ★ There is a need to coordinate state and federal genetics policy issues and to address community concerns about informed consent, genetic privacy and discrimination.

I. INTRODUCTION



Introduction

> *Genomics will be to the 21st Century what infectious disease was to the 20th Century for public health. It has the potential to change our thinking. Genomics should be considered in every facet of public health: infectious disease, chronic disease, occupational health, and environmental health, in addition to maternal and child health.*
>
> S. Gerard, M. Hayes, and M.A. Rothstein
> *J Law Med Ethics; 2002* Fall; 30 (3 Suppl):173-176

The sequencing of the human genome and other advances in genetic science are expected to provide new insights into the complex, interactive roles that genetic and environmental factors play in morbidity and mortality. This knowledge holds great promise for improving the public's health and for preventing disease, which is the primary responsibility of the Connecticut Department of Public Health (DPH). In its 1999 report, Looking Toward 2000 - An Assessment of Health Status and of Health Services, DPH recognized the advances that are occurring in molecular medicine and the need to incorporate them into its public health activities beyond newborn screening and reproductive services (Connecticut Department of Public Health, 1999). Planning for this effort has been made possible through federal funding from the Maternal and Child Health Bureau (MCHB) of the Health Resources and Services Administration (Grant No. 6 H46 MC 00192-02-02). Implementation of the recommendations developed in this plan will help DPH reach some of the national Healthy People 2010 objectives as well as various MCHB Title V Block Grant state and core objectives.

The planning process was coordinated by a DPH Genetics Planning Team ("Team") with guidance from a Genetic Stakeholders Advisory Group, a broad-based group of genetic professionals and family representatives. The Team used the "ten essential services of public health" (U.S. Department of Health and Human Services, 1994) and the MCHB four-tier pyramid model of essential components of a public health system (Maternal and Child Health Bureau, 2000) as a framework for the planning process.

This planning document is divided into nine chapters. Chapter I introduces the planning process. Chapter II looks at technological advances that are occurring in the field of genetics and the potential impact of genetic discoveries on the public's health across the lifespan. Chapter III presents an overview of the State and identifies demographic changes that are likely to affect future genetic service delivery. Chapter IV identifies health information data systems in DPH that may be enhanced by genomic information in the future and indicates the need for continued data integration. Chapter V looks at genetic health care services in Connecticut with an emphasis on services provided by DPH, such as newborn metabolic and hearing screening and case ascertainment and referral of children with special health care needs. Chapter VI deals with workforce development; educational activities for health care professionals, public health professionals, and the general public; available support groups; and financing mechanisms. Chapter VII addresses ethical, legal, and social issues, and public policy implications associated with the appropriate use of genetic technologies and information derived from genetic testing. At the end of chapters II - VII are concluding remarks that highlight the findings and recommendations that arise from identified needs. Chapter VIII describes the multiple strategies the Team used to obtain input for the plan, including surveys, educational workshops, and establishment of

internal and external stakeholders groups. Finally, Chapter IX provides recommendations and actions required to achieve those recommendations to integrate genomics into DPH public health activities. As a result of the Human Genome Project, there is a recognition that the study of single genes and their effects, i.e., genetics, is shifting toward genomics, defined as the study of the functions and interactions of all genes in an organism (Centers for Disease Control and Prevention, 2004). Genomics has also been described from a public health perspective as "the study of the sum of the gene-environment-host interaction that leads to disease – or disease prevention – in populations" (Patrick, 2002). The nature versus nurture debate is giving way to the view of nature and nurture. We now understand that most human disease is a result of complex interactions between genetic susceptibilities and environmental factors.

We use the term genomics in this document as noted above but recognize that genomics is also defined as the simultaneous analysis of the full complement of human DNA from an individual. Further, the term genomics can also be used to describe high-throughput technologies in which thousands of genes are examined in a given setting. We have elected to incorporate the term genomics in the title of this plan in order to convey the shift beyond single-gene disorders.

Genomics is relevant to many areas of public health. Traditional public health activities associated with genetics have pertained primarily to newborn screening and reproductive health. Through the activities of this project, DPH has already begun to align its activities to consider the genomic impact on chronic diseases, infectious diseases, environmental health, and epidemiology. However, a substantial commitment and much work remain. New policy constructs are needed to assure the safety and effectiveness of genetic tests and their appropriate use in clinical and public health practice. Additional policies are needed to protect the confidentiality of genetic information and prevent it from being used to discriminate or stigmatize, and to ensure equitable access to genetic services throughout the population. There is also a need to educate health professionals, health policy-makers, and the public about genetic technologies and information. Infrastructure development, from reliable data to a workforce that is competent in genomics, is necessary to ensure that genetic knowledge and technology are used to improve the health of Connecticut's citizens.

The CT Genomics Action Plan is intended to be a dynamic document that will evolve over time as new findings occur and as DPH priorities change. It serves as a foundation for advancing a new vision for the role of genetics in public health beyond maternal and child health programs. This new vision is a challenging one. It involves monitoring knowledge gained from genomics and facilitating where possible additional population-based research, deliberating the relevance of technologic advances to medical and public health practice, and, when appropriate, developing intervention programs. Yet implementation of recommendations is within reach with commitment, leadership, and additional resources.

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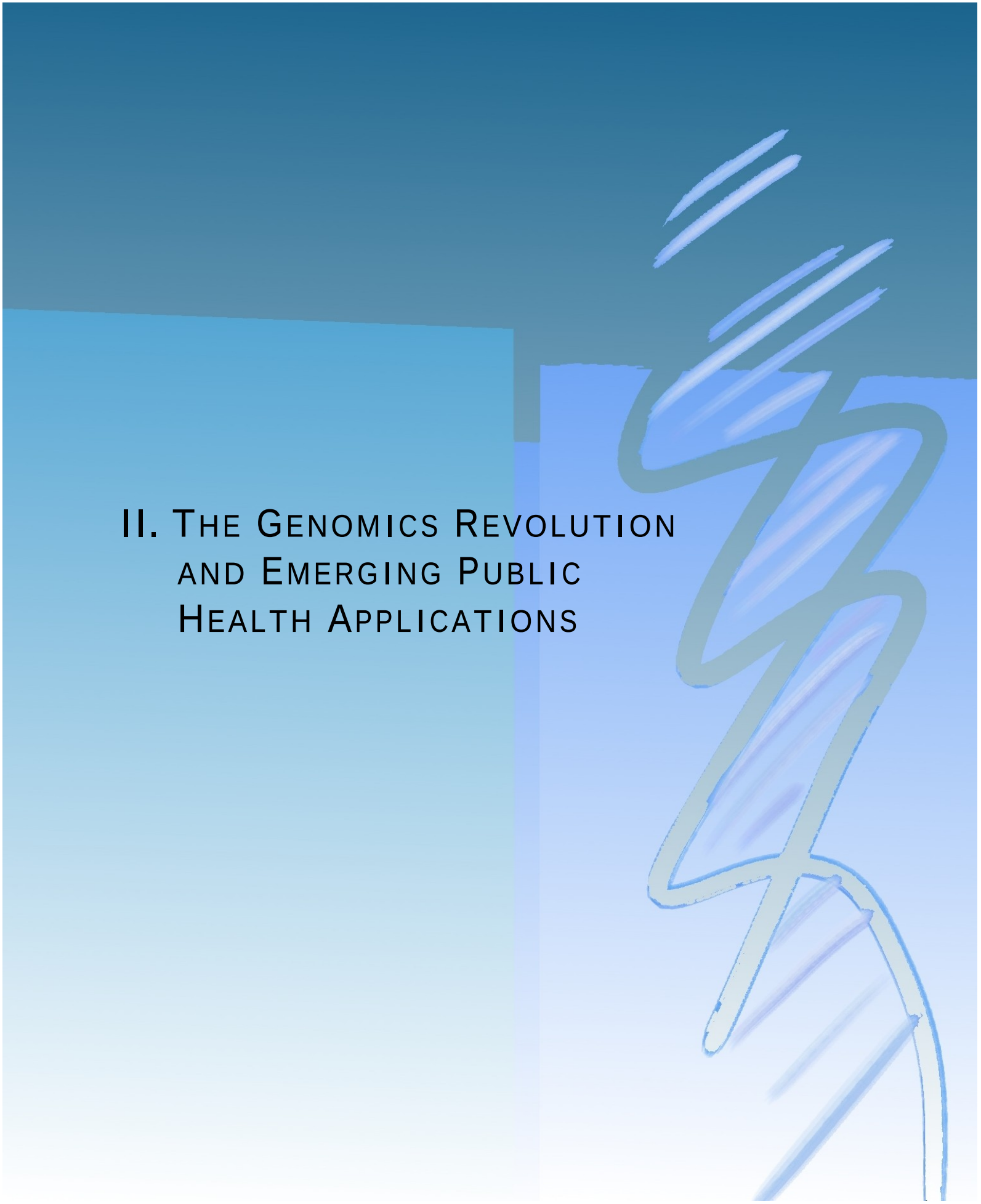

II. The Genomics Revolution and Emerging Public Health Applications

The Genomics Revolution and Emerging Public Health Applications

Introduction

The recent completion of the sequencing of the human genome and other advances in genetics and related technologies are expected to create significant changes in health care. Within this context there is new genetic information relevant to most health care and public health programs and across most diseases. The challenge facing DPH is to first determine the relevance of these genetic discoveries to public health practice and, when appropriate, integrate them into disease prevention and management strategies.

Human Genome Project

In 1977 scientists began the ambitious task of mapping, isolating, and decoding every gene in the human genome. Impetus to this endeavor arose with the establishment of the Human Genome Project (HGP). The HGP was an international research effort formally begun in October 1990 to determine the complete chemical sequence of the 3 billion base pairs of human DNA and to identify all of the genes in the human genome. The Project had a controversial history, the competitive nature of which fortuitously accelerated the project. Years ahead of schedule, working drafts of the human genome sequence were published in special issues of *Nature* (February 15, 2001) and *Science* (February 16, 2001). On April 14, 2003 the Human Genome Project was declared officially complete. The HGP was finished two and a half years ahead of time and cost $2.7 billion in 1991 dollars, which was significantly less than original spending projections. A major surprise from the Project was finding that the human genome consists of fewer than 35,000 genes, which is one third of the number previously predicted. Ongoing research has refined the number of protein-encoding genes to be in the range of 20,000-25,000 (International Human Genome Sequencing Consortium, 2004). Such results, however, do not lessen the remaining task of determining how genes function and interact.

There are still numerous obstacles to understanding the linkages between genes and diseases. The fact that genes can code for more than one different protein product, the specific functioning of proteins, the multitudinous interactions of genes, and the complexity of environmental factors affecting gene expression imply that much work remains to be done. Unclear is how humans develop and function, how human development and functioning varies from person to person, and how such variation produces disease and affects drug treatment.

It has long been known that simple monogenic diseases (caused by a single gene), such as β-thalassemia, often are not clinically homogeneous (Weatherall, 2000). Patients with the same genetic defect may show remarkably different clinical conditions ranging from mild to profound anemia, even though they have the same disease. Such clinical variability is apparently due to other modifying genes, along with environmental factors. What was thought to be a relatively simple monogenic disease is really a complex syndrome. This means that understanding multigenic disorders will be even more difficult with heterogeneity at multiple loci.

Impact of the Human Genome Project

The HGP is full of promise for the public's health. The potential exists for finding new ways of diagnosing, treating, and preventing disease by studying disease at the genomic rather than genetic level. Rather than just looking at single genes in isolation, genomics views all the genes as a dynamic system and tries to determine how they interact and influence biological processes and physiology.

Determination of biological basis of disease

Although familial inheritance patterns can be used to infer that certain disorders may have a genetic component, "gene discovery" methods can identify the biological basis of conditions. Thus the sequencing of the human genome should improve our understanding of disease mechanisms leading to a new classification of disease based on molecular mechanisms. The genetic definition of disease may even force a reassessment of what it means to have a disease.

Diagnosis and predictive testing for genetic disorders and susceptibility to disease

Knowledge of whether a person has a mutated version of a gene may be used to diagnose a genetic disorder in an individual or during a pregnancy, or it may be used to test an individual for a specific disorder prior to any symptoms being present. Pre-symptomatic testing will make medicine more predictive and consequently more preventative with appropriate interventions. It is this application of genetics that may have the most relevance to public health. One such example deals with genetic testing for breast cancer, which is discussed below under applications to DPH programs.

New treatments and improvements in drug efficacy

Gene Therapy

Knowledge of the molecular basis of disorders offers hope for treatments that can correct malfunctioning genes or the proteins for which they code. Gene therapy is one possible gene-based strategy, in which the gene is used as medicine. A carrier vehicle, such as a virus, is used to deliver a therapeutic copy of a gene to the patient's target cells. The generation of a functional protein product from the therapeutic gene restores the target cell to a normal state.

Gene therapy has been used with mixed results to treat X-linked severe combined immunodeficiency disease (X-SCID), commonly known as the "bubble boy" syndrome (Hacein-Bey-Abina et al., 2002). In January 2003, the U.S. Food and Drug Administration (FDA) suspended all gene therapy trials using retroviral vectors in blood stem cells after two children in gene therapy trials for X-SCID developed a leukemia-like condition.

Beginning in August 2003, the first of twelve patients approved by the FDA for participation in a phase I clinical trial received gene therapy for severe Parkinson's disease (Barclay,2003). A synthetic gene called GAD was injected into the patient's brain inside an engineered virus known as an adeno-associated virus (AAV), which transfers the gene from cell to cell. Unlike retroviruses, AAV has not been associated with an oncogenic event. Inside the brain, GAD produces a chemical called GABA, which is critical for maintaining normal motor activity. After a year, the first patient appears to have benefited significantly, but it's still too early to draw conclusions about the effectiveness of the

treatment. The hope is that gene therapy will prove to be safe and efficacious, and it will replace other surgical approaches, not only for Parkinson's disease, but also epilepsy and other neurologic disorders.

Although gene therapy research is continuing, its use raises ethical considerations. For example, what is normal and what is a disability and who decides? Do disabilities need to be cured? Because gene therapy is costly, who should have access to its use and who will pay for its use? The debate will continue as the research evolves and the public develops greater knowledge and expectations.

Drug Efficacy and Pharmacogenomics

Genetic testing can also be used to improve drug efficacy by identifying those who may respond well to a drug and those for whom a poor response or an adverse effect is likely. Traditionally pharmacogenomics refers to the general study of all the many different genes that determine drug behavior. Pharmacogenetics, a subset of pharmacogenomics, refers to the study of inherited differences in drug metabolism and response. The two terms are now being used interchangeably.

Understanding an individual's genetic makeup is important for tailoring drugs with potentially greater efficacy and safety. Instead of the standard trial-and-error method of matching patients with the right drugs, analysis of a patient's genetic profile may enable physicians to prescribe the most efficacious drug therapy with the appropriate dosage, and eliminate the likelihood of adverse drug reactions. For example, researchers have found that people with particular genetic variants (i.e., DNA polymorphisms) in enzymes that break down the blood-thinning drug warfarin are more likely to suffer serious bleeding if they take the medication (Higashi, 2002). Screening for such genetic variants may allow clinicians to develop dosing protocols to reduce the risk of adverse drug reactions in patients receiving warfarin or to advise that patients take a different blood-thinning drug. Another study has found that cholesterol-lowering drugs are less effective for some people because of a genetic variation in the HMG-CoA reductase gene, which governs cholesterol synthesis (Chasman, 2004). Study author Daniel Chasman writes that "there may be promise in the concept of 'personalized medicine' and the use of genetic screening to target certain therapies. Future studies must determine whether this difference [in effectiveness] can be offset by dose adjustment or the choice of an alternative therapy."

Interest has arisen in therapies based on mechanisms that target critical molecular pathways of tumors. Herceptin is a breast cancer drug designed for the 25% of breast cancer sufferers whose tumors carry multiple copies of the HER2 gene. The drug works by blocking certain genetic signals, thereby preventing the growth of cancerous cells. Another drug, Gleevec, is for leukemia patients who have a genetic variation that causes an overproduction of white blood cells. Like Herceptin, Gleevec is targeted to cancer cells with specific mutations. Challenges remain, however, for targeting the many common solid tumors with no obvious critical molecular driver (Green, 2004).

The continuing advances in molecular technology and information present extensive public health opportunities for understanding and promoting health, lowering mortality and morbidity, and preventing diseases.

HapMap Project

With the completion of the Human Genome Project come new challenges, some of which have already begun to be tackled. For example, in October 2002, an international research consortium launched the International "Haplotype Mapping" or HapMap Project. The project is a $135 million, three-year

effort to produce a map of common human genetic variations aimed at speeding the discovery of genes that contribute to cancer, diabetes, heart disease, asthma and many other common conditions. To create the HapMap, DNA will initially be taken from blood samples from people in Nigeria, Japan, China, and the United States. By comparing genetic differences among individuals, researchers expect to identify which differences are related to disease.

Because genetic variation has been shown to affect the response of patients to drugs, toxic substances and other environmental factors, mapping an individual's haplotype may also be used to help customize medical treatment. In addition, the HapMap may help pinpoint genetic variations that may contribute to good health, such as those that protect against infectious diseases or those that promote longevity.

As of the end of February 2005, the first draft of the HapMap reached completion with the mapping of one million markers of genetic variation, called single nucleotide polymorphisms (SNPs). Phase II of the project intends to improve upon the HapMap by adding data on an additional 2.25 million SNPs. The second phase will provide researchers with a denser map that should enable them to more precisely narrow gene discovery to specific regions of the genome.

Applications to DPH Programs

Infectious Diseases

Infectious diseases are a leading cause of morbidity and mortality in humans. Although improvements in water and food sanitation and the use of vaccines and antibiotics have made significant improvements in the control of infectious agents, vaccines have not yet been developed for many infectious agents, or the infectious agents acquire resistance to antibiotics, making them a continuing problem. Technological advances have provided new tools with which to study infectious agents, the diseases they cause, and the hosts' responses to infections. For example, identification of the SARS virus occurred in a matter of weeks aided by large, complex gene arrays, whereas identification of the AIDS virus took 2.5 years. Such rapid identification helped to speed efforts to diagnose, treat, and prevent the spread of SARS.

Genetic variation among humans appears to affect human susceptibility to some common infectious diseases (Cooke, 2001). For example, the rate at which individuals infected with the human immunodeficiency virus progress to AIDS and the probability of a person latently infected with TB developing TB disease differs among individuals. Human genetic susceptibilities have also been associated with reportable infectious diseases such as meningitis (Corvini et al, 2004).

A large number of genes appear to influence susceptibility to disease from infectious pathogens. Efforts are being made to identify genes that may modulate diseases such as malaria, tuberculosis, viral hepatitis, SARS, and HIV/AIDS. Identification of these genes may provide insights into pathogenic and protective mechanisms, identify new molecular targets for preventive and therapeutic interventions, and lead to more effective screening programs.

Current DPH infectious-disease reporting systems enable rapid detection of disease outbreaks; the goal is to ascertain cases quickly and to initiate appropriate control measures. In this era of heightened concern about bioterrorism, such reporting systems become even more critical to develop and maintain. Ongoing surveillance systems of diseases such as HIV/AIDS, hepatitis, and tuberculosis provide data that are used to identify populations who may be at increased risk for infectious diseases,

so that interventions, such as training and public education programs, can be employed to reduce the occurrence of such diseases.

As new infectious diseases emerge and persist, research pertaining to human and pathogen genomics and interactions promises to be a valuable weapon in the fight to control such diseases. As such, it is important for DPH to stay abreast of such developments.

Environmental Health and Toxicogenomics

Just as genetic variation among humans may affect susceptibility to common infectious diseases, genes may also influence susceptibility and vulnerability to other environmental health hazards, such as cigarette smoke, alcohol, and toxic chemicals. Environmentally associated diseases include cancer, pulmonary diseases, neurodegenerative disorders, developmental disorders, birth defects, and autoimmune diseases. In addition, the toxic effects of high internal doses of heavy metals such as lead and mercury may be linked to genetic susceptibilities (Lidsky, 2003; Godfrey, 2003).

Recent technological advances in human genomics have opened the door to the new field of toxicogenomics, which combines toxicology, genetics, molecular biology, and environmental health in an effort to study the response of living organisms to stressful environments. The goal of toxicogenomics is to find correlations between toxic responses to toxicants and changes in the genetic profiles of those exposed to such toxicants. Application of this knowledge will be used to enhance understanding and therapeutic management of human diseases caused by environmental pollutants or toxicants.

On April 16, 2003, the National Institute of Environmental Health Sciences (NIEHS) announced the completion of the first phase of the Environmental Genome Project (EGP) to characterize genes that confer susceptibility to environmental agents (National Institute of Environmental Health Sciences, 2003). The EGP has identified and re-sequenced 200 environmentally-responsive genes, identifying links to vascular disease, leukemia, prostate cancer, and other conditions. The second phase of the project will involve functional analysis of the various polymorphisms occurring in regulatory regions of genes. The third phase of the project will involve the development of animal models for use in studies of how environmental agents interact with specific polymorphisms to cause human disease. Altogether the EGP plans to re-sequence 554 environmentally-responsive genes identified by the NIEHS scientific community.

As part of the federal Children's Health Act 2000, approval was given for the National Children's Study (NCS), which will follow 100,000 children from before birth to age 21 to study how genetic and environmental factors affect the health and development of children (National Children’s Study, 2004). Federal agencies involved in this initiative include the National Institute of Child Health and Human Development, the National Institute of Environmental Health Sciences, the Centers for Disease Control and Prevention, and the Environmental Protection Agency. It is anticipated that preliminary results will be available in 2008-2009.

The DPH Environmental and Occupational Health Assessment Program evaluates health risks posed by environmental exposures so that appropriate interventions can be implemented. Research results from future gene-environment studies can be used to develop public health strategies that are targeted to help individuals avoid or reduce adverse exposures. In addition, policies can be developed to ensure that contaminant standards set for air, water, and food quality are set to protect not only the

general population, but also those who might be more susceptible to exposures due to their genetic make-up.

Chronic Diseases

The majority of non-infectious diseases arise from complex interactions of multiple genes and environmental exposures. That is, most gene variants do not confer increased risk of disease independently, rather disease risk increases due to combinations of specific alleles from several genes or when triggered by an environmental stimulus. Therefore, prevention of most chronic diseases is likely to require a more thorough understanding of their genetic and environmental causation. Research in human genetic susceptibility to many chronic diseases is rapidly growing for conditions such as testicular and ovarian cancer (Hemminki, 2004), childhood cancers (Stiller, 2004), Alzheimer's disease (Rocchi, 2003), osteoarthritis (Aigner, 2003), autoimmune disorders and type 1 diabetes (Vaidya, 2004), and hemochromatosis (Beutler, 2003). These diseases may play an increasing role in public health. Below is a discussion of three diseases that currently have a significant public health impact -- heart disease, breast cancer, and type 2 diabetes.

Coronary Heart Disease

Coronary heart disease (CHD) is the leading cause of mortality and hospitalization in Connecticut and contributes heavily to the overall burden of disease. The only inherited condition to predict the disease with nearly 100% certainty is homozygous familial hypercholesterolemia, a genetic condition with a prevalence of 1 case per 1 million people in the U.S. The vast majority of CHD, however, is the result of environmental influences alone, or a combination of genetic and environmental factors. Genetic mutations have been associated with risk factors for CHD, including lipid metabolism and transport, high blood pressure, and elevated blood plasma homocysteine levels (Nabel, 2003). Thus, predicting CHD is complex and will require much more research. Until more information becomes available, DPH will continue its surveillance activities and promotion of healthy lifestyles to prevent heart disease. In the absence of genetic information, family history may be used as a surrogate and could be incorporated into the Behavioral Risk Factor Surveillance System administered by DPH.

Female Breast Cancer

Breast cancer is the most common cancer diagnosed among women in Connecticut and the United States. It is the second leading cause of cancer-related death for females in Connecticut. In addition, Connecticut has the second highest incidence of breast cancer in the nation. Therefore, identification of breast cancer genes is a priority to enable the identification of individuals at high risk and to aid in the design of more effective therapies for breast cancer chemoprevention and treatment. Two important genes, known as breast cancer susceptibility genes 1 and 2 (BRCA1 and BRCA2), have already been identified. The BRCA genes are involved in DNA repair. In those with BRCA mutations, the repair process is defective. The lifetime chance of developing breast cancer for women who have BRCA mutations are substantially higher than are the rates for the general population. A recent study indicates that the lifetime risk of breast cancer for Ashkenazi Jewish women with BRCA mutations is as high as 82 percent (King, 2003). Even so, having an altered BRCA gene does not mean that one will get breast cancer, nor does it indicate at what age cancer may develop or how aggressively the disease might progress.

Mutations in BRCA1 and BRCA2 genes account for less than 1% of all breast cancer cases. There may be other genes associated with increased risk for breast cancer. About 5% to 15% of women with breast cancer report having a first degree relative with breast cancer. This implies that most breast cancer cases are nonhereditary and are the result of acquired mutations. Acquired mutations arise from either random mistakes when cells are in the process of cell division or from damage due to environmental agents. These agents can be endogenous (e.g., reproductive hormones) or exogenous (e.g., cigarette smoke). It is the interaction between these genes and environmental factors that determines an individual's overall breast cancer risk. Breast cancer, in most cases, is thus a complex multifactorial disease.

Genetic testing for breast cancer is not yet amenable to population screening because most people with a family history of breast cancer do no have a defective BRCA1 or BRCA2 gene. Testing for BRCA gene mutations among family members of a woman with a defective BRCA gene is an important tool for risk assessment and for developing management strategies. In the meantime, DPH continues to promote early detection through regular mammograms and to monitor research about possible behavioral risk factors, such as alcohol consumption and physical inactivity. DPH monitors such activities using its Behavioral Risk Factor Surveillance System. DPH also works with the Connecticut Cancer Partnership and its implementation plan to achieve cancer control in Connecticut.

Type 2 Diabetes

Type 2 diabetes is a serious metabolic disorder in Connecticut, affecting approximately 6% of the state's population in Connecticut, of who nearly one-third have not been diagnosed. It is characterized by defects in both insulin secretion and action. Formerly it was known as non-insulin-dependent diabetes or adult-onset diabetes because it most often occurred after the age of 40. Genetic involvement in type 2 diabetes is under research, but progress has been limited. It appears that various mutations in multiple genes may contribute to the overall risk. Until more information becomes available, the Diabetes Prevention and Control Program in DPH will continue with its current diabetes surveillance system and its promotion of diabetes control through diet and exercise.

Concluding Remarks

The Human Genome Project has laid the foundation for a revolution in understanding the role that genes play in human health and disease. Identifying and understanding gene functions and genetic variation has the potential to improve health through better diagnosis, targeted treatments, and identification of reduced efficacy of medications, or in other words, through personalized medicine. Disease prevention could also be revolutionized through assessing susceptibility to disease-causing environmental and infectious agents. Prevention may consist of (1) environmental interventions, such as improved air quality; (2) behavioral or lifestyle interventions, such as smoking cessation, healthy eating, and exercise; (3) diagnostic interventions, such as newborn screening or mammography; (4) chemoprevention, in which medication is given to reduce the risk of developing a specific cancer or in which preventive antibiotics for tuberculosis are only given to those who would predictably benefit from them; (5) prophylactic removal of target organs, such as mastectomy or oopherectomy; and (6) gene therapy.

However, identification of disease-causing mutations is likely to have a greater impact on Mendelian disease and some complex diseases than on others. Merikangas and Risch note that for certain diseases, such as type 2 diabetes or AIDS, resources might be better placed in environmental or

behavioral interventions that can have a major impact on public health rather than in gene-hunting (Merikangas, 2003).

Nevertheless, scientific knowledge about genetics is expanding beyond the maternal-child-health arena. Information is growing about genetic involvement in chronic, adult-onset conditions. Consequently genetic discoveries ought to be considered regarding public health activities related to common chronic diseases such as heart disease, cancer, and diabetes. Contributing to this need is the aging population and the fact that older people suffer more chronic diseases. DPH should maintain its surveillance systems that monitor these disorders and their risk factors, and seek ways to incorporate information gained from genetic advances.

There is also a need for DPH to broaden its disease-prevention activities to consider the impact of genomic advances on infectious diseases and environmental health related to human susceptibility to infectious agents and environmental exposures.

In order to expand genetics-related activities beyond newborn screening within DPH, a genomics program of high visibility and agency-wide influence is recommended. In addition, internal support for genomics issues needs to be cultivated in an effort to integrate genomics throughout the agency. Establishment of an internal working group is seen as a vehicle for information dissemination and advocacy within the Department, and work in this area has commenced through the development of an internal "Gene Team".

The Gene Team currently is composed of almost thirty members, representing a wide cross-section of departmental programs such as asthma, chronic disease, infectious disease, environmental and occupational health, tumor registry, newborn screening, and so on. It is anticipated that to enable the further development and ongoing facilitation of this important departmental internal resource, the Team would need to have the oversight of a dedicated Genomics Coordinator. Under the direction of an experienced genomics leader, the Gene Team could convene to continue planning for the integration of genetics within DPH by identifying genetics developments that have direct application to public health practice and, thus, on the work of the various programs within DPH.

As indicated in the DPH report, *Genetic Testing: A Plan for the Future* (Connecticut Department of Public Health, August 2000), "the GAC (Genetic Advisory Committee) and the DPH Sections of Laboratory Services, Family Health, Infectious Diseases, AIDS and Chronic Diseases, and Environmental Health will collaborate to develop a plan for testing for infectious and chronic diseases." Such collaboration is important for DPH to pursue.

Recommendations for integrating genetics into state health departments have been put forth in the December 2000 report, *Integrating Genetics into State Chronic Disease Programs*, from the Association of State and Territorial Chronic Disease Program Directors' Genetics Retreat (ASTCDPD, 2000). Although the recommendations are specific to chronic disease programs, they could be generalized to other departmental programs. Such recommendations include, but are not limited to, the following:

- Identify a state genetics coordinator who can interact with chronic disease program professionals, who can act as a resource of genetics information, who can advise on policy development, and who can analyze issues surrounding ethical, legal, and social implications of integrating genetics and public health practice.
- Include genetics in the agency's state health plan.
- Build the genetics competencies of health agency professionals.

- Work to incorporate genetics questions into the Behavioral Risk Factor Surveillance System.
- Expand the core data infrastructure of chronic disease programs to incorporate genetic information from population-based research and program services.

DPH is collaborating with federal agencies that are developing genetics activities that go beyond maternal and child health. For instance, DPH, in partnership with the Department of Environmental Protection, has received a grant from the Centers for Disease Control and Prevention to track chronic diseases and explore environmental exposures that could be linked to them. This Action Plan points to ways to foster more collaborations in the future.

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III. Overview of the State of Connecticut

Overview of the State of Connecticut

Introduction

As one of the wealthiest and best-educated states in the nation, Connecticut's unique population is likely to have a significant impact on the availability and use of genetic services and resources. Connecticut ranks nationally as follows:

- Twenty-ninth in population (3.4 million people).
- Third in median household income ($56,803, in 2003 inflation-adjusted dollars).
- Fourth in percent of population with a bachelor's degree or more (34.6%).
- Fourth in personal health care expenditures per capita ($4,656, in 1998).

Other demographics that play a role in health care in Connecticut include:

- Twenty-two percent of state residents are Hispanic, Black, or other minority group.
- Women of childbearing age (15-44) compose about twenty-one percent of the population.
- Twenty-one percent of the population consists of children under the age of 15.
- Ten percent of the population is uninsured.

Geography of Connecticut

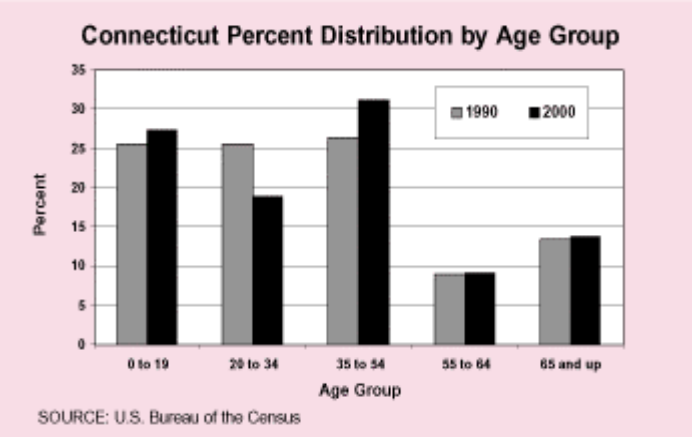
Connecticut is New England's second smallest and southernmost state. Its 5,009 square miles are bordered by the states of New York on the west, Massachusetts to the north, and Rhode Island on the east, and by Long Island Sound on the south. The southerly flow of the Connecticut River divides the state roughly in half. The coastal plain along the Sound and the central river valley are relatively flat and they contain most of the larger cities, including Bridgeport, New Haven, Hartford, and Stamford. Three major interstates cross the state. Interstate 95 runs along the southern coast parallel to the Sound. Interstate 91 runs north-south through the river valley, and I-84 traverses the state diagonally from southwest to northeast. Many of the State's acute-care hospitals are located along these interstates.

Population Growth and Diversity

During the decade of the 1990's, Connecticut's population grew a modest 3.6 percent to 3.4 million, making it the 29th most populous state in the nation. Connecticut has a population that is older, more educated, and more diverse. The median age rose a dramatic nine percent from 34.4 in 1990 to 37.4 in 2000, making it the seventh oldest state in the nation. The United States showed a similar trend but to a lesser degree (32.9 in 1990 to 35.3 in 2000). The increase was fueled mainly by the entry of the "baby boom" generation (those born from 1946 to 1964) into the 45-to-54-year-old age group. This trend is expected to continue into the foreseeable future thereby accelerating the growth of the elderly population (65 and over) (Prisloe, 2002).

The number of women 35 years of age and older, a group who routinely use prenatal genetic health care services, increased by 18 percent over the last decade.

Figure 1. Connecticut Population Distribution by Age Groups



Connecticut's growing racial and ethnic diversity is also reflected in the 2000 census. All racial categories of the non-white population have increased, as have the Hispanic or Latino population. Hispanics have overtaken non-Hispanic blacks as the state's largest minority group. Currently about 22% of Connecticut residents belong to a racial or ethnic minority, and projections indicate that by the year 2025, this percentage will rise to 31% (Campbell, 1996).

Table 1. Connecticut Racial/Ethnic Chart for 1990 and 2000

	White NonHispanic	Black NonHispanic	Am Indian/ Alaska Native NonHispanic	Asian/Pacific Islander NonHispanic	Hispanic
1990 (1)	2,753,210	267,005	6,329	49,689	212,677
2000 (2)	2,671,330	315,618	9,150	89,461	323,990

(1) Source: U.S. Census Bureau, PE-65. Release date 9/4/98
(2) Source: Stone C, Mueller L. 2004. State-level Bridged Race Estimates for Connecticut, 2000-2002. Connecticut Department of Public Health, Hartford, CT.

Socioeconomic Status

Connecticut residents are generally well educated. Eighty-eight percent of persons 25 and older are high school graduates and 35 percent have undergraduate degrees, which is the fourth highest percentage nationally (American Community Survey, 2003).

Connecticut is third in the nation in median household income at $56,803 (in 2003 inflation-adjusted dollars). Previously, in 2001, the state had been first in the nation. At that time Connecticut suffered a slumping economy leading to increased unemployment, state budget cuts, reduction in state services, and state tax increases. As of January 2005, job losses have yet to recover, making the future economic climate uncertain.

Although Connecticut is seen as one of the wealthiest states in the nation, income levels are not evenly distributed across the state. Fairfield County, which borders on New York, boasts affluent towns with

median household incomes in the $100,000 - $200,000 range. On the other end of the spectrum is the city of Hartford with a median household income of $28,000 (Connecticut Department of Economic and Community Development, 2000).

In Connecticut, 8.1 percent of the population lives below the federal poverty level compared to 12.7 percent of the nation's population. Of children under the age of eighteen in Connecticut, 10.8 percent live below the poverty level compared to 17.3 percent nationally (American Community Survey, 2003). However, the picture changes when poverty rates are viewed by town. While 38 out of 169 towns have child poverty rates less than 2 percent, Hartford's 41.3 percent rate is second highest in the nation among cities with populations exceeding 100,000 (Children's Defense Fund, 2002). New Haven, Bridgeport, and Waterbury rank 29th, 72nd, and 84th with rates of 32.6, 25.1, and 23.9 percent, respectively.

With regard to health insurance, 10 percent of Connecticut residents are uninsured compared to 15 percent nationally (Current Population Survey, 2004). Personal health care expenditures per capita are approximately $4,700 in Connecticut making it the fourth highest in the nation (Centers for Medicare and Medicaid, 2002).

Vital Statistics

The number of live births to Connecticut residents in 2000 was 43,026. This number is 7,000 less than the number of births in 1990. The birth rate has decreased from 69.3 to 61.2 per 1,000 population. The number of births, however, has steadily increased for Hispanic women over the last decade; the Hispanic birth rate in 2000 was 90.6. The birth rate for teenagers (ages 15-19) has decreased considerably from 38.8 in 1990 to 31.1 in 2000. In contrast the birth rate among women aged 35-to-44 has increased from 23.2 in 1990 to 30.5 in 2000, thereby continuing the trend toward childbearing at later ages. In 2000, 7.4% of infants were born low birth weight in Connecticut, up from 6.9% in 1990. The increase in multiple births over this time has contributed to these low birth weights. Infant mortality in Connecticut has decreased over the last decade from 7.4 in 1991 to 6.5 in 2000 (March of Dimes, 2003).

Heart disease continues to be the leading cause of death in Connecticut with 8,976 deaths in 2000. This is followed by malignant neoplasms with 7,038 deaths. The third leading cause is cerebrovascular disease with 2,003 deaths. Chronic lower respiratory diseases rank fourth with 1,524 deaths and accidents rank fifth with 1,170 deaths. These five leading causes accounted for 69 percent of resident deaths in 2000.

The next five leading causes of death include pneumonia (871 deaths), diabetes (683 deaths), septicemia (538 deaths), Alzheimer's disease (527 deaths), and nephritis (522 deaths). These five are responsible for an additional 10 percent of the total number of deaths. (Connecticut Department of Public Health, Health Care Quality, Statistics, Analysis, and Reporting, 2003). Of the ten leading causes of death in Connecticut, nine have a genetic component associated with them; accidents do not.

Concluding Remarks

The demography of Connecticut including the size, growth, distribution, and vital statistics of its population, bears directly on the current and future health care of the State's population, including genetic services. Factors to consider include:

- *The aging of the population.* As the elderly population continues to grow, the role of genetics, associated with diseases such as heart disease, cancer, diabetes, and Alzheimer's will intensify, and more genetics-related services are likely to be required. The health workforce also is aging and many health professionals are expected to retire at a time when demand for services will be increasing.

- *Connecticut's growing racial and ethnic diversity*. Opportunities will increase for epidemiologic studies to evaluate the frequency and role of genetic variants in different populations as racial and ethnic groups diversify. Providing culturally and linguistically sensitive genetic services are likely to be needed as will continuing efforts to eliminate racial and ethnic health disparities.

- *Childbearing at older ages*. With women delaying childbearing and having children at older ages (35 years or older), the need for obstetric providers trained to deliver prenatal genetic services is likely to grow.

- *The two Connecticuts.* Although as a state, Connecticut ranks high in socioeconomic status, extremes exist. Those with higher incomes and health insurance coverage are most likely to demand genetic testing and services as such services make their way to the marketplace. At the other extreme, those with lower incomes and those who lack health insurance may have difficulty accessing the genetic services that they might need.

DPH should continue to enhance its capacity to monitor ongoing demographic trends in its effort to assure the residents of the state equitable access to comprehensive, culturally appropriate genetic services.

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IV. Public Health Registries and Surveillance Systems

Public Health Registries and Surveillance Systems

Introduction

The Department of Public Health has a number of special population-based registries and surveillance systems that may be of use in genomic studies and may be enhanced by genomic information in the future. For example, the Tumor Registry is being used to facilitate population-based studies of genetic tumor markers and may be able to be used to establish a bank of tissue from selected tumors for further genomic studies. In the future, as genetic markers of selected cancers are identified and screening becomes possible, results of screening may be included in information contained in the registry. The following describes some of the DPH registries and surveillance systems that may have a role in genomics, both in facilitating genomic studies and in being enhanced by inclusion of genomic information.

Birth Certificate Data

In Connecticut, a birth certificate is filed with the registrar of vital statistics in the town in which a birth occurs within ten days after the date of birth. In addition, birth hospitals electronically transmit birth information to DPH daily. Approximately 43,000 birth certificates are filed annually. Birth certificates contain a wealth of information about the newborn, the mother, and about the pregnancy itself. Information about the newborn, such as birth weight, Apgar score, and presence of congenital anomalies or other abnormal conditions, enables babies with medical problems or abnormal conditions to be identified and investigated. Knowledge of births assists in the evaluation of newborn screening efforts and provides critical population data used in rate calculations, such as birth defects per 100,000 live births.

Birth certificates also provide information about the mother, including demographics, such as age, race, and educational attainment, and lifestyle risk factors, such as smoking and alcohol use. Pregnancy-related data, such as gestational period and adequacy of prenatal care, are also collected. These data can be used to study maternal health status and pregnancy outcomes, to identify vulnerable populations, and to develop public health initiatives designed to improve pregnancy outcomes.

Death Registry

The death registry contains mortality data pertaining to deaths occurring to Connecticut residents as reported on death certificates completed by funeral directors, attending physicians, medical examiners, or coroners. Sociodemographic information on death certificates is often based on reports by next of kin. Original records are filed in the state registration office. Approximately 30,000 deaths occur annually in Connecticut.

Mortality data contain cause of death information, including all diseases, conditions, or injuries that may have resulted in, or contributed to, death, as well as the circumstances or the event that produced any injuries. Mortality data are used to identify public health priorities, to focus public health promotion and disease prevention efforts, and to evaluate outcomes of prevention initiatives.

Linkage of Birth Records with Infant Death Records

Connecticut routinely links birth records with infant death records to evaluate the impact of newborn characteristics on death such as low birth weight or congenital anomalies, or maternal characteristics that may have affected the infant's death, such as lifestyle risk factors or level of prenatal care.

Tumor Registry

The Connecticut tumor registry is recognized as the oldest statewide tumor registry in the United States. It was initiated in 1941 and tracks cases of cancer diagnosed as early as 1935. All hospitals in Connecticut are required by law to report incident cases, along with information on follow-up and treatment. In 1981 the reporting mandate was extended to include private pathology laboratories. The registry has been part of the National Cancer Institute's Surveillance, Epidemiology, and End Results (SEER) Program since 1973. About 90% of the registry's funding comes from the SEER Program.

The National Cancer Institute uses SEER data to set priorities for research on the prevention and treatment of cancer in the U.S. population. The Connecticut tumor registry also provides a database for cancer surveillance efforts at the state and local levels.

The National Cancer Institute has estimated that the lifetime risk (0 to 85 years) of developing an invasive cancer is about 40%, i.e., about four out of every ten persons in the United States will develop some type of cancer at some time during their lives. The cancer sites with the highest lifetime risks are breast for females and prostate for males. During 1999, the most commonly occurring invasive cancers among Connecticut males were prostate (29%), lung (15%), and colon (8%). For females the highest incidence of cancers were breast (32%), lung (12%), and colon (9%) (Connecticut Department of Public Health, 2002).

Genetic testing is currently available for several genetic mutations that are linked to cancer. Mutations of the BRCA1 and BRCA2 genes are associated with an increased risk of breast and ovarian cancers. An increased risk for hereditary nonpolyposis colon cancer and familial adenomatous polyposis can also be determined by testing for certain genetic mutations. (Coughlin, 2000). Genetic testing is also available to identify families with Li-Fraumeni syndrome, which is a cancer predisposition syndrome.

Reportable Disease Registry

The Commissioner of DPH has the authority to annually declare lists of reportable disease and laboratory findings. While the lists mainly emphasize acute infectious diseases and are flexible, there are a number of diseases for which information has been reported for many years, such as AIDS, tuberculosis, sexually transmitted diseases, bacterial meningitis, and Lyme disease. The information reported is currently maintained in a series of disease-specific databases. An initiative is currently underway to integrate these databases into a single one, referred to as the Connecticut Electronic Disease Surveillance System, and to use this single database to enable electronic reporting from providers and laboratories.

Immunization Registry

The Connecticut Immunization and Tracking System (CIRTS) collects and manages statewide immunization data of all children who have not begun the first grade of school, including all newborns. Originally the Connecticut immunization registry, begun in 1993, included only children in Hartford. It was selected as the prototype for the state and expanded to include Medicaid managed care children. In 1994, the Connecticut state legislature passed legislation authorizing the Commissioner of DPH to establish a statewide registry. Beginning in 1998, all children born in the state of Connecticut were enrolled in CIRTS. Results from the calendar year 2003 National Immunization Survey show Connecticut first in the nation (94.6%) in vaccinating children 19-35 months of age when measuring vaccination status for 4:3:1:3 (4 doses Diphtheria/Tetanus/Pertussis, 3 Polio, and 1 Measles/Mumps/Rubella, 3 *Haemophilus Influenzae* type b) (Centers for Disease Control and Prevention, National Immunization Program, 2004).

Information contained in the immunization registry is confidential. Regulations stipulate the mechanisms under which data can be released to health care providers, parents or guardians, and directors of health.

CIRTS is valuable as a central repository for immunization information enabling providers, health directors, and public health administrators to track children's immunization status and provide outreach to those who lack access to regular medical care in order to reduce vaccine-preventable disease morbidity.

Newborn Screening System: Laboratory Newborn Screening

Since 1997, all babies born in Connecticut have been mandated by law to be screened for eight inherited disorders, which are generally metabolic in origin. They include phenylketonuria, congenital hypothyroidism, galactosemia, sickle cell disease, maple syrup urine disease, homocystinuria, biotinidase deficiency, and congenital adrenal hyperplasia. When the screened disorders are detected at or near birth, successful treatment or management can lead to prevention of death or serious physical or mental handicaps.

With the DPH Laboratory purchase of two tandem mass spectrometers, newborn screening has expanded to include additional amino acid disorders, organic acid disorders, and fatty acid oxidation disorders, including, but not limited to, medium-chain acyl-CoA dehydrogenase deficiency (MCADD), 3-hydroxy long-chain acyl-CoA dehydrogenase deficiency (LCHADD) and tyrosinemia. Expanded screening began in May 2004, and more than 40 disorders are being screened for as of January 2005.

Connecticut has a very effective genetics newborn screening program, which consists of testing, tracking, and treatment. Newborn screening blood sample collection is the responsibility of the birth facility. Written consent for the screening is not required in Connecticut, but protocols recommend that hospital staff inform all parents that the screening will be conducted. Parents may refuse screening if it is in conflict with their religious tenets or beliefs.

Heelstick blood specimens are transported via courier to the state laboratory where testing is conducted. Positive test results are reported to the DPH Family Health Section for follow-up. Staff are responsible for ensuring that abnormal laboratory results are appropriately and effectively reported

to the baby's primary care provider for disorder protocols, and if necessary referrals are made to designated state-funded regional treatment centers. Treatment centers provide comprehensive testing, counseling, education, treatment, and follow-up services. All laboratory results are tracked by DPH newborn screening program until diagnosis is normal or confirmation testing is completed.

In 2000, the Connecticut Department of Information Technology and the Office of Policy and Management, the lead agencies for the implementation of electronic forms in state government, developed an initiative to replace paper forms with electronic forms. They chose the Newborn Screening Program, administered by DPH, to be an early pilot program for electronic forms processing. The Newborn Screening System was developed and implemented to deliver information from birth facilities to the DPH Laboratory and to the DPH Family Health Section. The biographical and demographic data regarding the newborn and parent is common to the Laboratory and the Hearing Screening programs as well as the Birth Defects Registry. Screening data are accessible to respective program staff (Hearing, Laboratory, and Birth Defects Registry). These data are submitted from the birth facilities to the DPH Laboratory and the DPH Family Health Section over the Internet on a virtual private network (VPN). The VPN allows for the encryption of all data at the hospital and the de-encryption at the State. This encryption process preserves the security of the data and ensures compliance with Health Insurance Portability and Accountability Act requirements. Fast, secure communication enables timely diagnosis and treatment.

Newborn Screening System: Newborn Hearing Screening

Effective July 1, 2000, the Connecticut legislature mandated a universal newborn hearing screening program under which all infants are screened for hearing loss at birth. Hearing loss is the number one birth defect in the United States. Advances in the study of genetics suggest that 60% of all hearing loss cases are caused by genetic factors. Reliable DNA-based screening tests have been developed to detect common genetic forms of deafness such as Connexin 26 and mitochondrial deafness. In addition to non-syndromic forms of deafness in which hearing loss is the only clinical finding, a large number of syndromic forms are recognized in which there is associated involvement of other tissues or organs. Establishing an accurate genetic diagnosis can therefore be of great benefit to the patient and family in establishing an etiology and the prevention of adverse clinical sequelae.

Using a physiologic technologies testing mechanism recognized by the American Academy of Audiology or American Speech Language Association, birth facilities screen infants for hearing impairments prior to discharge. Results are transmitted to the DPH Family Health Section. Those infants not passing the initial screen are screened again prior to discharge. Those that do not pass the second screening are referred to an audiologist for further diagnostic testing. The audiologist refers infants with a diagnosed hearing loss to the Birth to Three System in the Department of Mental Retardation for early intervention services. The Connecticut Birth to Three System is responsible for implementing Part C (related to infants and toddlers) of the Individuals with Disabilities Education Improvement Act of 2004.

Connecticut does an excellent job of screening for hearing loss – 99.2 percent of infants were screened in 2002. In addition, the universal hearing screening program has exceeded the federal goal of referring infants with a diagnosed hearing loss to early intervention by 6 months of age, with an average age of referral being 3.7 months.

Demographic and hearing screening data are integrated within the Newborn Screening System and transmitted from the birth facility to DPH on a virtual private network.

Birth Defects Registry

DPH implemented a Birth Defects Registry Surveillance Program on January 1, 2004. Data collected under this program will be used to monitor the frequency, distribution and type of birth defects occurring in Connecticut and to identify environmental factors associated with birth defects that can be modified in an effort to prevent birth defects.

The timeliness of case ascertainment and referral to specialized health care services should improve dramatically by changing the source of cases from retrospective hospital discharge data to data from health care professionals at the point of diagnosis, which are being submitted via the Internet-based Newborn Screening System. Cases in the Registry will be limited, however, to newborns diagnosed before they are discharged from the birth facilities.

Concluding Remarks

As the validity and usefulness of new findings are documented, efforts to collect and add genetic information to the various DPH registries/surveillance systems should be considered. Ultimately, in order to eliminate duplication of effort in collecting data and to enhance data systems, data integration is needed. Currently efforts have begun to create a comprehensive database of linked child health data within DPH. The plan to do so can be found in the Appendices. Integration of laboratory newborn screening data, newborn hearing screening data, and birth defects data has already been partially implemented.

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V. Genetic Health Care Services in Connecticut

Genetic Health Care Services in Connecticut

Introduction

Currently Connecticut has both public sector and private providers who offer different components of genetics-related health care services including: clinical care, therapeutic care, counseling, screening programs, laboratory services, educational activities, family support services, outreach, and advocacy. The Family Health Section of DPH works in partnership with many of these programs, as well as with professional service organizations, but efforts are currently limited to newborns and children with special health care needs. There is no provision for services across the life span.

History of Genetic Services (National)

Long before there was a Human Genome Project, the federal government's Maternal and Child Health (MCH) program was leading the way. Through Title V of the Social Security Act, the MCH program began providing support to develop genetic services. This included the support of community-based child development clinics in the 1950s and state newborn screening programs for metabolic disorders in the early 1960s.

The 1972 Sickle Cell Anemia Control Act was the first federal legislation concerned with genetic disorders. This law called for grant support for newborn screening programs for sickle cell disease. However, it wasn't until the late 1980's that most states started performing sickle cell, or hemoglobinopathy, newborn screening.

The National Genetic Disease Act (Title XI of the Public Health Service Act) was passed in 1976, but funds to implement programs under the Act were not appropriated until 1978. At that time, the federal MCH program began to build and improve genetic services at the state level by providing funding for statewide genetic services programs with the understanding that states would continue these programs when federal support ended.

The Omnibus Reconciliation Act of 1981 replaced the National Genetic Disease Act, significantly reducing the funding for the expansion of genetic centers and the training of medical geneticists, counselors, nurses, and social workers. Training centers were forced to use their own internal resources to support fellowships and genetic training programs.

From the late 1970s to the early 1980s, several regional genetics networks came into existence to assess needs, coordinate services, and share resources. In 1983 the MCH program began funding these regional networks. Three years later, the Council of Regional Networks for Genetic Services (CORN) was established and supported by the MCH genetic service program to coordinate activities among the ten federally funded regional genetics networks representing all fifty states, to facilitate planning for genetic services, and to address national public health priorities in genetics. CORN was disbanded as of August 31, 1999.

In 1999, the MCHB Genetic Services Branch began two initiatives to facilitate the development of a public health infrastructure necessary to integrate appropriate genetic services and education into public health and health care delivery systems. First the National Newborn Screening and Genetics Resource Center (NNSGRC) was created to replace CORN. The NNSGRC is a cooperative

agreement between the MCHB and the University of Texas Health Science Center at San Antonio, Department of Pediatrics. The Center provides technical assistance to states for newborn screening and genetic services, and serves as an educational resource for health professionals, the public health community, consumers, and government officials.

The second initiative provides planning grants to states to develop state genetics plans and to build genetic service infrastructure within states. Connecticut has received such a grant and is in the process of planning for the growth of genomics in public health across DPH programs and throughout all phases of life – a critical new paradigm from traditional public health genetics.

History of Genetic Services (Connecticut)

In 1964, the Connecticut Statewide Newborn Screening (NBS) Program was implemented with the screening for phenylketonuria and galactosemia. Subsequently, six other screens were added to the panel: congenital hypothyroidism (1976); hemoglobinopathies (1990); maple syrup urine disease, homocystinuria, and biotinidase deficiency (1993); and congenital adrenal hyperplasia (1997).

Through the National Genetic Disease Act, funding in 1978 helped to establish genetic services at Yale University, the University of Connecticut Health Center and the Connecticut Department of Public Health.

In 1980, Connecticut became part of the New England Regional Genetics Group as part of an effort to coordinate genetic services and share resources among the New England states.

A 1985 line item in the State budget firmly established the Newborn Screening (NBS) Program in Connecticut. Yale University and the University of Connecticut Health Center/Saint Francis Hospital & Medical Center were designated as metabolic clinical centers. Separate legislative program funding was established for sickle cell screening with Yale University and the University of Connecticut Health Center. In addition, a statewide pregnancy exposure information service was created and based at the University of Connecticut Health Center.

In 1991**,** the State reauthorized the NBS Program. Three components of testing, tracking, and treatment were formally implemented in the NBS Program.

In 1995**,** linkage to Children with Special Health Care Needs (CSHCN) was established. For the first time since 1985, additional funding was provided in 1998 to the regional genetic treatment centers at Yale University and the University of Connecticut Health Center through the CSHCN's budget.

The 2002 Connecticut General Assembly enacted a bill to expand the newborn screening panel to include additional amino acid disorders, organic acid disorders, and fatty acid oxidation disorders, including, but not limited to, medium-chain acyl-CoA dehydrogenase deficiency (MCADD), 3-hydroxy long-chain acyl-CoA dehydrogenase deficiency (LCHADD) and tyrosinemia. Expanded screening began in 2004.

Genetic Services in Connecticut

The DPH Family Health Section provides coordination and funding of statewide genetic services and activities, information and referral on clinical genetics, newborn screening, sickle cell disease, community trait screening, maternal phenylketonuria (PKU), and human genetics education. Two major regional centers provide comprehensive genetic services for Connecticut's residents. They are located at the University of Connecticut Division of Human Genetics, located in West Hartford, and the Yale University Department of Genetics, located in the southern part of the state in New Haven. The Laboratory Newborn Screening Program has additional regional treatment centers for endocrinology and sickle cell at the Connecticut Children's Medical Center in Hartford and at Yale University in New Haven. Table 2 summarizes the providers and types of genetic services offered in Connecticut. Life-span services include preconception testing and counseling, prenatal testing and counseling, newborn screening, services for children with special health care needs, and adult genetic services related primarily to cancer.

As seen in the map in Figure 2, genetics clinics are located throughout the state and serve the most heavily populated areas as well as the poorest cities in Connecticut as previously described in the demographic overview of Connecticut. Most of the clinics outside of New Haven and Farmington are outreach clinics of either the Yale University School of Medicine or the University of Connecticut Health Center. Unknown, however, are the number and type (e.g. prenatal, pediatric, or adult) of patients served in each of these facilities.

A previous DPH report that assessed the genetic resources within the state of Connecticut identified a lack of resources in the eastern part of the state (Hromi, 2001). Working with DPH, the Yale University Department of Genetics and the University of Connecticut Health Center's Human Genetics Division now provide genetics services in New London and Norwich, respectively.

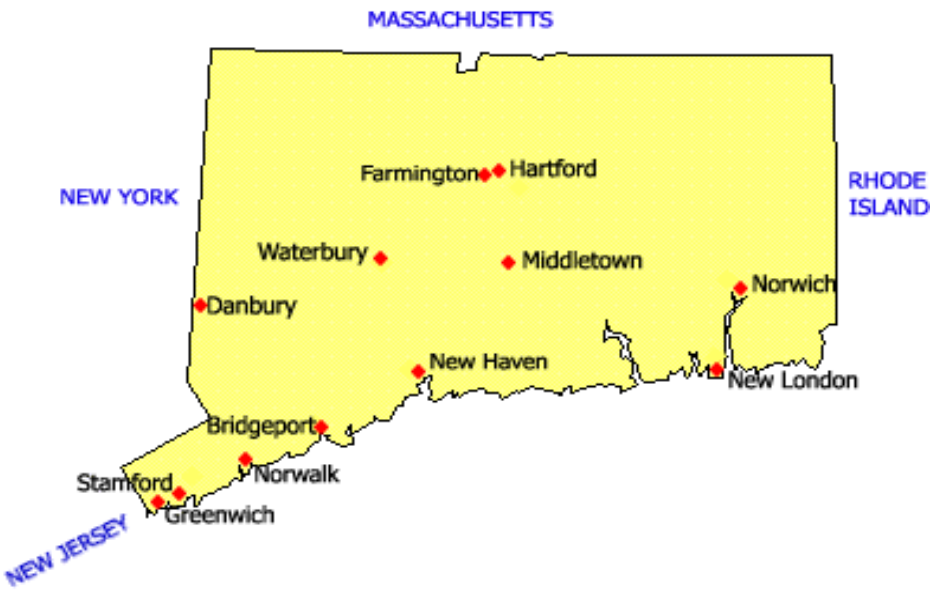
Eight laboratories in the state offer clinical genetic diagnostic and/or screening services. Three are associated with the University of Connecticut Health Center in Farmington. Three are located in New Haven at the Yale University School of Medicine. One is the molecular genetics laboratory of DIANON Systems in Stratford and the eighth laboratory is the DPH Laboratory in Hartford that performs all of the mandated newborn screening tests in the state.

Table 2. Connecticut Providers of Genetic Services

	Provider		
	Department of Public Health		Private Organization or Other State Agency
Genetic Service	**Direct Service**	**Contracted Service**	
Preconception testing & counseling			UCHC & Yale Genetics
Prenatal testing & counseling			UCHC & Yale Genetics
Newborn screening	State Lab Tracking Unit in FHD	Follow-up services at UCHC & Yale Genetics CCMC Sickle Cell Yale Sickle Cell	Yale & CCMC Endocrine Services
Service related to children with special health care needs			
Case ascertainment and referral	CSHCN Program		
Genetic clinics		UCHC & Yale Genetics	
Genetic counseling		UCHC & Yale Genetics	
Coordination of care		Stamford Hospital, Stamford Yale University School of Medicine, New Haven LEARN, Old Lyme Charter Oak Health Center, Hartford St. Mary's Hospital, Waterbury	
Adult genetics (primarily cancer related)			UCHC & Yale Genetics

NOTE: CCMC is the Connecticut Children's Medical Center
CSHCN refers to Children with Special Health Care Needs
FHD refers to the Family Health Division of DPH
UCHC is the University of Connecticut Health Center

Figure 2. Genetic Clinics in Connecticut*



*In: GeneTests: Medical Genetics Information Resource (database online). Copyright, University of Washington and Children's Health System, Seattle. 1993-2003. Available at http://www.genetests.org. Accessed December 29, 2003.

Preconceptional/Prenatal Testing and Counseling

Individuals who are planning to start a family or who are already pregnant and are concerned about the risk of passing genetic conditions or birth defects on to their children, can undergo counseling and testing to determine whether a pregnancy may be affected. Genetic counselors work with physicians trained in maternal fetal medicine to provide risk assessment, genetic evaluation and prenatal diagnosis, as well as psychosocial support. Such services are offered in various private clinics throughout the state. Connecticut data are not available regarding the number of patients served and the types of procedures and other services received.

Previously, only pregnant women with a family history of cystic fibrosis (CF) had been offered carrier screening. In more recent years, the American College of Medical Genetics (ACMG) has recommended that all Caucasians, not just those with a family history, be offered genetic testing, and has published guidelines for population-based CF carrier screening (Grody, 2001). Because members of other racial and ethnic groups are less likely to be carriers, testing should be made available only after testing limitations are provided to such individuals. Although ACMG recommends that preconception testing be encouraged whenever possible, most likely testing will occur in the prenatal setting. Regardless, population-based screening for CF carrier status is expected to grow substantially in the near future. Connecticut data for CF carrier screening are unknown.

Fetal genetic tests can now diagnose hundreds of genetic conditions such as cystic fibrosis, fragile X, deafness, and dwarfism; and high-resolution sonograms can detect birth defects early in a women's pregnancy. The American College of Obstetrics and Gynecology has begun endorsing use of these newer techniques that can be performed as early as 10 weeks, which is much sooner than traditional screening tests, such as amniocentesis, that are only available during the second trimester. Broader availability of new screening technologies will enable more women to find out about potential problems earlier in their pregnancies.

Although prenatal testing, such as amniocentesis or chorionic villus sampling, has been recommended for high-risk women, a new study suggests that all women, regardless of age, could benefit from prenatal diagnostic testing (Harris, 2004). Current testing guidelines were chosen back in the 1970s when age 35 was the approximate age at which amniocentesis was considered to be cost beneficial. New findings show that prenatal diagnostic testing can be cost effective at any age or risk level and perhaps guidelines should be revisited to consider these findings.

Connecticut Pregnancy Exposure Information Service

The Pregnancy Exposure Information Service provides information on all types of exposures during pregnancy, such as medications, infectious diseases, substances of abuse, and occupational and environmental exposures. This service is available to pregnant women and their partners, to those planning a pregnancy, and to health care providers. Free telephone counseling is offered to the majority of persons calling this service. Funded by a grant from DPH, the program is part of the Division of Human Genetics of the University of Connecticut Health Center.

Newborn Screening: Testing, Tracking, and Treatment

The Newborn Screening (NBS) Program for the state of Connecticut is administered through the DPH Family Health Section. The goal of universal newborn screening is early identification of newborns at increased risk for selected genetic diseases so that prompt medical treatment can be initiated to avert complications and to prevent irreversible problems and death. As of June 30, 2004, Connecticut was one of only 21 states to offer testing for a core group of nine genetic/metabolic disorders and hearing deficiency as recommended by the March of Dimes (March of Dimes, 2004). As of the end of September 2004, the March of Dimes had increased its newborn testing recommendations from nine to 30 diseases based upon a report prepared for the U.S. Department of Health and Human Services by the American College of Medical Genetics. Coinciding with these additional recommendations, DPH has expanded its newborn screening panel to include additional amino acid disorders, organic acid disorders, and fatty acid oxidation disorders. As of January 1, 2005, DPH exceeds the March of Dimes recommendations and screens for more than 40 disorders.

Since newborn screening began in 1964, there have been 841 (excluding hemoglobin traits) confirmed cases of the eight disorders tested prior to 2004. In addition, there have been 10,250 newborns identified with hemoglobin traits. The disease prevalence in Connecticut exceeds the national rate for PKU, galactosemia, and sickle cell disease. Table 3 provides a summary of the NBS statistics for Connecticut from 1964 through 2003.

Table 3. Connecticut Newborn Screening Statistics, 1964-2003

Disorder	Testing Initiated	Infants Tested	Confirmed Cases	Disease Prevalence in CT	National Incidence[a]
Classical PKU	1964	1,788,853	165	1 in 10,842	1 in 13,947[b]
Classical Galactosemia	1964	1,788,853	35	1 in 51,110	1 in 53,261[c]
Hypothyroidism	1976	1,213,384	303	1 in 4,005	1 in 3,044[d]
Sickle Cell Disease	1990	637,809	321	1 in 1,987 (All Births)	1 in 3,721/ 1 in 7386[e]
Hemoglobin Traits	1990	637,809	10,250	1 in 62 (All Births)	
MSUD	1993	468,996	1	1 in 468,996	1 in 230,028
Homocystinuria	1993	468,996	1	1 in 468,996	1 in 343,650
Biotinidase Deficiency	1993	468,996	6	1 in 78,166	1 in 61,319
Congenital Adrenal Hyperplasia	1997	274,239	9	1 in 30,471	1 in 18,987

Sources: National Newborn Screening and Genetics Resource Center and newborn screening literature. Published in the GAO-03-449 State Newborn Screening Programs.

[A]Preliminary data on disorder incidence presented by the National Newborn Screening and Genetics Resource Center at the 2002 Newborn Screening and Genetic Testing Symposium. Incidence rates are based on data from 1990 to 1999.

[b]Incidence rate is for clinically significant hyperphenylalaninemia, which includes classical phenylketonuria and clinically significant phenylketonuria variant.

[c]Incidence rate is for primary congenital hypothyroidism and does not include other forms of hypothyroidism.

[d]Incidence rate is for classical galactosemia and does not include other forms of galactosemia.

[e]Sickle cell anemia has an incidence of 1 in 3,721, while sickle hemoglobin C disease has an incidence of 1 in 7,386.

Although nineteen hospitals in Connecticut offer newborn screening for cystic fibrosis (CF), it is not currently mandated, but it may be in the near future. A 1997 National Institutes of Health (NIH) *Consensus Statement of Genetic Testing for Cystic Fibrosis* recommended against offering CF genetic testing to the general population of newborn infants because the panel found no definitive data to demonstrate medical benefit and cost savings associated with screening (National Institutes of Health, 1997). However, a recommendation that came out from a workshop held at the Centers for Disease Control and Prevention in November 2003 indicates that states should now begin to consider cystic fibrosis newborn screening (www.cdc.gov/ncbddd/cf/meeting.htm).

The DPH Laboratory performs all testing as required by state law and reports abnormal test results to the DPH Family Health Section for tracking and follow-up. Staff are responsible for ensuring that abnormal laboratory results are appropriately and effectively reported to the baby's primary care provider for follow-up procedures and referrals to the designated regional treatment centers per disorder protocols. The treatment centers provide: confirmatory testing, diagnosis, treatment, follow-up, and counseling services for genetic/metabolic disorders, endocrine disorders, and hemoglobinopathies. The genetic/metabolic and hemoglobinopathies services are provided through contractual agreements between DPH and the treatment centers. Increased public health awareness of genetic disorders and public health education are also key attributes of the DPH programs.

An examination of the communication practices between state newborn screening programs and the medical home (i.e., primary care physicians) indicates that parts of Connecticut's newborn screening system function well, but there are additional areas where improvements can be made (Sunnah, 2003). Testing and short-term tracking and follow-up appear to be well coordinated by DPH. However, DPH does not have a mechanism in place for linking prospective parents with a pediatric primary care physician before the child's birth. Early identification could simplify the communication of results to primary care physicians and facilitate the follow-up process. Although preliminary efforts are underway to build an integrated child-health information system within DPH, linkages with community-based primary care physicians would provide a mechanism for improved communications and information sharing among those involved in care for children. DPH also does not currently engage in long-term tracking of infants with conditions identified through newborn screening. Such tracking would enable DPH to monitor the clinical progress of children and the effectiveness of treatment, particularly in lesser-known conditions that are being added to the newborn screening panel. Data would be available to evaluate and help determine whether screening should continue for newly added conditions. Therefore, long-term tracking data is important to consider for inclusion in the integrated Newborn Screening System being implemented in DPH.

Laboratory Services

The DPH Laboratory provides numerous types of testing in an effort to reduce preventable health risks, such as those related to bioterrorist events, antimicrobial resistance, foodborne illness, and environmental threats. In addition it performs testing of newborn blood specimens to detect specific genetic disorders. Recognizing that much testing in the future will be at the molecular level and that the DPH Laboratory must retool to adapt to a changing health care environment, the DPH Laboratory Services Division and the Family Health Section prepared a plan for the future of genetic testing in August 2000 (Connecticut Department of Public Health, August 2000). With the acquisition of two tandem mass spectrometers, the Laboratory has expanded its newborn screening panel to test for over

40 disorders as of January 2005. The success and acceptance of these new services will pave the way for future expansion.

Newborn Hearing Screening

The purpose of the Newborn Hearing Screening Program is to provide early hearing detection and intervention in an attempt to minimize speech and language delays and promote normal speech development. Newborn hearing screening was implemented in Connecticut in July 2000. Already Connecticut has achieved a high screening level – 99.2 percent of infants were screened for hearing loss in 2002 and the average age of referral to the Birth to Three System in the Department of Mental Retardation was 3.7 months, which is well below the federal goal of 6 months.

Infants are screened for hearing impairments by the birthing facilities prior to their discharge, and results are reported to the DPH Family Health Section. Those not passing the initial screen are screened again prior to discharge, and if they fail to pass a second time, they are referred to an audiologist for further diagnostic testing. Because 50% of permanent congenital hearing loss is attributed to genetic causes, appropriate genetics consultation and management may be indicated. The major genetic treatment centers are the University of Connecticut Division of Human Genetics and the Yale University School of Medicine. DPH tracks infants from the initial screening, through diagnosis, and into enrollment in the Birth to Three System for early intervention.

Children and Youth with Special Health Care Needs

Public health interest in birth defects, developmental disabilities, and genetic conditions in children began in the early 1960s, partially in response to the epidemic of limb deficiencies that occurred due to maternal thalidomide exposure (Lenz, 1988). Coupled with the contribution of birth defects to infant mortality, surveillance programs were established. Legislation creating a birth defects surveillance program in Connecticut was passed in 1989. Initially the program was the responsibility of the Division of Epidemiology of the Department of Community Medicine at the University of Connecticut School of Medicine. Responsibility shifted to DPH in 1998.

In addition to children with congenital conditions such as birth defects and genetic disorders, children and youth with special health care needs (CYSHCN) include those who have or are at elevated risk for (biologic or acquired) chronic physical, developmental, behavioral, or emotional conditions and who also require health and related services (not educational or recreational) of a type and amount not usually required by children of the same age. Based upon a 2001 State and Local Area Integrated Telephone Survey (SLAITS), it is estimated that approximately 120,000 (14%) of Connecticut's children aged 0-17 have special health care needs. One in five households has a child with special health care needs.

In addition to primary care givers, these children may need the services of other medical specialists, surgical subspecialists, nutritionists, genetic counselors, public health and school nurses, physical therapists, occupational therapists, speech therapists, audiologists, psychologists, and social workers. The American Academy of Pediatrics believes that all children should have a medical home where care is accessible, family-centered, continuous, comprehensive, coordinated, compassionate, and culturally competent. Access to medical homes for CYSHCN is less than adequate, but improvement efforts are under way. As part of a follow-up effort from the 15-month National Initiative for Children's Healthcare Quality Medical Home Learning Collaborative, DPH has implemented a

Medical Home Training Academy to increase awareness among pediatric practitioners of "best practice models" for medical homes.

SLAITS-specific data for Connecticut also show that 19 percent of CYSHCN needed genetic counseling during the year prior to the survey and did not receive it. Additional analyses and assessments may reveal possible reasons for this unmet need, such as workforce shortages and insurance reimbursement inadequacies.

When compared to other states, Connecticut ranks in the top five states on 15 key indicators for CSHCN services related to child health, health insurance coverage, access to care, and family-centered care, and the impact that such children's needs have on their families (Blumberg, 2003). However, nearly a third of currently insured CSHCN in Connecticut have insurance that is not adequate to cover their comprehensive needs. Because Title V funds are limited to provide all the needed services for eligible CSHCN, Connecticut's General Assembly created a supplement for CSHCN under the Title XXI state Children's Health Insurance Program (SCHIP), known as HUSKY in Connecticut (Healthcare for Uninsured Kids and Youth) to improve access to more comprehensive services.

Adult Testing (Hereditary Cancer Program)

For those who are concerned about hereditary cancers, services are available to evaluate an individual’s risk for cancer, including breast, ovarian, colon, melanoma, and other forms of cancer. Family histories are reviewed and evaluated for the risk of predisposing cancer genes. Genetic testing and management options are advised based upon an individual’s cancer risk. Interpretation of laboratory data and psychosocial support are also provided. Such services are currently available at the University of Connecticut Health Center and the Yale Cancer Center. Unknown are the numbers served. DPH does not currently provide genetic testing services for adults.

DPH Genetic Advisory Groups

The DPH has representation on several genetics-related groups and committees that act as program consultants, advisors, and advocates as listed in Table 4.

The existing advisory groups serve specific programs. SASH is in the process of being replaced by a new advisory group for CSHCN. As genetics expands into other program areas within DPH as well as into other state agencies, broader oversight and/or advisory groups may be desirable. One example would be an Interagency Council for Genetic Services, similar to the one in Texas, which has representatives from the Department of Health, Department of Mental Health and Mental Retardation, Department of Insurance, University of Texas health science centers, entities that contract with the Department of Health to provide genetic services, and consumer groups.

Table 4. Genetics-Related Advisory Groups and Task Forces with DPH Representation

Group Name	Purpose
Genetic Advisory Committee (GAC)	THE GAC reviews and identifies emerging issues and revised procedures and protocols related to the genetic testing, tracking, and treatment of newborns. They act as an advisor to the DPH on newborn screening issues.
CT Newborn Hearing Screening Task Force	The Task Force provides interagency and provider collaboration to DPH on pertinent aspects of the Universal Newborn Hearing Screening Program
Steering and Advisory Committee for Children with Special Health Care Needs and HUSKY Plus Physical (SASH)*	SASH reviews the work of subcommittees looking at covered services, appeals, and quality assurance activities pertaining to Title V CSHCN and Husky Plus Physical programs. SASH also focuses on topics relevant to member agencies and providers servicing CSHCN and their families in an effort to increase access to services and supports and to avoid duplication.

* SASH was disbanded in 2002.

Concluding Remarks

Screening of newborns for genetic/metabolic disorders has occurred since the mid-1960s in Connecticut and it represents the first attempt at integration of genetic testing into public health. The Newborn Screening Program in Connecticut exemplifies successful coordination of services consisting of testing, tracking, and treatment. Recent advances in technology have made possible new forms of newborn screening programs, such as newborn hearing screening, which began in Connecticut in 2000. With 99.2 percent of infants being screened for hearing loss in 2002, the Connecticut Newborn Hearing Screening Program has quickly become a success story as well.

With the completion of the Human Genome Project and with advances in molecular technology, the impetus to expand diagnostic capabilities is escalating. Connecticut has already begun to respond to this growing need. With the acquisition of two tandem mass spectrometers, the DPH Laboratory has expanded its newborn screening efforts. Testing has increased from eight disorders to more than forty. The DPH needs to ensure that an adequate infrastructure is in place to meet this growing demand for services because the success and acceptance of these new services will pave the way for future expansion.

Expansion of DPH Laboratory services related to molecular testing will likely require that the Laboratory work with existing and new partners to build support for change; seek new resources to fund equipment, supplies, personnel, and training; and expand its current technology base to increase its computing and data storage capabilities.

Oversight of genetics-related activities in Connecticut is currently program specific, such as the Genetics Advisory Committee for newborn screening. Yet clients such as children with genetic disorders may require information or services from multiple state agencies, such as the Department of Public Health, the Department of Mental Retardation, the Department of Social Services, and the

Department of Insurance. Creation of an interagency council for genetic services may help guide the development and expansion of genetic services and assist in the coordination of those services. Advisory groups should also have participants representing consumers, health professionals, and payers. Broader representation may also be needed as genetics expands into other program areas within DPH.

Outreach and referral services are an integral part of Connecticut hospitals' strategic plans to provide genetic resources to their patients. Genetic services are available in the largest and poorest urban centers in the state and have expanded to support the underserved areas in the eastern part of the state.

The demography of Connecticut bears directly on the future of genetic services that are likely to be needed. The aging of the population, childbearing at older ages, and the growing ethnic diversity will likely affect the types and delivery of genetic services in the state.

Survey data indicate that 14 percent of Connecticut's children and youth aged 0-17 have special health care needs. Other than survey data from the MCH Policy Research Center's National Survey of State Title V Directors and the State and Local Area Integrated Telephone Survey (SLAITS), limited data are available regarding children and youth with special health care needs, particularly those with genetic disorders. Therefore, only generalities can be made regarding access to comprehensive services and coordination of care. Access to medical homes for CYSHCN is less than adequate, but improvement efforts are under way. As part of a follow-up effort from the 15-month National Initiative for Children's Healthcare Quality Medical Home Learning Collaborative, DPH has implemented a Medical Home Training Academy to increase awareness among pediatric practitioners of "best practice models" for medical homes. DPH is striving to enhance the quality of care and services provided to CYSHCN and their families.

It would be beneficial for DPH to take advantage of the opportunity to work with the Maternal and Child Bureau and SLAITS staff to identify topics that would enhance future SLAITS to address the needs within Connecticut.

Other than results from newborn screening, there is no currently available information on tests for genetic disorders done in Connecticut, such as the volume of tests that are provided or access to them, making it difficult to determine the unmet need for genetic services.

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VI. Genetic Resources in Connecticut: Workforce, Education, and Support

Genetic Resources in Connecticut: Workforce, Education, and Support

Introduction

In addition to the genetic services already discussed, additional resources are available that support and enhance genetic services. These include educational activities for professionals and consumers, family support services, advocacy organizations, and financing mechanisms.

Workforce and Professional Development

Appropriate integration of genomics into health care and public health in Connecticut will depend upon a workforce of health professionals with adequate education and training. Over time, direct health services related to genetics are likely to spread from specialty centers into most other areas of health services, thus requiring a wider workforce of health professionals to undertake new roles.

For instance, the responsibilities of pediatric primary care providers may ultimately need to expand, particularly with the growing emphasis on medical homes for children, the purpose of which is to ensure that medical care for children is accessible, continuous, comprehensive, family centered, coordinated, compassionate, and culturally effective. The medical home approach is one of the five key principles guiding the future of optimal pediatric care (American Academy of Pediatrics, 2000). Another principle emphasizes the need for pediatric training that embraces new areas that reflect the changing health care needs of children, including genetic issues.

Results from a recent direct-to-consumer marketing study about genetic testing for breast and ovarian cancer susceptibility show that health care providers often lack knowledge to counsel patients about such testing (Centers for Disease Control and Prevention, 2004). Educating providers to enable them to better respond to the complexities of genetic testing is therefore needed.

Although private sector efforts to educate physicians about genetics have focused on curricular changes, continuing medical education, and combined residencies and licensing exam changes, barriers to training include an overcrowded curriculum, complexity surrounding probability and risk inherent with genetic disorders, and a perceived lack of relevance of genetics to health care (Sarata, 2004). In addition, many physicians maintain that they don't see enough genetic diseases in their practice for ongoing training to be worthwhile. These are challenges that medical schools and continuing medical educational development programs will need to address.

An effective clinical genetics workforce will likely be a balance between generalists and specialists. Specialists are needed to provide direct genetic services related to counseling, testing, and test interpretation. Currently the number of genetic specialists (including genetic counselors) is very small, but appears to be growing in Connecticut as seen in Table 5. It is not surprising to see the larger concentrations of specialists in the North Central and South Central regions which are where the two regional genetic referral centers are located in the State. Although two genetics clinics have been opened in Norwich and New London in the Eastern region of the state since 2001, there is a general lack of specialists in the region, whereas the Southwest region of Fairfield County seems to have a larger percentage of specialists relative to their population.

Table 5. Geographic Distribution of Genetic Specialists in Connecticut

Uniform Service Region	Population (%) July 2002	Medical Genetics Specialists				Genetic Counselors			
		June 2001*		June 2004**		June 2001*		June 2004**	
		Number	Percent	Number	Percent	Number	Percent	Number	Percent
Southwest	19%	1	9%	4	20%	8	32%	13	33%
South Central	24%	6	55%	8	40%	8	32%	11	28%
Eastern	12%	0	0%	1	5%	0	0%	0	0%
North Central	28%	3	27%	5	25%	8	32%	9	23%
Northwest	17%	1	9%	2	10%	1	4%	6	16%
Total	100%	11	100%	20	100%	22	100%	39	100%

* Source: Hromi, 2001.
** Source: Certified by the American Board of Medical Genetics or American Board of Genetic Counseling. http://genetics.faseb.org/genetics/glisting_usa.html. Accessed June 2004.

Genetic counselors are important members of the genetics workforce. They provide information and support to families whose members have birth defects or genetic disorders, and to families who may be at risk for various inherited conditions. Provision of such counseling requires professionals who have received specific graduate training. Demand for such professionals is expected to increase as genetic testing evolves. Connecticut has approximately 40 board-certified genetic counselors (Table 5). As of December 2000 there were about 1,800 genetic counselors in the United States with membership in the National Society of Genetic Counselors, 70 percent of whom work primarily in prenatal settings (National Society of Genetic Counselors, 2004). There are only two dozen accredited masters-level genetic counseling programs in the United States, one of which is in New England. It is located at Brandeis University in Massachusetts.

Reasons for the low number of genetic counselors include limited funding for academic programs in genetic counseling as well as limited scholarship opportunities for students (Cooksey, 2000). Measures need to be adopted to promote interest and training in genetic counseling and to provide financial assistance for students in need.

Movements are underway in some states to license genetic counselors in order to assure that counselors are adequately trained and are accountable for meeting appropriate standards of practice (National Conference of State Legislatures, 2004). Because genetic information has the potential to dramatically affect personal reproductive and health decisions, there are concerns that inadequately trained counselors may provide incorrect or incomplete information to clients, thus leading to potential psychological and emotional harm. Licensure is one way of trying to protect the public. Licensure would also allow genetic counseling services to be eligible for insurance reimbursement, which has also been problematic in the past.

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In addition to increased genetics education for health professionals (both generalists and specialists), education and training are also needed to create a public health workforce in Connecticut that is capable of applying relevant genetic information into practice.

A report from the Institute of Medicine identifies genomics as one of the critical areas that training programs in public health should include in order to address emerging health challenges in the 21st century (Institute of Medicine, 2002). Connecticut's schools of public health should be encouraged to develop such programs. The University of Connecticut Health Center's Graduate Program in Public Health currently offers a course on Genetics and Public Health, so progress has begun in this direction (H. Swede, DPH; personal communication).

Within DPH, there are a limited, but growing, number of professionals who incorporate genetics into their programs. Historically most have dealt primarily with the newborn screening and children-with-special-health-care needs programs. Expanded newborn screening is creating even more of a burden on the limited personnel and resources available. Integration of genetic information into other programs will require additional infrastructure development. Staff knowledge and skills will need to expand and be upgraded regularly to keep pace with discoveries in genetics. For instance, chronic disease program staff will need to learn about the use of genetics to strengthen disease prevention activities. Educational opportunities thus need to be made available to provide training and program-development assistance. Work has started in this area with several DPH-sponsored educational workshops that have occurred under a HRSA-funded state genetics planning grant. A series of four workshops were offered to a broad team of DPH staff working in genetics-related programs. The topics were consistent with CDC-recommended standards for genetics competency among public health professionals. The workshops culminated in a statewide symposium in Public Health Genetics, which featured nationally recognized keynote speakers. Now that a foundation has been laid, continued education is needed to enable staff to keep pace with genetic advances and to incorporate genomic applications into health promotion and disease prevention activities.

To address the need for a workforce that is literate in genetics, various national organizations have developed competency sets. See Table 6. In addition, the National Association of Social Workers has developed standards for integrating genetics into social work practice (National Association of Social Workers, 2003). Needed, however, are strategies to train the workforce to develop the appropriate competencies.

Table 6. Competency Sets for Genetics-related Occupations and Professions

Competency Set	Worker Level	Resource
Genomics Competencies for the Public Health Workforce, May 2001	Leaders/Administrators Clinicians Epidemiologist/Data Manager Health Educators Laboratorians Environmental Health Workers	Office of Genomics and Disease Prevention, CDC http://www.cdc.gov/genetics/training/competencies/comps.htm
Competencies in Public Health Genetics, June 1999	MPH, MS, PhD Students	"Public Health Genetics in the Content of Law, Ethics, and Policy" Program, Institute for Public Health Genetics, Public Health Genetics Training Collaboration (CDC, HRSA funded) http://depts.washington.edu/phgen/DegreeTracks/competencies.html
Core Competencies in Genetics Essential for All Health Care Professionals, February 2000	Health Care Professionals Students	National Coalition for Health Professional Education in Genetics (RWJ, DOE funded) http://www.nchpeg.org
Medical School Core Curriculum in Genetics, 1995	Medical Students	American Society of Human Genetics http://www.ashg.org/genetics/ashg/policy/rep-01.htm

Public Education

Among the ten essential public health services identified by the Association of State and Territorial Health Officials (ASTHO) that should be considered when integrating genomics into public health, one clearly highlights the importance of public education (ASTHO, 2001); it calls on public health agencies to “inform, educate and empower people about health issues.” Consistent with this priority, all 17 of the state genetics plans across the country that have been developed and included on the National Newborns Screening and Genetics Resource Center (NNSGRC) website contain an educational component to heighten public awareness about genomics issues (NNSGRC, 2004). An informed public within the state is vital for its partnership with DPH in responding effectively to newly perceived issues that impact public health. Yet now, after the more than 3 billion base pairs of human DNA have been sequenced, the nation remains as uninformed about genomics as it was when

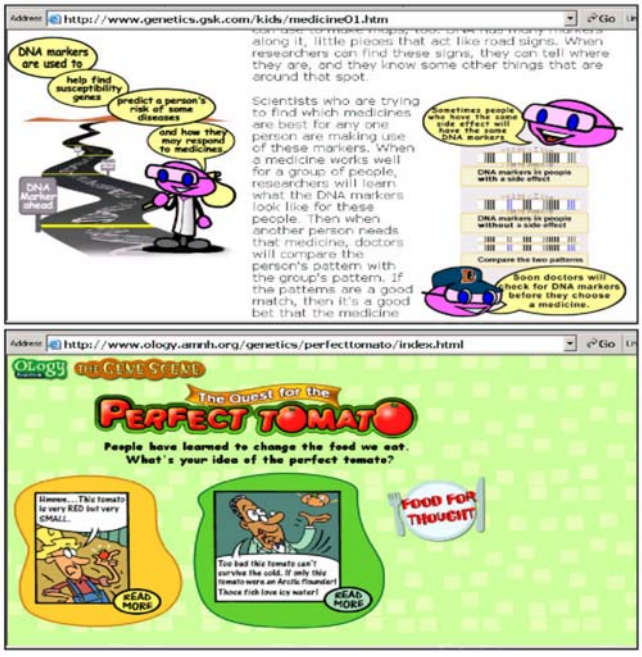
the human genome project began in 1990 (Singer, 2004). There is a need to heighten and maintain awareness of genomics and its potential impact on human health among the general public.

Education in genetics and genomics for the general public can be directed at two distinct target populations -- school-aged youth and the adult population. Among adults, subpopulations exist that include parents of newborns and consumer families of children affected by genetic conditions. Each of these three target groups is discussed below.

School-Aged Youth

Increased genetics awareness among the state's youth could be managed through the school systems. The Connecticut Department of Education (DOE) maintains benchmarks for scholastic achievement in a variety of areas from kindergarten through grade 12, and in the area of science, the Department has established benchmarks for topics that include life science (Connecticut DOE, 1998). Within this topic are benchmarks for genetics that, by high school graduation, prepare the young adult for competence in Mendelian genetics, the basic function of DNA in a cell, and evolutionary changes that can be traced with mutations in DNA. There is also a need for students to understand the basics of molecular biology techniques. Given the rapid increase in genetics information during this past decade, a group called Mid-continent Research for Education and Learning has published a set of recommended genetics benchmarks for K-12 curricula, which could be considered for Connecticut's students (Mid-continent Research for Education and Learning, 2003).

Although the benchmarks for science by the DOE and the Mid-continent Research group address the importance of genetics to biology, evolution, and life, they do not deal with the impact of genetics and inheritance on human health risk and outcome. High school students, just a step away from adulthood, may not be prepared to understand how the genetic makeup they inherited from their parents may affect their long-term health outcomes. Young adults also need to understand how their genetic makeup and that of their chosen mate, with associated health risks, could be passed on to future generations.



Resources are available for closing the gap in student understanding of the association between genetics and health, largely through the Internet. Many web-based educational opportunities directed at school-aged individuals currently exist. For instance, the pharmaceutical giant GlaxoSmithKline manages a website called "Kids Genetics," which contains specific information on genetics and disease risk, as well as other genetics-related information (GlaxoSmithKline, 2004). The federal Department of Health and Human Services also manages a website for youth, with links that describe the potential health implications that are possible as a result of information collected from the human genome project

(U.S. Department of Health and Human Services, 2004). Information relating to genetically modified foods is available from a kid's website through the American Museum of Natural History (American Museum of Natural History, 2004). Also, Cold Spring Harbor manages a website on a variety of topics designed for older students and teachers that includes health and human diseases related to genetics (Dolan Learning Center, 2004).

In addition to the use of websites, many other organizations are developing classroom activities that enrich the educational opportunities for students. For example, the Institute of Human Genetics at the University of Utah has created a set of classroom activities that are developmentally appropriate for grades K-12 (Genetics Science Learning Center at the Eccles Institute of Human Genetics, 2004). The exercises focus on genetic traits, teaching students how to make an inventory of their traits, compare them, and develop genetic trees. Other topics include gene therapy, stem cells, and human cloning. The Genetics Education Center, through the University of Kansas Medical Center, manages a website for educators on human genetics that includes information, resources, and programs for curricular enrichment (Genetics Education Center, 2004).

Using available Internet resources, and partnering with the DOE and biotechnology companies in the State, science education curricula within Connecticut could be updated and enriched with health-related topics in genetics. Students could enter the general public with a greater awareness of genetics and its potential impact on their health and that of their families, current and future. The informed young adults would also be better prepared to respond to new advances in genetics with foresight and would be less susceptible to misinformation.

General Adult Population

Once young adults leave the academic environment, opportunities for obtaining and communicating information about genetics are considerably diminished. Yet, this is the group most in need of genetics understanding, because it includes women and their partners making reproductive choices or seeking genetic testing at preconception. It also includes individuals who may hear regularly through the media about genetic susceptibility to cancer, hypertension, diabetes, and other complex disorders that could impact their lives or the lives of their families.

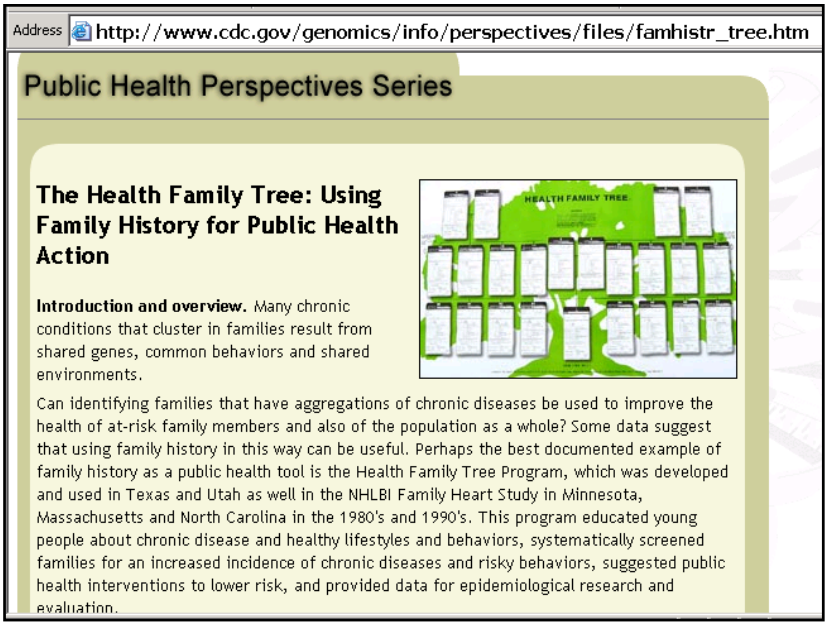
Another group among the adult general population that could be affected by genetics and genomics in the very near future are individuals under treatment for cancer and other chronic conditions. Targeted cancer therapies that direct medications to specific locations within the body are rapidly entering the field of individualized medicine (Green, 2004). Also, rapid advances in pharmacogenomics may soon be used by physicians to make drug choices for treatment that are tailored to the genetic makeup of the patient. Pharmacogenomics also promises to become widely available to tailor prescription medications to pregnant woman (Associated Press, 2004).

When a woman becomes pregnant and makes her first prenatal medical appointment, her obstetrician may recommend prenatal genetic testing for the presence of conditions such as Tay-Sachs, Down's syndrome, or neurological disorders. However, it is currently possible to measure as many as 250 birth defects of an unborn child (KidsHealth, 2004). Clearly, information available from prenatal genetic testing could become overwhelming for an expectant mother. Couples may not receive the counseling necessary to make informed decisions about taking these tests, and they may not receive the post-counseling services needed to act responsibly on the information obtained from the tests.

As genetic tests become more available, and as people undergo these tests, they will increasingly face decisions that require an understanding of the susceptibility of disease for themselves and for their families. There is currently no systematic, culturally sensitive program within Connecticut designed to inform the general adult population about the impact of genetics on human health. There is also no program designed to inform these adults about genetics services available to them within the state, and this group may not know where to go to obtain genetic testing, genetic counseling, and general information about genetics and genomics. Information about genetics and health from the media is sporadic and not designed to provide a general background in genetics needed to produce an informed adult population. The Internet currently contains a large, and often dizzying, amount of information about general genetics and genetic testing. A recent search on the commonly used *Google.com* revealed nearly three million search results for "human genetics, " and a subset search for "human genetics testing" revealed 1.4 million websites on the topic (Google, 2004). Before it can be considered a reliable resource for the general public, the most accurate websites contained in this vast expanse need to be identified and categorized for cultural sensitivity and ease of access for both the novice and more informed learner alike.

Beyond websites for those self-motivated individuals with computers and Internet access, there are few opportunities to learn about general genetics and its impact on health. The primary healthcare clinics may be one possible site at which individuals could receive information about genetics, but the healthcare worker may not be the appropriate source of information because of their limited confidence in genetic counseling, and because of the high demand within their offices for rapid turnover of services (Acton, et al, 2000).

Among clinical services, the CDC Office of Genomics and Disease Prevention recently initiated a program that encourages the use of Family Health History to assess genetic susceptibility to disease (Yoon, 2002). A collaborative between CDC offices and NIH, this initiative encourages the general public to develop family trees of their health, focusing on chronic diseases. The model for educating the public is based on a family tree tool developed in Utah and targeted toward high school students (Williams, 1998). Four states, Michigan, Minnesota, Oregon, and Utah recently received funding to study this model as a preventive health tool (Centers for Disease Control and Prevention, 2003). As a speaker at the April 2004 DPH Public Health Genetics Symposium, Dr. Muin Khoury, Director of the CDC's Office of Genomics and Disease Prevention, described the initiative and indicated that future funding is possible in this and other areas of genetics (L. Mueller, DPH; personal communication). Connecticut may want to pursue activities that raise awareness of family health history as a risk factor for disease. Connecticut may want to pursue activities that raise awareness of family health history as a risk factor for disease.



Families of Newborns

Among the many services provided by the DPH Family Health Section and the Laboratory, educational services to parents expecting children and parents of affected children rank high. Staff in the Newborn Screening unit are dedicated to educating health care providers, parents, and consumer families about newborn screening tests mandated within our state. Their efforts help to prepare over 40,000 families yearly for the tests infants receive soon after birth. Fact sheets for disorders included in the newborn screening panel are available on the DPH website, providing information for both the consumer family and the medical professional (Connecticut Department of Public Health, 2004).

Regional Efforts in Genetics Education

Consistent with the Title V Maternal and Child Health objective to "increase professional and public knowledge about how genetic diseases affect health," all states funded with Title V block grants should address the education of genomics and its relationship to health. In an effort to address this objective collectively, state representatives from the 6 New England states (Connecticut, Maine, Massachusetts, New Hampshire, Rhode Island, and Vermont) have established the New England Public Health Genetics Education Collaborative, within a Regional Genetics and Newborn Screening Collaborative (*HRSA-04-085 Heritable Disorders Program CFDA:93.110 Project 2, Regional Genetics and Newborn Screening Collaborative*). Funding for 3 years was awarded beginning on September 30, 2004. In preparation for the proposal and with Connecticut DPH leadership, the state representatives compiled a list of perceived needs in genetics and genomics education that addressed: 1) the target audience, including the general public, public health professionals, and medical professionals; 2) content, from general to specialized genetics topics; 3) whether the material currently exists or requires development; and 4) the degree priority for distribution (Table 7). From the table, it is clear that the six states share considerable interest in genomics education directed toward the general public and public health professional, as well as toward the medical professional. In addition, the six states share an interest to produce opportunities on genetics topics of general as well as specialized content. If funded, the collaborative group will develop educational resources that can be shared among the New England region.

Cultural Sensitivity

In a recent survey to assess the public's awareness of predictive genetic tests for cancer risk, 72% of Caucasians reported that they knew of such tests, compared to only 49% of African-Americans (Peters, 2004). Other recent studies echo this racial disparity in the knowledge of, and values, attitudes and beliefs toward genetic testing (Singer, 2004). These studies indicate that educational programs in genomics and health issues related to genetics must be culturally sensitive to the diverse ethnic and racial groups that reside in Connecticut.

Table 7. Perceived State Genetics Educational Needs

Perceived State Genetics Educational Needs
May, 2004

Target Audience: **C**=Consumer/Public, **MD**=healthcare professional, **PH**=public health professional
Genetics Content: **G**=General, **S**=Specialized/newborn screening
Process: **D**=item needs development, **R**=item developed but needs to be reproduced and distributed
Priority: **H**=High, **L**=Low

Opportunity	Audience			Content		Process		Priority	
Brochures	**C**	**MD**	**PH**	**G**	**S**	**D**	**R**	**H**	**L**
Pre-natal genetic testing at medical offices and clinics	x				x	x	x		x
General topics for healthcare workers, insurers & low literacy groups		x		x		x	x	x	
Newborn screening materials to healthcare providers		x			x	x	x		x
Distance Learning Modules	**C**	**MD**	**PH**	**G**	**S**	**D**	**R**	**H**	**L**
Newborn screening to pediatric practices, hospitals, community services, including MS/MS conditions		x			x	x	x	x	
Fact Sheets and Medical Protocols	**C**	**MD**	**PH**	**G**	**S**	**D**	**R**	**H**	**L**
Emergency protocols for metabolic disorders of newborns with confirmed conditions		x			x		x	x	
Disease-specific inserts and factsheets.	x	x			x	x	x	x	
Routine well-child protocols		x			x	x	x	x	
Translation Services	**C**	**MD**	**PH**	**G**	**S**	**D**	**R**	**H**	**L**
Newborn screening materials & fact sheets into other languages	x				x	x	x		x
Newborn screening materials & fact sheets into Spanish	x	x			x	x	x		x
Media Support for Presentations (CD, PowerPoint, websites, etc)	**C**	**MD**	**PH**	**G**	**S**	**D**	**R**	**H**	**L**
PowerPoint slides for community outreach presentations	x			x		x		x	
Instructional CD to provide access to specialized genetic educational material		x			x		x	x	
PowerPoint slides genetics 101, current and emerging issues, applications			x	x	x	x		x	
PowerPoint slides for professional presentations.		x		x		x	x	x	
Offer Conferences	**C**	**MD**	**PH**	**G**	**S**	**D**	**R**	**H**	**L**
Genetics and Public Health			x	x		x		x	
Public Health Genetics Competencies			x	x		x			x
What's new in newborn screening		x	x		x	x		x	
RI Annual Genetics and Public Health Conference		x	x	x	x	x		x	
Regional Conferences in Genetics		x	x	x	x	x		x	
K-12 Curricula	**C**	**MD**	**PH**	**G**	**S**	**D**	**R**	**H**	**L**
Lecture & experiential materials, standardized test questions, benchmarks for curricular development	x			x		x	x		x
Regional Ideas	**C**	**MD**	**PH**	**G**	**S**	**D**	**R**	**H**	**L**
Enhance NERGG and state websites	x	x	x	x	x	x		x	
Annual retreat among state representatives			x	x	x	x		x	
Other	**C**	**MD**	**PH**	**G**	**S**	**D**	**R**	**H**	**L**
Feasibility study of family health history model	x			x	x	x			x
Experts for Grand Rounds & advisory committees		x	x		x	x	x	x	
Clinical case review sessions		x			x	x	x	x	

This *Needs* Table shows the results of a collaborative effort among the six states, in which each state listed and prioritized the specific educational needs of its state. Most items in the table were identified by at least two states, and in some cases by as many as four states, highlighting areas of mutual interest and opportunities for future collaboration.

Other Resources

Birth to Three System

The Birth to Three program administered by the Connecticut Department of Mental Retardation is a statewide program of early intervention services for infants and toddlers, birth through age 2 years, who are at risk for developmental delays or disabilities. Federal legislation, the Individual with Disabilities Education Act (IDEA), Part C, defined the services that must be provided and establishes minimum eligibility requirements to ensure that all eligible children are identified and served appropriately (Individuals with Disabilities Education Act Amendments of 1997, 2005).

Local Interagency Coordinating Councils (ICC)

Connecticut's local ICCs are independent regional partnerships of families of young children who may have developmental delays or disabilities or health-related concerns, and members of local community groups, including early intervention providers, the medical community, local public school systems, regional offices of state agencies, and others (Connecticut Birth to Three System, 2004). ICCs advise appropriate agencies on the unmet needs in early childhood special education and early intervention programs for children with disabilities, assist in the development and implementation of policies that constitute a statewide system, and assist in coordination for implementation of a statewide system.

Regional Genetics Groups

DPH participates in several regional genetics groups where they can share experiences with other New England state representatives and health care professionals (see Table 8). These organizations serve as regional centers of genetic information.

Table 8. Regional Genetics Groups with DPH Representation

Group Name	Purpose
New England Regional Genetics Group, Inc. (NERGG)	NERGG is a consortium of genetic service providers, public health planners, consumer groups, and federal and state MCH personnel in New England whose purpose is to promote the availability of high quality human genetic services and to encourage genetic education.
New England Consortium of Metabolic Programs Planning Group	The Consortium provides an opportunity for collaboration and networking of healthcare professionals involved in identifying and treating those with metabolic disorders. They develop social support programs and educational materials, and provide training for students in medicine and related fields.

Professional Service and Educational Organizations

There are a number of professional genetic service and educational organizations that sponsor educational activities and provide family support services, outreach and advocacy. In addition to the discussion below, DPH has developed a formal directory of genetic resources, entitled *Genetics Resources: A Directory* that is being shared with health care professionals and consumers.

American Academy of Pediatrics, Connecticut Chapter (http://www.aap.org)

The American Academy of Pediatrics is a national organization of over 50,000 pediatricians dedicated to the health and well-being of infants, children, adolescents, and young adults. The Connecticut chapter promotes children's good health through working committees, lobbying efforts, and education of families and those who care for children.

American College of Obstetricians and Gynecologists (http://www.acog.org)

The American College of Obstetricians and Gynecologists (ACOG) is a leading group of professionals providing obstetric-gynecological care for women. ACOG advocates for quality health care for women, promotes patient education, and works to increase awareness among its members and the public of changing issues facing women's health care.

CDC Office of Genomics and Disease Prevention (http://www.cdc.gov/genomics)

The Centers for Disease Control and Prevention, Office of Genomics and Disease Prevention provides information about human genomic discoveries and how they can be used to improve health and prevent disease across the lifespan.

Connecticut Down Syndrome Congress (http://ctdownsyndrome.org)

The Connecticut Down Syndrome Congress (CDSC) is a network of over 350 parents and professionals statewide. The CDSC supports those affected, disseminates accurate information about the disorder, promotes public awareness, encourages quality services for those affected, and advocates for the rights of those with Down syndrome. They provide services such as educational meetings and conferences, quarterly newsletters, social activities, and scholarship funds.

Connecticut Perinatal Association (http://www.connperinatal.org)

The Connecticut Perinatal Association is a statewide, multidisciplinary organization concerned with perinatal health issues from preconception through infancy. Members consist of health professionals, including geneticists and genetic counselors, as well as legislators and consumers working together to support education for providers and consumers of perinatal health care and to promote initiatives toward improving the care of mothers and infants.

Genetic Alliance (http://www.geneticalliance.org)

The Genetic Alliance is an international coalition of individuals with genetic conditions and advocacy, research, and health care organizations working together to promote healthy lives for all those living with genetic conditions. Key programs include education on genetics issues, outreach to diverse and underrepresented communities on genetics issues, and consumer-centered public policies that speed research applications into accessible technologies and services.

March of Dimes, Connecticut Chapter (http://modimes.org)

The March of Dimes is a national voluntary health agency whose mission is to improve the health of babies by preventing birth defects and infant mortality. The March of Dimes funds programs of research, community services, education, and advocacy to save babies. Connecticut advocacy issues and priorities for 2004 include the following:

- Expand coverage under HUSKY (Connecticut children's health insurance program) for pregnant women over the age of 19 up to 300% of the federal poverty level.
- Expand the Connecticut Birth Defect Prevention and Surveillance Program.
- Increase smoking prevention and cessation programs for pregnant women.
- Advocate for public policy initiatives to reduce racial disparity in birth outcomes.

National Newborn Screening and Genetics Resource Center (http://genes-r-us.hthscsa.edu)

The National Newborn Screening and Genetics Resource Center provides information and resources in the area of newborn screening and genetics to assist health professionals, the public health community, consumers, and policymakers. It is a cooperative agreement between the Maternal and Child Health Bureau, Genetic Services Branch and the University of Texas Health Science Center at San Antonio, Department of Pediatrics.

Sickle Cell Disease Association of America (http://www.sicklecelldisease.org)

In 1983, the Sickle Cell Disease Association of America (SCDAA), Connecticut Chapter, Inc. was organized to support people living with sickle cell disease and their families through services like research, advocacy, and education. Additional services have since been added, including a nutrition supplement program, patient financial assistance, counseling, and community education. The SCDAA, Connecticut Chapter, Inc. is located in Hartford with a satellite office in New Haven. The Department of Public Health provides financial support to this organization for educational activities and to assist individuals with sickle cell disease transition from pediatric to adult primary care and hematology services.

The Southern Regional Sickle Cell Association, Inc., located in Bridgeport, was founded in 1985. Their mission is to provide education, screening, counseling, and support services to persons affected with sickle cell disease and hemoglobin trait. Financial support has recently been provided by DPH

for this organization to provide sickle cell trait testing and counseling, and to provide case management and transition services.

Spina Bifida Association of Connecticut (http://www.sbac.org)

The Spina Bifida Association of Connecticut (SBAC) strives to educate the public about spina bifida and to enhance the lives of all affected. It advocates the use of folic acid to help reduce the risk of having a pregnancy affected by a neural tube defect, such as spina bifida. SBAC supports those affected and their families through services such as educational meetings, quarterly newsletters, social activities, medical expenditure funds, and scholarship funds. SBAC also raises public awareness about the condition and how affected individuals can reach their full potential as productive members of their communities.

Financing Mechanisms

Children's health coverage is of particular importance to Connecticut. Nearly one fourth of all Connecticut births are financed by Medicaid. An estimated 71,000 children under 18 in Connecticut (8%) are uninsured (Mills, 2003). One in five children are enrolled in the state's Healthcare for UninSured Kids and Youth (HUSKY) program, which is an expanded Medicaid program (Solomon, 2004). Yet more than half of Connecticut's low-income children (defined as family incomes below 200 percent of the federal poverty level) remain uninsured even though they appear to be eligible for HUSKY (U.S. Census Bureau, 2004).

Although Connecticut requires universal newborn screening for certain conditions, funding for such a program is insufficient. DPH needs to assure the public that adequate financing mechanisms are in place to support such a comprehensive program, particularly in light of the recent expansion of the screening program. DPH also has the responsibility to assure an appropriate system of care for children with special health care needs as identified under the Title V Maternal and Child Health Block Grant.

Title V Maternal and Child Health Block Grant

The purpose of Title V is to improve the health of all mothers and children, including children with special health care needs (CSHCN). Title V funds are allocated among pregnant women, infants, children and youth ages 1-22, CSHCN, and administrative functions. At least 30 percent of the funds are budgeted annually for services for CSHCN.

State Children's Health Insurance Program

The Balanced Budget Act of 1997 created a new national children's health insurance program under Title XXI of the Social Security Act called the State Children's Health Insurance Program (SCHIP). The purpose was to enable states to initiate and expand health insurance coverage for uninsured children beyond the Medicaid eligibility levels. In Connecticut an umbrella program known as HUSKY (Healthcare for UninSured Kids and Youth) was created. Part A of HUSKY reflects Medicaid coverage for children through age 18 in families with incomes up to 185% of the federal poverty level (FPL) (Centers for Medicare and Medicaid Services, 2004). For Part B of HUSKY,

children in families with incomes between 185% and 300% of the FPL are eligible for insurance coverage by paying a sliding fee.

Because Title V funds are limited to provide all the needed services for eligible CSHCN, Connecticut's General Assembly created a supplement for CSHCN, referred to as HUSKY Plus. Children enrolled in HUSKY B are also able to apply for HUSKY Plus, which covers services for those with special behavioral or physical needs. The HUSKY Plus Physical program is jointly administered by the Connecticut Children's Medical Center in Hartford and the Yale New Haven Children's Hospital in conjunction with the Yale University School of Medicine. The two regional centers serve as the coordinating organizations, but services are provided by entities under contract to provide Title V services. The behavioral health services are organized by the Yale Child Study Center, which administers a statewide network of providers (Centers for Medicare and Medicaid Services, 2004).

As of May 1, 2004, the Connecticut Voices for Children reported that enrollment in HUSKY Part A was 213,377 and enrollment in Part B was 14,523 (Solomon, 2004). Over the past several years, enrollment has increased in both HUSKY A and B programs, as seen in Table 9, thus improving access to those who otherwise might be uninsured.

Table 9. Enrollment in HUSKY, 2000 - 2004

	As of May 1				
	2000	2001	2002	2003	2004
Part A[1]	176,352	177,547	194,536	208,580	213,377
Part B[2]		8,196	12,779	14,617	14,523

[1] http://www.ctkidslink.org/publications/covhuskya_kids.xls
[2] http://www.ctkidslink.org/publications/covhuskyb.xls

It is important that financial resources available through Title XIX (Medicaid), Title XXI (State Children's Health Insurance Program), Title V (Maternal and Child Health Block Grant), and other third party payers be blended and coordinated to guarantee necessary coverage for all children and adolescents diagnosed with genetic disorders and having special health care needs.

Concluding Remarks

The Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) has taken a lead role in developing recommendations pertaining to a variety of areas related to genetic testing, such as oversight of genetic technologies, marketing, and laboratories; genetics workforce, education and training; insurance coverage and reimbursement; and genetic discrimination in health insurance and employment (U.S. Department of Health and Human Services, 2002). As such, they are a valuable resource for information and ongoing developments in the genetics arena. At the June 2004 meeting, the SACGHS Education Task Force identified the need for genomics training programs and appropriately trained faculty (Reede, 2004). They recommended that education programs for genetics/genomics be supported and that this is an appropriate role for government to undertake. They also recommended that support be given to programs that enhance diversity among and cultural competency of health professionals. In addition, they encourage the incorporation of genetics/genomics into the certification and licensure process.

As genetic tests and therapies become more available, there will likely be a growing need for genetic counselors and medical geneticists to meet demands for testing and services. Although the numbers of these professionals in Connecticut have been growing, their distribution across the state is disproportionate to the population. Unknown, however, is a standard for determining an adequate ratio of specialists to the population being served. Nevertheless it's important to assure that sufficient numbers are appropriately trained and available. Measures need to be adopted to promote interest in the field of genetics, expand the limited number of genetic counseling programs that exist, and increase scholarship opportunities for students pursuing genetics-related careers.

Increasingly primary care physicians, physician assistants, nurses, psychologists, health educators, and social workers will need a working knowledge of genetics. Because these providers are frequently consumers' entry point into the health care delivery system, they must be able to provide quality genetic information, education, and available resources. Licensing requirements for health professionals should be reviewed so that genetic competencies can be added as needed.

In addition, the public health workforce needs to receive ongoing education about genetic advances in order to apply them to disease prevention and health improvement activities. Work has started in this area with several educational workshops, funded under the state genetics planning grant, that have been provided to DPH staff from a broad range of programs. Thus a foundation has been laid within DPH to raise awareness and stimulate interest in genetics. It's important not to lose the momentum that has been established.

National organizations, such as the Centers for Disease Control and Prevention, the National Coalition for Health Professional Education in Genetics, and the American Society of Human Genetics have developed competency sets and standards for creating a genomically literate workforce (see Table 6). Now strategies are needed to train the workforce to develop the appropriate competencies. As discussed in Chapter 12 of *Genomics and Population Health: United States 2003*, the Michigan experience in genomics training for public health practice provides training strategies and available courses that may provide a good starting point in this effort (Centers for Disease Control and Prevention, 2004).

Enhancing awareness of genetics and genomics and their impact on health among the general public is important for DPH to pursue. Education strategies for our youth differ from those directed toward the general public. Whereas partnerships with the DOE to enhance school curricula could address this priority for the youth, other more personal strategies may be needed to enhance genetics awareness among the general population. The good work conducted within the DPH Family Health Section to educate families about newborn screening needs to be continued. A genetics educational initiative at the federal level is being evaluated at the state level and needs to be monitored for possible implementation within Connecticut. In addition, efforts initiated within DPH to share resources for genetics educational opportunities among the six New England states needs to continue.

Educational materials will need to be continuously evaluated to keep current with the changing demographics of childbearing, cultural and demographic changes, and the ongoing developments in genetic science.

Because Connecticut requires universal newborn screening for certain conditions, DPH should assure that adequate financing mechanisms are in place to support such a program. DPH also has the responsibility to assure an appropriate system of care for children with special health care needs as

identified under the Title V Maternal and Child Health Block Grant. Yet, nearly a third of currently insured CSHCN in Connecticut have insurance that is not adequate to cover their comprehensive needs (Blumberg, 2003). It is important that financial resources available through state funding mechanisms, such as Title V and HUSKY, and by other third party payers be blended and coordinated to guarantee necessary coverage for all children and adolescents diagnosed with genetic disorders and having special health care needs. Health disparities can be reduced by directing publicly financed genetic services to populations most at risk or with the greatest need. DPH should continue to foster policies that improve reimbursement for comprehensive services and coordinated care.

The health care safety net tends to emphasize insurance coverage for children. Most low-income adults without children have no access to health insurance unless they are severely disabled. As adult genetic testing progresses, insurance barriers to access will become an even greater issue.

The American Academy of Pediatrics Task Force Report, *Serving the Family from Birth to the Medical Home: Newborn Screening: A Blueprint for the Future* has developed recommendations focused on newborn screening that could be generalized to genetic testing beyond newborn screening (American Academy of Pediatrics, 2000a). They recommend that state public health agencies:

- Adhere to nationally recognized standards for the validity and utility of genetic tests. States have a responsibility to review the appropriateness of existing tests, tests for additional conditions, and new screening technology and modalities.
- Set standards for laboratories, health professionals, and health care financing plans based on nationally recognized standards and guidelines for follow-up, diagnosis, and treatment.
- Assure that genetic screening is done appropriately through performance monitoring and quality assurance activities.
- Conduct oversight of program operations including test analysis and tracking, collection of data, laboratory quality, diagnosis and treatment at specialty clinics, and research by academic institutions.
- Monitor and evaluate program performance, including outcome evaluations.
- Establish and fund a genetics advisory board/committee that is multidisciplinary (includes advisors representing health professionals, payers, and appropriate government agencies) and has meaningful representation of consumers and the general public. The Committee will advise state officials and others on genetic testing and policies, including review of new tests under consideration by the state and in the development of pilot programs for new tests. The Committee will be involved in the ongoing evaluation of all aspects of the state's process for genetic testing. Oversight activities will include a review of: testing, follow-up, and treatment efforts; the impact on families receiving a false-positive testing result; and the state's process for handling consumer input, including grievances.
- Design and implement public, professional, and consumer education efforts.
- Provide support for coordination and integration of program activities, including information and services.
- Structure interagency coordination to maximize resources and to improve the efficiency and effectiveness of screening systems.

- Ensure adequate financing of all parts of the newborn screening system: screening, short-term follow-up, diagnostic testing, comprehensive medical care/treatment, and evaluation of the system. States should blend resources available through Title XIX (Medicaid), Title V (MCH Block Grant), Title XXI (State Children's Health Insurance Program), and private insurance to guarantee necessary coverage and financing for all children and adolescents with a condition diagnosed through the newborn screening system.

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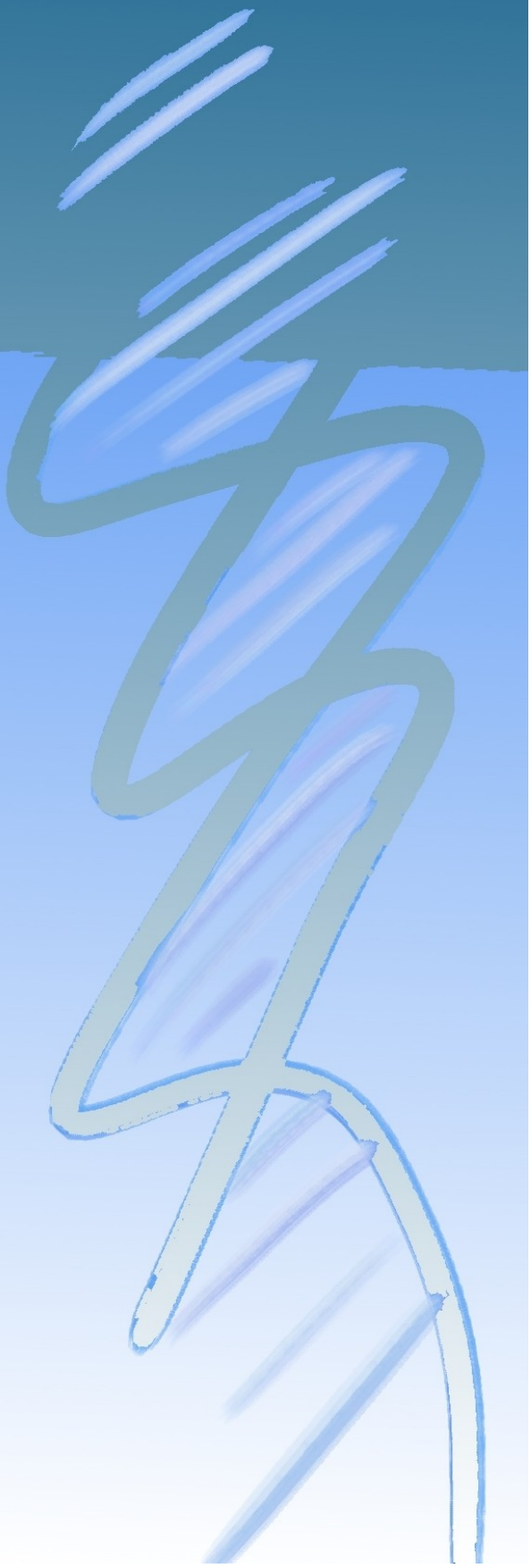
VII. Policy Issues in Genomics



Policy Issues in Genomics

Introduction

Federal and state public health policies strive to improve the health status of Connecticut's residents while providing necessary individual protections. As such, effective policies must be developed regarding the appropriate use of genetic technologies and information derived from genetic testing. Genetic information is personal and potentially prejudicial. Discrimination may be practiced by employers, insurers, the military, and others and must be guarded against. Therefore, laws are needed to ensure that access to personal genetic information is protected. Yet privacy also needs to be balanced with the potential implications that results may have for others, such as relatives, society (e.g. public health), those concerned with public safety, and medical researchers.

Ethical, Legal, and Social Concerns

Although the expanding use of genetic testing and screening should help to improve public health outcomes, the use of genetic information will simultaneously raise significant ethical, legal, and social concerns.

Recognizing that these concerns would arise as a result of the Human Genome Project, the National Human Genome Research Institute (NEGRI) established an extensive research program on the ethical, legal, and social implications of genetics research, referred to collectively as ELSI. The Task Force on Genetic Testing that was created under this program examined the values underlying the use of new genetic technology. They have provided extensive recommendations on the safety and effectiveness of genetic tests as well as overarching principles to guide future policy development (Holtzman, 1997). In addition, the Department of Health and Human Services Secretary's Advisory Committee on Genetics, Health and Society (SACGHS) is also charged with reviewing the impact of genetic technologies on society, including the clinical, ethical, legal, and social implications of genetic testing.

As genomics is increasingly incorporated into public health practice, it is also necessary to view ethical, legal, and social issues from a public health, i.e., population-based, perspective. Some are now referring to this as PHELSI (Public Health ELSI). While some view genetic information as uniquely personal, others view genetic data as just another type of population data to be used along with other surveillance data. With the power of genetic advancements to improve individuals' health, there is also the view that public health has the responsibility to ensure equitable and appropriate access to genetic services.

Ethical Issues

Underlying the discussion about ethical principles is the question of "genetic exceptionalism," which argues that information derived from genetic testing is unique to other health-related information and therefore should have exceptional status and specific privacy protections. One example is the difference between predictive genetic testing and ordinary diagnostic testing. Genetic testing has the potential to predict disabling conditions and discriminate against healthy individuals who may or may not develop a disease. Another important feature of genetic information is the familial risks indicated by the information, whereby information obtained about an individual may have implications for that

individual's relatives. A third unique aspect is the permanency of genetic information in that an individual cannot alter his/her genetic makeup and may face discrimination for a lifetime if found to have certain genetic traits. On the other hand, heightened focus on the exceptional nature of genetic information may enhance the stigma of genetic testing and increase the public's fear regarding its use. Ethical issues are often related to the potential psychological harm that may occur to individuals and family members as a result of genetic testing, to the anxiety that may result from inaccurate or misunderstood results, and to the potential misuse of genetic risk information in insurance and employment discrimination.

Increasingly ethical issues will continue to arise with expanded newborn screening initiatives, potential screening for the identification of individuals at risk for chronic diseases, prenatal screening and reproductive choices, privacy rights surrounding the use of family history, and the desirable use of newborn screening blood samples for other research applications. The public needs assurance regarding:

- Equitable access to genetic services, including counseling, testing, and treatment, and financial coverage for these services.
- Privacy and confidentiality of individual genetic information, particularly in relation to the insurance industry and employers.
- Informed consent, including protecting the right of the consumer to make decisions with regard to genetic testing and research, based on a clear understanding of the benefits and risks.
- Voluntary genetic testing.
- Protection of the rights of those living with genetically determined conditions.

Legal Issues

Ethical issues can also be viewed from a legal perspective by considering laws and policies that guide the use of genomic technology. The National Conference of State Legislatures maintains summaries of state legislation pertaining to genetics and the law. There is considerable diversity among the states and there tends to be a lack of consensus on whether federal, state, or mixed legislation is most appropriate (National Conference of State Legislatures, 2005). In addition, privacy laws pertaining to disclosure of public health data vary within and among states. Federal and state laws pertaining to genetic privacy, employment and insurance discrimination, and newborn screening are discussed in more detail later in this chapter.

Social Issues

Integration of genomics into public health practice also has social policy implications. Concerns exist that eugenics may reappear or that underserved populations may not receive fair treatment as occurred in the 1932-1972 Tuskegee Syphilis Study, in which poor black sharecroppers were denied treatment. There are also concerns that health disparities among different demographic groups may widen and that genetic testing could result in stigmatization and discrimination of the disabled and underserved. In her book, *Future Perfect*, Lori Andrews raises concerns that individuals in disadvantaged groups, such as people of color and individuals with disabilities, are most likely to have their individual decisions overridden on the grounds that it is for their own good or for the supposed good of society. She argues that allowing disadvantaged individuals to make informed choices could reduce the resulting stigmatization and inequalities (Andrews, 2001). She argues that allowing disadvantaged individuals to make informed choices could reduce the resulting stigmatization and inequalities.

Although it can be argued that individuals should be given the opportunity to make informed, educated decisions about their health care and enrollment in research studies, informed consent poses a public health limitation when an intervention is to be imposed on an entire group of the population. Therefore, social policies are needed to avoid discrimination, to address informed consent concerns, to build trust in research participation, and to reduce health disparities. Ongoing discussion about emerging ethical and social dilemmas needs to be promoted and coordinated.

Privacy and Confidentiality

Public concern regarding genetic testing is grounded to some extent in the fear of misuse of genetic information and of inappropriate access to such information. Privacy laws, as opposed to discrimination laws, regulate the processing (collection, maintenance, use, and disclosure) of personal information. Controversy exists whether genetic information should be protected generally as another component of health data, or by special privacy laws. Regardless, protections are needed that balance privacy concerns with use of genetic information for purposes of public health, public safety, clinical research, employment, and insurance reimbursement.

Historically DPH and its predecessors have been granted broad powers through statute to protect the public's health. As a result, identifiable individual information is required to be submitted for all reportable diseases to DPH. DPH also requires reporting to the Tumor Registry of cancer cases that are diagnosed in a number of settings. Newborn screening data are also disease specific and approved by statute for collection. It is important that DPH continue to balance public health interests with individuals' rights to privacy as needs arise for information about more diseases and conditions, particularly genetic-related information. Under Connecticut General Statute 19a-25, personal data procured by DPH for the purpose of reducing morbidity or mortality from any cause or condition is considered to be confidential.

A patchwork of federal and state laws exists to protect the confidentiality of personal health information. The federal Health Insurance Portability and Accountability Act of 1996 (HIPAA), effective April 2001, provides substantial protection at the federal level for health information, including genetic information, created or received by private health care providers, clearinghouses, and health plans. However, it does not regulate entities such as pharmaceutical companies, life or disability insurers, or employers. It also does not protect the actual tissue or blood sample that generated the genetic information.

According to the Connecticut Department of Public Health, Notice of Privacy Practices, the DPH is considered to be a hybrid entity, i.e., a covered entity whose business activities include both covered and non-covered functions under HIPAA; and "the only health information DPH receives, generates, and maintains that is governed by HIPAA is information at the DPH Laboratory" (Connecticut Department of Public Health, 2003). As a covered health care component, the Laboratory is required to maintain the privacy and security of personal health information, such as newborn screening data, pursuant to HIPAA requirements. Under Title 45, Code of Federal Regulations, Part 164.512(b) of HIPAA, components/entities covered under HIPAA may disclose the information to health care institutions and other providers for treatment and payment purposes, and to public health authorities authorized by law to collect and receive information for preventing and controlling disease, injury, or disability. Those HIPAA provisions as well as the Clinical Laboratory Improvement Amendments of 1988 (CLIA) and state statutes and regulations permit the DPH Laboratory to disclose newborn

screening data to the DPH Newborn Screening Program in the Family Health Section. Those programs, in turn, comply with state laws governing the confidentiality of health information.

Although the HIPAA privacy regulation sets a federal floor of privacy protections, it does not preempt state laws if such laws provide stronger privacy protections or cover entities not regulated under HIPAA. Connecticut General Statute 38a-999 requires insurance institutions that regularly collect, use, or disclose medical records to have protections to safeguard against the unauthorized disclosure of sensitive health information, including results from genetic testing and the fact that an individual has undergone a genetic test. As of May 9, 2005, 26 states had passed genetic privacy laws that require informed consent prior to disclosure of genetic information by health insurers, but Connecticut is not one of them (National Conference of State Legislatures, 2005). In addition, laws in 16 states other than Connecticut require informed consent before a third party can perform or require a genetic test or before they can obtain genetic information (National Conference of State Legislatures, 2005). Connecticut should, therefore, review its privacy and informed consent policies pertaining to genetic testing and disclosure of genetic information.

Discrimination

Irrespective of privacy and confidentiality issues, personal medical data including genetic data should not be used to discriminate unfairly against individuals. Discrimination prohibits unfairness by restricting the inappropriate use of specific personal information (e.g., age, sex, race, medical status). Although genetic testing holds great promise for improving the public's health, public fear of genetic discrimination in employment and insurance is high and may deter patients from receiving genetic testing (Hall, 2000).

Employment

Title VII of the Civil Rights Act of 1964 prohibits employment discrimination based on race, color, religion, sex, and national origin. However, it does not take into account medical status. The Americans with Disabilities Act of 1990 (ADA) restricts medical examinations and inquiries in the workplace, and forbids employment discrimination based on disabilities that have no influence on job performance. In 1995, the Equal Employment Opportunity Commission (EEOC) interpreted "disability" in the ADA to include genetic predisposition to disease, but several court rulings since then have questioned whether the Supreme Court would accept this EEOC interpretation. In February 2000 President Clinton banned genetic discrimination in the federal workplace and called on Congress to pass a federal genetic information nondiscrimination law for private sector employment.

By a vote of 95-0, the U.S. Senate passed the Genetic Information Nondiscrimination Act (GINA) (S.1053) in October 2003, but the bill did not make it to a vote in the House of Representatives before the end of the 2003-2004 congressional session. Beginning in 2005, the bill was reintroduced in the Senate as S.306 and passed in February 2005 by a vote of 98-0. The companion bill, H.R. 1227, was introduced in the House on March 10, 2005, and has been referred to three committees where it awaits consideration. With regard to employment discrimination, GINA is expected to prohibit discrimination in hiring, compensation, and other personnel processes; prohibit the collection of genetic information, and allow genetic testing only to monitor the adverse effects of hazardous workplace exposures; and require genetic information possessed by employers to be confidentially maintained and disclosed only to the employee or under other tightly controlled circumstances.

Currently genetic nondiscrimination-in-employment laws are in place in 32 states, including Connecticut. Although the laws expressly prohibit genetic discrimination in employment, the laws contain different exceptions where employers can potentially use genetic information in the workplace, particularly when it might be job-related (National Conference of State Legislatures, 2005). Factors that may be considered to be appropriate use include the relevance of the genetic information to job qualifications, health and safety issues, and employer liability.

Connecticut General Statute 46a-60 prohibits discriminatory employment practices generally. Under Connecticut General Statute 46a-60(a)(11), an employer may not request or require genetic information from an employee or applicant, and genetic discrimination is prohibited in hiring, firing, and privileges of employment. The law does not contain exceptions where employers can potentially use genetic information in the workplace. In light of the September 11, 2001 catastrophe at the World Trade Centers, attitudes may ultimately change whereby office workers would register their DNA for the purpose of being able to later identify their remains. Further study of permissible employer uses of genetic information may be desirable.

Health Insurance

Consumers worry that genetic risk for disease will be considered a "preexisting condition," and that a health insurer will deny them health insurance or make them pay higher premiums. A patchwork of federal and state laws governs health insurance discrimination based on genetic information.

The Health Insurance Portability and Accountability Act prevents health plans and insurers, in the *group* market, from refusing to enroll an individual due to that individual's genetic information. It also prohibits charging an individual (or family) in a group more than others in the group on the basis of the individual's (or dependent's) genetic information. The HIPAA also prohibits insurers in the *individual* health insurance market from refusing to enroll, for any health-related reason, a subset of individuals who are leaving the group market and who meet other prerequisites.

States have acted to fill in the gaps left by HIPAA. In Connecticut, Section 38a-816(19) of the Connecticut General Statutes governs unfair practices by health insurers whereby discrimination by *group* and *individual* health insurance providers on the basis of genetic information is an unfair method of competition and an unfair deceptive act. Insurance eligibility cannot be denied based upon genetic information, and the use of genetic information for risk selection and risk classification purposes is strictly prohibited. Connecticut General Statute 38a-476 similarly prohibits group or individual health insurers from treating genetic information as a preexisting condition in the absence of a diagnosis of the condition. Such prohibition extends to insurance arrangements such as multiple employer welfare arrangements, as defined in Section 3 of the Employees Retirement Income Security Act of 1974 (ERISA).

Because ERISA preempts state laws pertaining to self-funded employee benefit plans, there is a concern that genetic discrimination is not prohibited by such plans. An exception to the preemption rule protects the right of states to regulate "insurance," so if an insurance contract exists in an employee benefit plan, ERISA may not preempt and state protections may apply.

The Genetic Information Nondiscrimination Act (GINA) under review by Congress in 2005 would prohibit health insurance enrollment restrictions and premium adjustments on the basis of genetic information; prevent health plans and insurers from requesting or requiring that an individual take a genetic test; and prevent health plans and insurers from pursuing or being provided information on

predictive genetic information or genetic services prior to enrollment. GINA covers all health insurance programs, including those regulated by the federal government under ERISA, state-regulated plans, Medigap, and the individual market.

Life, Disability, and Long Term Care Insurance

While states such as Connecticut have enacted laws that prohibit the use of genetic information for risk selection and risk classification in health insurance, few states restrict the use of genetic information in life, disability, and long-term care insurance. Connecticut does not have any protections against genetic discrimination in these areas of insurance.

Because these types of insurance are purchased more frequently by individuals rather than by groups, personal genetic information may be more likely to factor into underwriting these policies. Insurers could force individuals to undergo genetic tests and use genetic information to deny insurance coverage or to charge higher premiums. That is, applicants may go uninsured because they may be predisposed to a genetic condition, even though they are currently healthy.

On the other hand, insurers worry that restrictions on the use of genetic information to underwrite life, disability, and long-term care insurance policies could result in adverse selection, which is a financial advantage that applicants gain by purchasing insurance based on risks known or suspected by them but unknown to the insurer. Insurers typically try to prevent adverse selection by reviewing medical records and classifying an individual's risk, whereby individuals at higher risk are charged higher premiums or even denied coverage.

Although several states prohibit insurers from requiring applicants to undergo genetic testing, insurers in those states are allowed the use of genetic test results if such testing has occurred. Connecticut does not prohibit life, disability, and long-term care insurance providers from requiring applicants to undergo genetic testing nor does it restrict the use of genetic information in underwriting policies. At some point, laws may be needed to ensure that the insurance market remains open to those with genetic predisposition to disease. At a minimum, informed consent policies may be desirable so that individuals are made aware of the meaning and risk of genetic testing before agreeing to submit to testing by an insurer. Laws may also be needed that require actuarial justification in setting premium rates based upon genetic information in order to prevent insurers from acting on the basis of misinformation. Further study of these issues is needed.

Newborn Screening

As a population-based public health service, Connecticut's newborn screening program is located in the state Department of Public Health, and is governed by state law. Because no uniform national policy exists, great variations exist among state newborn screening programs. States differ regarding newborn screening laws, conditions tested, advisory boards, processes for informing parents, exemptions, fees assessed, and blood sample storage policies and use of blood samples. Mounting pressure is being exerted on state legislatures to increase the number of conditions included in newborn screening programs. As of June 30, 2004, the March of Dimes recommended that programs perform nine core tests plus newborn hearing screening (March of Dimes, 2004). However, in September 2004, the March of Dimes increased its newborn testing recommendations to include the 29 core conditions being recommended by the American College of Medical Genetics (ACMG) (March of Dimes, September 22, 2004). The report, *Newborn Screening: Toward a Uniform*

Screening Panel and System, was prepared by ACMG for the Maternal and Child Health Bureau of the U.S. Health Resources and Services Administration. Although no uniform national policy exists for newborn screening as of May 2005, guidance will be forthcoming from the Department of Health and Human Services Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children.

As of June 30, 2004, Connecticut was one of only 21 states to offer testing for the recommended core group of nine genetic/metabolic disorders and hearing deficiency (March of Dimes, 2004). Testing for the ninth core condition, known as medium-chain acyl-CoA dehydrogenase deficiency (MCADD), was implemented in Connecticut beginning in 2004. Since that time, the newborn screening panel has expanded to include additional amino acid disorders, organic acid disorders, and fatty acid oxidation disorders. As of January 1, 2005, DPH screens for more than 40 disorders. Although recommended as one of the 29 ACMG core conditions to include in a uniform screening panel, cystic fibrosis is not currently *mandated* for screening in Connecticut. However, twenty hospitals in the state do screen newborns for cystic fibrosis so that approximately half of all newborns are screened annually. This creates a lack of uniformity within the state.

With expansion, fees for newborn screening have been increased from $18 to $28. However, these fees are much less than those collected in other states. The average fee collected in states that currently test for MCADD is $45 (based on data from the National Newborn Screening and Genetics Resource Center as of August 2004). Approximately $1.2 million is collected annually from newborn screening fees, of which only $335,000 is directly allocated to the State Laboratory for testing services. The remaining fees go into the State's General Fund, from which state agencies' budgets are allocated.

Newborn health screening, including the required tests, exemptions, and fees, is legislatively mandated under Connecticut General Statute 19a-55. Informed consent is not required, but parents may refuse screening for their infant if it is in conflict with their religious tenets or beliefs.

Storage and retention policies of newborn blood spots are not legally dictated. The DPH Laboratory stores the newborn blood spots for 5-6 months, at which time they are destroyed. Ethical issues may arise in the future regarding the additional use of newborn blood samples for epidemiological studies of genetic variation in populations. Unclear is ownership of the blood samples and whether informed consent would be required due to the impracticality of re-contacting subjects from population studies. While informed consent is accepted as a requirement for genetic research, no consensus exists regarding informed consent standards for the use of stored samples.

A Genetics Advisory Committee (GAC) advises DPH on newborn screening issues, such as the expansion of screening. Newborn screening policies have also been strongly influenced by the state's legislature. This raises concerns about available resources and the appropriate use of genetic technologies. The 2000 DPH report, *Genetic Testing: A Plan for the Future*, indicated that expanded screening using tandem mass spectrometry would require additional funding for capital equipment, personal services, educational needs, and data systems (Connecticut Department of Public Health, 2000). However, state budget cuts to DPH and genetic treatment centers have made expanded screening difficult to implement. Development of a more coordinated process with systematic advisory input is needed to assure appropriate use of genetic technologies and adequate financing to support comprehensive testing, tracking, and treatment within DPH. Lack of a coordinated process raises concerns for ongoing expansion of newborn screening as well as future screening for later-onset disorders, such as asthma and diabetes.

In February 2004, the national Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children was created pursuant to Title 26 of the Children's Health Act 2000. Its purpose is to advise and guide the Secretary of the U.S. Department of Health and Human Services regarding the most appropriate application of universal newborn screening tests, policies, guidelines, and programs for effectively reducing morbidity and mortality in newborns and children having or at risk for heritable disorders. The Committee is also to provide technical information that will assist state public health agencies to develop policies for newborn and child screening having or at risk for heritable disorders. DPH and its Genetics Advisory Committee may find guidance from this national advisory committee.

Genetic Screening of Populations

DPH currently provides population genetic screening only to newborns. However, as advances occur in genetic testing technology, policymakers can expect the demand to increase for genetic screening of certain gender-specific and/or ethnic subpopulations. Such screening already exists as in the example of carrier screening for Tay-Sachs disease as part of reproductive planning for parents who are descendents of at-risk groups such as Ashkenazi Jews, Cajuns, or Amish. So far, this has been beyond the purview of public health.

In addition, genetic testing for increased disease risk may eventually become possible for common chronic disorders, for determining susceptibility to common infectious diseases, for determining vulnerability to environmental health hazards such as cigarette smoking, and for determining drug efficacy due to the fact that people metabolize drugs differently. Questions are likely to arise as to whether the whole population, or only certain subpopulations, will benefit from screening or whether testing ought to only be recommended on a case-by-case basis.

According to a population-based survey in the United Kingdom, public interest in genetic testing for susceptibility to both heart disease and cancer is high (Sanderson, 2004). In the United States, commercial ventures have already begun to market genetic testing [for breast cancer] directly to consumers, but no process exists for review of the accuracy of advertising claims about the validity and utility of genetic tests (Centers for Disease Control and Prevention, 2004). The public needs to be educated so that they expect standards of accuracy for genetic tests comparable to those that exist in other diagnostic and prognostic testing efforts in clinical medicine, because a study of offspring of Alzheimer's disease cases found that 40 percent would be willing to accept high false-positive and false-negative error rates in tests (Bassett, 2004).

As genetic testing works its way into general health practice, policymakers will increasingly be confronted with the need to balance patients' desire to access new genetic technologies with ethical considerations and consumer protection. The role of government and public health agencies is unclear, particularly in areas concerning reproductive choice.

Although no policies or programs currently exist within DPH to address the many issues surrounding these genomic advances, there is likely to be a growing need for meaningful information and regulation with respect to the provision of genetic screening on a population-wide level. Ultimately DPH may need to assume responsibility for guiding the use of genetic technology for the benefit of Connecticut's residents. The Action Plan presented in Chapter IX begins to address this issue.

Concluding Remarks

While potential public health benefits of genetic testing and screening support their use, underlying risks to individuals and populations require awareness and responsibility. Following the public health three-core-function continuum, there will be an ongoing need to *assess* ethical, legal, and social implications surrounding the use of genetic technology and information and to develop *policies* and laws to *assure* the public that their interests are being protected and that genetic services, such as testing and counseling, are equitably available and accessible. There is a need for DPH to facilitate discussion and consensus building to guide genetic policy development in Connecticut.

As of May 2005, there is no comprehensive federal law that safeguards the privacy of health information, including genetic information. The federal Health Insurance Portability and Accountability Act only protects health/genetic information created or received by health care providers, clearinghouses, and health plans. It does not regulate entities such as pharmaceutical companies, life or disability insurers, or employers. Likewise there is no federal genetic nondiscrimination law that prohibits employment and insurance discrimination on the basis of genetic information. Although the Genetic Information Nondiscrimination Act (GINA) (S.306) unanimously passed in the Senate in February 2005, it is still under debate in the House of Representatives (H.R.1227). Connecticut laws have tried to fill in some of the gaps left by federal legislation, but holes still exist. Therefore, it is important to further review privacy, discrimination, and informed consent policies pertaining to genetic testing, disclosure, and use of genetic information in Connecticut. Policy recommendations that address the balance between privacy and the importance of population-based data to public health need to acknowledge the role, contributions, and authority of legally mandated public health activities, such as newborn screening.

No uniform national policy exists for newborn screening although guidance will be forthcoming from the Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children. Instead, states govern their own programs. Connecticut has expanded its screening panel to include more than 40 conditions, but cystic fibrosis is not one of them. There is no formal process in place in Connecticut to guide expansion of testing. Therefore, there is a need for DPH to establish a process for coordinating state genetics policy issues in general, which could then be adopted for newborn-screening expansion. As part of the process, it is important to bring together a broad array of representatives to advise, debate, and recommend policy options. Representatives should consist of stakeholders with a statutory, economic, or professional interest in genetic polices, including the public, as well as experts who can assess the policy implications of research findings and evaluate policy options.

In addition to developing policies and passing legislation, public health interests can best be promoted by actively engaging the public in policy development, by supporting public education in genomics, and by providing balanced information that tempers commercial marketing. As such, it is important for DPH to continue its leadership role.

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VIII. Determination of Public Health Genomic Needs: Process and Findings

Determination of Public Health Genomic Needs: Process and Findings

Introduction

Earlier chapters in this document refer to the many genetic advances in the past decade, due largely to the Human Genome Project and other research. This expanded knowledge base will continue to have an impact on public health policy and service delivery, as well as on health care and social services practice. To optimize the impact of this scientific knowledge, advances in genetics will need to be integrated into public health activities (Kaye, 2001).

Incorporation of genomic aspects of disease into public health practice has the potential to make the goals of health promotion and disease prevention more effective. Public health agencies are increasingly being called on to develop an infrastructure that is equipped to address the growing implications of genomics (Centers for Disease Control and Prevention, 2003).

To assist states in these efforts, recommendations have been developed and disseminated through means such as Genomics: A Guide for Public Health. This comprehensive guide was developed by the Genomics and Public Health Toolkit Workgroup under the leadership of ASTHO. The guide serves to assist state health departments plan for the effective use of genetic information and developments. Although recommendations vary somewhat depending upon the source, common areas include developing a comprehensive strategic plan for genetics; improving the genetics knowledge base of health departments; assessing and addressing genetic and general medical workforce needs; developing methods to share genetics resources across program areas; and developing both the state health department's internal capacity and community partnerships to assess and address the general public's and affected families' needs for information and privacy protection.

Similarly, the intent of the Connecticut Genomics Action Plan is to assist in the translation of genetic developments and technology into public health and health care practice, and to generate recommendations for long-range planning and infrastructure development regarding the state of the art in genomics in the coming decade. To do so effectively required an informed planning process with input acquired from various sources and populations on factors ranging from needs, service utilization, access and financing issues, and regulatory issues, among others.

Various strategies were utilized to solicit input, the foremost of which included:

- Convening a genetic stakeholders advisory group
- Conducting a statewide needs assessment
- Establishing an internal "Gene Team"
- Establishing a linkage with the Department Newborn Screening Unit's Genetics Advisory Council
- Structuring input opportunities through a Genetics Seminar Series and a statewide Genetics Symposium.

Genetic Stakeholders Advisory Group

It was recognized early on that a key strategy in the Department's efforts to develop and inform a state genomics plan would be to convene a group of genetics experts or "stakeholders." These individuals were seen as possessing a unique skill set, experience, or knowledge base in their respective fields relating to genetics. Such an approach has also been utilized in other states, such as Texas, Michigan, Washington, and Rhode Island.

The DPH administration assembled a list of members for inclusion in the group, with input from the Genetics Planning Team on the important aspect of affected family representation. The CT Genetic Stakeholders Advisory Group was composed of:

- 3 clinicians
- 3 academics
- 2 representatives from affected families
- 3 industry representatives
- 1 ethics and legal expert
- 1 genetic epidemiologist

The body was invited for an initial series of three half-day monthly meetings during the fall of 2003. The meetings were developmental in nature, each building upon the work of the previous session. Relevant materials such as articles and publications, and genetics plans from other states were disseminated. Additionally, structured "problem solving" hypotheses were posed for discussion, issue identification, and generating recommendations. Such questions included: What should be the role of the Department of Public Health with regard to genetics? What partnerships are needed to enable such developments? What infrastructure is needed to integrate genetics into DPH? An additional exercise posed a hypothetical situation to the group regarding adult cystic fibrosis carrier testing and related issues and public health implications. (See *Appendices* for full report.) The genetics expertise within the group was tapped for leading certain segments of issue identification and discussion.

The Genetics Stakeholders Advisory Group (GSAG) developed into a cohesive, well-operating body, benefiting greatly from the wide range of expertise that was evidenced. In addition to a draft mission statement (see *Recommendations*) and set of preliminary recommendations (see *Recommendations*, and *Appendices*), the group made additional important contributions to the genetics planning process. Of particular note is the recognition of the need for an informed, cohesive body able to convene periodically *on an ongoing basis* to consider and advise the Commissioner on presenting genetics issues, such as new testing and treatment, needed education and training, pending legislation, ethical considerations, and so on. Additionally, their recommendations included the development of the internal capacity for greater genetics presence among units in DPH to respond to the increasing impact of public health genetics across the lifespan.

The Genetics Stakeholders Advisory Group also assisted in the development of a draft mission statement for genetics integration, which reads as follows:

To integrate evolving genetic information and technology into effective public health actions which ensure equitable access to comprehensive and culturally appropriate genetic services in an effort to promote health and prevent disease and disability while maintaining personal choice and privacy.

Statewide Needs Assessment

Previous needs assessments, attitudinal surveys and questionnaires on genetics issues have been conducted at the regional and national level and are referenced elsewhere in this document. Such efforts have identified emerging issues of note to the development of a genomics action plan for Connecticut. However, there were several reasons for conducting a local, state-specific genetics needs assessment to inform this plan.

First, a Connecticut-specific needs assessment was viewed as a planning tool that would help both the Department and those partnering with it in this endeavor to identify the breadth and depth of need association with integrating genomics into public health practice. Secondly, it was seen as a means to determine the level and use of various related services; to determine the level of genetic "literacy" within the general public, affected families, and general medical practitioners; and to determine the extent and nature of workforce training needs. Lastly, and equally important, a needs assessment effort was seen as vital in setting relevant priorities among the recommendations for future action.

In identifying the populations possessing important genetics-related knowledge, feedback and experience, it was concluded that input would be solicited from families affected by genetic conditions, general medical practitioners, and genetics professionals in Connecticut. A consultant was enlisted to work with the Genetics Planning Team to develop the surveys, assist with survey administration, collect the data, and generate reports.

What follows is an overview of each survey effort and the beneficial outcomes of each.

For the **physician's survey** (see *Appendices*), 847 printed surveys were distributed in June 2004. Recipients were selected randomly from the DPH licensed physician database. Between 300 - 400 printed **consumer/family surveys** (see *Appendices*) were distributed to various sources (support groups, in-state associations, genetic service centers) in June 2004, and an *online* version was released through a statewide advocacy network in September 2004. For the **genetics professionals' survey,** an *online* needs assessment survey of Connecticut based genetics professionals was carried out during June of 2004. This Internet based survey (see *Appendices*), was administered to 218 individuals considered Connecticut based genetic health professionals. Names were obtained from the National Society of Genetic Counselors, American College of Medical Genetics and American Society of Human Genetics membership directories, as well as DPH offices.

Two of the three reports generated had a sufficient number of responses to be deemed a valid sample of the groups surveyed. The affected family returns were too low, however, to be considered valid, despite the additional release of the online survey.

Although full reports appear in the appendices, the following summaries illustrate key findings gained from the survey effort.

Genetics Professionals Survey: Eighty-two responses (45.3% response rate) were obtained and several themes were noted as major obstacles to the provision of genetic services in the state. The primary hurdles, as perceived by the genetics professionals, include lack of funding and insurance reimbursement for services, insufficient numbers of genetics professionals to support the state's clinical needs, and the need to increase both consumer and health professional educational efforts.

Medical Professionals Survey: One hundred and fifty-seven responses (18.8% response rate) were returned and qualified for analysis. The majority of respondents saw primarily adult patients only (52.2%), while 28% of respondents were pediatric-related physicians, and 19.7% saw both age groups.

In general, whether a respondent's patient population was pediatric, adult, or both had an impact on the response to many questions related to integration of genetic services into their practices. Those physicians who see both adults and children were often less likely to discuss testing options with their patients or ordering those tests. In addition, they were less likely to refer their patients to genetic centers for testing, counseling and test interpretation.

Several findings consistent with the **genetics professionals survey** were identified including the need for more professional education, better reimbursement for genetic services, and increased access to genetic services. About half of the respondents felt that statewide availability of affordable genetic services should be the state public health department's top genetics-related priority.

The respondents are interested in advancing their genetic knowledge, particularly in the areas of at-risk patient identification (71.9%), advances in genetic technology (65.5%), resources for genetic testing, evaluation and counseling (59.7%), and genetic screening issues (56.1%). They want to continue using the methods they have employed in the past to learn this information -- by reading medical journals, consulting with experts, and attending grand rounds and local medical meetings.

DPH Internal "Gene Team"

Establishment of an internal working group is seen as a useful vehicle for information dissemination and advocacy within the Department and work in this area has commenced through the development of an internal "Gene Team."

Presentations and relevant resource materials regarding the Genetics Planning Project and its potential implications were provided to DPH managers to elicit their understanding and support prior to the kick-off of the Public Health Genetics Seminar Series in the fall of 2003. For the Gene Team, this series served as an opportunity for both genetics education and orientation to emerging issues that may impact their work.

The Gene Team currently is comprised of twenty-eight members, with representation across departmental units focused on chronic diseases, infectious diseases, environmental health, surveillance, newborn screening, and planning.

Oversight by a dedicated Genetics Coordinator would greatly facilitate the ongoing activities of this Departmental resource and would give impetus to ongoing integration of genetic developments in the various DPH programs.

Symposium

Participants at a one-day Symposium on Public Health Genetics in April 2004 were given the opportunity to respond to questions posed by a public health hypothetical situation (see *Appendices*). Participants chose one of five breakout sessions, which were chaired by members of the Genetics Stakeholders' Advisory Group and other genetics professionals throughout the state, and discussed a series of directed questions related to one of four focus areas: Public Policy, Surveillance, Services, or Education. Notes of each session were recorded (see *Appendices*). At a joint session following the

breakout sessions, co-facilitators presented summaries of each breakout, with key issues presented and recommendations highlighted. Summary proceedings appear in the Appendices.

Questions that were considered during the breakout sessions included:

- How can equal, culturally sensitive access to testing, screening, and genetic counseling be assured?
- How can confidentiality and privacy of test results be ensured?
- 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?
- Who needs genetic information, and how can such information best be provided?

Summary of Identified Needs and Gaps

Each preceding chapter has presented information and unique features relating to the integration of genomics into public health. Common across chapters, however, are concluding remarks that coalesce the elements of need posed within each, and from these emerge common themes and cross-cutting issues addressed in the "Recommended Actions" chapter of this plan. They address the advances in genetics and related technologies that are causing significant changes in health care. As such these developments are posing new challenges to the public health field and are serving as the impetus to plan for the integration of genetic discoveries into areas such as disease prevention and management strategies -- areas not previously viewed as having a genetic connection.

Chapter II, the "Genomics Revolution and Emerging Public Health Applications," addresses the context and resulting pressures of the new and emerging advances in genetics. Important among them is the expanding body of scientific knowledge about genetics, reaching far beyond the current maternal-child health arena into areas such as chronic, adult-onset conditions.

The "Overview of the State of Connecticut" provided in Chapter III lays out important demographic and economic changes taking place that warrant attention as genomic developments for public health are considered. They include an aging population, more racial and ethnic diversity, childbearing at older ages, and socioeconomic disparities. These factors will result in major shifts in the health care needs of Connecticut's citizenry, and imply the need for expanded genomics service capabilities with consideration given to its equal accessibility.

From Chapter IV, the need for "Public Health Registries and Surveillance Systems" was explored together with the importance of integrating data systems. As new genomic developments uncover more associations between previously assumed unaffected conditions, so too will the specific data silos currently maintained need to become better integrated. Ultimately, data integration efforts will need to expand into other public health areas dealing with chronic and infectious diseases and environmental health, to help quantify the genetic basis of disease in Connecticut's population.

In describing the current "Genetic Health Care Services in Connecticut" in Chapter V, critical needs emerge. Although genetics-related health care services are provided within the private and public sectors, the public sector allocates most of its resources to prenatal health care services and newborn screening. Many of the local genetics networks and oversight boards cited have a similar focus, pointing to the need for a genomics infrastructure, capable of addressing complex genetic issues across the lifespan. Additionally, important gaps emerge in both the geographic access to services and with the development of new screening and testing technologies.

Chapter VI, "Genetic Resources in Connecticut: Workforce, Education, and Support," identifies the need for workforce development, public and professional education in genetics, and appropriate financing mechanisms for genetic services. As direct genetic health care services spread from specialty centers into physician's offices, the need grows for a wider workforce of health professionals to be educated and trained in genetics. With the demand for genetic counselors expected to increase, so too must efforts grow to promote interest and training in the field, and to assure their adequate preparation for service, perhaps through licensure. The need for increased genetics education to prepare for an informed public health workforce is also important to provide for the application of relevant genomic information into disease prevention and health improvement. Lastly, and of critical importance, as genetic tests and therapies become more available and are marketed to the general public, there is an increasing need for genetics education efforts targeted at primary care physicians, physician assistants, nurses, social workers, and the general public, i.e., the marketing targets.

Timely and pressing issues pertaining to the ethical, legal, and social concerns surrounding the expanding use of genetic testing and screening, and the use of genetic information are discussed in the "Policy Issues in Genomics" of Chapter VII. Tantamount to any discussion of these issues is an understanding that while potential public health benefits support the use of genetic testing and screening, underlying risks to individuals and populations require awareness and responsibility. Policies and legislation are needed to provide guidelines and protections, in tandem with public education in genomics. It is vital that public health generally, and DPH in Connecticut, take a leadership role in these efforts.

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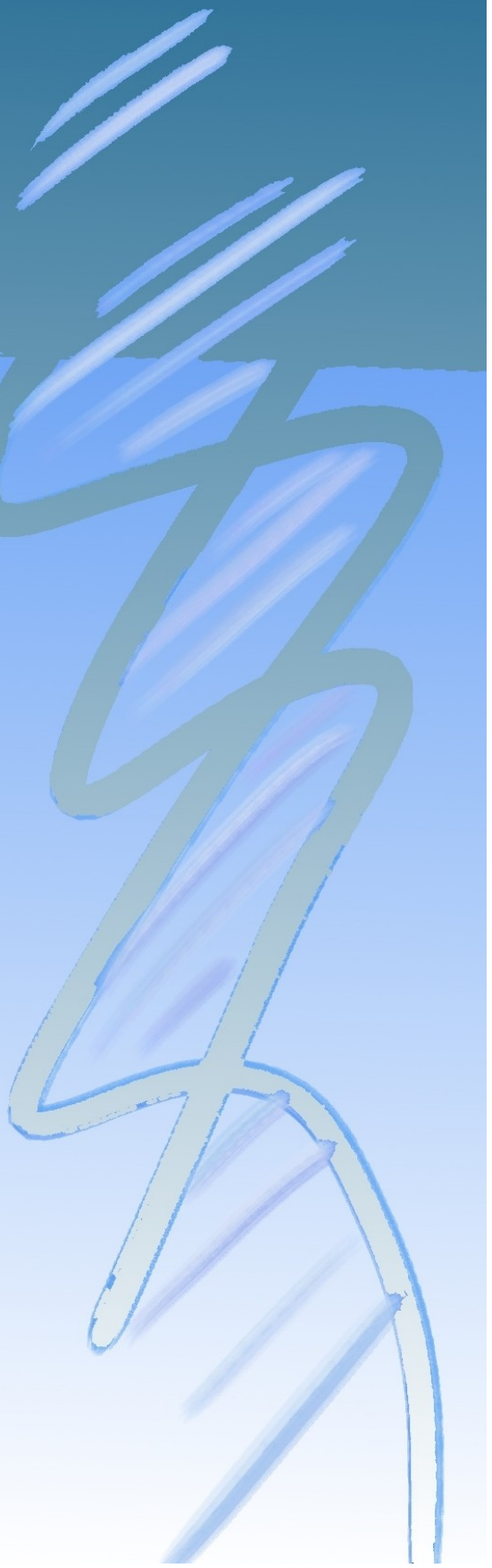
IX. Recommendations and Action Plan



Recommendations & Action Plan

The Connecticut Genomics Action Plan has presented an overview of the current state of genetics and a look to the future – with a key role for public health *genomics* nationally and in Connecticut.

It is a compelling picture of the necessity to create a significant and functional genomics presence within the Connecticut public health arena. The vision holds that by the year 2010, a genomics infrastructure could be active and fully functioning and capable of responding to the genomic needs of Connecticut's citizens.

The priorities, goals, and objectives that are anticipated to realize this vision within DPH follow. It is anticipated that short-term objectives will be targeted for completion within twelve to twenty-four months. The accomplishment of long-term objectives will require two years or more.

Priority I - Infrastructure

Establish a formal, stable, and sustainable infrastructure that promotes the integration of genomics into all relevant areas of public health across the lifespan.

Goal I.1

Create an Office of Genomics within DPH that has agency-wide reach and experienced Directorship.

Short-Term Objectives

I.1.a. Continue the Department's commitment to genomics through the creation of an interim Virtual Office of Genomics (see diagram, Infrastructure A.)

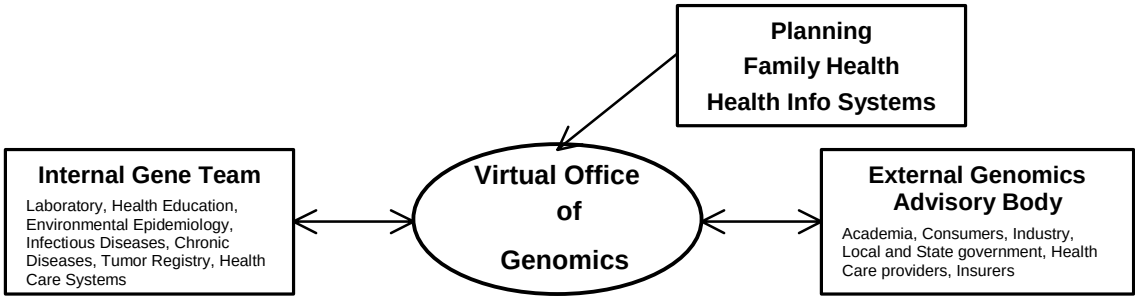
I.1.b. Establish position, and recruit a Director of Genomics, with broad-based experience in genetics to direct DPH genomics policy and activities.

I.1.c. Seek funding sources to fully implement the Connecticut Genomics Action Plan.

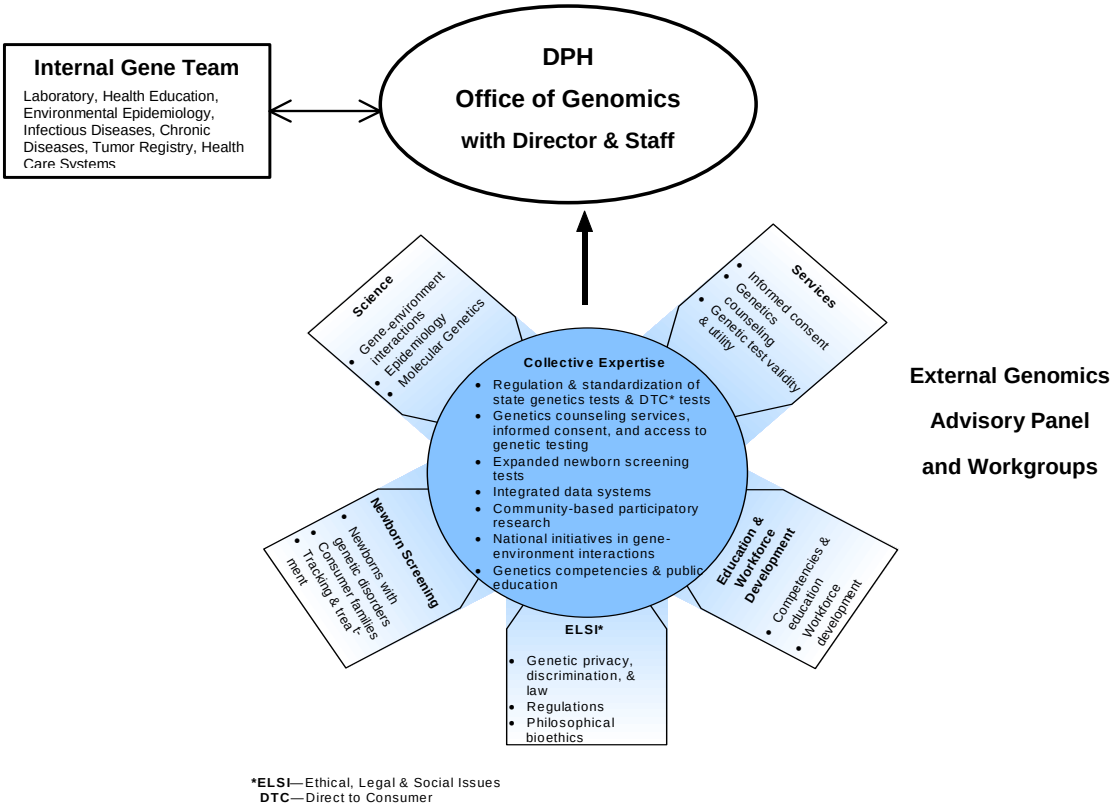
Long -Term Objectives

I.1.d. By year 2010, have developed an active, fully functioning Office of Genomics, operating across units to serve as a clearinghouse and central site for genomics within DPH (see diagram, Infrastructure B.)

Infrastructure A:
Continuing existing structure,
Transitional to Infrastructure B



Infrastructure B:
Ultimate Infrastructure



***ELSI**—Ethical, Legal & Social Issues
DTC—Direct to Consumer

Goal I.2

Establish internal and external interdisciplinary genomics advisory capacity within DPH.

Short-Term Objectives

I.2.a. Formalize the ongoing internal Gene Team for the purposes of internal genomic development, information dissemination and advocacy within DPH, and expanding the genetic competencies among Departmental staff.

I.2.b. Identify, recruit and formalize a multidisciplinary external Expert Genomics Advisory Panel with the capability of guiding DPH genomic integration and ongoing efforts. The Panel should consist of researchers, scientists, educators, health professionals, consumers and affected families, payers, community leaders, legal experts, ethicists, and representatives from advocacy groups and appropriate governmental agencies.

Goal I.3

Promote genomic public health interests by engaging the public and mobilizing community partnerships at the state and local levels to identify those communities that could benefit from genetic services and provide feedback about related needs and attitudes within the state, and by looking to key players at the national level for guidance and support.

Short-Term Objectives

I.3.a. Establish partnerships with local health departments, community groups and health service providers.

I.3.b. Outreach to other state agencies (Departments of Mental Retardation and Social Services, among others) and community providers to clarify and establish appropriate roles for each regarding genomic issues.

Long-Term Objective

I.3.c. Monitor community attitudes about genomics and genetic services, and facilitate consensus-building for genetic policy development.

Goal I.4

Develop policies and practices and support legislation that ensure quality genomics programs throughout the state, and that address the ethical, legal and social implications of the expanding use of genetic testing and genetic information.

Short-Term Objectives

I.4.a. Facilitate regular, ongoing review and discussion of ethical, legal and social implications for genomic policy development.

I.4.b. Establish a process for coordinating state genomic policy issues pertaining to genetic testing, disclosure and use of genetic information, guided by reviews of national/state privacy, discrimination, and informed consent policies.

Long-Term Objectives

I.4.c. Foster policies and support legislation that improve reimbursement for comprehensive genetic services and coordinated care.

I.4.d. Ensure regular, periodic dissemination of pertinent privacy regulations and policies to public health and other healthcare professionals who are impacted by them (cross referenced with Priority II/Education).

Priority II - Genomics Education

Educate the public about genomics, and ensure a public health and healthcare workforce that is competent in genomics, including the associated ethical, legal and social implications.

Goal II.1

Inform the general public and policymakers about genetics and its impact on health.

Short-Term Objective

II.1.a. Create a DPH Genomics Speaker's Bureau to reach a variety of audiences.

Long-Term Objectives

II.1.b. Assess community needs for genetic information/education services.

II.1.c. Develop and offer educational programs for school age youth that increase genetic awareness.

II.1.d. Develop and offer educational programs for the general public and disadvantaged groups that increases genetic awareness, including ethical and social implications.

Goal II.2

Develop, maintain, and assure availability of a public health and healthcare workforce that is competent in genetics.

Short-Term Objectives

II.2.a. Identify opportunities for including genetic information in the breadth of existing programs within DPH, having to do with chronic and infectious diseases, environmental and occupational health, family health and epidemiology.

II.2.b. Create and offer educational opportunities for workforce development among public health and healthcare workers, and build genomics literacy training into ongoing public health training.

Long-Term Objectives

II.2.c. Prepare students of public health and other healthcare areas for the role of genetics in professional practice.

II.2.d. Assure availability of a competent genetics workforce particularly genetic counselors and medical geneticists.

II.2.e. Partner with health departments in other states to develop educational materials that could be shared regionally.

II.2.f. Review licensing requirements of health professionals, both generalists and specialists, to consider incorporating genetic competencies.

Goal II-3

Develop and implement a regional strategic plan that addresses educational needs.

Short-Term Objective

II.3.a. Identify needed partners, and educational areas of common need.

Long-Term Objective

II.3.b. Develop regional response to areas of shared genetic education needs.

Priority III - Services

Assure equal access to, and appropriate use of genomic services across the lifespan.

Goal III.1

Assure high-quality, culturally competent genetic services, and help provide linkages for those needing services.

Long-Term Objectives

III.1.a Continue to assess the need for specific genomic services (public and private), identify ways to assess testing and other genetic services provided in state, and evaluate such services on an ongoing basis to identify and eliminate gaps.

III.1.b. Develop a strategic plan for ensuring high quality genetic services across the lifespan.

Goal III.2

Assure access to genetic services across the lifespan and across a broad range of conditions including infectious and chronic diseases.

Short-Term Objective

III.2.a. Assure access to genetic information that is culturally competent and effective in improving health.

Long-Term Objectives

III.2.b. Assure that all persons with genetic conditions have adequate public/private insurance to pay for needed services.

III.2.c. Assure seamless transition for children with genetic conditions to appropriate adult services.

Goal III.3

Ensure that an adequate capacity is in place to support the DPH newborn screening program and to address future needed capacity.

Short-Term Objectives

III.3.a. For the optimal provision of services, continue the implementation of the integration of all child health data.

III.3.b. Expand newborn screening resources to support comprehensive testing, tracking, and treatment options.

Long-Term Objective

III.3.c. Assure that all children with genetic conditions receive coordinated, ongoing, comprehensive care within a medical home.

Goal III.4

Expand DPH laboratory capacity to support comprehensive testing, tracking and treatment options for genetic conditions.

Priority IV - Information Systems Development & Integration

Develop a system of linked health databases that enables the monitoring of health status, and that could be enhanced with genetic information.

Goal IV.1

Develop a child health informatics profile (HIP-Kids) of child health databases within DPH.

Goal IV.2

Expand the HIP-Kids initiative to link with databases external to DPH and to incorporate health information across the lifespan.

Short-Term Objectives

IV.2.a. Develop a strategy to expand the HIP-Kids.

IV.2.b. Identify genetics information currently available within the HIP-Kids data system.

Goal IV.3

Seek ways to collect new genetic information from existing data sources for inclusion into HIP-Kids.

Short-Term Objective

IV.3.a. Incorporate genetics awareness questions into BRFSS.

Long-Term Objectives

IV.3.b. Analyze SLAITS data specific to Connecticut to assess needs of children with genetic conditions.

IV.3.c. Identify new ways to use existing infectious disease, chronic disease, and environmental health data systems to help quantify the genetic basis of disease and to identify populations at risk of developing a genetic-related condition.

Priority V - Improved Health Outcomes

Monitor health status to identify health problems linked to genomics.

Goal V.1

Use health data linked across divisions to identify genetic risk factors that can be incorporated into existing public health programs and that indicate needed development of new programs across the lifespan.

Short-Term Objectives

V.1.a. Assess the annual occurrence of newborn metabolic disorders and hemoglobinopathies, hearing disorders, and birth defects.

V.1.b. Analyze incidence, mortality, and morbidity data to support existing genetics-related programmatic activities aimed at early intervention, reduction of disease burden, and primary prevention of disease throughout the lifespan.

Long-Term Objectives

V.1.c. Monitor ongoing demographic trends such as: the aging population and its impact on chronic disease prevalence; growing racial and ethnic diversity; and the impact of delayed childbearing.

V.1.d. Encourage the use of genetic information in epidemiological analyses to associate genetics with disease and to support the development of novel genetics-related programs that reach across DPH divisions.

V.1.e. Analyze incidence, mortality, and morbidity data to identify environmental factors that may interact with genes to cause disease.

Goal V.2

Develop new strategies for linking genetics with adverse health outcomes within the state.

Long-Term Objectives

V.2.a. Assess the use of family history and other genetics programs in public health.

V.2.b. Develop a program that links adverse health outcomes with genetics that can be used to advise on the design of a needed response or intervention.

V.2.c. Assure the effectiveness of programs targeted at the prevention and reduction of disease burden of genetics-related diseases.

Goal V.3

Review and monitor the scientific merit and adverse health outcomes of genetic tests across the lifespan.

Long-Term Objectives

V.3.a Establish models for evaluating adult genetic tests.

V.3.b. Review promising genetics tests to support related legislative considerations.

V.3.c. Establish a model for ensuring informed consent for genetics tests.

Goal V.4

Ensure scientific accuracy of genetics materials**.**

Short-Term Objectives

V.4.a. Assess research findings for appropriate use in public health**.**

V.4.b. Ensure availability of updated genetics materials**.**

Long-term Objective

V.4.c. Become a resource for balanced information that tempers commercial marketing.

APPENDICES

Appendix A

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

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

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 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.

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.

STATE OF CONNECTICUT

DEPARTMENT OF PUBLIC HEALTH

J. Robert Galvin, M.D., M.P.H.
Commissioner

John G. Rowland
Governor

May 14, 2004

Dear Connecticut Resident:

I am writing to ask your help in completing a survey of individuals and their relatives who have health conditions or problems that may be inherited (passed down from one generation to another) or genetic (caused by a change in a person's chromosomes or genes). This survey is part of an effort to learn what are the current experiences and needs for those people living with these types of conditions. This information will be used as we develop a plan to help Connecticut families with inherited or genetic conditions get the care and services that would be useful to them.

You were chosen to complete the enclosed questionnaire because it is our understanding that either you or someone in your home has an inherited or genetic health condition or carries a gene that may cause a health condition either in themselves or their children. If this questionnaire does not apply to you or anyone else in your home, we are sorry to have inconvenienced you, but we would appreciate it if you would just return the uncompleted questionnaire in the enclosed envelope.

Your answers are completely anonymous. We do not ask you to include any information on the survey that will allow anyone to know who you are, and the envelope the questionnaire is returned in will be destroyed by our researchers. Completing the questionnaire and sending it back to us is completely voluntary. If you choose not to complete the questionnaire, there will be no penalty to you or your family. However, you can help us by taking about 30 minutes to complete the questionnaire then return it in the pre-addressed stamped envelope we have enclosed. **Please put the completed questionnaire in the mail by June 30, 2004.**

If you have any questions or comments about the survey or our efforts to provide useful services to you, please feel free to contact Beverly Burke, MSW, at the Department of Public Health. Her phone number is 860-509-7122 and her email is beverly.burke@po.state.ct.us.

Thank you very much for helping with this important activity.

Sincerely,

J. Robert Galvin MD MPH

J. Robert Galvin, MD, MPH
Commissioner

Your help is needed to help others

PHONE: (860) 509-7101 FAX: (860) 509-7111
410 CAPITOL AVENUE - MS#13COM, P.O. BOX 340308, HARTFORD, CONNECTICUT 06134-0308
Affirmative Action/Equal Employment Opportunity Employer

How the Department of Public Health Can Make Genetics Services More Useful to You?

Please take about 30 minutes to fill out this questionnaire. Your responses are anonymous – no one will know who you are. Then mail it back in the envelope by June 25, 2004. It is already addressed and stamped. If you have any questions or concerns, contact Beverly Burke, MSW at 860-509-7122, or by email at beverly.burke@po.state.ct.us.

Tell Us About Your Family's Health

These questions ask about health problems that may have been passed from parents or grandparents to their children. You might have the health problem yourself, or other family members may have the health problem. When answering the questions, think about just one person in your home with this health condition. The person could be one of your children, yourself, or another adult in your home. We will refer to this person as the "Family Member".

Some questions ask about "health care." This means care received at a health facility, doctor's office, hospital, or the office of another kind of health care provider. A health care provider may also be a nurse, or anyone who helps to treat the problems caused by the health condition. Another part of the questionnaire will have questions about other kinds of services, like case management or early intervention programs.

1. **Does anyone in your home have a health condition (affecting how their body works, how they learn or how they act) that may be passed down from one generation to another or is genetic in nature?**

   __ No, not as far as I know. **If you checked "No" you have completed the questionnaire. Please return it in the stamped envelope. Thank you for your help.**
   __ At least one person in my home has a genetic health condition or problem
   __ At least one person in my home carries a gene that can cause a health condition in themselves or their children, but does not have any health problems at this time.

   1b. Please list the name(s) of the health conditions or problems:
   _____

2. **How are you related to the Family Member (the person with the health condition) you listed above? (Check only one)**

   __ I have the health condition
   __ My husband, wife or partner has the health condition
   __ My child has the health condition
   How old is this child? _____
   __ My parent or my spouse/partner's parent has the health condition
   __ Another relative living in my home has the health condition
   How are you related to that individual? _____
   __ Other (please specify) _____

3. **In total, how many children younger than 18 years old in your home have this condition?** ____

4. **Has a doctor confirmed (knows for sure) that the Family Member has this condition?**

   __ No, a diagnosis cannot be made at this time
   __ Still seeing a doctor and having tests done to find out what is causing the problem
   __ Yes – please check which type of doctor below
   What type of doctor confirmed the final diagnosis? (Please check which doctor.)
   __ Regular doctor (such as a pediatrician, family doctor, gynecologist, adult doctor/internist)
   __ Genetics doctor, geneticist, or genetic counselor
   __ Specialist (such as cancer doctor, heart doctor, etc.)
   __ Other (please specify) _____

Please continue on the back of this page.

5. **When the Family Member with the health condition or problem was told about it, how much information (such as the cause, available testing, treatment, what to expect in years to come) were they or you given at the time? (Check only one)**

__ Nothing
__ Some information
__ A lot of information
__ I do not know or recall
__ Other (please specify) _____

6. **Where does the Family Member receive ongoing health care needed for this health condition? (Check all that apply)**

__ No ongoing care is needed for this condition
__ Local primary care doctor
__ Yale University Medical Center
__ University of Connecticut Medical Center
__ Connecticut Children's Medical Center
__Another hospital, which? _____
__ Other (please specify) _____

7. **Who is the main health care provider the Family Member receives care from? (Check only one)**

__ Doctor, What type? _____
__ Nurse Practitioner
__ Other (please specify) _____

8. **How happy are you with the care that the Family Member has received for this health condition from the main health care provider? Check the box for the answer that most closely reflects your feeling of satisfaction or dissatisfaction. If the question does not apply to you, check the "NA" box.**

Type of Care	Very unhappy	Unhappy	Happy	Very happy	NA
Time health provider has spent with the Family Member					
Amount of information given by health care provider	☐	☐	☐	☐	☐
Follow-up care given	☐	☐	☐	☐	☐
Overall sensitivity to your opinions and decisions	☐	☐	☐	☐	☐
Referrals given to other health care providers	☐	☐	☐	☐	☐
Referrals given to other non-health services	☐	☐	☐	☐	☐
Other aspects of care (specify)	☐	☐	☐	☐	☐

Please continue on the next page.

9. **Have you or the Family Member seen a genetics doctor or genetic counselor for this health condition? (A genetics doctor or genetic counselor is someone who helps to figure out if the health problem is something that can be passed down from one generation to another.)**

__ No, we were never told we should think about going to see these people
__ No, we were told about seeing these people, but we decided not to go
Why _____
__ Yes: Who told you about seeing the genetics doctor/genetic counselor? (Check only one)
__ a doctor
__ another type of health care provider, such as a nurse
__ a case manager
__ Someone in my home
__ Someone else (please specify) _____

10. **How happy are you about the care from the genetics doctor or genetic counselor that you or the Family Member has received? Check the box for the answer that most closely reflects your feeling of satisfaction or dissatisfaction. If the question does not apply to you, check the "NA" box.**

Type of Care	Very unhappy	Unhappy	Happy	Very happy	NA
Amount of time this provider has spent addressing family concerns or needs	☐	☐	☐	☐	☐
Amount of information given by health care provider	☐	☐	☐	☐	☐
Follow-up care given	☐	☐	☐	☐	☐
Overall sensitivity to your opinions and needs	☐	☐	☐	☐	☐
Referrals given to other health care providers	☐	☐	☐	☐	☐
Referrals given to other non-health services	☐	☐	☐	☐	☐
Other aspects of care (please specify)	☐	☐	☐	☐	☐

11. **If you could make changes to the health services that the Family Member as received, what would they be? For example, this might include how the Family Member is treated by the health care providers or the kinds of services that could be offered to the person.**

_____
_____

What Other Services, Besides Health Services Do You Use or Would Like to Have?

In this section we ask you to tell us what other kinds of services does the Family Member currently use and might want to receive because of the health condition. These services do NOT take place in a health center such as a hospital, private doctor's office, or other health care provider's office.

1. **What other services does the Family Member with the health condition use now? (Check all that apply)**

__ Early intervention (for example, Birth to Three)
__ Special education services for children older than 3 years
__ CSHCN/Children with Special Health Care Needs Programs
__ Case management coordination for services
__ Assistance with insurance coverage (Medicaid/Medicare/HUSKY)
__ Respite care
__ Support group (for example, parent meetings to discuss raising children with a medical condition)
__ Other (please specify) _____

Please continue on the back of this page.

2. **What additional services would you <u>like</u> to have access to? Please list any you would like to have, even if you don't know whether they are available.**

_____

_____

_____

How Does the Health Care Get Paid For?

These questions ask about how the health services used by the Family Member get paid for.

1. **How do the health services used by the Family Member get paid for? (Check all that apply)**

__ The Family Member does not receive any health care
__ Insurance is received through work/employer
__ Out of pocket or private insurance someone in the family pays for themselves
__ Medicare/Medicaid/HUSKY
__ Other state or federal aid
__ No insurance
__ Other (please specify) _____

2. **If the Family Member has health insurance coverage, has he/she had difficulty getting reimbursement for health services from the insurance company??**

__ I do not know
__ No
__ Yes Please describe the problem(s)_____
__ The Family Member does not have health insurance

Do You Have Concerns About Discrimination Because of the Condition?

In this section, we ask if the Family Member has experienced any type of discrimination due to the health condition. Discrimination is when you are not able to get something, such as insurance or a job, <u>because of the health condition</u> you or a member of your home may have.

1. **Has the Family Member experienced any discrimination because of the condition? (Check all that apply)**

__ No, not as far as I know
__ Yes, by an employer
__ Yes, by a company the Family Member tried to get a job with or school admission to
__ Yes, by a health insurance agency
__ Yes, by a life insurance agency
__ Yes, by those providing health services
__ Yes, other _____

2. **Have you, personally worried about discrimination in any setting because of the health condition?**

__ No __ Yes Why _____

Please continue on the next page.

How Do You, Personally Receive Information About the Family Member's Health Condition?

In this section, we ask how you get information about this health condition and genetics.

1. **Where do you get information about this health condition and genetics? (Check all that apply)**

__ I have not gotten any information about the health condition or genetics
__ Primary care doctor
__ Genetics doctor (geneticist) or genetic counselor
__ Other health care provider
__ Connecticut Department of Public Health
__ Support group
__ Family or friends
__ Local community leaders (such as clergy or religious counselor)
__ Internet websites Which sites? _____
__ Chat rooms/Internet discussions
__ Magazine/newspapers Which ones? _____
__ Television/radio
__ Other (please specify) _____

2. **Where would you prefer to learn about health care, genetics, and available services? (Check all that apply)**

__ I am not interested in obtaining information about these topics
__ Primary care doctor
__ Genetics doctor (geneticist) or Genetic counselor
__ Other health care provider
__ The Connecticut Department of Public Health
__ Support group
__ Family or friends
__ Local community leaders (such as clergy or religious counselor)
__ Magazine or newspapers
__ Television or radio
__ Lectures by medical experts
__ Telephone based lectures by medical experts
__ Internet websites
__ Internet based seminars or chats
__ Other (please specify) _____

3. **What additional information about the health condition would you like to receive?**

_____
_____

Tell Us About Yourself

This section has questions about your background. This will help us learn about the people who are filling out the questionnaire. Remember, the questionnaire is anonymous. We will not be able to identify you from your answers.

1. **In what county do you primarily live in Connecticut? (Check only one)**

__ Fairfield County
__ Hartford County
__ Litchfield County
__ Middlesex County
__ New Haven County
__ New London County
__ Tolland County
__ Windham County
__ Do not know what County
What town do you live in? _____
__ I don't live in Connecticut
Where do you live? _____

Please continue on the back of this page, it is the last page of the survey.

2. **What is the highest level of school you completed?**

__ Less than high school
__ High School
__ Some College/Associate Degree
__ Bachelor's Degree
__ Advanced Degree (please specify) _____
__ Other (please specify) _____

3. **What was the annual income for your entire household for the year 2003? (Optional)**

__ Less than $15,000
__ $15,000 to $34,999
__ $35,000 to $74,999
__ $75,000 to $99,999
__ $100,000 to $249,999
__ $250,000 or more

4. **What racial category do you consider yourself to be in? (Check only one)**

__ White, non-Hispanic
__ White, Hispanic
__ Black or African American
__ American Indian & Alaska Native
__ Asian
__ Native Hawaiian and Other Pacific Islander
__ Some other race alone (please specify) _____
__ Two or more races

What else would you like to say about the things we asked in this questionnaire? Please feel free to write down any thoughts you have about those topics or about this questionnaire.

_____
_____
_____
_____
_____
_____

You have now completed the questionnaire.

Thank you very much for taking the time to complete this questionnaire. Your answers will help us understand what people with genetics related health conditions in Connecticut are experiencing and what they need and want. We hope this will help us give better service to you, your family and your friends. Please return the questionnaire in the pre-stamped envelope by June 25, 2004.

STATE OF CONNECTICUT

DEPARTMENT OF PUBLIC HEALTH

J. Robert Galvin, M.D., M.P.H.
Commissioner

John G. Rowland
Governor

May 14, 2004

Dear Doctor:

I am writing to ask for your help in developing a statewide Public Health Genetics Plan. To help inform the Plan, we are conducting an assessment of the current practices, concerns, and needs of Connecticut-based physicians regarding genetic services and public health.

You are being asked to participate by completing the enclosed questionnaire because you are a practicing physician in the state of Connecticut who already addresses, or will soon be addressing issues of genetic testing and counseling with your patients. This questionnaire will provide us with the information we need to develop a state genetics plan that incorporates useful strategies to assist physicians with their patients who require genetic testing, counseling, or related services.

Your response to the enclosed questionnaire is completely anonymous. Upon receipt of your response, our researchers will immediately destroy the envelope in which the questionnaire is returned and there is no identifying information requested in the questionnaire. Participation in this questionnaire is voluntary. However, we would gratefully appreciate you taking about 15 minutes to complete the questionnaire, then **returning it in the enclosed pre-addressed stamped envelope by June 25, 2004.**

In appreciation of your time and participation in this assessment, we offer you a genetics educational tool, a newly developed CT Genetics Resource Directory. If you are interested in receiving this Directory, please return the pre-addressed stamped postcard with your name and address so that we can send the packet to you.

If you have any questions or concerns about the questionnaire or the development of the state Genetics Plan, please feel free to contact Beverly Burke, MSW, Connecticut Department of Public Health by phone at 860-509-7122, or via email at beverly.burke@po.state.ct.us.

Thank you again for helping us develop an effective Genetics Plan for the state of Connecticut.

Sincerely,

Robert Galvin MD MPH

J. Robert Galvin, MD, MPH
Commissioner

I need your expertise JR

PHONE: (860) 509-7101 FAX: (860) 509-7111
410 CAPITOL AVENUE - MS#13COM, P.O. BOX 340308, HARTFORD, CONNECTICUT 06134-0308

Integrating Genetics Into Your Practice and the Role of the Department of Public Health

Please take about 15 minutes to complete this anonymous questionnaire, then return it by **June 25, 2004** in the enclosed envelope or by Fax at 860-509-7160. If you have any questions or concerns contact Beverly Burke, MSW (860-509-7122, beverly.burke@po.state.ct.us.)

Medical Practice Section

1. **In what year did you receive your medical degree?** _____

2. **What is your primary area of medical specialty?** _____

3. **In which type of setting do you primarily practice? (Check only one)**

__ Tertiary medical center, Please specify _____
__ Community based hospital/clinic
__ Private practice
__ Health Maintenance Organization
__ Military hospital/clinic
__ Other (please specify) _____

4. **What age group is your patient population? (Check only one)**

__ I do not see patients as part of my work at this time
__ Predominantly pediatric (<18 years old)
__ Predominantly younger adult (Ages 18-50 years old)
__ Predominantly older adult (>50 years old)
__ Predominantly adults (18 years and older)
__ Both pediatrics and adult

(You have completed the questionnaire. Please return it in the stamped envelope. Thank you for helping us with this questionnaire.)

5. **On average, how many patients do you personally provide care for each week?** _____

6. **Which of the following languages do your patients primarily speak? (Check all that apply)**

__ English __ Hindi __ Russian
__ Chinese __ Japanese __ Spanish
__ French __ Korean __ Other (please specify) _____

Genetics in Medical Practice Section

1. **Of the patients that you personally provide care for, about what percentage are diagnosed with or you believe may have a medical condition that has an inherited or genetic bases?**

__ None __ < 1% __ 1-10% __ 11-25% __ 26-50% __ >50% __ Unknown

2. **About how many tests per year do you, personally order to rule out or confirm a genetic diagnosis? Please note that the test itself does not have to be DNA or chromosome based itself.**

_____ Genetic related tests ordered per year

3. **Of the tests referred to in the above question, which specific test(s) do you tend to order most often?**

Which tests? _____ For what diagnosis? _____

4. **A patient comes in with symptoms that may be due to a genetic disorder. How frequently are you likely to take the following actions? (Please check the option that best represents your response. NA= Not applicable)**

Actions	Never	Seldom	Sometimes	Usually	Always	NA
Take a detailed family history	☐	☐	☐	☐	☐	☐
Discuss possibility of a genetic diagnosis with the patient	☐	☐	☐	☐	☐	☐
Discuss genetic testing options, if available	☐	☐	☐	☐	☐	☐
Order the testing yourself	☐	☐	☐	☐	☐	☐
Discuss the test result interpretation with the patient without referring to a genetics center for follow-up	☐	☐	☐	☐	☐	☐
Refer to a genetics center for testing and/or counseling	☐	☐	☐	☐	☐	☐
Refer to a genetics center for test result interpretation	☐	☐	☐	☐	☐	☐
Discuss possible risks for other family members	☐	☐	☐	☐	☐	☐

Please continue on the back of this page.

5. **A patient comes in <u>with concerns about a family history</u> of an inherited disorder for which there is accepted genetic testing options. The patient does not have symptoms. How frequently are you likely to take the following actions? (Please check the option that best represents your response NA= Not applicable)**

Actions	Never	Seldom	Sometimes	Usually	Always	NA
Discuss detailed risk assessment for the patient	☐	☐	☐	☐	☐	☐
Discuss genetic testing options	☐	☐	☐	☐	☐	☐
Order the testing yourself	☐	☐	☐	☐	☐	☐
Discuss the test result interpretation with the patient without referring to a genetics center for follow-up	☐	☐	☐	☐	☐	☐
Refer to a genetics center for testing/counseling	☐	☐	☐	☐	☐	☐
Refer to a genetics center for test result interpretation	☐	☐	☐	☐	☐	☐
Discuss possible risks for other family members	☐	☐	☐	☐	☐	☐

6. **How <u>confident</u> are you with your ability to perform the following activities? (Please check the option that best represents your response.)**

Activities	Not at all confident	Not so confident	Somewhat confident	Extremely confident	Can't comment
Identify the genetic aspect of a patient's medical condition	☐	☐	☐	☐	☐
Take a three- generation family pedigree or equivalent tool	☐	☐	☐	☐	☐
Interpret the family history's contribution to a patient's genetic risk	☐	☐	☐	☐	☐
Order the appropriate genetic test	☐	☐	☐	☐	☐
Interpret genetic test results	☐	☐	☐	☐	☐

7. **About how many referrals to a geneticist/genetic counselor have you made in the last six months?** _____

8. **If you do not usually refer patients to a genetics center for consultation, how often is each of the following the reason? (Please check the option that best represents your response.)**

Reasons	Never	Seldom	Sometimes	Usually	Always	NA
Distance for the patient is too far	☐	☐	☐	☐	☐	☐
No counselor available at my facility or in my HMO	☐	☐	☐	☐	☐	☐
I don't see the benefit of the referral for the patient	☐	☐	☐	☐	☐	☐
I can provide the service myself	☐	☐	☐	☐	☐	☐
My patient's inability to pay for the genetics services	☐	☐	☐	☐	☐	☐
Other reason (please specify)	☐	☐	☐	☐	☐	☐

9. **What genetics services do you want for your patients that are not readily available? (Check all that apply)**

__ Nothing. We have what we need
__ Faster access to counseling appointments with genetic counselors
__ Faster access to geneticist evaluations
__ Broader availability of genetic services (please specify) _____
__ Better health care coverage for genetic services
__ Educational materials in multiple languages (which languages?) _____
__ Interpreters who are trained to discuss genetics
__ Consumer advocates who are trained to discuss genetics
__ Other (please specify) _____

Please continue on the next page.

10. **What gaps in the provision of genetic services are you concerned we may be facing in the coming years?**

_____

_____

Genetics and Public Health Section

1. **Below is a list of concerns regarding the integration of genetics into public health programs. Rate the significance of each of these concerns to you. (Please check the option that best represents your response.)**

Concerns	No Significance	Minor Significance	Moderately Significance	High Significance	Don't know No opinion
Need for population-based data	☐	☐	☐	☐	☐
Need for proven disease prevention measures	☐	☐	☐	☐	☐
Need for appropriate technology	☐	☐	☐	☐	☐
Current genetics knowledge level of health care professionals	☐	☐	☐	☐	☐
Genetics knowledge level of consumers	☐	☐	☐	☐	☐
Availability of genetics professionals	☐	☐	☐	☐	☐
Need for genetic testing cost/benefit data	☐	☐	☐	☐	☐
Need for policies and standards to guide genetic testing	☐	☐	☐	☐	☐
Need for more legislative protections for genetic information	☐	☐	☐	☐	☐
Improved integration of child health data (e.g. newborn screening and birth certificate data)	☐	☐	☐	☐	☐
Balance between patient rights and imposing limits on research	☐	☐	☐	☐	☐
Other (specify)	☐	☐	☐	☐	☐

2. **Based on your current knowledge and professional experience, what do you see as the top <u>three</u> priorities for public health genetics planning in Connecticut? (Check only three)**

__ Collection of population-based data about genetic conditions (e.g. prevalence rates for birth defects)
__ Statewide availability of genetic services (e.g., testing, evaluation, counseling)
__ Affordability/financial coverage of genetic services (e.g., insurance reimbursement)
__ Quality of services/resources
__ Coordination of genetics activities and service delivery across local and state agencies
__ Cultural sensitivity of genetic services/educational resources (e.g., multi-language patient literature)
__ Dissemination of scientific genetics information regarding testing, management and health promotion
__ Health care provider education
__ General public education
__ Genetic privacy and discrimination
__ Ethical use of genetic technology
__ Incorporation of new technology into public health practice (e.g. tandem mass spectrometry for newborn screening)
__ Other (please specify) _____

Genetic Information Education Section

1. **How have you received previous genetics training/education in the past? (Check all that apply)**

__ I have received no training/ education about genetics
__ Course work during medical school
__ Rotation in genetics during my internship/residency
__ Fellowship training
__ Post-training course in genetics
__ Session/lecture at medical conference(s)
__ Grand rounds presentation(s)
__ Journal articles
__ Consultation with genetics experts
__ Other (please specify) _____

Please continue on the back of this page. It is the last page of the survey.

STATE OF CONNECTICUT

DEPARTMENT OF PUBLIC HEALTH

J. Robert Galvin, M.D., M.P.H.
Commissioner

John G. Rowland
Governor

May 14, 2004

Dear Genetics Professional:

I am writing to ask for your help in developing a statewide Public Health Genetics Plan. To do so, we are conducting an assessment of the status of genetic services in the state, and of your input regarding the type of support needed by you and your patients. The information obtained through the enclosed questionnaire will be used by the Department of Public Health to develop the Genetics Plan.

Your response to the enclosed questionnaire is completely anonymous. Upon receipt, our researchers will immediately destroy the envelope in which the questionnaire is returned and there is no identifying information requested in the questionnaire. Participation in this needs assessment is voluntary. However, we would gratefully appreciate it if you would take approximately 10 minutes to complete the enclosed questionnaire and return it, by **June 25, 2004**, in the pre-addressed stamped envelope.

In appreciation of your time and participation, we offer you a new CT Genetics Resource Directory. If you are interested in receiving this directory, please return the pre-addressed stamped postcard with your name and address so that we can send it to you. The postcard will not be linked to your questionnaire.

If you have any questions or concerns about the questionnaire or the development of a state genetics plan, please feel free to contact Beverly Burke, MSW, Connecticut Department of Public Health by phone at 860-509-7122, or by email at beverly.burke@po.state.ct.us.

Thank you again for helping us develop an effective state genetics plan.

Sincerely,

J. Robert Galvin, MD, MPH
Commissioner

PHONE: (860) 509-7101 FAX: (860) 509-7111
410 CAPITOL AVENUE - MS#13COM, P.O. BOX 340308, HARTFORD, CONNECTICUT 06134-0308

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Appendix C

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Monday, August 16, 2004

Design Survey

Show All Pages and Questions

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Theme: Copy of Spring Day Add Edit Delete

Integrating Genetics and Public Health in Connecticut
A State Genetics Plan Questionnaire

Edit Title Edit Nu

Add Page

1. Current Role in Genetics Field Edit Page Delete Page Copy/Move Add Logic

Add Question Add Page

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What degree(s) have you obtained? (Check all that apply)

- MD/DO
- PhD/DSc/DrPH
- MS/MPH/MSc/MSW
- RN/PA
- Other (please specify)

Add Question Add Page

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In which health-related area(s) do you work? (Check all that apply)

- Prenatal
- General Pediatrics
- General Adult
- Cancer
- Public Health
- Clinical Laboratory
- Research Laboratory
- Teaching
- Other (please specify)

Add Question Add Page

Appendix C

Edit | Delete | Copy/Move | Add Logic

In which health-related area do you primarily work? (Check only one)

- Prenatal
- General Pediatrics
- General Adult
- Cancer
- Public Health
- Clinical Laboratory
- Research Laboratory
- Teaching
- Other (please specify)

Add Question | Add Page

Edit | Delete | Copy/Move | Add Logic

What is your primary role at work? (Check only one)

- Clinical geneticist
- Clinical genetic counselor
- Clinical Nurse
- Clinical laboratory geneticist
- Clinical laboratory genetic counselor/nurse
- Researcher
- Teacher/Professor
- Administrator
- Other (please specify)

Add Question | Add Page

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Do you see patients on a clinical basis?

- Yes
- No

Add Question | Add Page

2. Untitled Page

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Add Question | Add Page

Edit | Delete | Copy/Move

On average, how many patients do you see per week with a genetic disorder, at risk for a genetic disorder or who has a pregnancy at risk for having a genetic disorder?

Appendix C

Add Question | Add Page

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On average, how many new patients do you see per week?

Add Question | Add Page

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For each racial category please enter the approximate percentage of your patient population they represent.

	Estimated percentage of patient population
White, non-Hispanic	
Hispanic, non-White	
Black or African American	
Asian	
Native Hawaiian or other Pacific Islander	
American Indian or Alaska Native	
Other	

Add Question | Add Page

3. Current Practice Issues

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Add Question | Add Page

Edit | Delete | Copy/Move

Do you feel there are enough genetics centers and professionals to serve the current genetic needs of Connecticut's patients?

	Yes	No	Do not know
Genetic Centers	○	○	○
Geneticists	○	○	○
Genetic Counselors	○	○	○
Other Genetic Health Professionals	○	○	○

Add Question | Add Page

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If, in the previous question, you responded that there are not currently enough other genetics health professionals, please list which types of professionals you feel are needed.

1.

2.

Appendix C

3.
4.

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About what percentage of your patients' genetic services are covered by insurance? Check the box under the percentage that best represents your patients' current experiences for each type of activity.

	<1%	1-25%	26-50%	51-75%	76-99%	100%	I have no idea
Genetics consultation with the geneticist	○	○	○	○	○	○	○
Genetic counseling by a genetic counselor	○	○	○	○	○	○	○
Test procedures (e.g. ultrasound, amniocentesis)	○	○	○	○	○	○	○
Genetic related testing	○	○	○	○	○	○	○
Other	○	○	○	○	○	○	○

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If, in the previous question, you checked a percentage for the "Other" choice, please specify which genetic service(s) you were referring to.

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About how often each week do you spend working to obtain insurance coverage for genetics related services for your patients?

Never	Rarely	Sometimes	Often	Always
○	○	○	○	○

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Appendix C

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What non-medical services do you regularly refer your patients to?

	Never	Rarely	Sometimes	Often	Always
Long-term counseling	○	○	○	○	○
Social services (Medicare/Medicaid)	○	○	○	○	○
Early Intervention	○	○	○	○	○
Consumer support groups	○	○	○	○	○
Other	○	○	○	○	○

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What non-medical services do you regularly refer your patients to?

	Never	Rarely	Sometimes	Often	Always
Long-term counseling	○	○	○	○	○
Social services (Medicare/Medicaid)	○	○	○	○	○
Early Intervention	○	○	○	○	○
Consumer support groups	○	○	○	○	○
Other	○	○	○	○	○

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4. Untitled Page

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As a genetics service provider, what services or resources do you think are currently in greatest need in Connecticut? (Please check your top three needs)

- [ ] Higher lovel of insurance reimbursement for genetics services
- [ ] Stricter privacy and confidentiality policies/laws
- [ ] Weaker privacy and confidentiality policies/laws
- [ ] Legal protection for genetics service providers
- [ ] Non-genetics health care provider education regarding genetics and related services
- [ ] Public and consumer education regarding genetics and related services
- [ ] Additional clinical genetic services
- [ ] Additional clinical laboratory facilities in Connecticut
- [ ] Decreased patenting restrictions on testing
- [ ] Public health genetics programs

Appendix C

- More consumer support groups
- Other (please specify)

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5. Genetic Information Education

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Add Question | Add Page

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How do you prefer to obtain genetics-related information? (Please check no more than three choices.)

- Grand rounds
- Local meetings/conferences
- National meetings/conferences
- Audiotape based programs
- CD/DVD pased programs
- Internet websites
- Internet based programs (scheduled times)
- Internet based programs (on demand timing)
- Medical journals or books
- Consultation with genetics experts
- Popular media (TV, radio, magazines)
- I am not interested in receiving genetics related information
- Other (please specify)

Add Question | Add Page

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Please list any topics about which you would like to receive more genetics and/or public health-related information.

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How can the CT Department of Public Health facilitate your continuing education?

Appendix C

Add Question | Add Page

6. Future Issues for Genetic Services

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What gaps in the provison of genetic services is Connecticut facing in the coming five years?

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How might these gaps be addressed?

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What role do you think the State should play in addressing these gaps?

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Please tell us your other thoughts about the current and future provision of

Appendix C

genetic services in Connecticut.

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7. Thank you very much for completing this questionnaire. Edit Page | Delete

Your input is extremely valuable. In appreciation of the time and input you ha
we would like to send you the new Connecticut Genetics Resource Directory. (
DONE button and enter you mailing address on the screen that appears.

Add Question | Add Page

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Connecticut Department of Public Health
Statewide Public Health Genetics Plan
Non-Genetics Physician Needs Assessment Report
September 2004

Summary:

As part of the Connecticut Department of Public Health's development of a state-wide Genetics Plan, a needs assessment survey of Connecticut based physicians was a carried out in June of 2004. One hundred and fifty-seven responses (18.8% response rate) were returned and qualified for analysis. The majority of respondents saw primarily adult patients only (52.2%), while 28% of respondents were pediatric related physicians ad 19.7% saw both age groups.

In general, whether a respondent's patient population was pediatric, adult or both had an impact on the response to many questions related to integration of genetic services into their practices. Those physicians who see both adults and children were often significantly less likely to practice genetic services such as discussing testing options with their patients or ordering those tests. In addition, they were less likely to refer their patients to genetic centers for testing, counseling and test interpretation.

Several findings consistent with the Genetics Professional Survey were identified including the need for more professional education, better reimbursement for genetic services and increased access to genetic services. About half of the respondents felt that statewide availability of affordable genetic services should be the state's public health departments top genetics related priority.

The respondents are interested in advancing their genetic knowledge, particularly in the areas of at-risk patient identification (71.9%), advances in genetic technology (65.5%), resources for genetic testing, evaluation and counseling (59.7%), and genetic screening issues (56.1%). They want to continue using the methods they have employed in the past to learn this information, including: by reading medical journals, consulting with experts, grand rounds and local medical meetings.

Method:

A paper-based self-administered 4-page survey, consisting of questions regarding one's medical practice, concerns about the current state of genetics and public health, and genetic educational experience (Appendix A) was mailed via first class to 847 physicians licensed to perform medicine in the state of Connecticut. This list was randomly generated from the Physicians and Surgeons licensed in the state of Connecticut database. As an incentive, the physicians were offered a copy of the new Connecticut Genetics Resource Directory for completing the survey. To receive the directory, the physicians were instructed to complete the self-addressed postcard enclosed with the survey mailing and return it separately to maintain confidentiality.

Analysis was performed using SPSS statistical software package. Descriptive statistics were used to assess the respondents' medical practice, their use of genetics in their practice, views regarding the current state of Connecticut's genetic services and their interests in genetics education. Inferential statistics (Independent t-test, ANOVA) were employed to identify potential differences in response between older and younger physicians and physicians with primarily adult or primarily pediatric practices. Quantitative analysis was also employed to identify themes based on the open-ended survey questions.

Results:

Of the 847 surveys mailed, 9 were returned undelivered. One hundred and seventy-two were returned. Of those 172 returned surveys, fifteen respondents reported that they did not see patients and were, therefore, excluded from the analysis as determined by the survey protocol. The final analysis was performed on 157 returned surveys, a return rate of 18.8%.

Physician Practices

As shown in Table 1, the mean year of graduation from medical school was 1982 (sd = 10.7) with earliest reported year being 1945 and the most recent 2002. Over half of the respondents report practicing in some area of pediatrics. About 20% of the respondents are Ob/Gyn practitioners and 19% are family practitioners or in general medicine. Nearly 60% of the respondents work in a private practice while 17.8% and 16.6% work in a tertiary center or community hospital, respectively. Over half (5.2.2%) of the respondents see only pediatric patients, while 19.7% see both children and adults. Only 28% of the respondents see exclusively adult aged patients. The respondents see an average of 77.2 patients per week (sd=45.6).

Table 1. Physician Practices

Graduation Year (n=156)	Mean:	1982	
	sd:	10.7	
	Earliest:	1945	
	Latest:	2002	
Primary area of medical specialty (n=157)		n	%
Pediatrics		69	43.9
Obstetrics/gynecology		31	19.7
Family practitioner/General medicine		30	19.1
Pediatric specialties		11	7.0
Maternal fetal medicine/neonatal		5	3.2
Allergy		2	1.3
Emergency medicine		2	1.3
Infectious disease		2	1.3
Geriatrics		1	0.6
Gynecological oncology		1	0.6
Occupational/environmental medicine		1	0.6
Psychiatry		1	0.6
Reproductive medicine		1	0.6
Practice Setting (%) (n=157)			
Private practice:	59.9		
Tertiary center:	17.8		
Community hospital:	16.6		
Other setting:	4.5		
HMO:	1.3		
Patient Population (%) (n-157)			
Pediatric only:	52.2		
Adult only:	28.0		
Pediatric and Adult:	19.7		
Patient Visits/Week (n=154)			
Mean:	77.23		
sd:	45.6		
Mode:	100		
Min:	2		
Max:	300		

While 97.5% of the respondents report that they see primarily English speaking patients, 49% also see primarily Spanish speaking patients. Refer to Chart 1 for a complete listing of languages spoken by the respondents' patients.

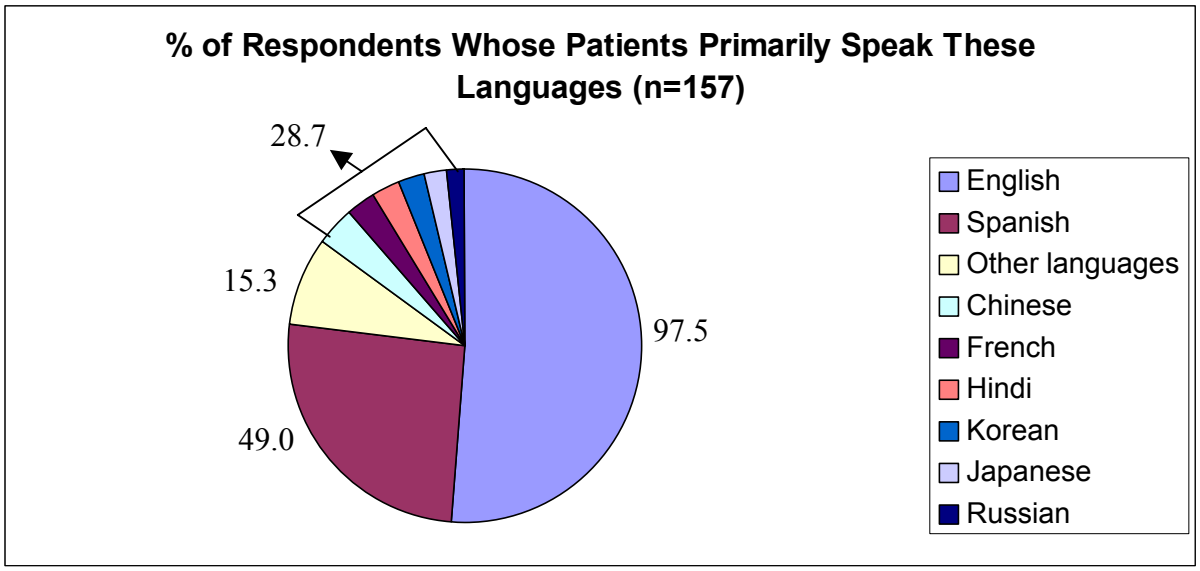

Chart 1. Percent of respondents whose patients' primary languages are shown. The other languages include Portuguese, Japanese, Russian, Polish, Albanian, Creole, German, Italian, and Vietnamese.

Genetics in Practice

Respondents were asked to estimate what percent of their patients have a disease with a genetic basis. Nearly 48% said between 1-10% of their patients do. Twenty-three percent thought that less than 1% of their patients' diseases are genetic in basis. Only 2.6% estimated a genetic basis of disease at greater than 50%.

The respondents also reported ordering an average of 44.4 (sd = 114.6, median = 6, range = 0 - 1000) genetic tests per year. Note that the tests did not have to be limited to DNA or chromosome based tests to be included. Of the 150 who answered this question, five respondents ordered between 400 – 1000 tests per year. If these five results were excluded from the analysis as outliers, the mean number of tests ordered would drop to 26.2 (sd = 44.8, median = 5, range = 0 – 210).

Assuming that the respondents see an average of 3850 patients/year (77/wk X 50 work weeks/yr), and an estimated 5% of the patients have a genetic bases as the cause of their disease (average of 0-10%) then the respondents each see about 193 patients with a genetic disorder per year. But they only order an average of 44 genetic tests.

Table 2. provides a list of the genetic tests most frequently ordered by the respondents. Chromosome analysis was by far the most frequently ordered test. Respondents did not specifically state whether the tests were done as part of prenatal testing or as part of a diagnostic workup of a patient.

Table 2. Tests ordered by respondents to diagnosis genetic disorders (n=127)

Tests	Frequency
Chromosome analysis	48
Hemoglobin electrophoresis	18
CF mutation	15
Fragile X DNA	10
Maternal serum screening	10
FISH	9
CF Sweat Test	8
Amniocentesis/CVS	7
BRCA1/2 DNA testing	7
Factor IV Leiden	5
Autoimmune antibodies	3
Hemochromotosis screening	3
Lipid profile	3
Amino acids/Organic acid panels	2
Celiac panel	2
ECHO	2
Metabolic screening	2
Tay Sachs test	2
Adrenal steroid test	1
Allergy test	1
CA 125	1
Cancer testing	1
CBC	1
Cholesterol panel	1
Connexin 26	1
CPK analysis	1
DMD gene test	1
Dystrophin	1
Gonadotrophin test	1
HLA analysis	1
Lysosomal enzyme panel	1
Periodic fever gene test	1
PKU repeat testing	1
Preimplantation genetic diagnosis	1
Protein S test	1
VonWillebrands test	1

Respondents were asked to assess the frequency with which they are likely to take various actions when a patient presents to them with symptoms due to a genetic disorder. Chart 2 illustrates the physicians' likely behaviors. While 89% will usually/always take a detailed family history and 77% and 71% will usually/always discuss the diagnosis and testing options, respectively, with their patients, only 40% will usually/always order testing themselves. Even less, only 15% will that routinely discuss the test interpretation with their patients. Sixty-two percent usually/always refer to a genetics specialist for testing and counseling, and 46% for interpretation. Approximately 59% of the respondents will usually/always discuss the family members' inheritance risks with their patients (Chart 2). On the other hand, when a patient comes to them with concerns about a family history of a genetic disorder, but has not symptoms themselves (Chart 3), the respondents are far less likely to discuss the risks with the patient and somewhat less likely to discuss testing options or risk to other family members than if the patient had symptoms. They are also much less willing to order testing or the non-symptomatic patient. However, they are similarly likely to refer for testing, counseling and test interpretation. .

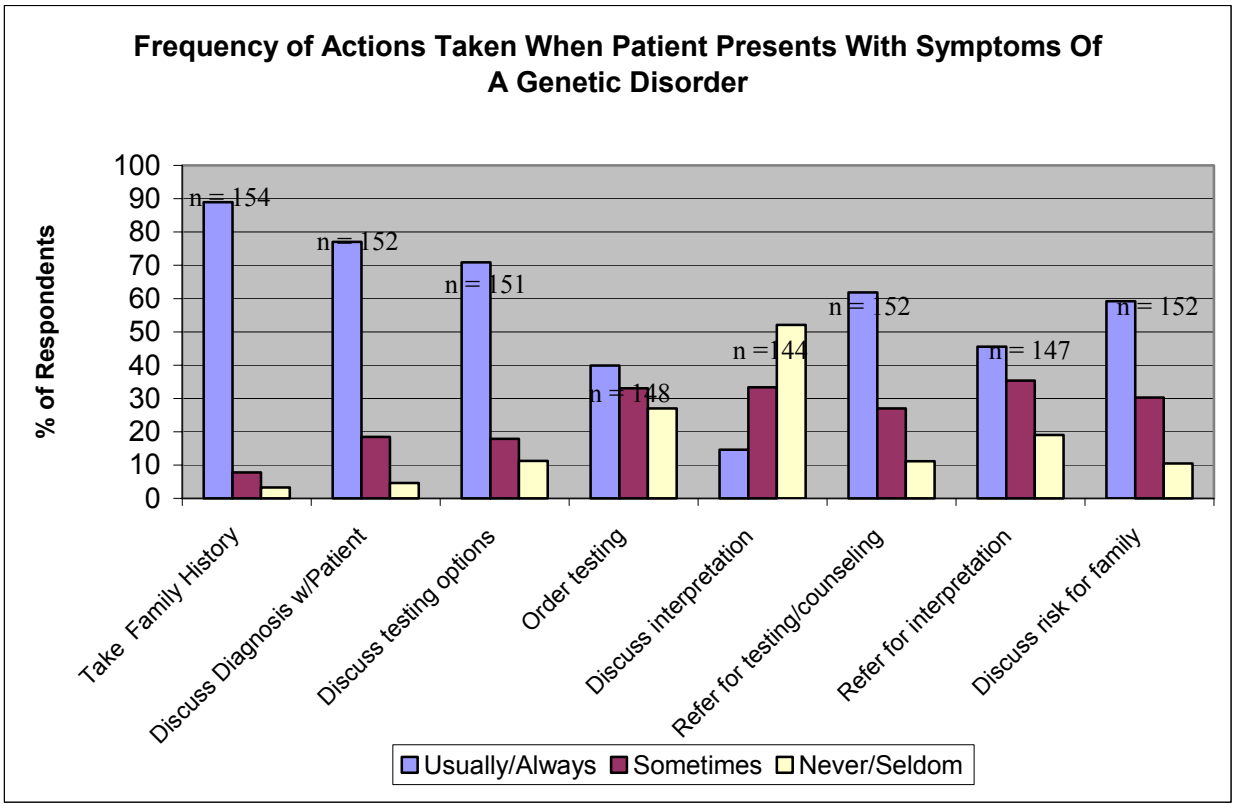

Chart 2. How frequently the respondents take these actions when a patient with symptoms that may be due to a genetic disorder presents to them.

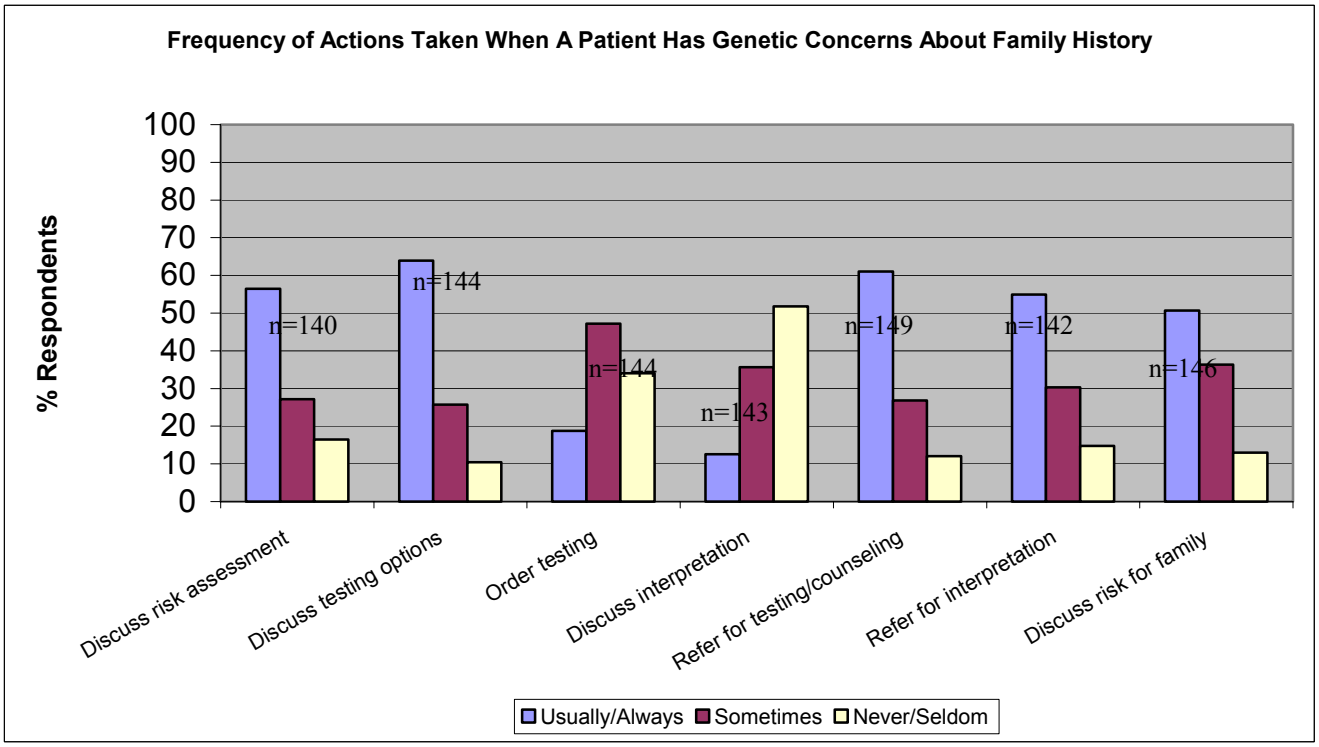

Chart 3. How frequently the respondents take these actions when a patient comes in with concerns about a family history of an inherited disorder for which there are accepted genetic testing options. The patient does not have symptoms.

Although respondents responses regarding what genetic service related activities they were likely to perform, between 76% and 53% reported being extremely confident/somewhat confident about their abilities to perform these activities (Chart 4).

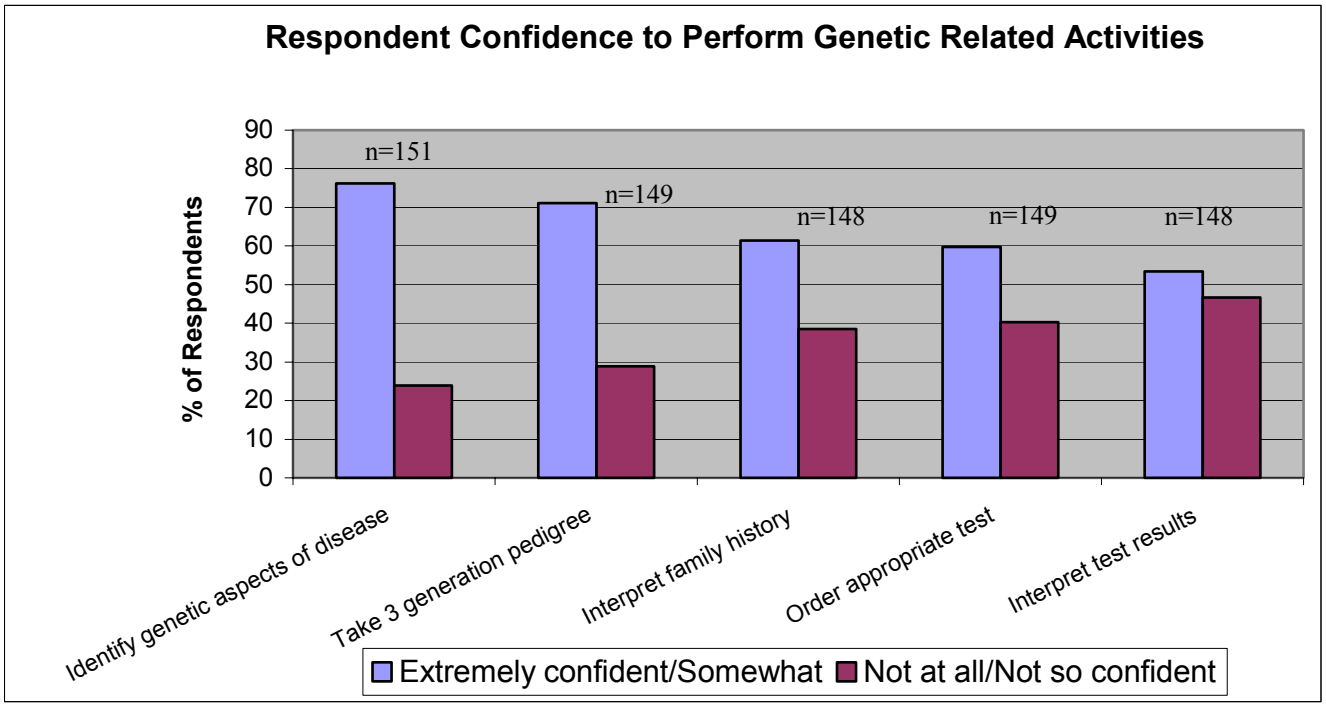

Chart 4. Respondents' levels of confidence regarding their ability to perform these activities.

The respondents reported making an average of 11.4 genetics referrals in the past six months (n=150, sd=45.4). However, a few physicians made between 100 and 500 referrals. If those three respondents were excluded from this analysis, the mean number of referrals would be 6.2 (sd=11.9, median of 2) or 12.4 referrals per year.

Expecting referrals to be fairly low, the respondents were asked how often the items in Chart 5 were their reasons for not referring more frequently. The reasons provided were seldom or never the cause for the respondents to not refer a patient to a genetics center. Although 47% said they felt they could either sometimes, usually or always provide the service themselves and 41% reported that their patients could not pay at least some of the times they might have referred them.

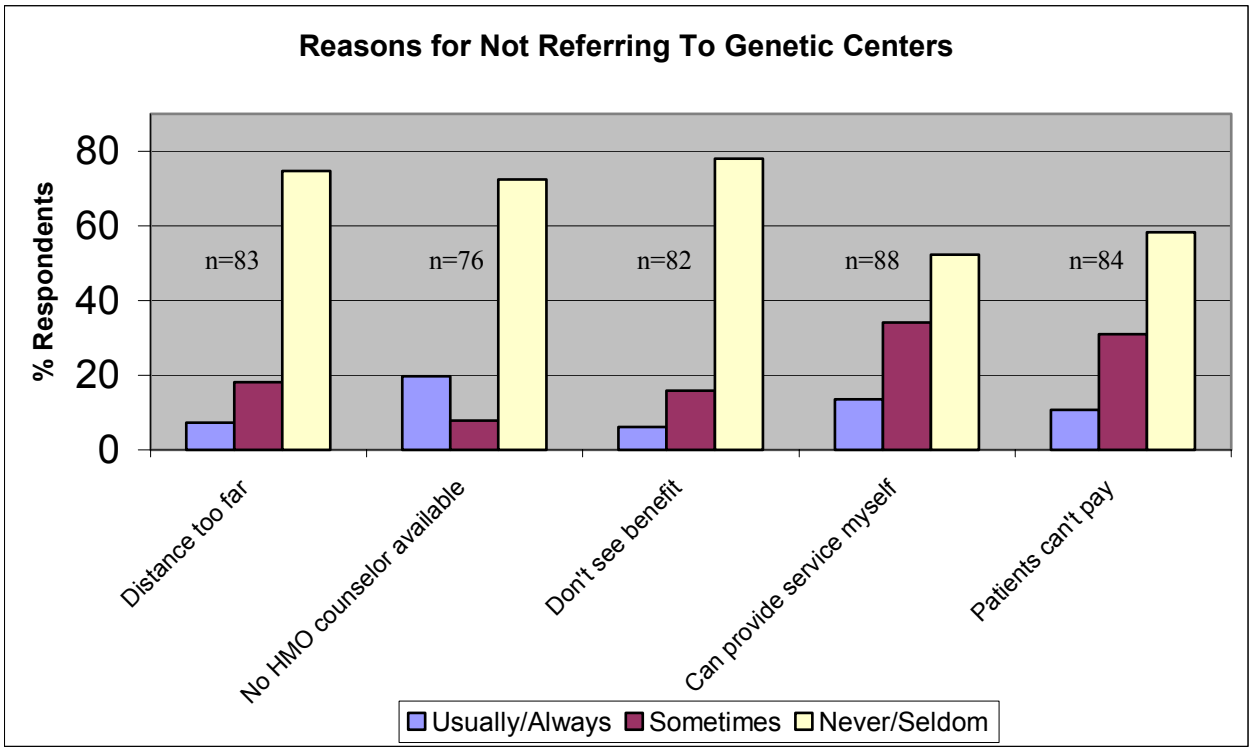

Chart 5. Why physicians do not usually refer to a genetics center.

When asked what services they would want for their patients that was not already readily available the respondents were interested in faster access to counseling (36.9%), better health care coverage (35.5%), and faster access to genetics evaluations (34.8%). Several specified needing genetics clinics throughout the state, including in rural Western Connecticut. However, 31.2% of the respondents felt that there were adequate genetic services for their patients (Chart 6). Individuals requested genetic counselors that spoke languages in addition to English, information be excluded from HMO forms because of insurance discrimination concerns, and information for patient resources.

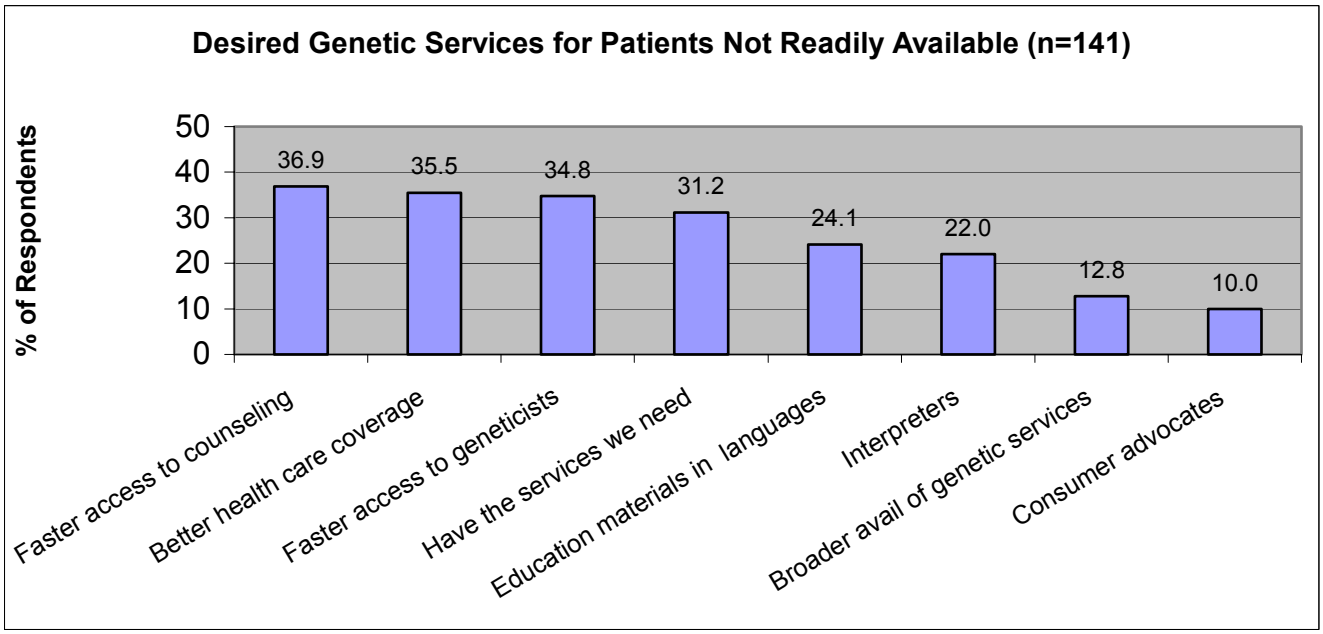

Chart 6. Genetic services respondents want for their patients that are not readily available

Several genetics related practice questions were analyzed comparing responses from physicians who see primarily children versus those who see primarily adults and those that see both children and adults. Several significant findings were observed (Table 3). In general, physicians that see both children and adults are less likely to perform the various genetic related activities, and think the level of health care providers' current genetics knowledge to be a concern. They also think the availability of genetics professionals is a greater concern than physicians seeing only children or only adults. Of the 31 respondents who said they see both children and adults, 23 said they were family practitioners and five said they practiced in an area that sees both adults and children (e.g. emergency, allergy, infectious disease). On the other hand, the other physicians practice in specialties specific to either children (pediatrics) or adults (Ob/Gyn). Therefore, it is likely that practicing medicine in an area that is less specialized or not likely to require immediate attention to genetic issues (emergency care) is the reason for their decreased involvement with genetic related activities. There is a significant number of more tests ordered annually by the pediatric group than the other two (p=.000), although this data point does not coincide with the lack of significance found in comparing the frequency or ordering between the pediatric and adult groups. Pediatric respondents ordered 94.5 tests per year as compared to 18.6% for adults and 10.8 for respondents that see combined age groups.

Table 3. Significant differences between physicians who see primarily children, primarily adults and those who see both age groups.

Category		**Likelihood of Activity** **P=primarily children** **A=primarily adults** **B=both children & adults**	**Significance** P values	**CI**	**N**
If patient has symptoms	Frequency of taking a detailed family history	P less likely than A	0.003	-.47,-.08	154
	Frequency of discussing testing options	B less likely than P B less likely than A	0.049 0.043	.00,.78 .01,.71	151
	Frequency of ordering genetic testing	B less likely than P B less likely than A	0.013 0.000	.09,.99 .26,1.07	148
	Frequency of referring for testing and counseling	B less likely than A	0.02	.15,.85	152
If patient has family history, but no symptoms	Frequency of referring for testing and counseling	B less likely than P B less likely than A	0.006 0.001	.12,.93 .19,.93	149
	Frequency of referring for test interpretation	B less likely than P B less likely than A	0.022 0.009	.05,.92 .10,.89	142
Confidence to do these activities	Identify genetic aspects of a patient's condition	B less confident than A	0.041	.01,.45	151
	Interpret family history contribution to a patient's genetic risk	P less confident than A P less confident than B	0.03 0.019	-.46,-.02 -.60,-.04	148
Concerns regarding integration of genetics into public health	Patients' inability to pay for genetic services	B less concerned than A	0.006	-.98,-.13	104
	Need for appropriate technology	B less concerned than P	0.036	.01,.30	131
	Current genetics knowledge of health care professionals	B less concerned than P B less concerned than A	0.028 0.038	.01,.21 .00,.19	133
	Availability of genetics professionals	B less concerned than P B less concerned than A	0.023 0.042	.02,.39 .00,.34	136

Interestingly, there were no significant differences in responses to frequency of implementation of genetic related services between respondents who graduated before 1982 or after.

Physicians identified several areas they consider future potential gaps in the provision of genetic services (Table 4). Notably, 25% of the responses referred to the inadequate number of genetic service providers in the future, 17.5% fear insurance inadequacies and 15% are concerned about the lack of genetics knowledge among health care providers.

Table 4. Future gaps in the provision of genetic services

Issues of concern for the future provision of genetic services	# of responses	% of responses
Inadequate number of genetic service providers	10	25
Inadequate insurance coverage	7	17.5
Genetics knowledge of providers	6	15
Insurance discrimination	4	10
Proper counseling/interpretation Need to counsel pts about testing and provide information/test interpretation – how to do that	3	7.5
Lack of availability of testing services	3	7.5
Provision for prenatal diagnosis and treatment options for patients	1	2.5
Financial survivability of genetic services	1	2.5
Lack of information about rare diseases identified in Human Genome Project	1	2.5
Ethical concerns that testing way out paces treatment, patients may not be insurable	1	2.5
Media hype about available info obtained from genetic testing	1	2.5
PH financial support of genetic programs	1	2.5
Inconsistencies in the newborn screening being offered	1	2.5
Total	40	100

Genetics and Public Health

Chart 7 indicates which issues, from a list proved on the survey, the respondents consider to be of moderate or high significance versus of no or minor significance when integrating genetics into public health. According to the respondents, all presented issues are of moderate or high significance (n=141). Health care professional knowledge of genetics was shown to be of most significance to the respondents (97%, n = 133), followed by the need for appropriate technologies (93.9, n=131), the need for proven disease prevention measures (93.3% n=135), and the need for policies and standards to guide genetic testing (91% n=133).

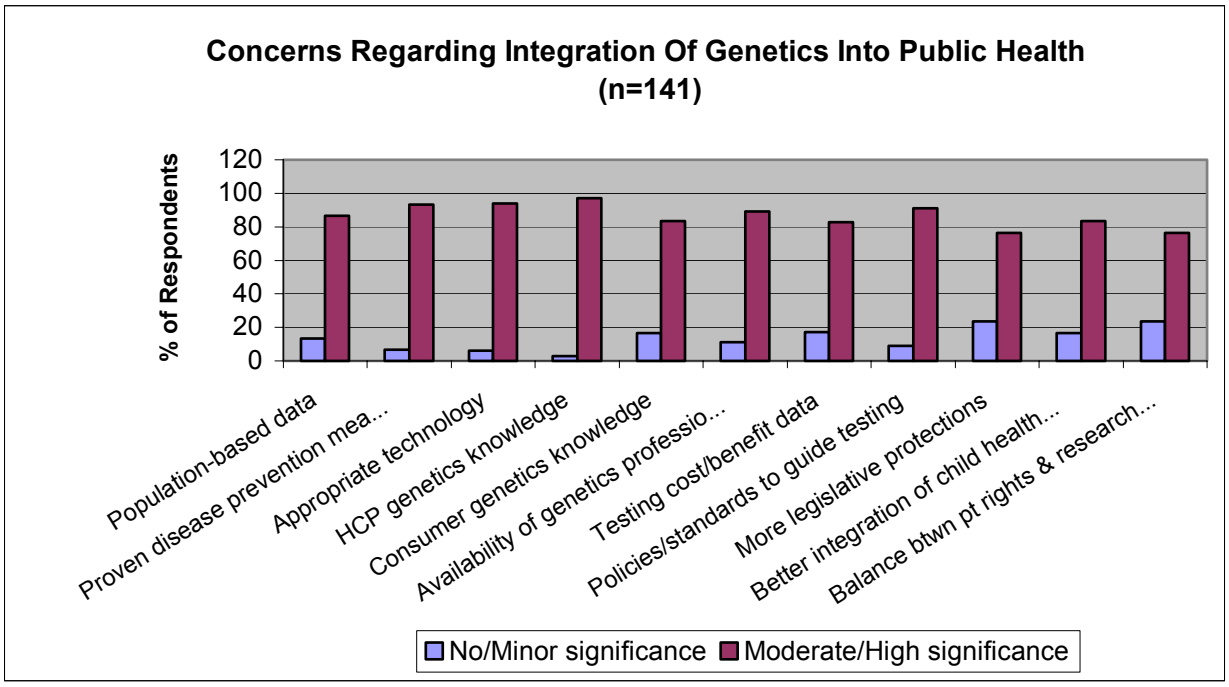

Chart 7. Respondents' level of concern regarding the integration of genetics in to public health activities.

Chart 8 shows the comparison of responses by those physicians who said each issue was of either moderate or high concern, allowing for distinctions between the two categories. The need for proven disease prevention measures seems to be of highest significance for respondents with 97 out of the entire 157 respondents to the survey identifying this as being highly significant.

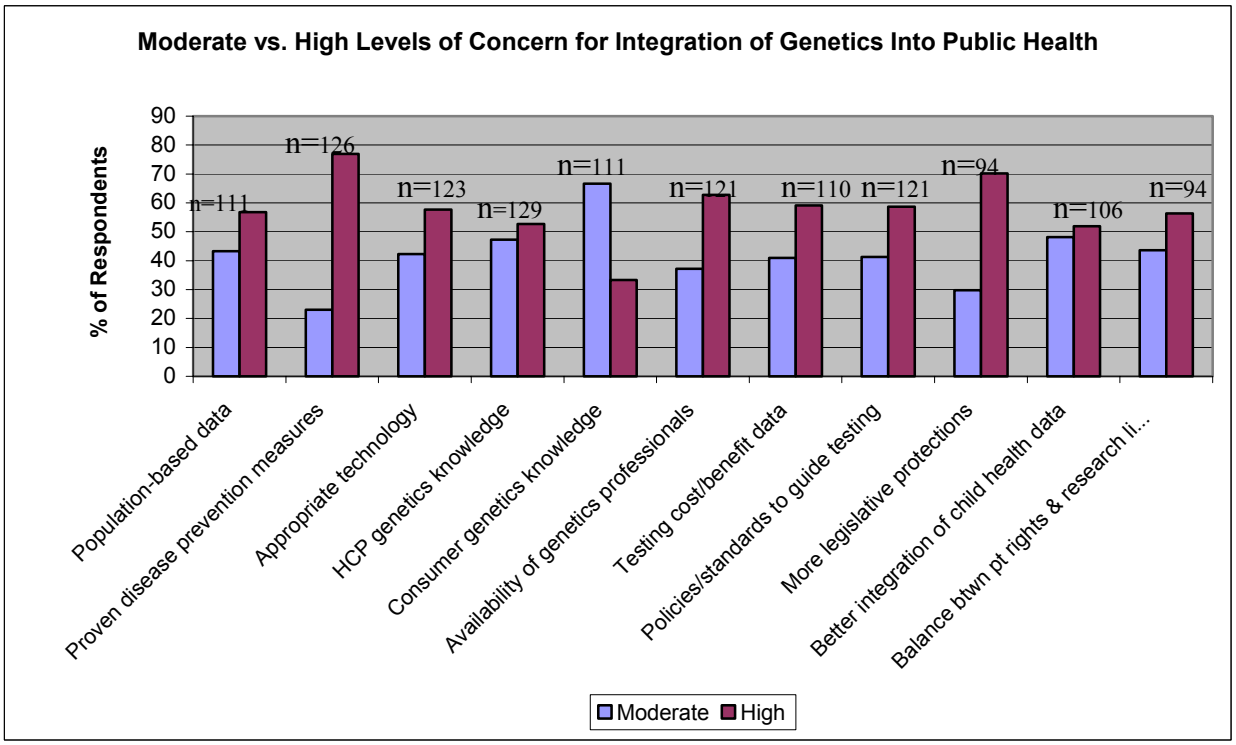

Chart 8. Comparison of moderate vs. high levels of concerns regarding the integration of genetics into public health. These data represent only those respondents who had a moderate or high level of concern for these issues.

However, when asked what their top three priorities for the state's public health genetics planning efforts were, more than half of the respondents report that statewide availability of genetic services was a priority, followed by affordability of genetic services (48%) and health care professional genetics education (34%). Note that many respondents gave more than three answers and all were recorded. Refer to Table 5 for a summary of results.

Table 5. Top priorities for Connecticut's public health genetics planning efforts (n=147)

	% of Responses	% of Respondents
Statewide availability of genetic services	17.4	53.1
Affordability/financial coverage of genetic services	15.8	48.3
Health care professional genetics education	11.1	34.0
Collection of population based data about genetic diseases	8.9	27.2
Incorporation of new technology into public health practices	8.5	25.9
Quality of services/resources	8.0	24.5
Dissemination of scientific genetics information regarding testing, management and health promotion	6.2	19.0
Ethical use of genetic technology	6.2	19.0
Genetic privacy and discrimination	5.6	17.0
Coordination of genetic activities and services delivery across local and state agencies	4.7	14.3
General public education	4.0	12.2
Cultural sensitivity of genetic services/educational resources	3.1	9.5
Other priorities for public health planning	0.4	1.4

Genetics Information Education

Respondents were asked about their the amount of genetics education they have received, which modes of education they have used and would prefer to use, and about which topics they would like to receive more training and information. Table 6 lists in ascending order the most common ways in which respondents gained their genetic educations. The vast majority received some training in medical school (84%). However, four individuals said they have never received genetics training. They graduated from medical school in 1945, 1961, 1976 and 1989. Approximately 63% obtain their genetics information from journal articles and over half via lectures, conferences grand rounds and consultations with experts.

Table 6. Methods used by respondents to learn genetics at any time in the past (n=150)

Methods of Education	% of Respondents
Course during med school	84.0
Journal articles	63.3
Session/lecture at med conferences	57.3
Grand rounds	56.7
Consultation with genetics experts	56.7
Rotation in genetics during residency	31.3
Fellowship training	10.0
Post-training course in genetics	8.0
I have no genetics training	2.7
Other method of learning	2.0

In the past 12 months, however, most of the respondents (65.8%) gained their information via medical journals and 54.6% via consultations with genetics experts (Table 7). Nearly half relied on grand rounds and only 28% on local meetings, 25% on national meetings.

Table 7. Methods used to obtain genetics information in the past year (n=152)

Methods of Learning	% of Respondents
Medical journals	65.8
Consultations with experts	54.6
Grand rounds	48.7
Local medical meetings	28.3
National medical meetings	25.0
Internet websites	20.4
Popular media	11.8
No information	8.6
Audiotape programs	8.6
CD/DVD programs	6.6
Internet programs	3.3

When asked for their preferred methods for obtaining genetics related information, more than half of the respondents selected the top four ways in which they currently get their information (Table 8). Notably, more are interested in using grand rounds and local meetings considerably more than they did in the past year.

Table 8. Preferred methods for obtaining genetics related information (n=153)

Preferred Methods of Learning	% of Respondents
Medical journals	60.1
Consultations with experts	58.8
Grand rounds	57.5
Local medical meetings	51.0
National medical meetings	30.7
Internet websites	30.1
CD/DVD programs	20.9
Internet programs - on demand timing	20.3
Audiotape programs	5.9
Not interested in more information	3.9
Internet programs - scheduled times	3.9
Popular media	3.3

Seventy-one of the respondents are interested in information about identifying at-risk patients (Table 9). In addition, advances in genetic technology (65.6), resources for genetic testing, evaluation and counseling (59.7%) and genetic screening (56.1%) are also topics of great interest. Respondents are interested in prenatal, carrier and newborn screening, less so in screening for adult-onset disorders.

Table 9. Preferred genetics related topics for additional training (n=139)

Topics	**% of Respondents**
Identifying at-risk patients	71.9
Advances in genetic technologies	65.5
Resources for genetic testing, evaluation and counseling	59.7
Genetic screening	56.1
Prenatal	66
Carrier	64
Newborn	62
Adult on-set disorders	50
Ethical, legal, social issues	33.1
Pedigree development and analysis	14.4
Specific disorders	10.1
Cultural sensitivity training	7.2

Table 10 shows a list of statements provided by respondents at the conclusion of the questionnaire. These comments are limited to those that had not yet been addressed by previous responses.

Table 10. Previously unstated comments

Final Comments
Cancer genetics is an issue too, not just prenatal genetics.
Most providers won't spend time on genetic activities because of low reimbursement.
Yale not consumer friendly, University of Connecticut is very responsive with quick turnaround.
Don't know what services are available in CT.
Need advocacy for rare genetic disorder testing.
Americans are living longer. Medicine is out of bounds of reasonable care. Don't need more medical advances or tax based state health initiatives
Training for providers on how to assess and influence patients' willingness to modify behavior to prevent modifiable pathologic consequence (e.g., DM, CVD, HTN)

Discussion

A needs assessment survey, mailed to 847 Connecticut based physicians yielded a response rate of 18.8% (n=157). The physician's addresses were randomly selected from the list of Physicians and Surgeons licensed in the state of Connecticut. The relatively low response rate is most likely due to the single mailing without follow-up, although an incentive to receive a Connecticut Genetics Resource Directory was provided. According to the literature, this is a low response rate, probably due to the lack of follow-up built in to the study.[2,3]

This group of physicians, most of whom graduated between 1971 and 1993, practiced in a private pediatric practice, seeing an average of 77 patients per week. In addition, nearly 50% of the physicians said they have patients that speak primarily languages other than English, particularly Spanish. This is the most

common, by far, non-English language spoken by patients, suggesting that any consumer education or informational materials should be written in Spanish as well as English.

As expected, Chromosome analysis was the most commonly ordered genetic related test, either ordered prenatally or to rule out chromosome defects in symptomatic patients. Screening for hemoglobinopathies, including sickle cell anemia is the second most frequently ordered genetic related test, although it is included in the state's newborn screening panel. Cystic fibrosis, which is the third most commonly ordered test, is not part of the legislated newborn screening panel but is probably ordered as a result of the 2001 American College of Obstetrics and Gynecology guidelines on CF screening (1). There are a few respondents who order hundreds of genetic tests annually (>400). However, the rest order an average of 26.2 tests/year, but make only 12 referrals over the same time-period. There is a significant difference in how many tests the pediatric physicians order annually as compared to those who see only adults or both age groups.

According to the respondents, they either usually or frequently employ many of the genetic services related activities when they suspect a patient is symptomatic with a genetic disorder, or if a patient has concerns about a family history suggestive of a genetic disorder (except to a lesser extent than the former scenario). Nearly 90% always or usually take a family history, which is considered the key tool to identifying potential inherited disorders. The exception to the respondents' implementation of genetic services is their willingness to discuss test interpretations with their patients. But it appears that these same physicians don't always, or even usually refer to a genetics center for test interpretation (referral is 45.6% if the patient is symptomatic, 55% if the patient has a family history only). This is particularly striking because just over 50% of the respondents say they are either extremely or somewhat confident with their ability to interpret genetic test results, and begs the question – How is the interpretation of genetic test results being translated to patients?

As expected, the average number of genetic testing referrals to a geneticists for evaluation or genetic counselor for counseling was (with the exception of three outliers who ordered hundreds of tests a year) minimal – 12.4 in the past year, while ordering 26.2 tests per year, presumably without first referring to a genetics expert. Most physicians rejected the reasons given as the cause for not referring for testing. Unfortunately, there was no indication as to why they didn't refer more frequently. It could simply be that testing wasn't considered clinically appropriate at the time or lack of knowledge about the appropriateness of the referral.

However, the respondents listed faster access to genetic counseling (36.9%), followed by better health care coverage (35.5%) as the most desirable genetic services for patients that weren't currently available. On the other hand, geneticists said public/consumer education was most critical (70.4%). That was followed by higher reimbursement rates for patients (57.7%)[(4)]. It is not unexpected that the two groups would identify a different top priority since their involvement with the patients differ. It is notable that both groups considered health care coverage to be the number two issue for improving patient access to genetic services.

Many physicians were concerned about the lack of services in the more rural areas of the state, even though they did not generally consider distance as a reason for not referring their patients now. Genetics education for health care providers was third top priority. Fifth on their list was the need for additional clinical services, but this group did list the need for more geneticists and genetic counselors as a major gap in the provision of services for the coming years. The physicians ranked consumer education 11th out of 12 listed priorities, followed by the need for culturally sensitive genetic services and educational materials.

Advances in genetics are coming at a rapid pace adding a huge educational burden to physicians' current workload. While 84% of the respondents did learn something about genetics in medical school, journal articles were the second most common learning tool and actually the most common way in which the physicians learned about genetic advances in the past year. This method is their preferred way to stay

abreast of new developments in the field followed by consultation with experts, grand rounds, and local meetings, all these selected by over 50% of respondents. National meetings, websites and other technological media ranked far lower. Clearly, the way to reach the physicians, if not all other health care providers is by publishing in their specialty journals, improving access to genetics experts and giving lectures at the state's local hospitals.

But what do they want to learn? About 72% of the respondents said they wanted to learn more about identifying at-risk patients. This is interesting considering that 76% of them said they were confident in their ability to identify these patients. They wanted to learn about advances in genetic technologies, genetic testing, evaluation and counseling resources and were specifically interested in genetic screening issues facing all the life cycle stages (prenatal, newborn, carrier, adult-onset).

One of the most significant characteristics separating the respondents was whether their practice included primarily pediatric cases, adult cases or a significant proportion of both groups. Does the respondent's patient population correlate in any way to their involvement with genetic services? The level of several of the genetic related activities did vary significantly based on the age of their patient population. In general, those respondents that saw both age groups were less likely to incorporate genetics into their practice, specifically discussing testing options, ordering genetic tests and referring their symptomatic patients to genetic services. The respondents seeing both age groups order an average of 10.8 genetic tests annually, while the pediatric group orders 94.5 tests and the adult only group orders 18.8 tests per year. There was also a significant difference in their behavior with regards to referring their patients with only a family history to testing and counseling or for test interpretation. There was little difference in their confidence levels – only in their ability to identify genetic aspects of a patient's condition as compared to the adult only physicians. Perhaps the reason for these differences is explained in the fact that these physicians are either family practitioners (23/31 in this group) or a specialty not yet faced with diagnostic testing for genetic disorders (allergy, emergency medicine, infectious disease).

Physicians who see both adults and children are also significantly less concerned about four of the eleven identified issues regarding the integration of genetics into public health, specifically patients' ability to pay for services, need for appropriate technology, the current genetic knowledge of health professionals and the availability of genetic professionals. Perhaps since they do not use these services at the same rate as the other groups of physicians it is expected that their patients have not experienced these problems to the same degree as the others' patients. In general, the respondents felt that all the identified issues did have moderate to high significance. However, a closer look reveals that 70% or more of the respondents said the need for proven disease prevention measures and more legislative protection of genetic information had a high level of significance to them. The medical profession continues to be wary of genetic discrimination from health insurers and employers even though very little evidence exists to support these concerns. But when asked to identify what issues should be the state's top priorities when developing its genetics plan, only 17% listed genetic privacy and discrimination. A likely reason for this discrepancy is that the respondents were asked to select the priorities based on their personal experience, and probably have not had patients face genetic discrimination after all.

Conclusion

In conclusion, the respondents to this survey:

1. Represented pediatric, adult and combined medical practices, with over half working in a private practice setting. The average year of graduation from medical school was 1982 (range from 1945-2002). However, there seemed to be no difference in responses regarding the implementation and use of genetic services between those graduating prior to 1982 or since.
2. As could be predicted, the most frequently employed genetic test is a chromosome analysis for either prenatal diagnosis or to rule out possible chromosome abnormalities in patients. Screening

for sickle cell anemia, and cystic fibrosis are also commonly ordered tests. However, only an average of about 26.2 tests were ordered by each respondent in the past year (excluding the three outliers) with approximately only 12% of referrals to a genetics center for any reason. Pediatricians order significantly more genetic tests than the other two groups (94.5 vs. 18.8 for adults and 10.8 for combined ages).

3. Respondents reported that they usually or frequently performed many of the genetic related activities such as taking a family pedigree (90%), discussing the possible diagnosis and testing options with the patient, and referring for testing and counseling. They are also willing to talk about risk to other family members. They are somewhat less likely to discuss the test interpretation with their patients, particularly if the patient is non-symptomatic, with only a family history. Instead, they will refer to a genetics center, but not at a rate high enough to compensate for the lack of test interpretation in their office.
4. Significant differences in practice were found between physicians based on their patient population's age range. Pediatricians and physicians who see only adults were more likely to employ these activities than respondents who see patients from both age groups. Perhaps this is due to the more general nature of this group's practices (e.g. family practice) that inhibits them from incorporating these practices.
5. At least 70% of respondents feel that a high level of significance should be attributed to both the need for proven disease prevention measures and more legislative protection of genetic information. However, they did not feel strongly about making genetic privacy and discrimination a priority for the public health genetics planning process.
6. The physicians are interested in increasing their genetics knowledge, particularly with regards to their ability to identify those at risk, advancements in new genetic technologies, available genetic resources and issues of genetic screening. They want to continue to learn the way they have been in previous years; by use of medical journals, consultation with experts, and local meetings and conferences.

Study limitations include not knowing how representative this sample group is of the Connecticut medical profession, and therefore how generalizable the results are to the rest of the state's physicians. The response rate is also low, at only 18.8%. Yet, there are key findings that coincide with anecdotal data, such as how the physicians want to learn about advances in genetics, and their concerns that there are insufficient genetic services available across the state and that reimbursement for these services needs to improve, especially if they will be expected to refer patients.

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Connecticut Department of Public Health
Statewide Public Health Genetics Plan
Genetics Professional Needs Assessment Report
August 11, 2004

Summary:

As part of the Connecticut Department of Public Health's development of a state-wide Genetics Plan, an online needs assessment survey of Connecticut based genetics professionals was a carried out in June of 2004. Eighty-two responses (45.3% response rate) were obtained and several themes were noted as major obstacles to the provision of genetic services in the state. The primary hurdles include lack of funding and insurance reimbursement for services, insufficient numbers of genetic professionals to support the state's clinical needs, and the need to increase both consumer and health professional educational efforts.

Method:

An Internet based survey (Appendix A), via Surveymonkey.com, was administered to 218 individuals considered Connecticut based genetic health professionals. Names were obtained from the CT DPH offices, and the National Society of Genetic Counselors, American College of Medical Genetics and American Society of Human Genetics membership directories. A cover letter (Appendix A) was emailed to the health professionals on June 3, 2004 with a link to the survey. Two subsequent reminder emails were sent to this group on June 15th and June 24th.

Analysis was performed using both the Surveymonkey.com results summary tool and SPSS statistical software package. Descriptive statistics were used to assess the respondents' demographics, genetics practice, and views regarding the current state of Connecticut's genetic services. Quantitative analysis was also employed to identify themes based on the open-ended survey questions.

Results:

Two hundred and eighteen emails were sent, of which thirty-one emails were undeliverable. One respondent reported that he was no longer practicing. Of the remaining 186 delivered emails, there were 82 responses for a response rate of 45.3%.

Demographics

Of those respondents, there were 80 responses to the question about what level of degree they had. Results are shown in Figure 1.

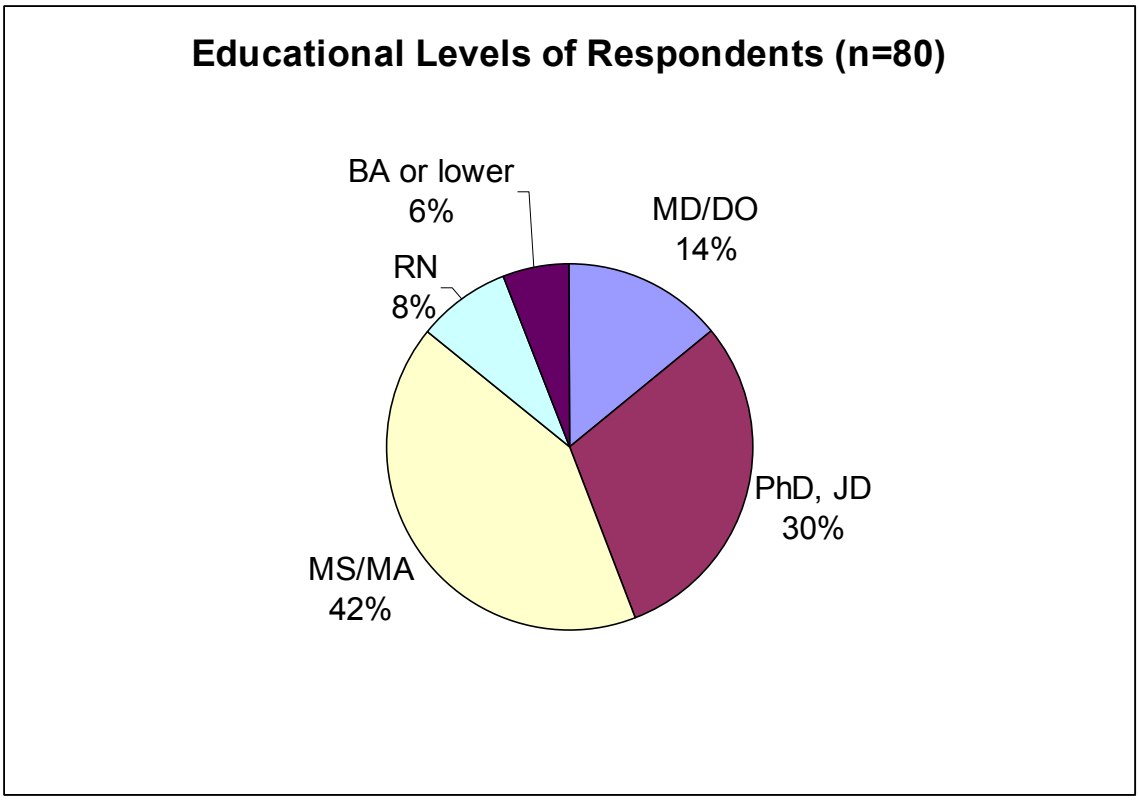

Figure 1: Degree level of respondents (n=80)

Respondent worked in multiple genetics related settings as outlined in Table 1. They were allowed to list as many settings as applied. Therefore, 80 individuals responded that they work in an average of 2.2 different settings.

Table 1. Genetics-related areas in which respondents work (n = 80)

Health Related Work Areas	**Frequency**	**Percentage of Responses**	**Percentage of Respondents**
Teaching	32	17.9	40.0
Prenatal Diagnosis/Maternal Fetal Medicine	26	14.5	32.5
Public Health	25	14.0	31.3
Research Laboratory	23	12.8	28.8
Clinical Laboratory	20	11.2	25.0
Cancer Genetics	15	8.4	18.8
General Pediatrics	12	6.7	15.0
General Adult	11	6.1	13.8
Specialty Clinic	11	6.1	13.8
Other Work Area	4	2.2	5.0
Total	**179**	**100**	**224**

While 40% of the respondents have some teaching responsibilities, prenatal diagnosis/maternal fetal medicine, public health, and the clinical laboratory are the three areas in which over half of the respondents are primarily involved (Figure 2).

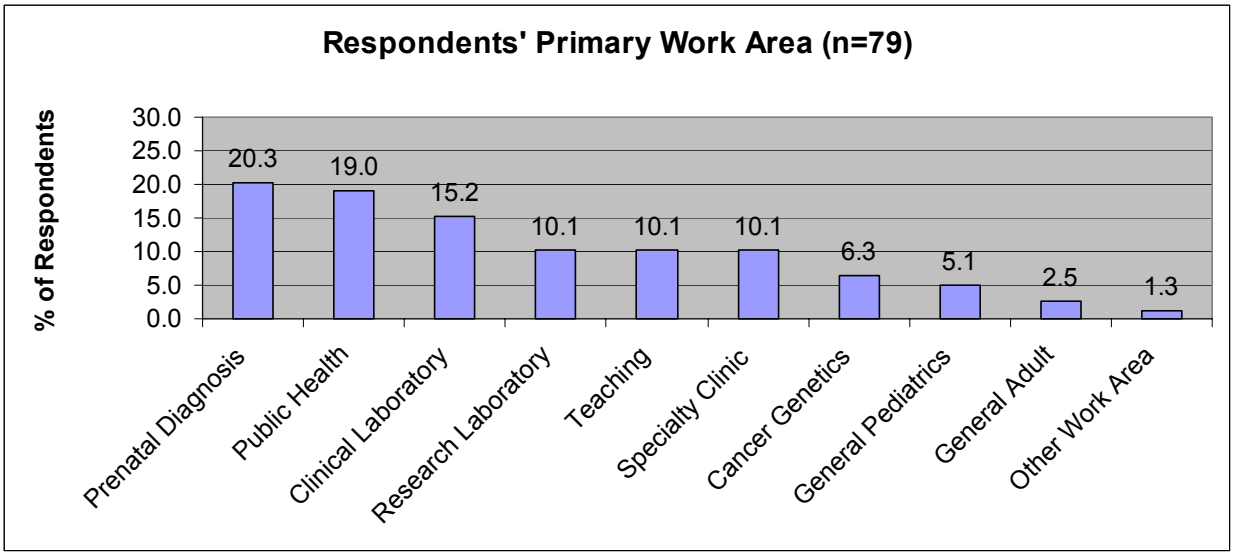

Figure 2. Respondents' primary work areas (n=79)

Of the 82 respondents, 80 categorized their primary role as shown in Figure 3. Eleven of those respondents listed themselves in the "Other" category, which included public health roles (epidemiologists, nurse consultants, service coordinator, public educator, lab inspector), and as non-geneticist physicians.

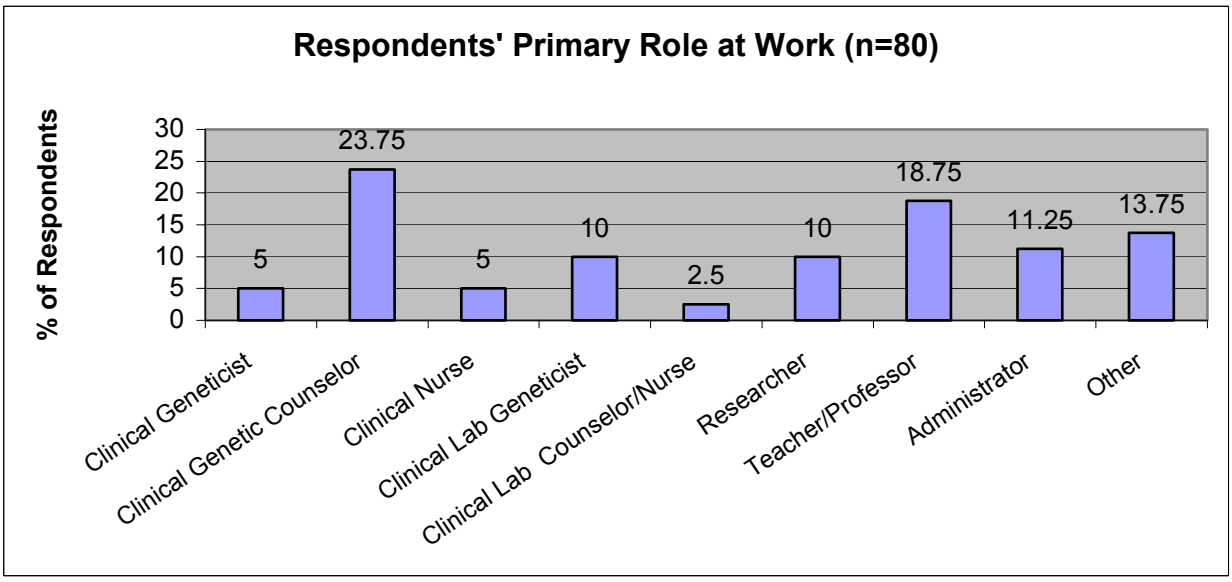

Figure 3. Respondents' primary roles at work (n=80)

The Connecticut Genetics Patient Population

Just over half of the respondents (53.8%, n =43) do not see patients on a clinical basis, which should note when interpreting the data. However, the respondents that do see clinical patients (46.2%, n=37) see on average approximately 9 patients per week (ranging from occasionally to as many as 40 each week). Nearly all of these patients are new to the clinicians, with virtually no repeat visits.

A racial breakdown of these patients is shown in Table 2. It shows that the vast majority of clinicians' practices are composed of White, Non-Hispanic patients. Fifty-six percent of the 34 clinicians that responded to this question said that this racial group made up between 70-100% of their patient

population. All the clinicians report seeing at least some Black/African-Americans and Hispanics, with the majority reporting that these groups make up between 1-10% of their patient population.

Table 2. Racial breakdown of patients seen by clinicians by percentage (n=34)

	Percentage of Practice That Each Racial Group Represents											
Racial Groups	**0**	**1-10**	**11-20**	**21-30**	**31-40**	**41-50**	**51-60**	**61-70**	**71-80**	**81-90**	**91-100**	**Response Total**
White, Non-Hispanic			3%	6%	6%	6%	12%	12%	35%	12%	9%	**34**
Black or African American		61%	24%	12%	3%							**33**
Hispanic, non-White		55%	35%	3%	3%	3%						**31**
Asian	3%	87%	10%									**30**
Other Pacific Islander	76%	24%										**17**
American Indian or Alaska Native	70.5%	29.5%										**17**
Other	38%	62%										**8**

The majority of the respondents (n=35) did not know what percentage of their patients' use of genetic services was covered by insurance. As shown in Table 3, test procedures such as ultrasounds and aminocentesis were generally covered, with 19 of the 20 respondents who did know this information, reporting that at least 51% of their patients' procedures were covered. Genetic testing was covered at a more inconsistent rate, ranging from 1% to 99%. This information was obtained from only 24 responses. As with genetic testing, clinical services performed by geneticists and genetic counselors were inconsistently reimbursed, also ranging from <1% (in 3 cases for genetic counselors) to 100% (in one case for both geneticists and genetic counselors).

Table 3. Reimbursement rate estimate for genetic services

	<1%	**1-25%**	**26-50%**	**51-75%**	**76-99%**	**100%**	**Don't Know**	**n=**
Genetics consultation with the geneticist	0%	3%	3%	9%	24%	3%	59%	34
Genetic counseling by a genetic counselor	9%	6%	15%	9%	18%	3%	41%	34
Test procedures (e.g. ultrasound, amniocentesis)	0%	3%	0%	9%	39%	9%	39%	33
Genetic related testing	0%	15%	21%	9%	27%	0%	27%	33

Figure 4 shows the amount of time the respondents spend working to obtain insurance reimbursement for their services. About 34% say they often or always help their patients obtain reimbursement. However, there was no determination of how many hours per week to which that equates.

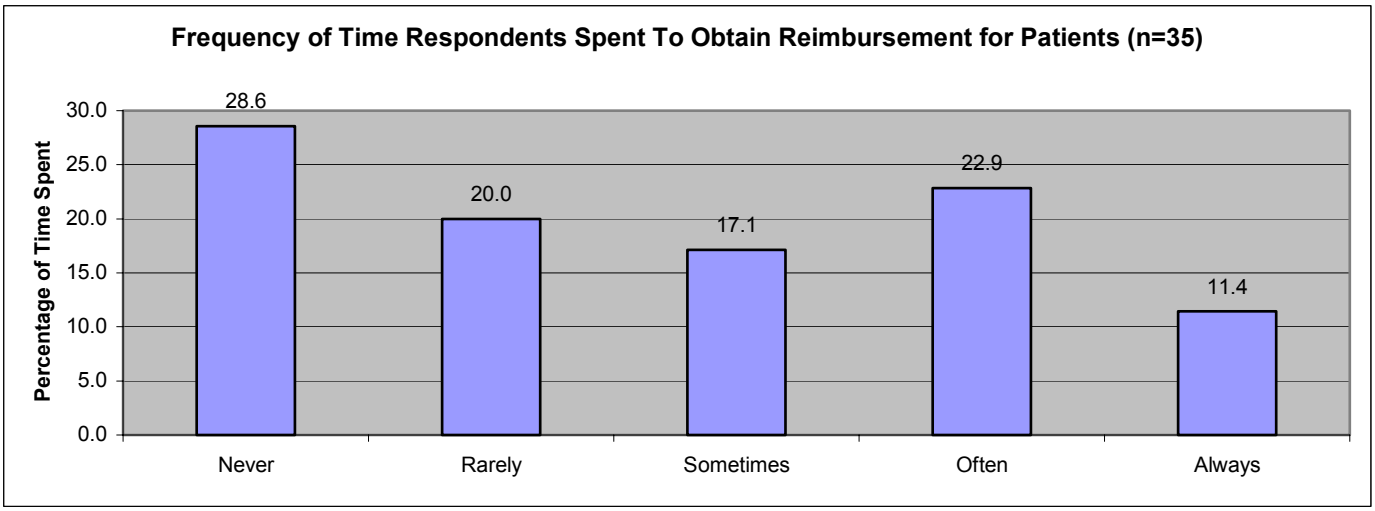

Figure 4. Frequency of time spent by genetic professionals working to obtain insurance reimbursement for their patients. (n=35)

The Current State of Genetic Services

Respondents were asked about the State's status of genetics services, particularly whether there were sufficient centers and professionals to support the residents' genetic needs. Of the 37 professionals that see patients clinically, 36 responded to this question. While the majority felt there were enough genetic centers, over 72% felt there were neither enough geneticists nor genetic counselors to support the demand for services. Over half were unable to say whether there were enough other types of genetic health professionals to meet the need (refer to Figure 5). Other types of genetic health professionals that respondents report needing were nurses and nurse practitioners with genetics training (4 responses), lab technicians (1 response), nutritional counselor (1 response), epidemiologist (1 response), genetic educator (1 response), and support staff (1 response).

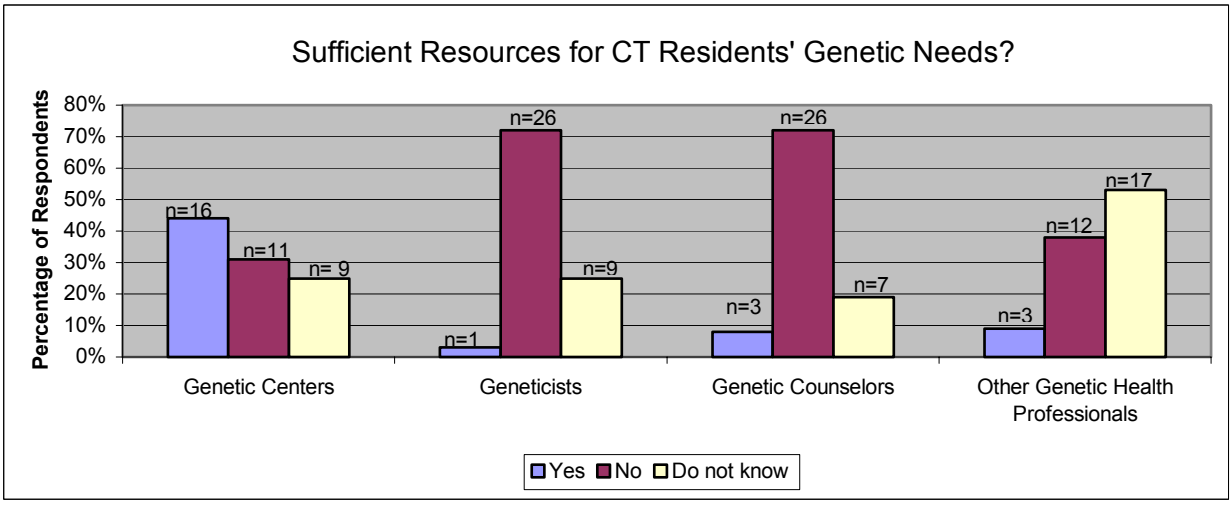

Figure 5. Are there enough centers, and genetic health professionals to support the genetic needs of Connecticut's residents?

One of the activities Genetics professionals, particularly genetic counselors carry out is the referring of patients to non-medical services such as long-term counseling or consumer support groups. Figure 6 shows the frequency with which the respondents (n=35) refer to a variety of non-medical services.

Consumer support groups receive the highest referrals (approximately 55% of the respondents say they often make this referral. Five respondents said they either sometimes or often refer to other non-medical services, but did not specify which services.

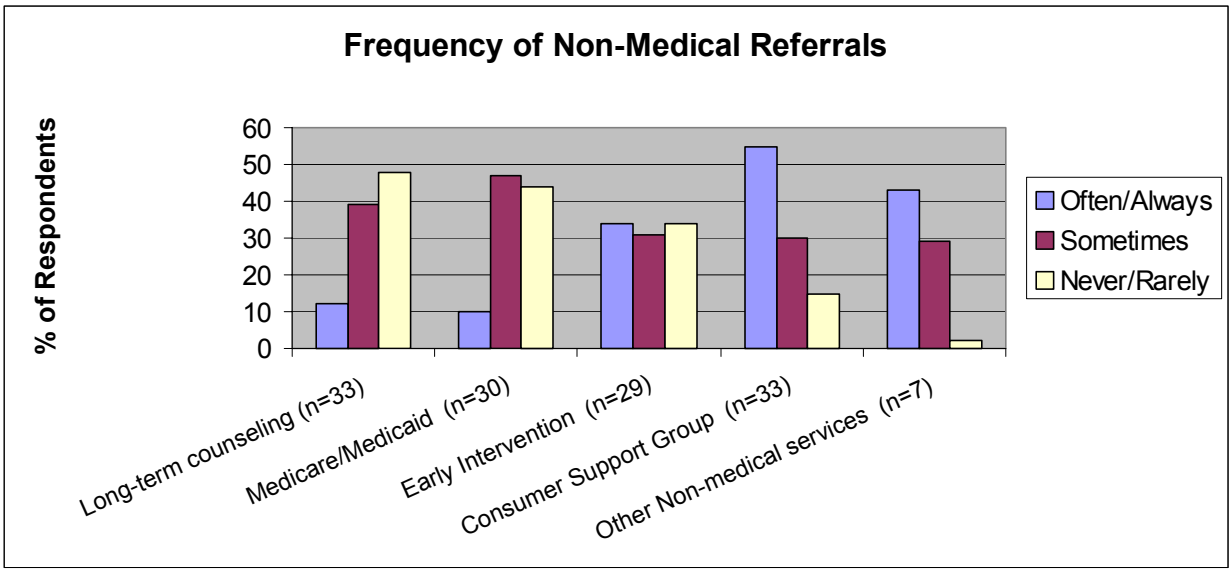

Figure 6. How frequently do genetic professionals refer to non-medical services

Respondents were asked to identify up to three resources they felt were currently in greatest need in Connecticut in order to improve the delivery of genetic services. According to Figure 7, over 70% the genetic health professionals responding to this question feel that public/consumer education regarding genetic services is most critical, followed by a higher level of insurance reimbursement (57.7%) and education of non-genetic health care providers regarding genetics services (53.5%) and public health genetics programs (42.3%). One respondent suggested the formation of a state genetics advisory committee to function as the coordinating group for all genetics testing and implementation of the state genetic plan is a significant need. One respondent suggested that legislators are a specific group that requires genetics education.

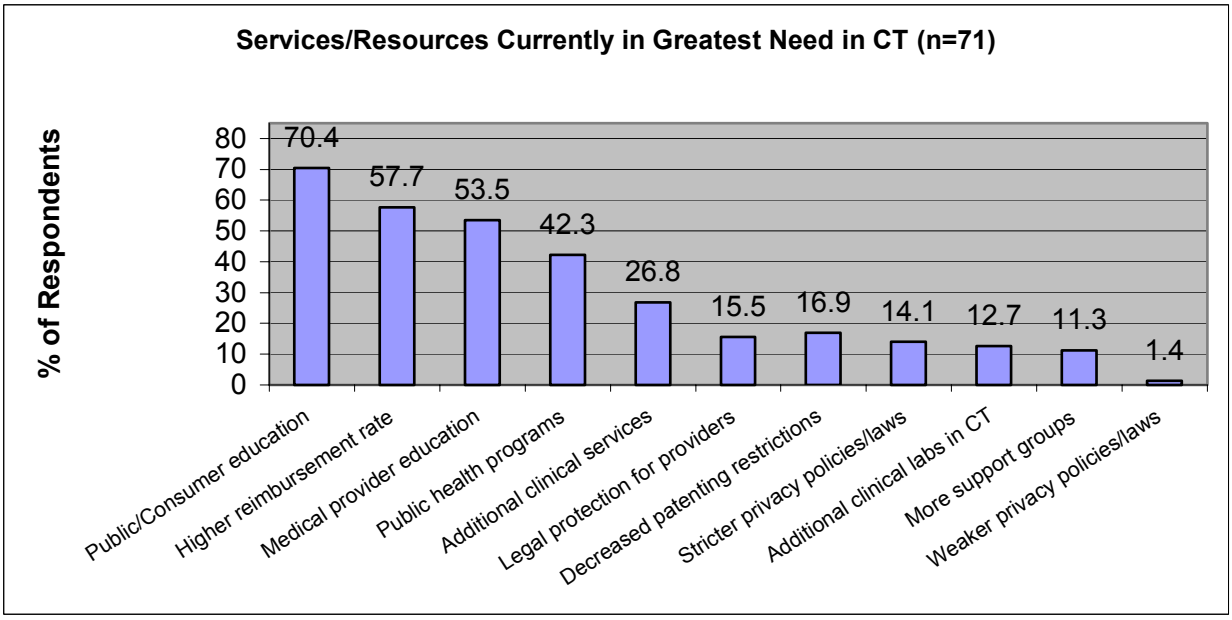

Figure 7. Services/Resources the state needs most to improve the delivery of genetic services (n=71)

Genetics Professional Continuing Education Preferences

When asked how they prefer to obtain their genetics related information, the genetics health professionals chose medical journals (68% of respondents), and national and local meetings or conferences (64%, 53% respectively). Selected Internet websites were also a preferred method for obtaining information (49%). However, other high tech or self-paced methods, such as CD/DVD based programs, Internet based programming (e.g., lectures, chats) or audio taped programs were of little interest. Refer to Figure 8 for detailed information.

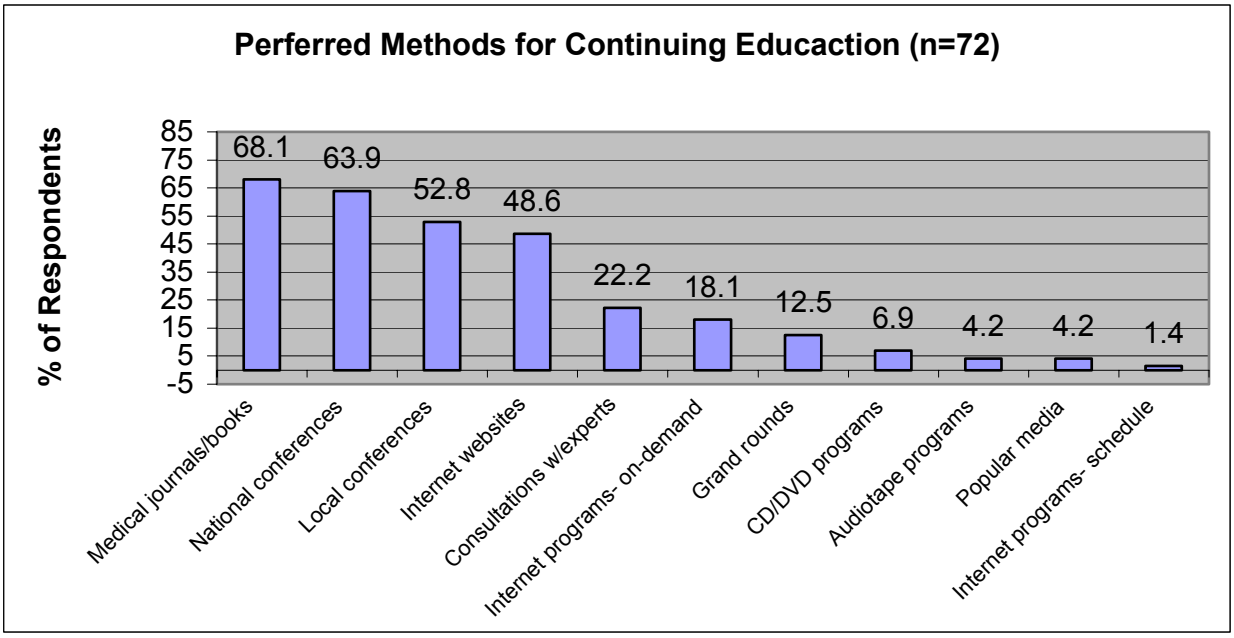

Figure 8. Preferences for obtaining medical genetics continuing education (n=72)

The respondents requested many topics, with the majority focusing on insurance related issues. Genetic screening programs, particularly newborn screening, and availability of genetic tests were also listed as topic of interest. The list of requested topics are shown in Table 4. Respondents were able to select multiple options.

Table 4. Requests for genetics related information (n=30)

Topics	Frequency of Request (n=30)
Insurance issues	6
Available genetic tests, testing and counseling services	5
Screening programs (particularly newborn screening)	5
Mental Illness/psychiatry and genetics	3
Patient confidentiality issues	3
Chronic disease and gene determinants	3
Public health role in genetic testing	2
Available patient resources and education approaches	2
Cancer genetics	2
Genetics of hearing loss	2
Population analysis of genetic susceptibilities	1
Taking family histories of adopted children	1
Stem cell and gene replacement therapies	1

Inborn errors of metabolism	1
Quality control and assessment of genetic testing	1
Counseling skills	1
Anti-aging technologies	1
Gene impact on non-genetic (environmental) diseases	1

Forty-one genetics professionals weighed in on how the CT Department of Public Health can facilitate their continuing education, many of them interested in attending local conferences with local or national experts. Websites with links to resources and new information was also cited as a desirable approach to maintaining their education. The need for CEU opportunities for nurses and genetic counselors was requested often. Table 5 lists the recommended ways in which the CT DPH can help facilitate the genetics professionals' continuing education.

Table 5. Respondents' suggestions for ways the CT DPH can help facilitate continuing education (n=41)

Approach to Maintaining Education	**Frequency of Request (n=41)**
Local workshops, conferences, lectures around the state (preferably one-day events)	17
CEU opportunities for nurses and genetic counselors	10
Use Internet to disseminate educational materials and resource information (e.g., feature a genetics topic on the CT DPH website or email reminders to visit various existing websites)	9
Educate non-genetics health professionals	2
Provide grants for attending national conferences	2
Support genetics course at UCONN MPH program	1
Educate CT Medicaid to bring them up to date on ICD9/CPT codes for genetics	1
Get local genetics experts involved	1
Newsletter with relevant, concise articles	1
Target and distributed info sheets focusing on specific disorders	1
Develop a genetics task force for people interested and involved in genetics to meet	1
Annual article in the CT Epidemiologist	1
Create Special Interest Groups	1
Education consumers	1
Teleconferences and audio-conferences	1

Many responses were given to the question of what gaps in the delivery of genetic services exist in the state, with 50 respondents weighing in. By far, the most frequently cited gap was the insufficient number of genetic health professionals, primarily genetic counselors and geneticists to deliver the services (21 comments). In addition, the lack of reimbursement or financial resources to cover the cost of delivering genetic services was often cited (19 comments). Table 6 provides a summary of the noted gaps.

Table 6. Gaps cited by respondents (n=50)

Gaps in the Delivery of Genetic Services	Frequency of Request
Insufficient numbers of genetic professionals (geneticists, prenatal genetic counselors, cancer genetic counselors, lab personnel) 15 – lack of genetic counselors, 6 – lack of geneticists, 2 – lack of lab personnel, 2 – lack of genetic centers in general	21
Lack of good reimbursement and financial resources to pay for genetic services including for: centers that are extension of DPH public policy, metabolic centers to address needs of extended newborn screening)	19
Lack of health professional education in areas such as: Metabolic diseases, Importance of genetics in disease	5
Need for legislation to protect patients' rights (privacy)	3
General lack of recognition of complex genetic disorders including: Lack of counseling/psychosocial support for patients and their families, Increased sensitivity by clinical staff to the needs of families	3
Need to address adult onset diseases that have large genetic component	2
Public education re: pros/cons of genetic testing and services	2
Equitable public access to genetic services for: un- or underinsured, culturally diverse population, geographically remote areas	2
No state registry for birth defects/genetic disorders	1
Need for DPH to contribute more funding to centers that are extensions of public policy	1
Downsizing of the CT Pregnancy Exposure Information Service	1
No requirement for taking family histories, especially in adoption cases	1
Need for ongoing surveillance systems to track susceptibility to certain biohazards	1
Need to show when new discoveries reach clinical efficacy and systems to implement them	1
Need to expand Connexin-26 screening	1
Poor planning by local and state communities for children with special needs as a result of genetics	1
Lack of services for adults with genetic diseases	1
Lack of culturally diverse staff	1
Little communication among the genetic facilities in the State	1

Quite a lot of suggestions were offered on how to address these perceived gaps in the provision of genetic services. As expected based on the list of gaps, the majority of suggestions included funding of programs (15), improved reimbursement for services (5) and education of both the consumer (12) and medical (4) communities. An expanded list of suggestions is found in Table 7.

Table 7. How to Address the Gaps in the Delivery of Genetic Services in Connecticut (n=48)

How to Address the Gaps in the Delivery of Genetic Services
State and Federal Funding for items such as: Genetics professionals (geneticists, genetic counselors), Additional genetic centers, Various support services for patients Required treatments not covered by insurance In-state testing labs Training of medical technologists Consumer and health professional education Low protein food and formulas CPEIS
Public education by Distributing publications in schools, supermarkets, Offering news stories on TV, magazine, other public media, Implementing a direct mail campaign, Providing seminars to cultural groups and underserved populations
Educate the medical community using: Grand rounds, DPH sponsored nursing supervision meetings, Seminars at local hospitals
Improve insurance system by: Passing legislation regulating the coverage of genetic services, Providing relief from exorbitant malpractice insurance premiums, Educating the insurance companies, Involving genetics professional in updating ICD9/CPT codes, Developing administrative role to address insurance issues
Grow and maintain strong genetics programs in the state by: Recruiting and hiring more genetics professionals, locate them at testing sites, Supporting licensure for genetic counselors, Creating a genetic counseling training program in the state, Ensure health professionals and staff are culturally diverse, Creating incentives for institutions to provide genetics services as a public health issue. Supporting the importance of genetic services in the management of patients, Establishing genetic services programs with lifespan approach (birth to geriatrics)
Create and manage a strategic planning or advisory committee that includes consumers and genetic experts. Create forums for consumers to express viewpoints to health professionals & policy makers.
Maintain a registry of children with special needs and carry out better screening for children with genetic disorders Develop a long-term storage of DNA for individuals with unconfirmed syndromes
Require standardized medical history questionnaire, especially for adoption cases. Encourage senior citizens to pass down family history information
Develop strategies for bioterrorism with members of the public health BT community

When asked how the State might be able to help in these efforts, respondents said the State should take a leadership role and be the organizing force. In addition to implementing many of the suggestions provided in Table 7, respondents recommended that the State work with national groups, lobby Congress and Medicaid/Medicare, and work with local educational institutions and private organizations to implement the programs.

The respondents reiterated, time and again the need for the State to provide funding for services, staff and patient support; recruit culturally diverse genetic experts and staff, initiate educational programs for consumers, the insurance industry and the medical profession; and pass legislation to support these plans.

Discussion:

The findings derived from this needs assessment of genetics professionals in Connecticut has provided some insight into their current practices, their perceived obstacles to practicing and suggestions for improving the delivery of genetic services in the state. Almost half of those identified as CT based genetics professionals responded to the survey, although only half of those say they see patients as part of their work. Therefore, only 35 or so individuals answered the questions regarding clinical practices. Nineteen of the respondents were genetic counselors and four were clinical geneticists. The remaining clinicians were non-genetics physicians involved in the field (e.g. maternal fetal medicine), laboratory geneticists or other health professionals.

Of those respondents that do see patients, the majority are in the prenatal field, one of the medical areas that was reported to be suffering most from a lack of genetic counselors. Cancer genetic counselors were also noted to be in demand, with insufficient professionals to address the population's needs.

Several respondents expressed concern about the lack of access to genetic services that will exacerbate the "tiers of health care that are currently in effect", caused primarily by insurance inequities among the under represented populations. However, while many of the respondents claimed that reimbursement and lack of funding for services was a major deterrent, the majority was unable to say what percentage of their patients' services are reimbursed. Yet, it was still clear that there are considerable inconsistencies in reimbursement rates for the counseling and genetics consultations. According to the study, 30% and 36% of the patients get between 50-100% reimbursement for counseling and a geneticists' consultation, respectively.

Race disparities are often another concern regarding inequitable access to services. However, it seems from the data that Black/African-Americans and Hispanics compose about the same, if not greater proportion of the genetics professionals' patient load as their percentage of the state's population (1). While Black/African-Americans are 10.2% of the state's population, the 33 clinicians that provided this data reported that this group makes up between 1-40% of their population. Similar data exists for the Hispanic population, which makes up 9.5% of the state's population, though the 31 responding clinicians say their patient population is comprised of between 1-50% Hispanics.

Clearly, but not surprisingly, the three most significant issues for genetics professionals are the need for more genetic professionals, increased funding and improved reimbursement, and educational efforts for all (consumers, health professionals, insurance companies).

Regarding the cost of services, respondents felt that it was unrealistic to rely on commercial and market factors to regulate the use of genetic services. Reimbursement for services is generally lower than the cost of providing these services, which forces institutions to reduce their genetics staffs and ability to offer genetic testing and counseling to the growing number of patients that could benefit from it. This will only increase as more tests become available for adult onset disorders such as heart disease and psychiatric disorders. Approaches to remedy this problem varied, but it is clear that genetics professionals are frustrated with the current system of access to and reimbursement for genetic services.

Respondents also felt that an educational campaign was necessary to improve the understanding of genetics' impact on health and availability of the service. While the genetics professionals preferred medical journals, national and local conferences, and the Internet for their continuing education, they suggested the use of mass media and local seminars to educate the public and health communities.

There were several limitations to this study that affected the analysis and interpretation. The response rate was only 43%, although two reminder messages were sent. However, the surveys were fully completed by respondents and consistent themes were observed. In addition, a large percentage of respondents do not see patients on a clinical basis. This leads to the speculation that the sample group included individuals whose roles were not primarily in the clinical genetics arena. The group not only included members of the three primary genetics organizations (NSGC, ACMG, ASHG), but also the list of attendants at a recently held genetics and public health conference in Connecticut. The conference list was likely to include several public health, rather than genetics professionals. Only 29% of the respondents reported their primary role as geneticist or genetic counselor.

In summary, the respondents consistently identified reimbursement/program funding, education, and the lack of sufficient genetics personnel as issues currently undermining the State's ability to offer a strong genetics service to its residents and prepare itself for the increasing demands for genetic testing and counseling. These concerns resonate with those expressed by genetics professionals around the country. However, each state must find its own strategy for dealing with this growing public health issue.

References

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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 sub-group.
- 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.

Appendix E

Plan to Create a
Child Health Informatics Profile
of
Linked Databases
within
The Connecticut Department of Public Health

Prepared By

The Genetics Planning Project Team

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DEFINITIONS

CHIP – Child Health Informatics Profile – electronic set of personal portfolios on children born in Connecticut that contains each child's comprehensive health and biographical information reportable to DPH.
Data Integration – process of combining two or more databases into a single, real-time, interactive database.
Data Linkage – process of combining two or more unique, retrospective databases in an *ad hoc* manner into a meta-database by matching unique identifiers within the databases.
Data Warehouse – the meta-database that results from data linkage, that is centrally located, that is automatically fed data regularly from multiple databases, and that generates data marts containing subsets of information extracted from the warehouse.

CGS – Connecticut General Statutes; **CSHCN** – children with special healthcare needs; **CEDSS** – Connecticut Electronic Disease Surveillance System; **DOIT** – Department of Information Technology; **DPH** – Department of Public Health; **HIC** – Human Investigations Committee; **RCSA** – Regulations of the Connecticut state agencies; **WIC** – Women, Infants, and Children.

EXECUTIVE SUMMARY

Goals and Objectives

The goal of this plan, developed by the Genetics Planning Team, is to create an electronic database within DPH that contains high-quality, comprehensive child health data that greatly simplifies data sharing among staff in DPH and with healthcare providers and researchers outside DPH. Two specific objectives are identified to achieve this goal by the year 2010.

Objective 1. Develop a Child Health Informatics Profile (CHIP) of child health data. The CHIP, with varied levels of access control, should contain high-quality retrospective data on newborns and children, consisting of results of metabolic and infectious disease tests, congenital abnormalities, hearing screening results, birth and death records, immunization status, and other health data. It should be capable of expansion for tracking health information across the lifespan, and being converted into a fully integrated database. Its creation should not interfere with existing databases in the Department. It should also simplify data sharing for assurance and assessment public health functions across Divisions by use of a customized and user-friendly reporting system.

Objective 2. Investigate legislative issues in data sharing and remove barriers to sharing data contained in the CHIP, while maintaining personal privacy. A legislative advisory group of individuals within and outside the Department should be created to consider the current limitations of data sharing within DPH and with healthcare providers outside DPH, and to recommend legislation necessary to remove needless barriers to sharing data contained within the CHIP, while remaining sensitive to issues of personal privacy.

Background

Within DPH, fourteen health-related databases mandated by the Connecticut General Statutes (CGS) and controlled by Regulations of Connecticut State Agencies (RCSA), are managed and stored at separate, local sites. Eleven of the databases contain child health data with personal identifiers. These data are not linked, resulting in multiple independent data "silos" within DPH containing child health data. Given this infrastructure and the large number of child health-related databases within DPH, a data warehouse may fit best the short-term needs of the Department to create a single, comprehensive child health informatics Profile (CHIP). A data warehouse links two or more distinct, retrospective databases while maintaining the integrity of the original databases. A data warehouse is also stored at a central location, and is automatically and periodically fed retrospective data from numerous distinct databases. Data extraction, translation, and cleansing occur regularly at this central location, and the data warehouse is secure, limiting access to sensitive information. Creation of a data warehouse requires an initial investment of time and money, but with the combination of the large number of databases required to generate a CHIP within DPH, a data warehouse becomes more cost effective. Also, data extracts, or data marts, are created from the data warehouse, and contain only the amount of information that is needed for an end user. Data marts need not have data components that compromise confidentiality, and each could be in a format that is tailored to meet the needs of an individual user. A web-based query system that is user-friendly is also possible.

Regardless of the method chosen to build a CHIP, each must fulfill the following requirements to be included in a growing comprehensive data system: 1) the database must contain unique identifiers that can be matched, 2) the database must contain data that enhance information, and 3) the database must be stored on a software platform compatible with the software chosen to build the CHIP. Of the fourteen mandated health-related databases within DPH, eleven fulfill these criteria. These databases are: The Child Health Profile, which, once completed, will consist of Laboratory newborn screening, Birth Defects Registry/ Children with Special Healthcare Needs (CSHCN) Registry, and Newborn Hearing Screening databases; Birth Records; Death Records; Childhood Immunization Registry; Connecticut Electronic Disease Surveillance System (CEDSS) databases, which, once competed, will subsume the AIDS/HIV and Hepatitis B & C databases; Women, Infants, and Children (WIC) database; Lead Surveillance database; and Tumor Registry.

Benefits, Potential Uses, and Challenges

Completed objectives would yield a CHIP containing comprehensive child health information, and a set of recommended legislative recommendations that remove needless barriers to sharing the data contained within the CHIP. The following future enhancements would be possible: incorporate genetics services and more genetic test information as it becomes available; include health information across the lifespan and health information from databases outside DPH, including Medicaid, the Birth-to-Three program, The Department of Mental Health and Addiction Services, health and expansion to consider child wellness; convert the data system into a fully integrated, interactive database in real time; and provide accessibility to researchers and qualified healthcare providers.

A CHIP would greatly enhance public health assurance and assessment activities within Connecticut. Assurance activities enhanced by a CHIP include: 1) better coordination of medical services to all children and especially CSHCN through linkages with qualified medical home environments; 2) reduced health disparities among childhood disease prevention activities through better outreach to the “hard to reach” populations; and 3) reduced need to disclose confidential information that is now needed to generate local linkages. Enhanced assessment activities include: 1) an increased ability to evaluate population-based health activities within the Department; 2) improved data quality through better data validation and coordinated data improvement efforts; 3) enhanced comprehensive data accessibility to support grant activities, health programming, and to support data requests from sources outside DPH; and 4) enhanced analyzing, interpreting, monitoring, and reporting activities by staff because less time would be needed to manage data. Creation of a CHIP also makes possible for the first time the support of new program activities that reach across divisions within DPH.

With the anticipated importance of genetic tests and their relation to a broad range of disease susceptibilities, a comprehensive set of linked health data, such as a CHIP, is important for future public health activities. Current legislation does not apparently preclude the creation of a data system of linked child health, and data extracts containing the same amount of information fed into the meta-database could certainly be channeled back to the individuals who manage the original databases. Data extracts that do not contain personal identifiers could also be readily shared. Sharing data extracts containing personal identifiers with individuals across divisions or outside DPH, however, could raise privacy issues. Specific legislation sensitive to community

issues of privacy may be required to allow data sharing of the information contained within the proposed CHIP.

CHIP Workplan

Objective A. Develop a CHIP of child health data (Years 01 – 04).

Step 1. Obtain an executive charge, create a Data Committee, and identify necessary characteristics of a completed CHIP.

Step 2. Contract with an external group to develop a technical strategic plan.

Step 3. Identify and secure sustainable funding to support the project.

Step 4. Identify and contract with an external group to develop the CHIP.

Step 5. Develop a pilot meta-database.

Step 6. Build a CHIP with additional DPH databases.

Step 7. Train DPH staff to use and maintain the CHIP.

Step 8. Investigate and develop a web-based query system for use within DPH and to selected individuals outside DPH.

Objective B. Investigate legislative issues in data sharing and remove barriers to sharing data contained in the CHIP, while maintaining personal privacy (Years 01 – 03).

Step 1. Create and charge a legislative advisory group.

Step 2. Investigate data sharing within DPH and outside DPH.

STATEMENTS

Shortly after each birth in Connecticut, data necessary to generate a birth certificate are collected, along with newborn immunization status, obvious physical disabilities, hearing screening results, and positive results of newborn laboratory screening. These data are reported to DPH and maintained in separate databases. Recent advances in electronic information technology make it possible to link these disparate databases into a single database containing comprehensive child health information, which we call a Child Health Informatics Profile (CHIP).

MISSION STATEMENT

Consistent with HRSA Title V objectives (Maternal and Child Health Bureau, 2004), the mission identified in this Plan is to create an interactive, real-time data sharing system that could be used by public health and clinical healthcare professionals to monitor the health status of Connecticut's children and adolescents, and to use family health and healthcare history to coordinate healthcare services. This ultimate child health database would: 1) maintain neonatal laboratory tests, birth, and immunization records for each child in Connecticut; 2) facilitate coordination of regular healthcare services for each child in Connecticut; 3) support services for those children with special healthcare needs; and 4) enhance public health assessment functions. This mission is consistent with the overall mission of the Connecticut DPH "…to protect the health and safety of the people of Connecticut and actively work to prevent disease and promote wellness…[and] to collect and analyze health data to help plan policy for the future" (Connecticut Department of Public Health, 2004).

GOAL STATEMENT

The goal of this Plan is to create an electronic database within DPH, fully functioning by the year 2010, which we call a Child Health Informatics Profile (CHIP). The CHIP would contain high-quality, comprehensive child health data from separate databases within DPH, and would be capable of sharing its contents with staff inside DPH for assessment and assurance public health functions. It would also have the capacity to share aggregated information to selected researchers outside DPH, and to provide individual tracking information to qualified medical homes. The CHIP would be compatible with other child-related databases stored at other state agencies, and would have the capability of expansion to include health and genetic information across the lifespan. While maintaining personal privacy, needless barriers to sharing data contained within the CHIP would be removed.

OBJECTIVES

Two specific objectives toward achieving the above goal, which should be accomplished within four years, are:

Objective A. Develop a Child Health Informatics Profile (CHIP) of child health data from separate databases housed within DPH.

Objective B. Investigate legislative issues in data sharing and remove barriers to sharing data contained in the CHIP, while maintaining personal privacy.

BACKGROUND

DATA COMBINATION METHODS

Within DPH, fourteen mandated health-related databases containing personal identifiers are managed and stored at separate, local sites, and eleven of these contain child-health data (see **Appendix A).** Data are not linked, resulting in multiple independent data "silos" within DPH containing child health data. Given this infrastructure and the large number of databases within DPH, there are three discrete methods with increasing degrees of sophistication that could be used to combine data: 1) local data linkage, 2) a method of data linkage called data warehousing, and 3) real-time data integration, expanding the database of an existing database. A fourth, "hybrid" method is possible that involves a combination of these three discrete methods. Definitions for data linkage and data integration vary, and for the purposes of this plan, *data linkage* is the process of combining two or more unique, retrospective (not in real time) databases by matching unique identifiers within the databases. *Data integration* is the continuous process of combining two or more databases into a single, real time, interactive database.

Data linkage at the local level is the process of linking two databases by matching unique identifiers in an *ad hoc* manner, with the original databases remaining intact **(Table 1)**. The resulting linked database is subsequently cleansed of missing data and errors, and linkage of additional databases to the growing meta-database is handled similarly. This linkage technique

Table 1

Desirable Features of
Data Linkage and Data Integration Methods

	Data Linkage Methods		
Desirable Features	**Local Data Linkage**	**Data Warehousing**	**Data Integration**
1. Database Maintenance			
Data are managed at a central location		✓	✓
Data are cleansed & stored at a central location		✓	✓
Database updates are easy & automatic		✓	✓
Integrity of the original databases are preserved	✓	✓	
Combined databases are stored on diverse platforms		✓	
Data are in real time			✓
2. Resources			
Database is created in relatively a short timeframe	✓	✓	
Cost effective for a small number of databases	✓		
Cost effective for a large number of databases		✓	✓
3. Usefulness to Personnel			
Data are accessible to a large number of end users		✓	✓
Minimal end user training is required	✓	✓	
Custom data sets are available to end users		✓	✓

works well for a limited number of databases that exist on compatible platforms familiar to the individuals who manage the databases **(Table 1)**. The process is inexpensive, but as the number of linked databases grows, this *ad hoc* process becomes increasingly more cumbersome and requires increasing amounts of resources. Also, confidentiality issues surface each time a database is shared, so periodic updates, which require repeated, manual linkages, are cumbersome. Some of the information contained in the meta-database is likely to be irrelevant to some end users, requiring complex extraction procedures. Of the three discrete methods, local data linkage is the most limiting.

A data warehouse, like local data linkage, links two or more distinct, retrospective databases while maintaining the integrity of the original databases. This method improves many of the disadvantages associated with local data linkage **(Table 1)**. For instance, a data warehouse is stored at a central location (SAS, 2002), and is automatically and periodically fed data from numerous distinct databases **(Figure 1**; *Dbase***)**. Data extraction, translation, and cleansing occur regularly at this central location, and the data warehouse is secure, with limited access to sensitive information (Greenfield, 2002a).

Creation of a data warehouse requires an initial investment of time and money (Greenfield, 2002b; **Table 1)**. The technique may not be cost-effective for a small number of databases, but with the combination of a larger number of databases, a data warehouse becomes more cost effective. Also, data extracts are created from the data warehouse that contain only the amount of information that is needed for an end user. These extracts, called data marts **(Figure 1**; *Dmart***)**, are used by end users for data mining, analysis, and reporting (Greenfield, 2002c). Data marts simplify data sharing because the extracted data need not have data components that compromise confidentiality. Also, each data mart could be in a custom format that is tailored to meet the needs of an individual user **(Table 1)**. Although a data warehouse is composed of dated information, current technology is moving quickly toward the capability of rolling data warehouses in real time (Haisten, 1999).

Whereas data linkage occurs with dated events, a fully integrated database is updated immediately with data entry **(Table 1)**. Individuals at local sites access elements of the database, modify and add personal information from their stations, and send information directly back to the database, avoiding duplication of data entry and updating the database with each modification. However, the cost of creating a fully integrated data system is high, requires that all individual databases be converted into a single software platform, and disrupts the integrity of the original databases **(Table 1)**. Development of integrated databases can take many years, and in the short term may not be the most cost-effective and comprehensive method for Connecticut.

Other states across the nation are creating child health profiles, each using methods especially suited to meet their needs. The method of choice has depended in each case on the unique environment within each state, the platform on which database were created, the amount of data available in each database, and available resources. The states of Utah, Oregon, Hawaii, and the District of Columbia are using data warehouse technology (*G. Land; personal communication*). Rhode Island, however, is using the "hybrid" method to link its health data, which involves a combination of data integration and data warehouse technology (*A. Zimmerman-Levitan; personal communication*). Missouri pioneered the use of a data warehouse to link health data, and has developed a warehouse of metabolic and hearing screening test

results linked to an integrated database that manages birth and immunization records of all children in that state (Land, 2002). The state has as its ultimate goal the creation of a single, real-time database, and has been working toward this goal for the past 15 years. Regardless of the method used, the successes of Missouri, Rhode Island, and other states in creating child health profiles within their states indicates that a similar project in Connecticut could also be successful.

Table 2

Databases Containing Child Health Information
Selected Features

Database	Database Software	Unique Identifiers					Data components that would contribute to a CHIP
		AN	CN	MN	DB	SS	
Newborn Screening *(stored at DOIT)*	DBase	X	X	X	X		Birth defects, biographical data, hearing screening results
DPH Laboratories	*DBase	X	X	X	X		Newborn screening test results
CYSHCN Registry/ Birth Defects Registry	ORACLE	X	X	X	X	MC	Birth Defects, CSHCN status Biographical data
Newborn Hearing Screening	*ACCESS	X	X	X	X		Newborn hearing results, biographical data
Birth Records	ASCII		X	X	X	MC	Biographical data family behavioral risk factors
Death Records	ASCII		X	X	X	C	Biographical data, cause(s) of death
Immunization Registry	*DBase		X	X	X		Immunization status, dates of vaccinations, identity of medical home
Hepatitis B & C	*ASCII		X		X		Hepatitis test results, social & behavioral risk factors
AIDS/HIV	*ASCII		X		X		HIV test results, social & behavior risk factors
WIC Food Program	FoxPro		X		X	M	Family income, eligibility status, use of funds
Lead Surveillance	*FoxPro		X	X	X		Services & treatment, blood lead level
Tumor Registry	ASCII		X		X	C	Type of tumor, site of tumor, diagnosis date, census tract

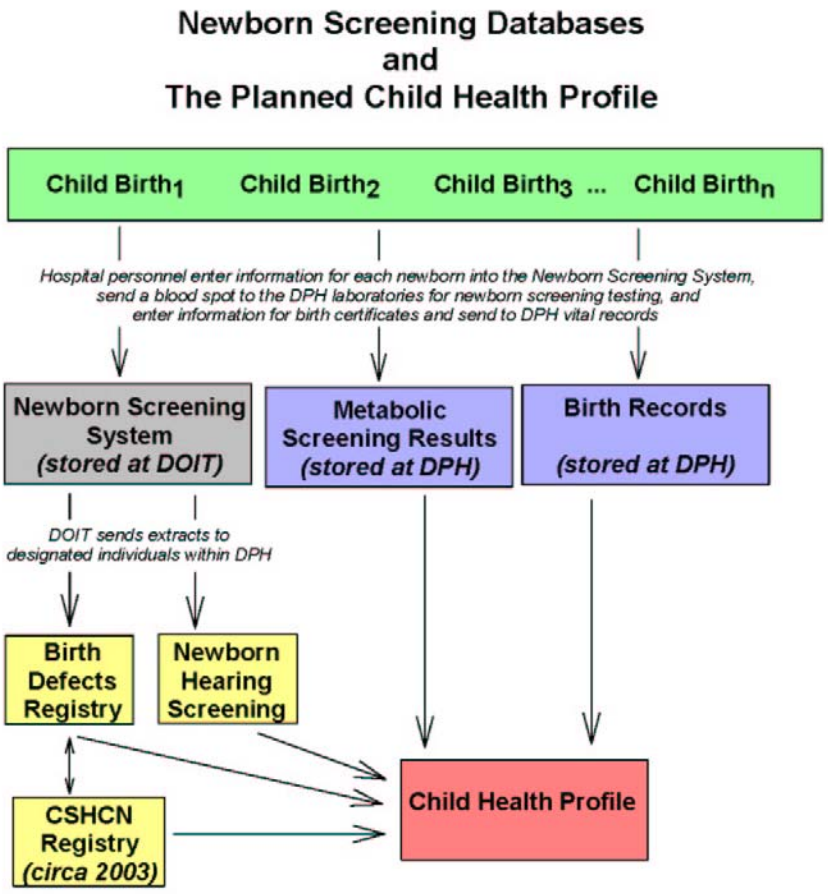
*Platform conversion to ORACLE is planned (see **Database Compatibility Survey**).
CSHCN – Children with Special Healthcare Needs, WIC – Women, Infants, and Children
AN –Accession Number; CN – Child's Full Name; MN – Mother's Full Name; DB – Date of Birth; SS-Social Security Number (Mother's (M), Child's (C))

CHILD HEALTH DATABASES WITHIN DPH

Of the fourteen health-related databases mandated by the Connecticut General Statutes (CGS) and Regulations of Connecticut State Agencies (RCSA), eleven contain information on newborns and children (*D. Maselli; personal communication*; **Table 2**). Databases containing newborn health information are populated from a twelfth database stored within the Department of Information Technology (DOIT), on a Dbase platform (*N. Athukorala; personal communication*; **Figure 2**). This master database, partially in use, is called the Newborn Screening System. It relies on hospitals to voluntarily create a record of every child born within their facility. Biographical information, birth defects, and hearing screening results are currently available from the database. At the time a child is entered into the Newborn Screening System, a unique accession number is created and stored in the database (**Table 2**), and this number is carried into other newborn databases as data are extracted for use within DPH.

A few drops of blood from each newborn are collected onto filter paper and sent to the state laboratory to be tested for an array of genetic disorders (*F. Larson; personal communication*). Results from one of eleven state-mandated newborn genetic tests, are stored in the Gemini database at the DPH laboratories. The biochemical tests screen for genetic disorders including phenylketonuria, hypothyroidism, galactosemia, sickle cell disease, maple syrup urine

Figure 2



disease, homocystinuria, biotinidase deficiency, and congenital adrenalhyperplasia, and for the presence of HIV antibodies (RCSA 19a-2a-15, CGS 19a-5; *D. Mayo; personal communication*). The state recently began screening for other metabolic disorders, including medium-chain acyl-CoA dehydrogenase deficiency, and long chain 3-hydroxyacyl CoA dehydrogenase deficiency *(Public Act 02-113, 2002)*, and the technology used for these tests makes possible future tests of other amino acid and fatty oxidation disorders.

The Children with Special Healthcare Needs (CSHCN) Registry (RCSA 19a-2a-3,CGS 19a-50, 19a-53, a91-54, and 19a-56a), and Newborn Hearing Screening database (CGS 19a-59-1), are retrospective extracts obtained separately from the Newborn Screening System, and are extracted from DOIT daily to specific individuals within the Family Health Division of DPH **(Figure 2)**. The Newborn Hearing Screening database contains the test results of newborns tested for hearing loss, risk factors for hearing loss, and audiological referral information *(D. Maselli; personal communication*). The CSHCN Registry, which is currently not being updated with child health information, contains the identity of children with special healthcare needs, as well as information about referral to early interventions offered to each child. The CSHCN Registry was recently transitioned into the Birth Defects Registry, and it contains information about children detected at birth with birth defects *(C-F. Liu; personal communication*). Data from the Birth Defects Registry, as well as reports from future medical home environments, are planned to re-population the CYSHCN Registry.

Within a week of a child's birth, hospital personnel report information contained in Birth Records to DPH, and birth certificates are drawn from these records **(***C. Whopper; personal communication;* **Table 2; Figure 2)**. This database contains a wealth of information on each newborn, as well as on the parents, and is considered one of the most important databases within the Department (see **Appendix A**). Maternal risk factors such as tobacco and alcohol use are recorded, as well as weight gain during pregnancy and the frequency of prenatal care. Race and ethnicity of the mother and father are also recorded, along with birth order of the infant and any congenital abnormalities or birth-related conditions. Information about WIC usage and Medicare enrollment are entered into the birth record, and plans are under way to include additional WIC usage information of the mother during pregnancy (*L. Mueller; personal communication*). If a child dies, Death Records contain information about the primary cause of death, other secondary causes of death, as well as the age at which the child died. The birth and death vital records data are also important to monitor pregnancy outcomes and adverse health effects associated with maternal risk behaviors.

Additional databases within DPH contain valuable child health information **(Table 2)**. Positive test results for the infectious diseases AIDS (AIDS/HIV; RCSA 19a-2a-22) and Hepatitis B & C (RCSA 19a-2a-12) are currently maintained in separate databases (*A. Roome, B. Baume; personal communication*). The Immunization Registry contains vaccination information on children and neonates (CGS 19a-7h). The Women, Infants, and Children (WIC) database contains information on individuals requiring federal food assistance (RCSA 19a-2a-18), and data on family income and eligibility status (*T. Young; personal communication*). The Lead Surveillance database (RCSA 19a-2a-6) contains records of children exposed to lead, and information such as blood lead level and medical treatment is recorded (*K. Frost, J. Peng; personal communication*). Finally, although cancer afflicts only about 150 children aged 0-19 annually, the Tumor Registry maintains information on cancer in children, as well as in adults (*H. Swede, R. Capozzi; personal communication*).

The creation of a CHIP composed of the child health databases within DPH, regardless of the method used, requires that each databases: 1) contain unique identifiers that can be matched as databases are linked; and 2) contain data that enhance information in a growing data system. All the databases containing child health information within DPH fulfill these two requirements **(Table 2)**. The CSHCN Registry, Birth Defects Registry and Newborn Hearing Screening databases contain the same unique accession number that is created by the Newborn Screening System to identify each newborn entered into that system. Additional information such as the child's full name, mother's full name, or date of birth, could be matched among the other databases. Each of the databases contains information that would contribute uniquely to a CHIP, and that would prove useful as future genetic tests become available. All the databases are either stored on software platforms compatible with a large number of data systems software, or can be converted to compatible formats through data extracts, indicating that a fully functioning CHIP could be developed in the short term with data warehouse technology (**Table 2**).

Table 3

Current Database Linkages within DPH[1]

Selected Linked Databases	CEDSS	Immunization Registry	Child Health Profile	Cancer Registry	Birth Records	Death Records	WIC	Lead Surveillance	[2]Laboratory Results
CEDSS	■							▒	▒
Immunization Registry		■			▒	▒			
Child Health Profile			■		▒				▒
Cancer Registry				■		▒			
Proposed CHIP	▒	▒	▒	▒	▒	▒	▒	▒	▒

Patterned Shaded Cells - full linkage; Shaded Cells - linkage of selected data elements; Blackened Cells - N/A

[1] - Although linkage with external databases such as CHIME, Medicaid/Medicare, and Birth-to-Three may be important, our focus for this discussion will be on linkable databases within DPH.

[2] - Laboratory Results include newborn screening, infectious disease

CURRENT EFFORTS IN DATA SHARING

Three data sharing projects are currently in progress and near completion within DPH. These projects will soon lead to: 1) the Child Health Profile within the Division of Family Health; and the 2) the CEDSS database, and 3) the Immunization Registry within the Division of Infectious Diseases. All fulfill important functions within their respective units, and include some degree of linkage with other databases within DPH. All rely on information from external sources such as medical homes, hospitals, or laboratories. A fourth project is in the early planning stages. Each of these projects are discussed more fully below.

The Family Health Division within DPH, which is working with DOIT to develop the Newborn Screening System, also has responsibility for the Hearing Screening, Birth Defects, and CSHCN databases, which are stored within DPH. Individuals within the Division are near completion of a project to combine information from these three databases into a single Child Health Profile (*C-F. Liu; personal communication*; **Figure 2**). All three databases are stored in ORACLE, so a fully integrated database is possible, but the data extracts obtained from the Newborn Screening System are retrospective, which precludes creation of a real-time database. Work was also recently completed to match children in the database with their birth records, obtaining valuable biographical information and confirming congenital defects *(***Table 3**). The Birth Records database is located outside the Family Health Division.

The amount of information extracted from Birth Records to augment the Child Health Profile is limited to those biographical fields that support the programs within the newborn screening program (*C-F. Liu; personal communication*). For instance, at the time data are entered by hospital personnel into the Newborn Screening System, some infants have yet to be assigned a first, middle, or last name by their legal guardian. By matching the subsequent Child Health Profile with Birth Records, the database will be able to maximize the quality of its data. Once fully functional, the database will identify children who test positive for newborn screening tests, refer them quickly to treatment, and track them throughout childhood. The database will play a vital role in public health assurance functions for CSHCN, allowing the Division to respond rapidly and efficiently to newborns identified with special needs.

Matched birth record extracts are also used to augment the Immunization Registry (*D. Fraiter; personal communication*) (**Table 3**). In addition to Birth Records, this Registry also matches biographical information from Death Records to its database (*M. Tommasi, N. Caruk; personal communication*). The data are used to identify those children who are deceased and who should be deactivated from the database. The Immunization Registry is being partially implemented and is providing access to a small percentage of medical home environments, allowing qualified medical providers to view and update the vaccination records of Connecticut's children. Once fully implemented, this capacity will aid tracking activities within the Immunization Program and will provide a valuable public health assurance function. For reasons similar to those with the Immunization Registry, the Tumor Registry is linked to Death Records by local data linkage process requiring manual updates (**Table 3**).

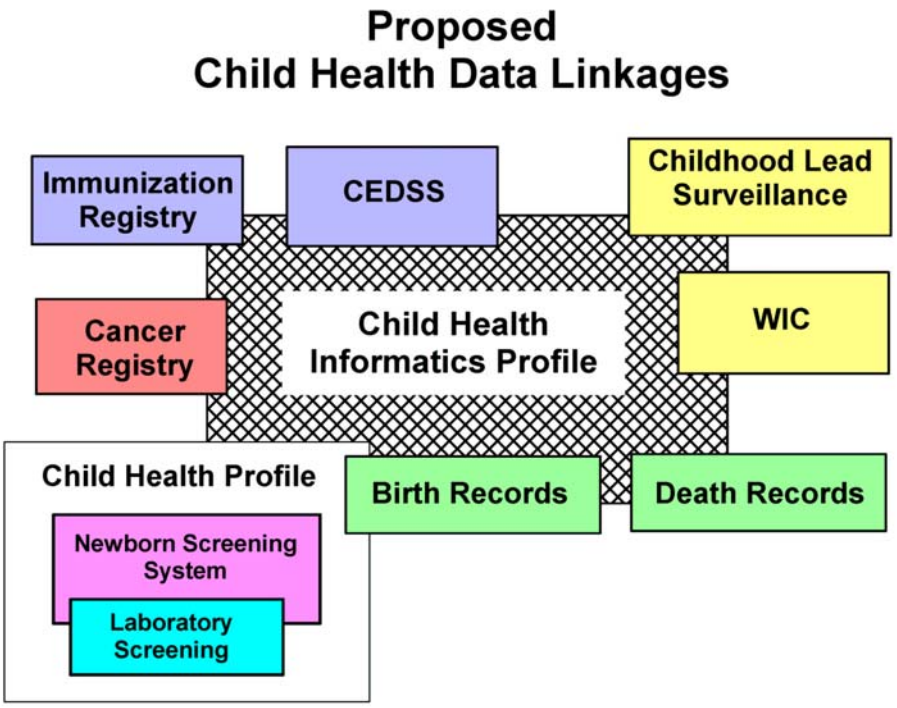
Another project underway that will streamline disease tracking is the Connecticut Electronic Disease Surveillance System (CEDSS), which is targeted for completion by January, 2005 (*N. Barrett, G. Archambault; personal communication*) (**Table 3**). An extensive project that will track reportable infectious diseases throughout the state, the project relies on a combination of integration and data warehouse technology. Although reported diseases such as

AIDS and hepatitis B & C are currently stored on separate databases, these databases will be subsumed by CEDSS. Future plans include the incorporation of lead surveillance data. The CEDSS database will fulfill an important infectious disease tracking function capable of the rapid response needed for emerging epidemics, but the system will contain additional information that may not be useful for a database of child health information. Also, like the Child Health Profile, only children entered into CEDSS will be linked to Birth Records, and public health assessment activities will not be possible with this data system. In addition, only data fields needed to support CEDSS disease activities will be extracted from Birth Records.

A fourth data sharing project in the early stages of planning is an Environmental Public Health Tracking Program (EPHTP) tracking system (Centers for Disease Control, 2003a; *K. Frost; personal communication*). Its goals are to link environmental hazards, environmental exposure, and health effects for public health assessment and tracking activities. Although parts of this effort may be associated with CEDSS (*G. Archambault; personal communication*), the plan is not yet complete.

Current data sharing efforts underway within DPH support specific programmatic assurance, or tracking, activities, and are tailored to meet the needs of those divisions within DPH that are developing the data systems. A CHIP, however, could benefit many divisions simultaneously.

Figure 3



Extracted child health data from across divisions, linked together by unique identifiers, are proposed in this Plan (**Figure 3**), expanding the current Child Health Profile to include health data from all eleven of the databases within DPH that contain child health information. The data system would allow assessment activities, as well as assurance activities, because all births in the state would be entered into the meta-database.

Data integration efforts across the country are receiving support from several national programs. A program called All Kids Count *(http://www.allkidscount.org)*, directed by the Public Health Informatics Institute *(http://www.phii.org*), with funding from the Robert Wood Johnson Foundation, has as one of its goals "to develop an action agenda for child health information systems that meet medical care and public health needs and to develop resources and tools that assist public health agencies in developing information systems that help ensure timely delivery of child health services and follow-up" (All Kids Count, 2004). The Public Health Informatics Institute is a component of The Task Force for Child Survival and Development *(http:www.taskforce.org*), and the organization provides leadership and resources on data integration efforts and works closely with the Maternal and Child Health program of HRSA (*D. Ross, D. Linzer; personal communication*). A variety of publications on the topic of child health database linkage projects are available through this agency (Public Health Informatics Institute, 2004). Data integration is also encouraged through the Title V Maternal and Child Health block grant.

To provide a user-friendly method to access resulting child health meta-databases, a web-based query system developed in Missouri and financed by CDC is offered free of charge to interested states (*G. Land; personal communication*). These resources are helping to make data integration a reality for many states. Locally, the University of Connecticut Health Center, through its burgeoning School of Public Health, is examining ways to integrate data within the medical center to facilitate child health research (*R. Aseltine; personal communication*). In addition, DataConnect (*http://www.chdi.org*), a nonprofit state organization, is helping to facilitate child well-being data that reaches across state agencies within Connecticut (*S. Wilson; personal communication).* Interactions with these two organizations have been initiated. With support from the national and local level, there is no better time for DPH to develop and implement a CHIP of comprehensive child health data.

DATABASE COMPATIBILITY SURVEY

SUMMARY

To determine if health databases within DPH are compatible with data sharing, a survey of all individuals who manage the databases was developed and conducted during in winter, 2003. All data managers of the fourteen mandated health databases within DPH were surveyed by written surveys and follow-up interviews. The results indicate that all the databases as currently structured could be linked by data warehouse technology to generate a comprehensive CHIP in the ORACLE platform, and that a fully integrated meta-database may be more cumbersome to develop. The databases are stored on as many as 8 different software platforms, but all platforms are compatible with ORACLE software. Also, it is expected that nearly half of the databases will be shifted into ORACLE software in the future, simplifying creation of a data warehouse or facilitating a fully integrated data system. Although some training in ORACLE may be required, the concept of data warehousing is familiar to 93% of the data managers, which should facilitate its development. Unique identifiers within the databases could be matched, and most of the databases have data dictionaries that would aid database linkage. The survey results also indicated that datasets extracted from a data system for use by different program areas will need to be in a variety of formats to assist the diverse needs of end users. Because enhancements of all databases in the Department are continually ongoing, additional surveys are needed to update the current status of these databases.

INTRODUCTION AND METHODS

To deelop a plan for integrating child health data within DPH, information is needed about each database that includes details of the data contained within each of the mandated databases that contain child health information and that contain unique identifiers, and information about the ways in which the data are currently used. To gather information on these two criteria, a survey was developed and conducted of the data managers who manage the health-related databases within DPH. The survey was distributed by email to each of 14 data managers, and a follow-up personal interview of each was also conducted. All responded to both the survey and the interview. The survey, which took four weeks to complete, was initiated on January 16, 2003. Specific summary responses to the survey are included in **Appendix A**. The databases from which information was solicited were: Laboratory Newborn Screening, Newborn Hearing Screening, and CSHCN; Birth Records and Death Records; Immunization Registry; AIDS, AIDS Epidemiology, Hepatitis, and Infectious Disease; Tumor Registry; Lead Surveillance and Occupational Health; and the WIC Food Program. Information on the database containing childhood asthma that will began in Fall, 2003, was also collected, but was excluded from the results because the database does not currently include personal identifiers. Additional information in the survey related to uses, benefits, and challenges of a developed CHIP, and are discussed elsewhere (see **Benefits, Potential Uses, and Challenges of a CHIP**).

RESULTS AND DISCUSSION

All the data contained within databases managed in the Agency are secured by password protection, and, in addition, 50% of the databases are maintained on a private network. To those with access through passwords, the complete dataset becomes available to 80% of the databases. Only two databases permit limited access beyond a password. Data within the databases largely contain information on newborns (71%), or children and adolescents (79%). Seven of the fourteen databases contain information on both newborns and children, and six contain data across the lifespan.

All fourteen databases surveyed contain unique identifiers that could be matched when combining databases. The databases surveyed are currently stored on as many as eight different software platforms; most commonly used is FoxPro (3 responses), followed by ACCESS (2 responses) and EpiInfo (2 responses). Among all the databases, however, 57% are expected to change software platforms in the future. Six will be stored in ORACLE, and 2 will be stored in dBase. Ninety-three percent of the databases surveyed have accompanying data dictionaries that contain information on the data stored within them; only 1 database reportedly does not contain such a document. Of those data dictionaries that exist, all contain data element names, and 85% contain data types, data lengths, and data values.

Of the fourteen health-related databases in the Agency, 50% are managed by individuals who also function as end users. Three of the databases are overseen by a coordinator who neither manages nor uses the data, while 64% of the databases are maintained by individuals whose responsibilities for the database do not include use of the data contained within the database. End users analyze data within the databases using various software packages, and most databases are compatible with multiple packages. Of these packages, SAS is used most commonly (24%). Other packages used less commonly include ACCESS, SPSS, FoxPro, and EpiInfo (14%). Also, although 86% of the data managers do not currently use ORACLE software with their databases, 93% percent of the data managers report being familiar with data warehousing. Most data managers report verbally, however, that their familiarity with the software package does not include data warehousing.

Health-related data are used within DPH for a variety of purposes, and many are used for multiple purposes. Sixty-four percent of the databases are used for screening, and 57% are used for policymaking; a smaller number of databases (29%) are used for programs, services, or surveillance. Of the four databases that reportedly have a single use, 3 are used for surveillance and 1 is used for programs. One of the databases, the Tumor Registry, is reportedly used for surveillance, registry, policy making, programs and services, and generating reports.

These results indicate that most databases within the Department could be useful in preparing a comprehensive database of child health data, and data dictionaries are available to aid database linkage. All databases within the Department are compatible with ORACLE software, however, data extracted from a comprehensive database system would need to be tailored to the diverse needs of end users, and would need to be in various formats for analysis. The results also indicate that some data managers could benefit from training in ORACLE and data warehouse management.

BENEFITS, POTENTIAL USES, AND CHALLENGES

To evaluate the potential uses of a CHIP within DPH across divisions, a Round Table Discussion was organized and held on September 9, 2004. This event was coordinated by the Genetics Planning Team, in close collaboration with Lloyd Mueller, Division of Health Information Systems and Reporting, and Marcie Cavacas, Division of Family Health. Keynote speakers began the meeting and consisted of Garland Land from the Missouri Department of Health and Human Services, and Amy Zimmerman-Levitan, from the Rhode Island Department of Health. Following these speakers, presentations were heard from individuals within DPH who represented three current data sharing projects. These data sharing projects were CEDSS, the Immunization Registry, and the Child Health Profile, presented by Gary Archambault, Nancy Caruk, and Chun-Fu Liu, respectively. Although there was little time remaining for participants to enter into a lengthy dialog about data integration within DPH, a follow-up survey was conducted of the participants, and summary responses to a set of questions are presented below. These responses, and those obtained from a survey of database managers within DPH (see **Database Compatibility Survey**), and personal interviews, contributed to the comprehensive lists of benefits and programmatic uses, as well as challenges to a CHIP within DPH. These items are listed below.

BENEFITS AND POTENTIAL USES OF A CHIP

Potential uses of a CHIP containing child health data across divisions within DPH are shown below, loosely divided into either general benefits, or uses specific to individual programs. The extensive and far-reaching list of benefits and potential uses contributed by many throughout the Department demonstrates that a considerable amount of interest in data linkage exists within DPH. Further evidence of this was the nearly unanimous call for further discussion following the Round Table Discussion in September, 2004. This Plan is a response to that call for action.

General Benefits

1. Improved case management would be possible by creating linkages with qualified medical home environments, to improve healthcare services and assessment for children in the state, and to coordinate quality services for children at risk for multiple adverse health outcomes.
2. Access to birth and death records would be possible, which are considered especially valuable databases (see **Appendix A**), and which could be used for population-based assessment, as well as for assurance activities, such as determining gestational age of individual children.
3. Comprehensive data would enhance public health programming and planning, and support new, cross-division programs.
4. Data could inform the five-year Title V (MCH) Needs Assessment, which is a federal requirement, and could also provide supporting statistics for other grant applications.
5. Unduplicated counts of children served within Connecticut could be obtained to support the

annual Title V (MCH) block grant report. These data could be used to document past program activities, and to discuss future planned activities.

6. Comprehensive data could be more accessible to users within DPH, and requests for comprehensive health data and statistics by local health departments and children's advocacy groups could be more easily accommodated.
7. Quality assurance for data validation and overall data integrity could occur, and data improvement efforts could be better evaluated, producing better quality data.
8. Outreach and tracking activities for "hard to reach" populations could be maximized, and disparities in childhood disease prevention could be more easily addressed.
9. Automated matching algorithms could ensure that data are up-to-date, with less duplication, and would streamline data collection and analysis, reducing the resources required to gather comprehensive information on individual children. This would allow analytic staff to better focus on analysis, interpretation, monitoring, and reporting activities.
10. The end user's need for confidential information to link data and generate aggregate statistics could be eliminated. Person-level matched data would already exist, reducing the amount of confidential information provided to investigators.

Family Health Program Uses

1. Children with special healthcare needs (CSHCN) could be monitored for the use of WIC food supplements to ensure maximum development throughout childhood.
2. CSHCN who are eligible but not listed in the registry could be identified.
3. The Medicaid status of CSHCN at birth could be assessed to ensure enrollment in the WIC program.
4. The CSHCN could be assessed for parental education level, history of and co-morbidity of pregnancy, and maternal obstetrical history.
5. Deceased children could be identified to discontinue tracking efforts.
6. Neonatal mortality that is associated with congenital abnormalities could be identified.
7. Information about potential risk factors for birth defects and genetics disorders could be evaluated.
8. Possible newborn genetic tests that identify children at risk for chronic or infectious diseases could be monitored.

Women, Infants, and Children (WIC) Program Uses

1. Health outcomes related to WIC participation could be evaluated.
2. WIC participation could be assessed for targeted outreach and planning activities.
3. Primary prevention activities could be maximized by increasing early enrollment in WIC and monitoring child growth and development.

4. Enrolled children of mothers using WIC during pregnancy could be identified.
5. Children with metabolic disorders could be identified, and food supplementation could be tailored to meet the nutritional needs of these children.
6. Children born of mothers on Medicaid could be identified to ensure continued eligibility.

Lead Surveillance Program Uses

1. CSHCN who are more vulnerable to the toxic effects of lead exposure could be monitored more aggressively.
2. The demographic factors that result in lead exposure among children could be identified.
3. Possible newborn genetic tests that identify children at greater risk for lead toxicity could be identified and monitored.
4. Children enrolled in the WIC program could be monitored for lead exposure.

Infectious Disease and Immunization Program Uses

1. Immunization rates could be increased by identifying target populations in need of vaccinations.
2. Vaccine effectiveness could be better evaluated.
3. The link between infectious disease and susceptibility for cancer could be investigated.
4. Children with reportable diseases who are eligible for CSHCN status could be identified.
5. Maternal risk behaviors that are associated with susceptibilities to infectious disease could be identified among children with reportable diseases.
6. Co-morbidities among children with reportable diseases could be identified.
7. Possible newborn genetic tests that identify children at greater risk for infectious disease could be identified and monitored.
8. The immunization status of children enrolled in WIC could be evaluated.

Chronic Disease and Cancer Program Uses

1. Possible newborn genetic tests that identify children at greater risk for chronic disease could be identified and monitored.
2. CSHCN could be monitored for development of chronic disease during childhood.
3. Predictive information about the prevalence of chronic disease in the future adult population could be obtained for program planning.

CHALLENGES TO CREATION OF A CHIP

Below is a list of challenges cited by individuals who participated in the Data Compatibility Survey (see **Appendix A**) or the Round Table Discussion in September, 2004, and those who contributed information during follow-up interviews. The work plan outlined in the next section (see **CHIP Workplan**) addresses these potential challenges.

1. A sustained commitment at the executive level would be necessary (**Objective A, Step 1**).
2. A dedicated consensus of core individuals across the Department, that includes the Division of Data Processing, would be needed (**Objective A, Step 1**).
3. Standards in data coding to allow future expansion and linkage between agencies is needed (**Objective A, Step 2**).
4. Issues in data quality need to be addressed (**Objective A, step 2**).
5. Resources would be required that include people, time, and money (**Objective A, Steps 2 & 3**).
6. Confidentiality issues would need to be addressed, with legislative support (**Objective B**; see also **Issues in Confidentiality**).

ISSUES IN CONFIDENTIALITY

Historically, health data within DPH have been managed and maintained within separate program units, and, until recently, there have been limited resources to combine databases. With the anticipated future importance of genetic tests and their relation to a broad range of disease susceptibilities, a comprehensive set of linked health data will become increasingly important for future public health surveillance activities. Section 19a-2a, part 10 of the CGS permits the Commissioner of DPH to develop "specific uniform methods of keeping statistical information, including a client identifier system" (see **Appendix B**). The CHIP planned here is one such client identifier system, permitting the Commissioner to combine databases within the Department to maximize health information.

Creation of a comprehensive meta-database such as a CHIP would eliminate the need for unique identifiers in most instances in which data is shared, because the unique identifiers needed for local data linkages are eliminated. For those situations in which personal identifiers are required, however, data sharing with individuals within or outside DPH could be problematic. The Commissioner of the Department is limited in these cases by distinct statutes, and some of the databases maintained within DPH are restricted by additional regulations that protect confidentiality **(Appendix B)**.

Among the legislation that protects privacy, the Commissioner, in a statement of his powers and duties (CGS 19a-2a), is restricted by the Personal Data Act (CGS 1), in addition to CGS 17a-688 and RCSA 19a-2a-23 **(Figure 4)**. These legislative restrictions permit the release of medical information for scientific research and program development, but the information cannot include personal identifying information. Although many of the databases maintained within DPH contain no restrictions beyond those stated for the Commissioner (including Birth and Death Records, Newborn Hearing Screening, Newborn Screening, Lead Surveillance, Tumor Registry, and the WIC databases; **Figure 4**), the infectious disease databases are restricted by additional statutes and regulations. Reportable diseases are reported to the Department and are protected by RCSA 19a-36-A5 **(Figure 4; Appendix B)**. This regulation maintains confidentiality during disease investigation. The AIDS/HIV database is protected by CGS 19a-581 and 19a-592, which restrict disclosure of personal information without specific written permission. In addition, data within the CSHCN Registry are protected by CGS 10a-56b. These statutes and regulations need to be investigated before data within a CHIP can be shared.

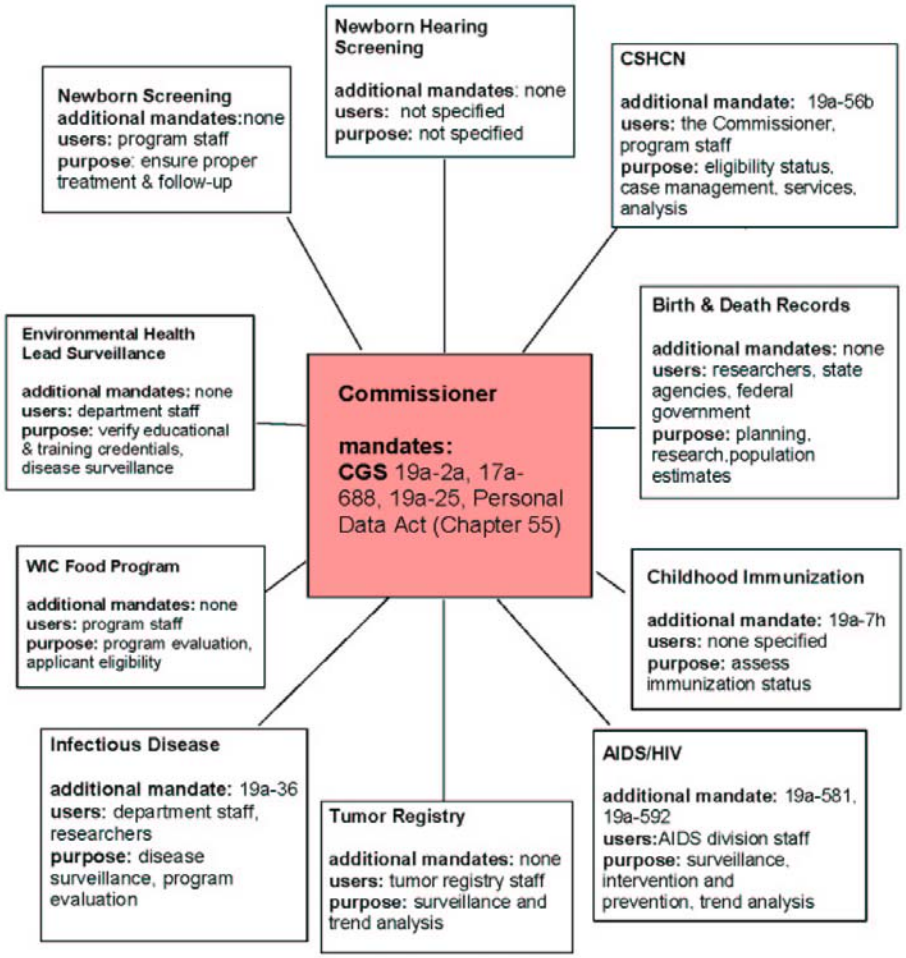
The Childhood Immunization Registry contains additional statutes and regulations that, rather than further restrict data sharing, stipulate the conditions under which personal data may be shared (CGS 19a-7h; RCSA 19a-7h-4). Information for medical and scientific research can be shared with other governmental or private research agencies, under the condition that the agencies agree not to disclose the information further (**Appendix B**). The Registry, through RCSA 19a-25-1 through 19a-25-4, is further permitted to share health information with healthcare providers and others designated by the Commissioner to prevent and control disease. These regulations allow the contents of the Registry, which includes vaccination dates and immunization status of Connecticut children, to be shared with healthcare workers.

Decisions by DPH to share *ad hoc* information are controlled by the Human Investigations

Committee (HIC) within the Department. The committee meets regularly to consider proposals for data sharing. Although it is expected that the role of HIC would apply equally to CHIP, its policies in data sharing would need to be evaluated in relation to the meta-database. A recently formed Data Sharing Committee is considering issues of data sharing within the Department, and its recommendations may impact data sharing of a CHIP. This possibility needs to be investigated.

In response to potential liability issues associated with electronic use of personal identifiers, the Health Insurance Portability and Accountability Act of 1996 (HIPAA) was adopted, and contained a set of HIPAA Privacy Rule that protects privacy of health information Office of Civil Rights, 2003. A report was recently generated to guide its impact on public health (Centers for Disease Control and Prevention, 2003b). The impact of this rule on data sharing within DPH, and with a CHIP, needs to be investigated.

Figure 4
Confidentiality Statutes Associated with Mandated Databases within DPH Containing Child Health Data



An important purpose of a CHIP containing comprehensive child health data is to coordinate activities among select individuals within the Department who monitor the health status of children in the state. Legislation that prohibits these activities should be investigated, and modified, if necessary, to allow DPH staff to perform public health assessment and assurance functions. In addition, legislation such as that provided for the Childhood Immunization Registry, could be proposed that allows data sharing outside DPH with qualified medical home environments to improve the health status of all children in Connecticut. An objective of this Plan is to examine and recommend removal of needless barriers to these public health functions, remaining sensitive to community issues about personal privacy (see **Objective B, CHIP Workplan**).

CHIP WORKPLAN

OBJECTIVE A. DEVELOP A CHIP OF CHILD HEALTH DATA (YEARS 01 – 04).

Recommended steps toward developing a CHIP are widely available, and include considerations such as data hardware and software, personnel, database infrastructure, and staff training (O'Neil, 1997). The sequential steps that could lead to the CHIP, and that could be completed in three years, are 1) create a Data Committee charged with the development of the meta-database, 2) plan the CHIP infrastructure and strategy, 3) develop a pilot meta-database, 4) enlarge the pilot meta-database into a full CHIP, and 5) train staff to maintain the CHIP **(Table 4)**. The sequence of action steps discussed below conforms to these steps. The time line and steps outlined below presuppose that a data warehouse is the best method of choice for development of a CHIP. If a fully integrated database is the preferred method of choice, however, the timeline would need to be broadened considerably.

Step 1. Obtain an executive charge, create a Data Committee, and identify necessary characteristics of a completed CHIP (Year 01, month 0-3).

A strong commitment at the executive level is believed to be one of the most important indicators to a successful project of this magnitude (Dodge, 2000). Development of the CHIP proposed here would require the cooperation of a wide range of groups within DPH, including multiple units within the Bureau of Community Health, the Bureau of Administrative and Support Services, and the Commissioner's office. An individual charged from the executive level should be designated as the lead on the project, and should have the ability to build consensus throughout the development phase of the project, should have considerable experience with health databases within DPH, and should be able to interact with Information Technology specialists as well as programmatic and epidemiological staff.

All units that maintain the individual databases targeted for inclusion into a CHIP should, in turn, charge representatives to create a Data Committee. This Committee should help guide development of the CHIP through active participation on the Committee, chaired by the lead person on the project. Individuals such as those in attendance at the Round Table discussion on data integration would be ideal representatives for the Committee. They would include staff working directly with the Laboratory Screening, CSHCN Registry, Birth Defects Registry, Newborn Hearing Screening database, Birth and Death Records, Childhood Immunization Registry, CEDSS database, Tumor Registry, WIC Food Program database, and Lead Surveillance database. In addition, individuals from within the Data Processing Division would be necessary. The Data Committee should be active throughout the four-year period needed to generate the fully functional CHIP, and should seek input from stakeholders outside DPH to ensure that the meta-database has interoperability (Public Health Informatics Institute, 2004). The first task for the Data Committee should be to identify specific programmatic needs for a CHIP, and to develop a comprehensive list of characteristics that describe the ultimate CHIP product.

Table 4

Time Line of Project Completion

Objective	Time							
	Year 01		Year 02		Year 03		Year 04	
	1	2	1	2	1	2	1	2
A. Develop a Child Health Informatics Profile (CHIP) of child health data								
1. Obtain executive charge, create a Data Committee, & identify necessary characteristics of a completed CHIP	✓							
2. Contract with an external development group to develop a technical strategic plan	✓							
3. Identify & secure sustainable funding to support the project.	✓	✓	✓	✓	✓	✓	✓	
4. Identify and contract with an external group to develop the CHIP,		✓	✓	✓	✓	✓	✓	✓
5. Develop a Pilot Meta-Database System.			✓	✓	✓			
6. Build a CHIP with additional DPH databases.				✓	✓	✓	✓	✓
7. Train data managers to use & maintain the CHIP.					✓	✓	✓	✓
8. Investigate & develop a web-based query system for use within DPH and to outside researchers.	✓	✓	✓	✓	✓	✓		
B. Investigate legislative issues in data sharing and remove barriers to sharing data contained in the CHIP, while maintaining personal privacy.								
1. Create and charge a legislative advisory group.	✓	✓						
2. Investigate data sharing within DPH and outside DPH.		✓	✓	✓	✓	✓		

Step 2. Contract with an external group to develop a technical strategic plan (Year 01, month 3-6).

After the Data Committee has identified the characteristics of a completed CHIP that fit the collective needs of the Department, the Committee should hire an Information Technology professional or group. This contracted group should have expertise in data warehouse technology and other forms of data integration, preferably of health-related data, and should have expertise with the ORACLE platform. This individual or group should be contracted to design a three-year technical strategic plan for development of a CHIP, recommending the best method for DPH. The group may recommend a data warehouse, but other recommendations are possible. It is also possible that the group may identify an existing architecture from other projects within DPH that could be used for the CHIP. The strategic plan should include a time line and resources needed to achieve the project, and include the considerations described below

a) Hardware and Software Needs, and Infrastructure of the CHIP. The CHIP, with its accompanying data, should be stored on servers in the Department's Data Processing Division. The disc space required to store the CHIP will need to be estimated. Space may be available on the server within DPH to store the data, and personnel house the server in a secure location with limited access. If space is not sufficient on one server, then other servers within DPH and/or DOIT may share the load, distributing the CHIP among several servers.

It is expected that the CHIP would be created and maintained in ORACLE software. The

software supports database management, and is capable of maintaining large, complex databases. The software also boasts the ability to create, manage, and maintain data warehouses, and the software company is considered the "world's leading supplier of information management software" (Sun Microsystems Computer Company, 1995) The Department holds a license for ORACLE 9i, and inclusion of the Internet Application Server should provide the capability needed to generate and maintain data warehouses (*ORACLE Sales Support, personal communication*). Advanced Security, Oracle Partitioning, and OLAP are additional options that could enhance a data warehouse.

In addition to its availability as a data warehouse builder and manager, ORACLE being chosen by the Department to maintain future databases. Although only one database within DPH is currently maintained in ORACLE, as many as six may be housed in ORACLE within the next year **(Table 2)**. The software package is also compatible with the storage platform of all the databases housed within DPH, and the custom data marts supplied to end users could be constructed in data formats familiar to those users. Also, as more databases shift their storage platform to ORACLE, it would become possible to roll a fully developed data warehouse into a real-time, integrated database. Finally, the Data Processing Division within DPH is familiar with the software, which should facilitate development of the CHIP in ORACLE.

For the above reasons, it is anticipated that the contracted development group would recommend creation of the CHIP in ORACLE. If, however, the group recommends use of some other software platform, then more than 40 other software applications are available for data warehousing and could be used to develop the CHIP. Among these software platforms are DB2, SAS, and Microsoft SQL Server (Greenfield, 2002e).

b) Data Discovery. An important technical task toward development of the CHIP would be to acquire the data dictionaries from each of the databases involved in the data linkage. A comparison of the data dictionaries should reveal those data elements that are shared among the databases **(Table 2)**. Some data elements within the individual databases may not be useful for the meta-database, and these may need to be eliminated as data are fed into the CHIP. Also, some data elements specific to the CHIP may need to be created, such as the date when data are fed into the meta-database, and the identity of the data source.

Since the objective of the CHIP is to create a set of linked health data for each child identified in the meta-database, information from any dated file could be fed into the meta-database. This includes archived data such as social security numbers. Additional data elements such as this may be identified for inclusion into the CHIP.

c) Data Quality. Data linkage requires the matching of unique personal identifiers. Although social security number is a preferred personal identifier, many of the child health databases mandated by the state do not contain this information **(Table 2)**. The first and last name of the child, coupled with the date of birth and the mother's full name, may be the next best choice for matching databases. Several problems are associated with this option. The actual spelling of a child's name can vary dramatically, so multiple children with very common names, and spellings, may be difficult to distinguish if coupled data are missing. Data entered shortly after birth may not contain a first name, and data entry errors compound matching processes that depend only on a child's full name. Sometimes, a child's last name, which reflects one parent's last name, is changed after birth to that of the other parent. In addition, many children are recorded in databases by his/her guardian, which, in less stable families, can

change as the child is shifted from one relative to another. The Data Committee and contracted development group should consider these issues as the CHIP is planned.

One of the major technical barriers envisioned by database managers within DPH is the matching of unique identifiers (**Appendix B**). Although plans are underway to link at least 95% of birth records from 1991 to 2001 with the child's social security number *(L. Mueller; personal communication*), these unique identifiers are not currently available in the eleven databases that should be used to create the CHIP. Therefore, data linkage should initially be performed with the first and last name of each child, matched with the child's date of birth and the mother's last name. A technique similar to this is being used to perform the *ad hoc* linkage of the CSHCN registry with birth records, using the mother's first and last name, child's date of birth, and baby's sex to link data. The resulting dataset apparently contains about 95% successful linkages (*B. Silverman, personal communication*). Starting with the database believed to contain the most accurate information, the data elements used to link its data with those of any other database should be chosen to maximize the amount of successful linkages. It is possible that matched data from one set of databases could be used to match another database to the growing meta-database. For instance, the newborn databases may not contain a child's first name, and so their linkage with birth records is an important first step to add the child's first name to the growing meta-database. Subsequent linkages with other databases could then include a child's first name.

Some of the databases proposed for linkage in the CHIP are "event-based" databases, such as the cases located in the Tumor Registry (*H. Swede; personal communication*). In these databases, a single person with multiple events would be entered into the database multiple times. The technical strategic plan must consider mechanisms for de-duplication.

d) Standardized Coding. The development group should consider how data elements within the CHIP would be coded. This should involve planning a data dictionary that contains the codes for sex, race, ethnicity, and even date of birth. These codes are not currently consistent among the databases in DPH. Codes for these and other variables within the CHIP, however, should be consistent with Health Level 7 (HL7, 2004), a universal coding scheme developed for health-related databases. Attention to coding in the planning phase would facilitate future data sharing efforts with databases outside DPH.

e) Database Security. Ensuring confidentiality is a major issue with data collected and maintained within DPH. A CHIP such as that proposed here would contain sensitive information that must be protected. As the CHIP is planned, the development group should consider security features that ensure confidentiality, and should consult with individuals within the Data Processing Division who specialize in this task (*J Cianci; personal communication*). Data should be maintained in a physically secure area within DPH, with limited access by personnel. In addition, the contracted development group should consider who would have access to the data, and what precautions should be taken to ensure that no breach in confidentiality occurs.

f) Database Maintenance and Disaster Recovery. A CHIP is an electronic file, and its integrity is subject to electrical spikes and other disasters. As it plans the CHIP, the contracted group should consider mechanisms for disaster recovery and should also establish protocols for database backup and maintenance. A large database could be shared by two or more computer servers using parallel server technology, and, if one server experiences a failure, the information could be automatically shifted to another server that remains operational (Dodge, 2000). This possibility should be considered as the CHIP is planned.

In addition, the contracted development group should consider how and when the data would be physically backed up. The CHIP could be backed up at the end of each day, a process called a cold backup (Dodge, 2000). If the server loses the CHIP data sometime during the day, however, any data entered during that day would be lost. A hot backup is one in which data are backed up each time the CHIP is fed data from a database. The group should consider these options, as well as other back up issues such as incremental backups and corrupt block detection, keeping in mind the resources and disc space required for each process.

Step 3. Identify and secure sustainable funding to support the project. (Years 01 – 03).

Funding is a major determinant of success for data linkage projects and the cost of a CHIP will also require sustained funding. Although considerably less costly than a fully integrated data system, a data warehouse would also be costly. If developed *de novo*, the total cost of a fully functional CHIP could be as much as one million dollars (*G. Land; personal communication*). If current database architectures such as those already develop with CEDDS, the Immunization Registry, or the Child Health Profile, were used for the CHIP, the cost could be considerably reduced. A sustained level of support throughout the project period would be required. The needed funds identified by strategic technical plan should be shared among programs across the Department who would benefit from the CHIP. External funds should also be sought. Resources may become available through the Public Health Informatics Institute, which recently received 3.2 million dollars to advance public health information infrastructure (Public Health Informatics Institute, 2004c). Additional funding from the Maternal and Child Health program of HRSA, and the immunization program through CDC, are possible.

The US Department of Health and Human Services recently convened a group to discuss strategies for building a national health information infrastructure, which developed a set of national recommendations that included "promoting the development of state and local population health information capacities" (US Department of Health and Human Services, 2001). The recommendations also called for "securing funds for state and local health departments to develop their health information capacities." In the near future, with the creation of a new National Coordinator of Health Information Technology, and a plan, released earlier this year (National Coordinator for Health Information Technology, 2004), funding through this Office may become possible.

Another organization called Turning Point (*http://www.turningpointprogram.org*) is funded jointly by the RW Johnson Foundation and the WK Kellogg Foundation, and it seeks to strengthen the public health infrastructure (Turning Point, 2003). Among the services boasts is the ability to help public health agencies "develop population data that supports decision-making about public health priorities" (Turning Point, 2004), and funds through this organization could be pursued. Finally, grant funds may be available by partnering with state academic institutions. For instance, a federal grant opportunity was recently released that will "support the development and demonstrate the feasibility of programs that have high potential for advancing population research" (National Institutes of Health, 2004). Currently working with the University of Connecticut Health Center, it is possible that a partnership with the Center may help finance development of the CHIP (*R. Aseltine; personal communication*).

Step 4. Identify and contract with an external group to develop the CHIP (Years 01 – 04).

Using the strategic plan developed in **Step 2** above, the Data Committee should solicit bids and recommend a development group that is best qualified to implement the technical strategic plan completely. The development group ultimately contracted to develop the CHIP should be responsible to this Committee, and in addition to developing the CHIP, should train existing personnel within DPH to manage and maintain the meta-database. The group should contain experts in data warehousing, preferably of health-related data, and should be experts in using the software platform ORACLE.

Step 5. Develop a Pilot Meta-Database (Years 02-03).

a) Software Programming. Once the previous steps have been completed, software programming to extract, transform, and load data could begin. A pilot meta-database should be developed within Year 02 of the project. The pilot meta-database should contain no more than three databases identified by the Data Committee as having the highest priority among the databases within DPH that contain child health information.

Software programming should be developed to feed each database into the pilot meta-database and to translate the data into ORACLE. Also, mechanisms for feeding data into the database should be developed so that the process, once completed, can be performed automatically at regular intervals. The pilot meta-database should be programmed to obtain extracts of the three databases at regular intervals. The process should not interfere with the original databases.

b) Custom Data Marts and Quality Assurance. The contents of the pilot meta-database should contain information of the highest possible quality, and quality assurance should be conducted by the data managers who maintain the databases. Throughout the creation of the pilot meta-database, data marts containing the information fed into the database should be channeled back to these data managers. They should compare the data marts to the original databases that fed the pilot database, and data errors in the data marts should be corrected. Any entries that cannot be matched should be forwarded back to the data managers, who should research the data for possible errors, and for missing and redundant data. The data managers should also research other issues such as novel spellings, and corrective steps should be taken. Through this series of data feedings and data extractions, the programming needed to generate a high quality CHIP could be developed.

As the pilot meta-database nears completion, the Data Committee should plan the data marts needed for regular DPH activities. Some end users may need the data contained within the CHIP for reports (see **Appendix A**). Others may want to perform analysis. The format of data marts created from the CHIP should suit the specific needs of each end user. Representative end users should be surveyed for their needs, and for the format needed to perform their tasks. Some end users may want to generate their own reports and analyses. Others, however, may appreciate ORACLE's ability to generate reports and perform analysis.

Step 6. Build a CHIP with additional DPH databases (Years 03 - 04).

With the completion of the pilot meta-database, additional databases should be added to complete the CHIP **(Table 4)**. The addition of the remaining 8 databases should involve 3 to 6 months each of effort. However, linkage of three databases to create the Child Health Profile is near completion. Also, Birth Records are linked to three of the databases, and Death Records are linked to two. These previous linkages would aid future linkage of Birth and Death Records to the growing CHIP. By the end of Year 04, a comprehensive CHIP should be completed. Data marts should continue to be created throughout Years 03-04 for individuals who work with the databases that are fed into the CHIP, and the data managers for each of these databases should help ensure the best data quality possible for the CHIP.

Step 7. Train DPH staff to use and maintain the CHIP (Years 03 – 04).

Throughout the development of the CHIP, data managers should have been involved in assuring the quality of the data fed into the meta-database and the quality of data marts created from the meta-database. After completion of the CHIP, these individuals should continue to be involved. Although individuals with the Data Processing Division of DPH should take responsibility for maintaining the CHIP, the data mangers should assume responsibility for the interaction of their databases with the meta-database. They should be involved in the regular feeding of their databases into the CHIP, and they should continue to monitor the quality of data generated from the database. They should alter, if needed, the feeding frequency of their data into the CHIP, and they should monitor the loading, translating, and cleansing of their data within the CHIP. They should continue to develop customized data marts that suit the needs of the end users within their unit.

Although most data managers are familiar with data warehousing and have some knowledge of ORACLE, it is anticipated that they may require training to assume responsibility for maintaining their own database as it interacts with the CHIP. The contracted development group, in addition to developing the CHIP, should have the responsibility to train data managers in the fundamentals of data warehousing and ORACLE programming. In addition, individuals within the Data Processing Division may benefit from training in ORACLE data warehouse programming, ORACLE systems administration and disaster recovery, and in development of customized data marts.

Step 8. Investigate and develop a web-based query system for use within DPH and to selected individuals outside DPH (Years 01 – 03).

The fully developed CHIP would contain a very large amount of data. Custom datamarts could be generated and sent to end users regularly for report and surveillance activities. Spontaneous queries could arise, however, particularly while investigating novel concepts. For these situations, a use-friendly query system would be ideal. One such system with a web front has been developed in Missouri (*G. Land; personal communication*). Called MICA, the system is made available to other states across the country, free of charge. Funded by CDC, the program pays for travel and accommodations to train individuals to run the software, and provides all software freely. At the end of the three-day training period, each state carries back

to their state a completed web-based query system. The MICA system is being investigated within DPH (*L. Mueller; personal communication*), and, if feasible, could be used by end users to obtain specific information contained in the CHIP.

OBJECTIVE B. INVESTIGATE LEGISLATIVE ISSUES IN DATA SHARING, AND REMOVE BARRIERS TO SHARING DATA CONTAINED IN THE CHIP, WHILE MAINTAINING PERSONAL PRIVACY (YEARS 01-03).

Databases maintained within the Agency are mandated by state or federal statutes, and these same statutes, and others, also limit how the data within the databases can be used. Although legislative barriers restrict data sharing among at least 50% of the databases, many of the databases are already shared, at least in part, inside and outside the Agency, indicating that some precedence exists for sharing data (**Appendix A**).

Step 1. Create and charge a legislative advisory group (Year 01, month 0-6).

The advisory group should be staffed with individuals from a wide range of disciplines. Experts in law and personal ethics, as well as healthcare providers and individuals serving on HIC within DPH should be included in the group. Representatives from Government Relations within DPH should also participate. This advisory group should meet regularly to discuss specific issues, and to report recommendations to any changes in legislation.

Step 2. Investigate data sharing within DPH and outside DPH (Year 01 – 03).

Access to data contained within the newly developed CHIP should not be problematic when personal identifiers are eliminated. Requests for data marts containing this sensitive information however, should be handled in a manner consistent with the legislative mandates that control data with unique identifiers. The Human Investigations Committee (HIC) within DPH currently deals with issues in sharing sensitive data. It is unclear, however, what issues may become evident with sharing sensitive CHIP data with units inside DPH, and with healthcare providers across the state.

The legislative advisory group should first examine the current legislation that permits and limits data sharing of the databases maintained by DPH. The group should then examine the practical implications of these legislative mandates in relation to data sharing within the Department, and to healthcare providers around the state. Issues may include the following:

a) How do the statutes controlling use of specific health databases relate to those statues that refer to the Commissioner? The Commissioner of DPH has primary responsibility for establishing the policies and guidelines for sharing information, and, through the Connecticut General Statues, has the power and duty to develop "specific uniform methods of keeping statistical information…including a *client identifier system*" (CGS 19a-2a, part 10). One such client identifier system may be the CHIP outlined in this Plan. This client identifier system, however, is subject to confidentiality restrictions stated in CGS 17a-688. In addition, data stored

in the DPH are controlled by the Personal Data Act (CGS 1).

Although some mandated databases contain no language that limits their use, others are restricted by specific privacy statutes and regulations **(Figure 3)**. It is not known if the databases without specific restrictions are limited by the same statutes and regulations referenced in CGS 19a-2a (CGS 17a-688), or if other statutes and regulations restrict them. The legislative advisory group may identify the priorities of CGS 19a-25, CGS 19a-581, and RCSA 19a-25-1 through 19a-25-4, 19a-36-14, and 19a-36-A5, relative to CGS 17a-688 and the Personal Data Act.

b) What is the role of HIC and HIPAA in sharing CHIP data and what protocols should be developed for data sharing within the Department*?*

c) What constitutes disclosure within DPH*?* Although several statutes restrict disclosure of personal identifiers **(Figure 3)**, the term is not clearly defined in the context of developing a CHIP. Each database in DPH contains personal identifiers, and when matched, broaden the amount of information on any given person, but does not identify otherwise unidentified individuals. Also, only a limited number of individuals involved in the creation of the CHIP should have access to *all* the health information, because each individual database within the Department would be fed directly into the CHIP. Further, data marts, or databases containing limited information, should be channeled back to each unit managing an individual database, and the amount of information contained within each data mart should be tightly controlled. The definition of disclosure as it relates to a CHIP needs to be addresse

d) How should indivdual statutes limiting data sharing be interpreted? The "silos" of newborn, child, and adult health databases maintained within DPH are mandated by a series of distinct statutes that stipulate who may use each database **(Figure 3)**. For instance, Newborn Screening stipulates that "program staff" may use the data within the database. The language of other statutes appears to limit data usage to department staff. This language is not interpreted consistently. The legislative advisory group may recommend consistent interpretation of the language within each of the statutes that mandate health databases.

e) How can needless restrictions to data sharing with healthcare providers be removed? While most of the Connecticut General Statutes mandating health databases limit use of data to program staff, RCSA 19a-25-3 allows information to be shared with healthcare providers. The Commissioner's responsibility to maintain confidentiality, identified in CGS 17a-688, also allows information sharing for research, audits, and program evaluation. These statutes allow DPH to share data containing patient identifiers under conditions in which the individuals obtaining the data will not share the information with other individuals. In addition, RCSA 19a-7h-4, which accompanies the mandate to maintain the Childhood Immunization Registry, permits data sharing with healthcare providers. The role of these mandates as they pertain to a CHIP should be clarified to permit future access of the CHIP to qualified healthcare providers, while preserving personal privacy and remaining sensitive to the cultural sensitivities of Connecticut's communities.

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Appendix A

Survey Summary Results

of

Health-Related Database Managers at

The Connecticut Department of Public Health

February, 2003

Database Manager Survey--Summary Responses

Of 14 database managers

1. Identifying Information.

2. What is your role as data manager?
64% Collect, validate, and maintain
50% Use
50% Other: database coordinator, software developer, report generator, administrator
7 people report their role in both collecting and using the database

3. What population(s) is (are) included in the database?
71% Newborns
79% Children/Adolescents
64% Adults (18+ years)
Seven databases contain information on both newborns and children, and six databases contain information on all ages. Only 3 databases contain information on only newborns.

4. How is data access controlled?
50% Private network
100% Password protection
21% Other: statutes, network access, limited access to server area

5. What state statutes regulate the collection and/or confidentiality of the data?
58% Statutes 19a-53, 19a-54, 19a56a, 19a7-1, 19a7-2, 19a-7h-1:19a-7h-5, 19a-581:19a-592, 19a-25, 19a-25-1:19a-25-4, 19a-110-a:19a-110-c, 19a-215, 31-40a, 19a-36, Fed Reg 7 CFR Ch 11 Part 246 Subpart G 246.26(d).

6. Do confidentiality restrictions pose a barrier to data integration/linkage/sharing? (Yes / No) If yes, please explain.
79% Yes: Explanations were: parental consent, B23 consent, hardware at provider site, client identification, surveillance staff, Fed Regulations, approval of division manager
21% No

7. Assuming there are no technical or legal barriers to data integration, do you foresee any other barriers that might
preclude such an effort? (e.g., resources, expertise, lack of commitment, etc.)
8 resources/staff/training
2 expertise
1 lack of commitment
2 unique identifiers
2 statutes
1 de-duplication, migration of children into state, planned new database, database not online

8. What unique identifiers, if any, are currently being collected? (e.g., Social Security Number)
10 first and last name
3 mother's name
6 accession or state id number
5 date of birth
2 social security number
2 medicaid number
3 address
Others reported: phone number

9. Is a data dictionary available? (Yes / No)
93% Yes
7% No

If yes, does it contain:
93% Data element names
79% Data types (e.g., numeric, alpha)
79% Lengths of data (e.g., Last Name allows up to 20 characters)
79% Data values (e.g., M = male for data element SEX)

10. Please identify the method of coding missing values (e.g. all missing values are coded as blanks or as 9999)
7 999
2 888
11 blank
1 Unknown

11. What software is currently used to collect data and maintain the database?
2 ACCESS
3 FOXPRO
2 Epi Info
Other software reported: VRV2000, Prodas, HARS, CTR, ORACLE, MVS Cobol, DB2, EBCDIC (ASCII)

Is there any plan to change software in the future, and, if so, what will it be?
57% Yes
43% No
Of those who responded "Yes," 6 will in ORACLE, 2 will be in dBase

12. What *technical* obstacles do you foresee to linking databases retrospectively?
50% No unique identifiers
36% Incompatible data (i.e. data element names, types, lengths, values)
43% Incomplete or dirty data
29% Other (specify): duplication, misspellings, different unique identifiers, legal issues, non-standardized data values

13. What *technical* obstacles do you foresee to integrating databases in real time?
43% No unique identifiers
36% Incompatible data (i.e. data element names, types, lengths, values)
43% Incomplete or dirty data
36% Data are not timely
43% Other (specify): duplication, misspellings, validation errors, different unique identifiers, legal issues, non-standardized data values

14. Are you familiar with the concept of data warehousing? (Yes / No)
93% Yes
7% No

15. Do you currently use ORACLE software? (Yes / No)
14% Yes
86% No

16. What is the primary use of the data? (More than one can be checked)
79% Screening

29% Surveillance
29% Registry
57% Policymaking
36% Programs and services
Other: reports (3), intervention (1), legal certification (1)

17. What software do you use to analyze the data?
7 SAS
3 Access
3 SPSS
3 FoxPro
3 Epi Info
2 Systat
2 ArchView GIS
2 Match Maker
Other responses: Stata, Prodas, Excel, DB2 SQL

18. Would integration/linkage with other databases enhance current usage? If yes, identify which database(s) from Appendix A would be most useful and explain how it (they) would enhance the current database. (For example, linkage between the laboratory newborn screening database and vital records birth certificates would help to assure that all Connecticut newborns are screened for mandated genetic disorders.)
3 Birth Records
3 none
2 immunization records
2 Newborn Screening
2 AIDS database
Others: Tumor Registry

19. Does the database currently allow access to individual identifiers for all users? (Yes / No)
64% Yes
14% No

20. **What programs/units, if any, *outside* your unit have either total or partial access to the database or have database extracts routinely made available to them?**
Birth to 3, Yale, DPMS, CSHCN, NBS, Immun Reg, DSS, NCHS, SSA, DCF, CLPPP, MD offices, CHC, Health Depts, Pediatric Centers, AIDS Epidem, Lead Environmental Management Unit, U. Conn Division of Environmental and Occupational Medicine. Tumor Registry regularly provides extracts to a list of participants.
5 responded "None."

21. Are you obligated to report on the subpopulation of Medicaid clients? (Yes / No)
7% Yes
86% No

22. Would it be useful to link to databases maintained by agencies other than DPH? (Yes / No)
50% Yes
36% No
2 did not respond

If yes, which ones?
43% Medicaid Eligibility File in Department of Social Services
29% Birth to Three in Department of Mental Retardation
29% Other: Department of Education, CHIME, DSS, DCF, Hospital deaths

Appendix B

Child Health Data Systems within DPH

Mandates and Confidentiality

A Summary of:

Public Acts

Connecticut General Statutes (CGS)

Regulations of Connecticut State Agencies (RCSA)

November, 2002

MANDATED HEALTH DATABASES WITHIN DPH, SUMMARY

Public Act 02-113. An act requiring the screening of newborns for metabolic diseases. This act sets a $28 minimum fee the Department of Public Health (DPH) must charge hospitals for its newborn screening program. By law, the DPH commissioner must establish a fee that covers all program expenses, including initial testing; tracking to assure that infants who initially test positive are referred for comprehensive testing and parent counseling; and treatment. DPH previously set the fee at $18. The act also requires DPH to buy two tandem mass spectrometers to screen newborns for metabolic disorders. The law requires screening for eight named conditions, including phenylketonuria, biotinidase deficiency, hypothyroidism, and "other inborn errors of metabolism." It also requires the DPH commissioner to adopt regulations specifying the conditions to be tested for. The act requires these regulations to include, by January 1, 2003, testing for amino and organic acid disorders and fatty oxidation disorders, including medium-chain acyl-CoA dehydrogenase (MCAD) deficiency and long chain 3-hydroxyacyl CoA dehydrogenase (LCHAD) deficiency. And it requires testing for other metabolic diseases. EFFECTIVE DATE: October 1, 2002 for the fee increase and testing changes; July 1, 2002 for the mass tandem spectrometer purchase. LCHAD and MCAD are genetic deficiencies that result in the body's inability to break down fatty acids as a usable energy source. LCHAD can result in dangerously low blood sugar levels, poor muscle tone, and heart problems. It can also cause medical complications in the pregnant mother, including liver failure. Children with MCAD can develop seizures, respiratory failure, and heart problems. Treatment for both is based on diet.

RCSA 19a-2a-3. Children with special healthcare needs system. The system provides program personnel with data about CSHCN who receive service. The official responsible is the director of child and adolescent health Personal data are obtained from the referring person or agency, the family of the child, and providers of service to the child. Legal authority is provided by sections **19a-48** through **19a-55**, **19a-59**, and **19a-61**, and includes children with cerebral palsy, hearing deficiencies, diabetes, and those who tested positive for newborn blood tests. The specific data collected include: name of child; names of parents or guardians and siblings; address of child; birth date of child; race of child; sex of child; family income; employers of parents or guardians; family medical insurance information; medical information regarding the child; and information regarding services provided for the child, including amount paid on fee-for-service care. Data are used by program staff to determine eligibility, provide case management services, and prepare statistical reports.

RCSA 19a-2a-6. Environmental health data system. This system is used to document reduced morbidity and mortality, and improve living conditions for state residents as a result of: educational programs; regulatory programs; and passive programs that address hazards through alteration of the environment. The collection, maintenance and use of personal data in the environmental health data system is authorized by sections: **19a-110** (collects additional information on lead exposure: birth date, race, ethnicity, information about tests and test results, and healthcare provider) through **19a-111d** (collects additional information on lead exposure that includes birth date, race, ethnicity, information about test and test result, and the name of the healthcare provider); **19a-421**; **19a-426**; and **20-435** through **20-439**. The data collected include: educational; employment history; inspectional; training and work experience; and medical. Records are obtained from: applicants for professional registration, certification or licensure; and persons with high blood lead levels. The data are used by department staff to verify educational credentials, verify training or work credentials, and to perform disease and environmental surveillance.

RCSA 19a-2a-8. Vital records data system. The purpose of the vital records data system is to provide support for health status surveillance, health program development, and individuals seeking certified copies of their birth certificates or other personal data records as provided by the Connecticut General

Statutes. The responsible official is the registrar of vital records. Data are obtained from hospitals, funeral directors, and town clerks. Legal authority is provided by Sections: **19a-41; 19a-41-2; 19a-41-4; 19a-44**; **19a-45**; **7-41a**; **7-47**; **7-47b** (which asks institutions to maintain addition information, such as date of death and details of body release); **7-48**; **7-60**; **46b-68**; and Section 19-6a-2 (this section of the Regulations does not apparently exist). Data collected include: name or hospital medical record number; social security number, name of mother, address; race; sex; ethnicity; marital status; religion; and social and medical risk factors. Data are collected from: newborns; manned persons; deceased persons; and adopted persons. Data are used by: genealogical researchers; state agencies; the federal government; and researchers. Data are used for: community-based planning; statistical research regarding health status; and population estimates made by the U.S. census bureau and department.

RCSA 19a-2a-10. Connecticut tumor registry data. The purpose is to provide cancer incidence and survival data for Connecticut; data for cancer control program evaluation; data for epidemiological studies of cancer in Connecticut; and data for the National Cancer Institute. The director of the tumor is the official responsible for the tumor registry data system. Data in the tumor registry data system is routinely obtained from: hospitals, death certificates, private pathology laboratories, and reports from other state central cancer registries. Legal authority is provided by sections **19a-72, 19a-74**, 19-6a-2 (may not exist), and **19a-73-1 through 19a-73-7**. Data obtained by the system include: name; social security number, date of birth; address; race; ethnicity; sex; place of birth; social and medical risk factors; and health outcomes. The data system is used by: the department's Occupational Health Division; the department's Environmental Epidemiology Division; authorized researchers; and the National Cancer Institute. The data are used for: community based health planning; program development; statistical research; and program compliance evaluation.

RCSA 19a-2a-12. Infectious disease epidemiology data system. The purpose of the data are to monitor the incidence and trends in diseases, and evaluate health education and healthcare programs. Data are obtained from reportable disease reports from healthcare providers and healthcare facilities including medical laboratories; reports from the Department of Correction, reports from schools; reports from local directors of health; and data from department health counselors and educators. Collection, maintenance and use of personal data in the infectious disease epidemiology data system data are authorized by Sections: **19a-215; 19a-262**, and **19a-36-A1** through **19a-36-A6**; and **19a-36-A11**. Data input includes: name; address; age; race; sex; occupation; and behaviors that put the individual at risk for infectious diseases. Data are obtained from persons with specific reportable diseases. Data are used by: the department and authorized researchers for disease surveillance and evaluation of health education and intervention programs.

RCSA 19a-2a-13. Bureau of laboratory services data system. Data are collected document and maintain laboratory analysis reports. Personal data are routinely obtained from: physicians; private and public laboratories; directors of health; sanitarians; various state agencies; the United States justice department; state and local police; and other department bureaus and centers. Authority is provided by Sections: **19a-25** through **19a-30**; **21a-274**; and **21a-283**. Data are provided by: patients; physicians; directors of health; other in-state laboratories; alleged criminal perpetrators; and principal parties in environmental and consumer protection actions. Data are maintained on: newborns, patients of medical practitioners, and alleged criminal perpetrators. Data are used by: physicians; lawyers; officials of state agencies; judiciary department staff; laboratory supervisors; and federal agencies. Data are used for: diagnosis of disease; appraisal of environmental conditions; and testimony to support laboratory findings in court.

RCSA 19a-2a-15. Newborn screening system. The purpose of the newborn screening system is to track infants found to have a serious problem as a result of a blood test done right after birth. The director, maternal infant health division is the official responsible for the newborn screening system. Data in the newborn screening system is received from any laboratory carrying out a newborn screening test. Legal

authority is provided by section **19a-55**. Data collected include: name of infant; name and age of mother; sex of infant; birth date of infant; address of mother and father; telephone number of parents; place of birth of infant; and medical information on the infant, as well as information on services received by infant. Data are collected on: newborn infants, and parents of newborn infants. Data are used by program staff to ensure that the infant received proper treatment and follow-up.

RCSA 19a-2a-18. Supplemental food program for WIC. The purpose of the WIC [Women, Infants, and Children] system is to provide the WIC program with data regarding participants. Participant information is maintained for documentation of certification of eligibility as well as to enable the issuance of WIC checks to eligible participants. Nutrition surveillance information is maintained to track the health status of certain individuals, and maintain documentation on food stores and pharmacies who apply to become authorized program vendors as well as currently authorized vendors. The state WIC director is the official responsible for the WIC system. Authority is provided by section **19a-59c** of the Connecticut General Statutes. Data collected include: names; sex; address; race; telephone numbers; medical information; date of birth; and family income, as well as names of parents and guardians. Data are obtained from: program participants; and vendors who apply to be authorized program vendors. Data are used by program staff for program accountability, program evaluation, and eligibility determination of applicants and vendors.

RCSA 19a-2a-22. AIDS/HIV data system. Data are collected for disease surveillance. Data are received from physicians, institutions, laboratories, infection control practitioners, AIDS coordinators in various healthcare facilities or private practice, and from death certificates. Authority for the AIDS/HIV data system is provided by Sections: **19a-2a**; and **19a-581** through **19a-592**. Data collected include: name; address; date of birth; sex; various diseases experienced by these individuals; risk categories; laboratory tests; and date of death, if applicable. Data are collected from: adults with CDC-defined AIDS; and all children who are HIV exposed or infected. Data are used by staff epidemiologists employed by the AIDS division to monitor the occurrence and progression of HIV/AIDS disease in Connecticut; to target populations for intervention; to evaluate the effect of HIV/AIDS prevention initiatives; and to project the number of cases that will occur in the future and plan for healthcare resources.

CGS 19a-7f. Childhood immunization schedule. The standard of care for immunization for the children of this state shall be the recommended schedule for active immunization for normal infants and children published by the committee on infectious diseases of the American Academy of Pediatrics-or the schedule published by the National Immunization Practices Advisory Committee, as determined by the Commissioner of Public Health. The commissioner shall establish, within available appropriations, an immunization program which shall: (1) Provide vaccine at no cost to healthcare providers in Connecticut to administer to children; (2) provide all parents in this state with the recommended immunization schedule for normal infants and children; (3) inform all healthcare providers of changes in the recommended immunization schedule; *(4) assist hospitals, local health providers and local health departments to develop and implement record-keeping and outreach programs*; (5) assist in the development of a program to assess the vaccination status of children who are clients of state and federal programs; (6) access available state and federal funds; (7) solicit, receive and expend funds from any public or private source; and (8) develop and make available public health educational materials.

CGS 19a-7h. Childhood immunization registry. Regulations. The Commissioner of Public Health or his designee may, within the limitations of available resources, establish and maintain for the purpose of assuring timely childhood immunization an ongoing registry of all children who have not begun the first grade of school including all newborns. The registry shall include such information as is necessary to accurately identify a child and to assess current immunization status. Except as specified, all personal information including vaccination status and dates of vaccination of individuals shall be confidential pursuant to section **19a-25** and shall not be further disclosed without the authorization of the child or the child's legal guardian

CGS 19a-50. (Formerly See. 19-20). Children crippled or with cardiac defects. Payment of "clean claims". The Department of Public Health is designated as the state agency to administer a program of services for children who are crippled or suffering from conditions which lead to crippling or suffering from cardiac defect or damage and to receive and administer federal funds which may become available for such services.

CGS 19a-53. (Formerly Sec. 19-21). Reports of physical defects of children. Each person licensed to practice medicine, surgery, midwifery, chiropractic, naturopathy, podiatry or nursing or to use any other means or agencies to treat, prescribe for, heal or otherwise alleviate deformity, ailment, disease or any other form of human ills, who has professional knowledge that any child under five years of age has any physical defect shall, within forty-eight hours from the time of acquiring such knowledge, mail to the Department of Public Health a report, stating the name and address of the child, the name and address of the child's parents or guardians, the nature of the physical defect and such other information as may reasonably be required by the department.

CGS 19a-54. (Formerly See. 19-21a). Registration of physically handicapped children. Each institution supported in whole or in part by the state shall report to the Department of Public Health, on a form prescribed by said department, the name and address of each child under twenty-one years of age who is physically handicapped for whom application is made for admission, whether such child is admitted or rejected.

CGS 19a-55. (Formerly Sec. 19a-21b). Newborn infant health screening. Tests required. Exemptions. The administrative officer or other person in charge of each institution caring for newborn infants shall cause to have administered to every such infant in its care an HIV-related test, as defined in section **19a-581**, a test for phenylketonuria, hypothyroidism, galactosemia, sickle cell disease, maple syrup urine disease, homocystinuria, biotinidase deficiency, congenital adrenal hyperplasia and such other tests for inborn errors of metabolism as shall be prescribed by the Department of Public Health.

CGS 19a-56a. (Formerly Sec. 10a-132b). Birth defects surveillance program. Collection of birth defects data. Advisory committee. There is established a birth defects surveillance program to monitor the frequency, distribution and type of birth defects occurring in Connecticut on an annual basis. The Commissioner of Public Health shall establish a system for the collection of information concerning birth defects and other adverse reproductive outcomes. In establishing the system, the commissioner may have access to identifying information in hospital discharge records. Such identifying information shall be used solely for purposes of the program. Management of personal data shall be in accordance with **Chapter 55**. The commissioner shall use the information collected pursuant to this section and information available from other sources to conduct routine analyses to determine associations that may be related to preventable causes of birth defects.

RCSA 19a-59-1. Newborn hearing screening program. Each institution that provides childbirth services shall develop and implement a universal newborn hearing screening program which shall at a minimum include a mechanism for monitoring the institution's compliance with the newborn hearing screening program which shall include, but not necessarily be limited to, the following information: name of each newborn infant; date of birth; date infant received hearing screening or documentation of parent refusal for newborn hearing screening; method of screening; results of screening; person performing screening; and to whom referral for further evaluation was made, if applicable.

CONFIDENTIALITY STATUTES AND REGULATIONS, SUMMARY

Directives to All State Agencies

CGS 4-190 through CGS 4-197. Personal Data Act (Chapter 55). These statutes describe the procedures all state agencies must follow when personal data are gathered.

Directives Specific to the Commissioner of DPH

CGS 19a-2a. Powers and duties. Describes the duties of the Commissioner, and among those duties, ."the commissioner shall have the power and duty to: … 10) specify uniform methods of keeping statistical information by public and private agencies, organizations and individuals including a client identifier system, and collect and make available relevant statistical information, including the number of persons treated, frequency of admission and readmission, and frequency and duration of treatment. The client identifier system shall be subject to the confidentiality requirements set forth in section **17a-688** and regulations adopted hereunder." Section **17a-688** refers to the conditions under which data may be released: "the commissioner may use or make available to authorized persons information from patients' records for purposes of conducting scientific research, management audits, financial audits or program evaluation, provided such information shall not be utilized in a manner that discloses a patient's name or other identifying information."

CGS 17a-688. Records, keeping and confidentiality of. Disclosure permitted, when. Describes conditions under which data may be disclosed. The Commissioner can make data available for scientific research, financial audits, and program evaluation if the purpose is not to disclose a patient's name.

Directives Specific to Health Databases within DPH

RCSA 19a-2a-23. Maintenance of personal data. This section discusses issues of personal data maintenance as it relates to the Personal Data Act, and dictates under what conditions personal data may be maintained. Sections of the General Statutes that related to personal data include: the **Personal Data Act**, **Chapter 55**; section **4-196**; the Freedom of Information Act, Sections **1-15** and **1-18** to **1-21l** inclusive; and any other state or federal statutes or regulations concerning maintenance or disclosure of personal data kept by the department. All department employees who have written access to personal data must take reasonable precautions to protect personal data. Data should not be duplicated unnecessarily, and personal data sent through interdepartmental mail should be sealed in envelopes or boxes and marked "confidential." Manual personal data systems should be locked and kept in controlled access areas. Automated personal data systems should: be located in a limited access area; require visitors to sign a visitor's log; limit regular access to operations personnel; and prevent disclosure of personal data to unauthorized individuals.

RCSA 19a-7h-4. Release of information by the immunization registry. Describes the conditions under which information from the immunization registry may be disclosed to healthcare providers.

CGS 19a-25. (Formerly See. 19-6a). Confidentiality of records procured by the Department of Public Health or directors of health of towns, cities or boroughs Notwithstanding the provisions of **Chapter 55,** the Department of Public Health may exchange personal data for medical or scientific research with any other governmental agency or private research organization. The state, governmental agency or private research organization shall not further disclose such personal data. The Commissioner of Public Health shall adopt regulations and procedures to ensure the confidentiality of such disclosures.

RCSA 19a-25-1. Disclosure of Health Data. Definitions. This section includes a list of definitions, as used in RCSA **19a-25-1** through **19a-25-4**, inclusive.

RCSA 19a-25-2. Disclosure of aggregate health data, anonymous medical case histories, and reports of the findings of studies of morbidity and mortality. The department may publish, make available, and disseminate aggregate health data, anonymous medical case histories, and reports of the findings of studies of morbidity and mortality, provided such data, histories, and reports: are prepared for the purpose of medical and scientific research; and do not include identifiable health data.

RCSA 19a-25-3. Disclosure of identifiable health data. The department may not disclose identifiable health data unless: the disclosure is to healthcare providers in a medical emergency as necessary to protect the health, life, or well-being of the person with a reportable disease or condition (**19a-215**); the disclosure is to healthcare providers, the local director of health, the department, another state or public health agency, including those in other states and the federal government, or other persons when deemed necessary by the department in its sole discretion for disease prevention and control (**19a-215**) or to reduce morbidity and mortality, and every effort shall be made to limit the disclosure of identifiable health data to the minimal amount necessary to accomplish the public health purpose; the disclosure is to an individual, organization, governmental entity in this or another state or to the federal government if necessary for medical and scientific research.

RCSA 19a-25-4. Use of health data for enforcement purposes. Notwithstanding any provisions of sections **19a-25-1** to **19a-25-3**, inclusive, the department may use aggregate health data, identifiable health data, and studies of morbidity and mortality to perform its statutory and regulatory responsibilities and to secure compliance with or enforcement of any laws. Disclosure of personal data may occur only if required by law, and only to a tribunal, administrative agency or court with jurisdiction over the enforcement action. Disclosure under this section does not constitute a waiver or release of the confidentiality that protects such data.

CGS 19a-36 A5. Confidentiality of data. All epidemiologic information which identifies an individual and which is gathered by the state or local health department in connection with the investigation of reported cases or suspected cases of disease or during the investigation of outbreaks of disease shall be kept in compliance with current confidentiality statutes.

CGS 19a-56b. (Formerly Sec. 10a-132d). Confidentiality of birth defects information. All information collected and analyzed pursuant to section **19a-56a** shall be confidential. The commissioner shall prepare detailed policies and procedures for maintaining confidentiality of program information. The commissioner shall maintain an accurate record of all persons who are given access to the information in the system. All research proposed to be conducted using identifying information in the system established pursuant to section **19a-56a** or requiring contact with affected individuals shall be reviewed and approved in advance by the commissioner.