
Integrating Genetic Services into Public Health – Guidance for State and Territorial Programs from the National Newborn Screening and Genetics Resource Center (NNSGRC)

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Key Words

Public health · Genetics · Guidance · Policy ·
Assessment · Assurance

Introduction

During the past decade, there have been many advances in genetics, most resulting from mapping the human genome through the Human Genome Project and other research initiatives. Research in the next decade will bring an understanding of genetic/biologic risk and protective factors, and a description of the influence of the environment on genetic variation. Tests will continue to be developed to identify both individuals with genetic disorders and asymptomatic individuals with a genetic predisposition to (a) particular disorder(s).

¹ On behalf of the Genetics Advisory Committee of the National Newborn Screening and Genetics Resource Center (see Appendix B for a complete listing of committee members contributing to this paper).

This expanded knowledge base in genetic medicine is having, and 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. Although the ability to incorporate genetic medicine into public health and health care practice depends ultimately on the capacity to achieve equal access to health care and social services, optimal health also depends on the ability to utilize the latest scientific knowledge in health care practice – in this case, genetic medicine or knowledge about disease causation, illness management, and health promotion.

Unraveling the interaction between genes, environment, and behavior will require partnerships between the public health and health care communities. Genetic medicine may offer new tools for both individual and community assessment that are useful for integrating primary care and public health activities. For example, by identifying asymptomatic persons at increased risk for acquiring certain disease(s) (clinical assessment), health care

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1422–2795/01/0043–0175\$17.50/0

Accessible online at:
www.karger.com/journals/cmj

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professionals and public health programs may be able to target intervention resources (assurance) more effectively. Such collaborations should also lead to evaluation of the clinical utility and validity of genetic tests from information within the health care system, thus contributing to the overall efficiency and effectiveness of health services. However, in addition to utilizing the public health core functions of assessment, policy development and assurance [1], public health and health care communities will also need to address the economic, legal, political, social, moral, and ethical issues surrounding the use of genetic information and technology.

This guidance document was developed to address requests from state public health professionals and health care practitioners for guidance in planning successful integration of genetic medicine into public health and health care services. It was developed as a cooperative effort among experts serving on an advisory committee to the National Newborn Screening and Genetic Resource Center. Committee members represent a significant number of organizations and groups with interests in the successful implementation of state genetics plans (see Appendix B). The guidance incorporates ideas previously suggested in the Council of Regional Networks for Genetic Services' (CORN) document *Guidelines for Clinical Genetics Services for the Public's Health* [2]. It also reflects consideration of numerous public health initiatives including books such as *Genetics and Public Health in the 21st Century* [3], and other reports such as *Translating Advances in Human Genetics into Public Health Action* [4], and *Integrating Genetics into State Chronic Disease Programs* [5]. Although targeted toward states and territories in the US, information in this guidance may have applications locally and internationally. (Note: The term *state* will be used generically throughout this document to refer to both states and territories.)

Background

Historically, federal, state, and local maternal and child health programs in the United States have provided leadership essential to the development of genetics for the public's health [6, 7]. A 1998 survey by the Council of State and Territorial Epidemiologists indicated that state health department program managers are becoming increasingly aware of new information in genetic medicine, its potential for health promotion and disease prevention, and the potential for its public health impact [8]. Based on the results of this survey, the Council recommended cer-

tain measures to allow state departments of health to plan for reasonable and effective use of new genetic information, genetic tests, and health promotion and disease prevention measures targeting genetic disorders. As a point of reference for planning, these recommendations are listed here:

- Develop a comprehensive strategic plan for genetics.
- Increase funding for new areas in genetics.
- Develop methods to share existing and future resources in genetics with all program areas.
- Conduct early needs assessments; plan for policy and program development as new genetic information regarding specific disease areas and prevention measures becomes available.
- Consider the need for state regulation of genetic testing in private laboratories; determine appropriate policy to prevent the undesired use of genetic tests results; and analyze new genetic tests for applicability to public health programs.
- Improve the genetics knowledge base of health departments.
- Develop methods to share information among state health departments.

The American Public Health Association's list of essential public health services [9] should also be considered in the planning process:

- Monitor health status to identify (individual, family and) community health problems.
- Diagnose and investigate health problems and health hazards in (among individuals, families and) the community.
- Inform, educate, and empower people about health issues.
- Mobilize community partnerships to identify and solve health problems.
- Develop policies and plans that support individual (family) and community health efforts.
- Enforce laws and regulations that protect health and ensure safety.
- Link people to needed personal health services and assure the provision of health care when otherwise unavailable.
- Assure a competent public health and personal health care workforce.
- Evaluate effectiveness, accessibility, and quality of personal and population-based health services.
- Research for new insights and innovative solutions to health problems.

While maternal and child health programs continue to be recognized for their leadership role in developing genetics in public health, it is imperative that all public health programs be strengthened as genetics is integrated into them. These efforts will require a commitment to sharing old and new resources to address identified gaps, and should encompass integration of population-based screening, genetic risk assessment, health promotion and disease prevention strategies within chronic disease, cancer, environmental health and other programs. To foster the integration process, many states have either developed, or are in the process of developing, a state genetics plan (copies or links to genetics plans are available through the NNSGRC or its website: <http://genes-r-us.uthscsa.edu>). The intent of a state genetics plan is to augment the translation of genetic medicine and technology into public health and health care practice. When engaged in the planning processes, each state must take into account its own population factors, fiscal milieu, provider characteristics, and legislative and regulatory environments. However, there are common themes that should be addressed in all state plans. Additionally, to meet the needs of the population, states may need to consider coordinating genetics activities with neighboring states. States should use planning resources to:

- Establish programs to eliminate barriers to information about genetics and genetic services.
- Optimize health status and health outcomes through effective use of genetic medicine.
- Assure access to quality genetic health services and information.
- Improve public health, health care, and social services systems within the state through the use of genetic medicine.

Coordination of genetics activities across local and state agencies and health department divisions, as well as within programs, should ensure accountability and visibility of genetics throughout local and state public health systems.

The following broad outline addresses common themes and provides a suggested process for developing a state genetics plan. Steps that could be followed in the planning process are provided in table 1. Since genetics programs and departments of public health are unique to each state, the process for the development of local and state genetics plans will vary. The process in table 1 is offered to provide policy makers with elements to consider in tailoring inclusive plans, that is, a process which will assure participation of key stakeholders [10].

Table 1. Suggested steps for developing a local/state/territorial genetics plan

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- 1 Identify leadership within public health, social services, and genetics communities to take responsibility for the process and the product
 - 2 Identify internal stakeholders whose participation is essential to the development of a plan that will achieve integration of genetics within existing programs (regulatory agencies, health care financing agencies, children with special health care needs, birth defects surveillance, newborn screening, chronic disease, cancer, immunizations, vital statistics, etc.)
 - 3 Identify external stakeholders whose participation is essential for the successful implementation of the plan (insurance industry, biotech industry, consumers, clergy, legislators, genetic services and other health care practitioners, the academic medical community, ethics and legal communities, etc.)
 - 4 Ensure involvement of families, consumers, and the public in the planning process
 - 5 Form a broadly based Advisory Committee representative of the above stakeholders
 - 6 Review available genetic assessment data as they pertain to the four levels of the MCHB pyramid and core public health functions
 - 7 Assess the necessity of and support for additional needs assessment(s)
 - 8 Complete and review the additional needs assessment(s)
 - 9 Set initial priorities for the community, state or territory, taking into account the MCHB/Genetics pyramid and core public health functions
 - 10 Review the initial priorities with stakeholder groups as represented on the Advisory Committee
 - 11 Draft the plan, taking into account the comments of stakeholder groups and incorporating mechanisms to evaluate the plan
 - 12 Review the draft plan with the Advisory Committee and finalize the document
 - 13 Submit the plan for public comment and address recommendations, as appropriate
 - 14 Implement the genetics plan
 - 15 Develop a schedule to evaluate and revise the plan at defined intervals
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Illustrating the Maternal and Child Health Bureau Pyramid and Institute of Medicine Public Health Model

The US Maternal and Child Health Bureau (MCHB), Health Resources and Services Administration (HRSA), Department of Health and Human Services (DHHS) has developed a four-tier pyramid model to outline the essential components of a public health system [11]. In this guidance, the pyramid model is used as a framework to illustrate the relationships of essential components of a

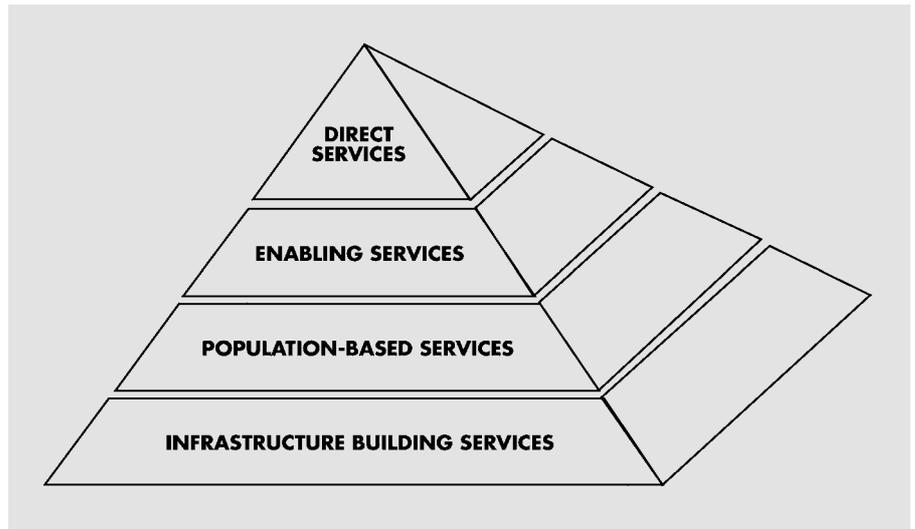


Fig. 1. MCH pyramid showing essential public health system components.

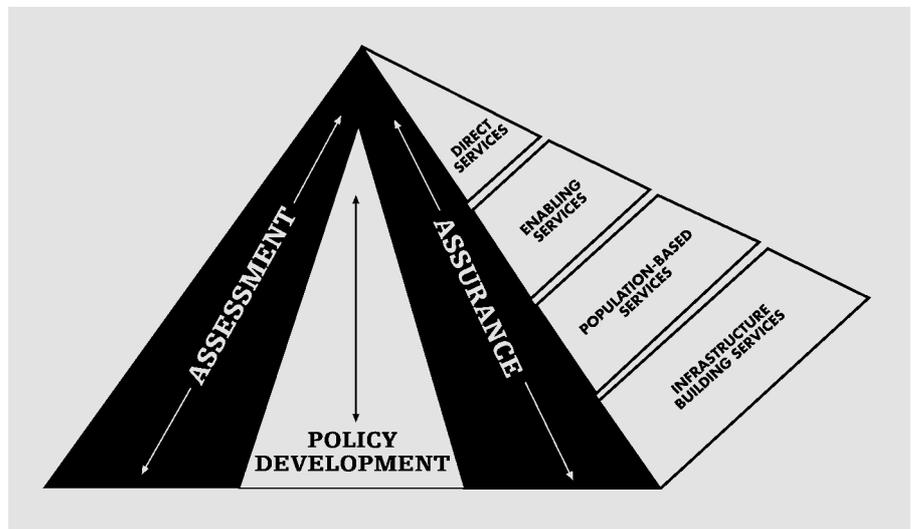


Fig. 2. IOM core public health functions integrate into all levels of the MCH pyramid.

state genetics plan: infrastructure building services, population-based services, enabling services, and direct health services (fig. 1).

The elements included in each tier of the pyramid incorporate the Institute of Medicine’s (IOM) public health core functions (assessment, policy development, and assurance) [1]. Together, these three public health core functions form a process that permeates the pyramid and can be applied to each layer (fig. 2).

Defining the Relevant Public Health Core Functions

The public health core functions can be used in a logical thought process for developing public health programs in general, and the relevant definitions and examples for genetics programs are given below.

- Assessment includes monitoring, analyzing, and evaluating data regarding: (1) the health, well-being, socio-cultural and educational status of individuals (e.g. case studies) and populations; (2) community concerns; (3) resources available and/or needs; (4) access, avail-

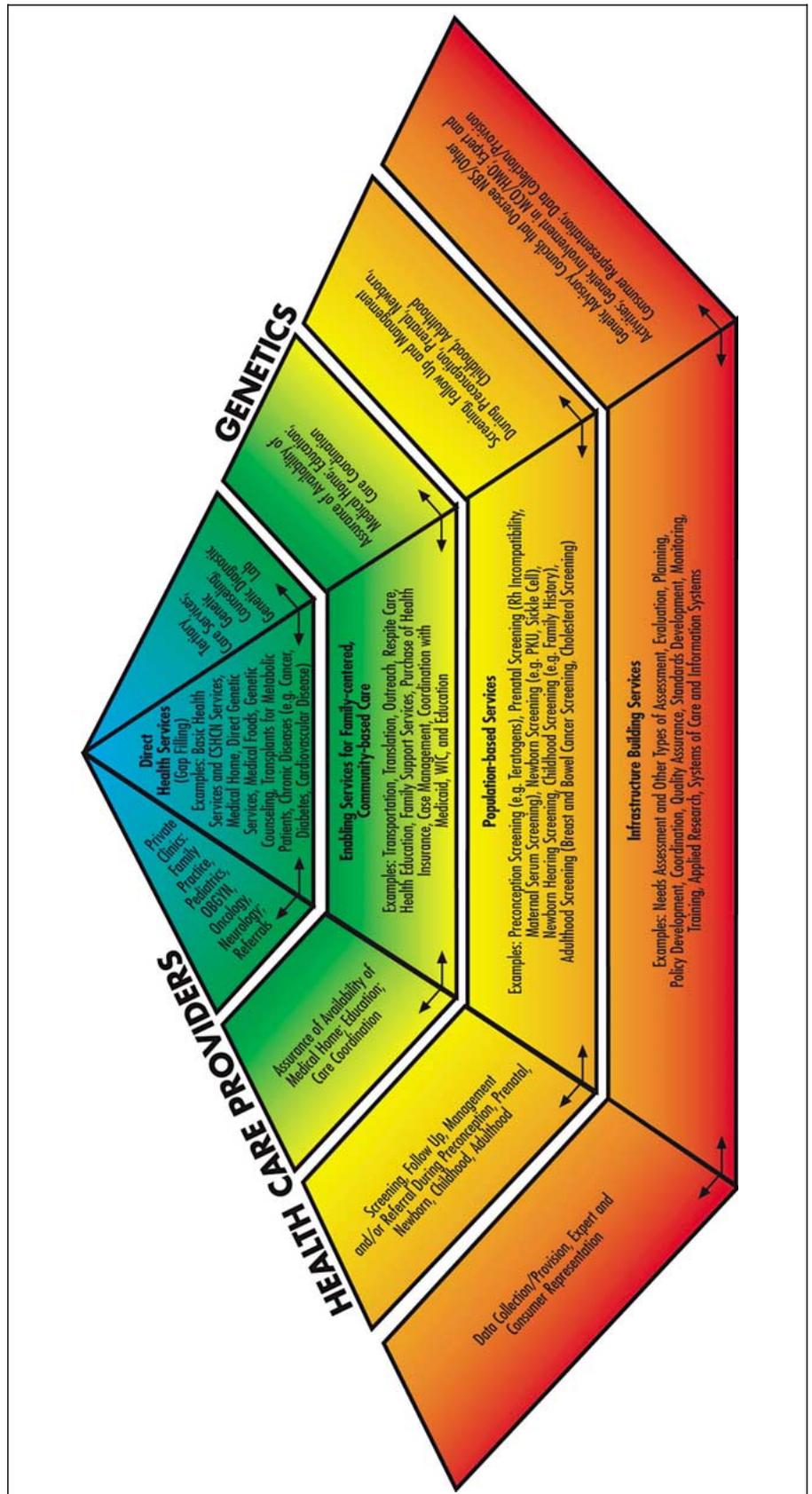


Fig. 3. The genetics pyramid.

ability, utilization, affordability, and satisfaction with services; (5) genetic, environmental and social risk and protective factors; and (6) systems of care and information systems. Assessment also includes gathering statistical data and conducting epidemiological and/or other investigations (e.g. surveillance and monitoring) as part of the regular, ongoing collection, analysis, and sharing of information.

- Policy Development includes the consideration of alternatives for the best possible use of shared resources, creation of short- and long-term plans with goals, objectives, performance measures, indicators and/or benchmarks aimed at health promotion and protection, and disease prevention. The policy development process should include diverse individuals and organizations for decision-making about the relative importance of health, social, educational, and other issues. Policy development also includes creating appropriate definitions and establishing necessary guidelines (including codes, regulations, standards – including standards of care, laws, etc.).
- Assurance includes informing populations about relevant health and social services, cultural, educational, and other issues, and ensuring that patients, families, and communities have access to appropriate, cost-effective, quality and timely services and information that enhance family and community relationships. Assurance includes patient, family, provider and public education, licensing and certification, and the implementation and maintenance of guidelines, codes, regulations, standards, and laws, including service utilization review, treatment outcomes research, and performance-based program evaluations, to achieve continuous quality improvement. Assurance may also include the actual provision of services.

Developing the Genetics Pyramid

If all aspects of genetic medicine are incorporated into the MCHB pyramid, then the model expands to include the elements depicted in figure 3, with public health at the center, flanked by corresponding activities of genetics and health care providers. In considering genetic medicine (broadly), it is obvious that the genetic components are inseparably linked to each of the pyramid's levels. It is also important to note that any activity may occur at more than one level of the pyramid. For example, when viewed as a system, components of a newborn screening program can be found at each level of the pyramid. Thus, using the

pyramid as a planning tool challenges planners to consider all aspects of a program or system of care in the planning and development processes. Although public health systems in different locales vary, the overall structural plan should encompass the four levels of the MCHB pyramid as described below. States must determine how best to achieve this within their own structures.

Infrastructure Building Services comprise the foundation of the MCHB/Genetics pyramid of health and social services. Activities are directed at improving and maintaining health status by providing support for development and maintenance of comprehensive health and social services systems. These activities include development and maintenance of health services standards/guidelines, training/education, data, and planning systems. Examples include needs assessment, evaluation, planning, policy development, program coordination, quality assurance, standards development, monitoring, training, applied research, information systems, and systems of care. In the development of systems of care, there should be assurance that all systems are family-centered, community-based, and culturally competent. Public health professionals, health care providers, genetic service specialists, consumers and consumer groups should all be actively involved in infrastructure building.

Population-based Services include preventive interventions and personal health services, developed and available for the entire population of a state. Disease prevention, health promotion, and statewide and community-wide outreach are major components. Common population-based services related to genetics include: preconception screening and counseling (e.g. family history, teratogens, folic acid); outreach/public education; prenatal screening and counseling (e.g. Rh incompatibility, maternal serum screening); newborn screening (e.g. phenylketonuria (PKU), sickle cell diseases, hereditary hearing loss); childhood screening (e.g. physical examination, medical and family history); and adulthood screening (e.g. breast and bowel cancer screening, cholesterol screening, hemochromatosis screening). Other population-based services include lead screening, immunization, sudden infant death syndrome counseling, oral health, injury prevention, and nutrition. These services generally are available to a mother, child, or other individual receiving care in the private or public system, in a rural clinic, health maintenance organization (HMO), military base, hospital, or the Indian Health Service, whether or not an individual is insured. Provision of population-based services in genetics is accomplished through coordination between public health programs

and other health systems and the partnerships among public health professionals, health care providers, and genetic service specialists.

Enabling Services allow, provide access to, or permit the derivation of benefits from the array of basic health and other health care services. These may include, for example, such services as transportation, translation/interpretation, outreach, respite care, health education, family support, health insurance, case management, coordination of/with Medicaid, and the special supplemental nutrition program for women, infants, and children (WIC) of the US Department of Agriculture's Food and Nutrition Services. Enabling services are required especially for the low-income, disadvantaged, geographically or culturally isolated, and those with special and complicated health needs (i.e. traditionally underserved). For many of these individuals, the enabling services are essential; without them, service access is not possible. Enabling services most commonly provided by agencies for children with special health care needs include transportation, care coordination, translation/interpretation, home visiting, and family outreach. Family support activities include parent support groups, family training workshops, advocacy, nutrition, and social work.

Direct Health Services generally are delivered individually from a health professional or social services professional to a patient. The setting is usually an office, clinic, emergency room, or hospital. Service providers may include geneticists, genetic counselors, primary care physicians, registered dietitians, public health or visiting nurses, nurses certified for obstetric and pediatric primary care, medical social workers, social workers with expertise in foster care and adoptions, nutritionists, dentists, sub-specialty physicians who serve children with special health care needs, and other sub-specialty physicians, audiologists, occupational therapists, physical therapists, speech and language therapists, specialty registered dietitians, and/or laboratory personnel.

Basic services include what most consider as ordinary medical care: inpatient and outpatient medical services, allied health services, laboratory testing, x-ray services, dental care, and pharmaceutical products. State Title V programs (Title V Grants to States for Maternal and Child Welfare, Social Security Act of 1935, H.R. 7260) in the US support services such as prenatal care, including prenatal genetics, by operating programs directly or by funding local providers. For children with special health care needs, these services include specialty and subspecialty care for those with genetic conditions, birth defects, chronic illness, HIV/AIDS, and other conditions requir-

ing sophisticated technology; access to highly trained specialists; or an array of services not generally available in most communities.

Applying the IOM Core Public Health Functions to Selected Layers of the Genetics Pyramid

Involvement and leadership from within the state public health and genetics communities are fundamental to the successful integration of genetic services into public health programs and health care and social services systems. Leadership requires not only the ability to create a vision, but also the will and wisdom to achieve it. At all levels, leaders are needed who have the knowledge, commitment, and capacity to build and maintain the infrastructure that supports the integration of genetic services into all areas of the MCHB pyramid. Planning activities must be as open as possible and involve all stakeholders. By evaluating each layer of the MCHB/Genetics Pyramid with respect to the public core functions, it is possible to systematically develop sound public health policy in genetics. For the sake of brevity, this section focuses on infrastructure building as an example of applying this process. This section also gives some limited examples of population-based services as additional examples of this process. A more detailed analysis can be applied to population-based services and other levels of the structure and the reader is referred to Appendix A for a more in-depth discussion of this process.

Infrastructure Building Services

Infrastructure building services should be designed to form the foundation upon which the other service levels are built.

Assessment

Health of communities and populations at risk is assessed and monitored to identify genetic health and health care problems and priorities.

In the future, it is likely that knowledge and technology will allow community health problems to be assessed and monitored in the context of genetic risk and protective factors. Information collected about genetics could be specific to genetic variants, health status, and demographics, and related to interventions or environmental triggers. The knowledge base created will require understanding: (1) relationships between DNA-based risk and protective

factors and their interactions with each other; (2) interactions between genetic and environmental factors; (3) disease pathogenesis; (4) strategies for reducing morbidity and mortality, and (5) relationships existing between phenotype and genotype. Creation of this knowledge base may necessitate large-scale, population-based studies that are beyond the scope of individual states. In such cases, states should consider positioning themselves to obtain and use this information.

Necessary requisites for the development of this information will include partnerships between health care and social service systems and public health programs, and the development of integrated information (data) systems. The tools that are developed from this knowledge base and partnership will enable ongoing assessment activities and a view of genetic information from a public health perspective.

Information Systems Development

Information systems should be developed with the following aims: integration of public health and personal health services data; linkage of clinically useful data to improve access to high quality services; and continuous quality improvement.

A number of information integration initiatives have targeted the enhancement of state and local data collection, storage, and analysis with concurrent elimination of duplicative efforts and information. However, their collective success has been limited in part by the inadequacy of an articulated long-range vision. This vision should be shaped and shared by key public health partners, with communication and coordination across federal, state, and local agencies and national organizations. There is a need for feasible plans of action that accelerate specific data capacity-building initiatives in public health systems with an understanding that data collection serves multiple functions: (1) to assist with evaluating and understanding direct health service needs; (2) to provide information for epidemiological purposes; (3) to monitor quality assurance, and (4) to form a basis for establishing scientifically sound policy.

Genetic knowledge and technology should be used to facilitate data and program integration with the goal (and capability) of identifying and responding to areas of need. The data collection system must capture relevant information and have the appropriate security in place to maintain individual privacy and confidentiality of all information recorded. The data collection system should encompass such elements as risk factor identification and identification of individuals with specific genetic condi-

tions. Thus, there should be: (1) adequate population-based data collection and validation mechanisms; (2) presentation of the data in a useful format, and (3) appropriate analysis of the data obtained. For example, surveillance and service data should be analyzed together to ensure that adequate and appropriate services (including genetic) are being delivered to individuals. Data on safety and efficacy of genetic interventions should also be collected for quality assurance.

A data collection system capable of identifying community genetic health and health care problems and systems of care is far larger in scope than a categorical registry and may require substantial time and effort to construct. However, efforts could begin by integrating existing surveillance systems (e.g. newborn screening programs, birth defects registries, vital records – birth and death certificates, children with special health care needs programs, immunization registries, cancer registries) with genetic service information resulting in a statewide surveillance system for genetics and genetic services. Public agencies traditionally have underfunded and undervalued such comprehensive data collection systems.

Applied Research

The genetic service system should be current and grounded scientifically in both basic and applied research.

While it is generally not the role of states to engage in basic research, a state genetics plan should address the identification of research priorities to benefit the public's health. Partnerships between the state health agency, genetic service providers, universities, and national organizations should be considered to identify appropriate areas such as environmental triggers, prevention strategies, and effective interventions. Human subject protection guidelines should be followed. Research in public health genetics likely will require the commitment of new resources.

Policy Development

Public policies should be formulated to prevent harmful uses of genetic information and to protect against discrimination.

Health policy must offer reassurance that genetic information and technology will be used only to improve health outcomes and that protection will exist to preclude genetic discrimination. Formulation of public policy should be reflected through program planning for a statewide system of genetic services integrated into public health and health care practice. Such a system should take into account the current health service delivery system

(public and private) for genetic services and the prioritized needs of the population. Development of public policies needs to occur through an informed process with input from a broad-based spectrum of individuals and organizations with differing health concerns and professional backgrounds, including special interest and consumer groups. Policy development should provide guidance for the:

- use of new genetic tests, technology, and treatments;
- provision of care for individuals identified by genetic screening, including guidance for diagnosis, treatment and prevention programs;
- prevention of unsanctioned uses of genetic information, and
- prevention of discrimination based on an individual's genetic constitution.

Legal Framework and Regulatory Infrastructure

Statutes and regulations should exist that protect the public interest while supporting the development and provision of comprehensive, quality genetic services.

An underlying policy debate concerns the argument that genetic information is unique and deserving of special consideration. Genetic information is considered permanent, identifiable, predictive, familial, and potentially prejudicial. The state genetics plan should address legal issues with the goal of protecting the public from unethical use of genetic information, research, or services. New legislation may be necessary to address the following: insurance discrimination, employment discrimination, disclosure of information, informed consent requirements, privacy of personal genetic information, storage and use of biological samples, penalties for violations, regulation of clinical professions providing genetic services such as counseling, conduct of genetics research, and use of laboratory samples for genetic testing. The state genetics plan should address the employment of qualified personnel responsible for monitoring compliance and enforcing statutes and regulations actively to ensure the public's safety. It should address the issue of leadership to secure an adequate legal framework, including the authority to regulate genetic services (laboratory and clinical). The education of health care professionals, governmental authorities (legislators and the judiciary), and the public on the protections afforded by existing state and national statutes should be part of the state plan. In addition, the state plan should include methods to educate the public, health care professionals and governmental authorities, including legislators and the judiciary, on the protections afforded by existing state and national statutes.

Assurance

Access to appropriate, beneficial, and cost-effective care should be assured for all populations, including health promotion and disease prevention services.

Access to health care lies at the heart of the ability to incorporate genetics knowledge from mapping the human genome into health care and public health practice. The infrastructure must be in place and partnerships forged to ensure that there are systems to increase knowledge and awareness of genetic risk factors and their relationship to preventing disease, to maintaining good health, and to regaining health through interventions. Major components of assuring access include reducing the cultural and linguistic barriers to health services and providing culturally competent health education for the general public. Quality of services should be monitored through relevant and meaningful performance measures. States should develop expertise and resources necessary for licensure, certification, or registration of genetic counselors, clinical geneticists and other health care and public health professionals qualified to provide genetic counseling and services. Systems should be developed to assess the quality of genetic service centers such as those for prenatal diagnosis, hereditary cancer screening, treatment of rare disorders, and specialized genetics laboratories.

System of Integrated Genetic Services

Genetic services should include integration of genetic medicine into public health programs and health systems.

Since most diseases are influenced by genetic risk factors, advances in genetic medicine should be integrated into health promotion and disease prevention activities in both public health and primary health care. Genetic services historically have included those services aimed at prevention, health education, and integration with primary care and sub-specialty care. Many states have organized family-based services that include general genetics clinics, as well as clinics for specific genetically based conditions, such as metabolic disorders and hemoglobinopathies or hemophilia. Other genetic services have included population-based efforts within newborn screening programs and prenatal screening programs. Many states have organized specific laboratory services for cytogenetics, newborn screening, molecular genetics, prenatal screening, and biochemical genetics. In addition, states have patient and family education activities associated with these efforts. Appendix A provides a detailed exposition on such an integrated system.

Funding for Genetic Services

Genetic services should be available, accessible, and affordable to all individuals who need or desire them.

Genetic services are currently funded by an array of public and private mechanisms including Title V, Medicaid, Children with Special Health Care Needs (CSHCN), state tax revenues (sometimes as a budget line item), public and private health insurance, and self-pay. Several states have mandated that private payers provide coverage for mandatory population-based screening (e.g. newborn screening and hearing screening) as well as corresponding clinical or related services (e.g. dietary formula for newborns and food products for infants and children). The incorporation of genetic medicine into public health and health care systems is creating a new (and additional) workload that requires new financial resources. A state genetics plan should identify and coordinate funding sources within the state for provision of individual genetic services. The state genetics coordinator may work with other departments or agencies within the state to eliminate gaps in funding for genetic services. Additionally, it should identify the amount of funding necessary to ensure that qualified personnel and facilities are available, accessible, and responsive to the public. Funds for data and service integration, coordination, and monitoring should be provided to enable states to develop programs and services to reduce the mortality and morbidity resulting from inherited conditions. State health systems also need to develop the funding capacity for medical genetics and its supporting workforce in new areas that have not had funding (particularly chronic disease, environmental, and occupational health programs) without impacting existing programs [12].

Training and Education of Health Professionals

A well-prepared community of health care and public health practitioners with genetics expertise should be available in numbers sufficient to meet the needs of the public. These practitioners should be capable of communicating the benefits, risks, limitations, and implications of genetic testing and accurately interpreting and appropriately utilizing genetic information in clinical and public health practice.

As the availability of genetic screening and testing increases and testing becomes more commonplace, it is likely that increased numbers of genetic studies will be ordered and interpreted by primary care providers. These health care providers will be increasingly called upon to interpret the meaning of genetic information for individual patients and to help those patients make informed deci-

sions about testing and treatment. To ensure that genetic tests are administered and interpreted in an appropriate manner, counseling and education will need to be essential components of care offered by health care providers. Thus, providers must be trained to perform genetic educational and counseling functions appropriately and to understand when it is best to refer patients to specialized genetics practitioners.

Public health professionals with expertise in applying epidemiological, social, and behavioral science to the field of genetics can play an integral role in providing quality genetics programs by conducting epidemiological studies associating genetic data with health outcomes. To this end, community programs and partners should be identified who are interested in cooperating with and/or conducting community assessments regarding detection, diagnosis, treatment, and prevention of genetic diseases.

The scarcity of health care providers and public health professionals with genetics expertise makes it essential that strategies to increase their numbers and the funding to support them, particularly among minorities, be included in a state genetics plan. This could include scholarships and support for medical geneticists and genetic counselors who accept positions in state agencies or state-funded genetic service projects. Genetics training must be increasingly incorporated into curricula at medical schools, nursing schools, schools of public health, post-graduate training programs, and continuing medical and nursing education. Other avenues for impacting the pool of qualified personnel include state-funded institutions such as universities, professional licensing agencies, and higher education coordinating boards.

A needs assessment study to evaluate the current and future service capacity of genetic service providers, including genetic counselors, should be a component of a state genetics plan. One strategy to increase numbers of genetic service providers is to specify the number of necessary genetic service providers in a genetics plan's quality of care requirements. The state genetics coordinator should also work with necessary departments and agencies to ensure that health plans that apply for licenses to operate within the state have a minimum number of genetic service providers. Genetics plans should also address informing policymakers and the public on issues related to training and educating health professionals.

Evaluation of the Genetic Services System

High-quality, culturally competent genetic services should be available and accessible to all who need or desire them. Health outcomes should be improved by the use of genetic testing and interventions.

Genetic services in the US are currently being provided in a variety of settings (such as community, migrant care and military clinics, academic health centers, public health departments, public health and private laboratories, and private medical practices). Some state programs currently provide linkage between screening and services, coordination of primary and specialty care, and integration of community services for pediatric and adult individuals with genetic conditions. There is a general impression that these programs of early identification, with appropriate follow-up services, improve the health status of pediatric and adult individuals with genetic conditions. Generally, provision of services takes place in the absence of sufficient assessment and quality assurance information. Concern by the genetics community is not necessarily that screening or testing will be done but whether or not:

- Genetic testing will be offered, performed, and interpreted appropriately with pretest and posttest counseling;
- Access to genetic screening tests will be equitably available;
- Analytical and clinical validity of tests will be properly established;
- Appropriate and timely interventions will be offered once an individual or family is identified;
- Privacy will be protected, and
- Sufficient resources will be provided to support these activities.

Quality of services, personnel providing them, cultural competency of the services, and use of surveillance and population-based epidemiological studies are important components of evaluation. The state genetics plan should assure that a system is in place to provide ongoing evaluation of the genetic services system on a variety of levels. Evaluation of policies and quality of genetic testing is needed to ensure the appropriateness and quality of population-based genetic testing. Planning, implementation, and evaluation are needed to ensure that genetic tests and services are incorporated into population-based interventions. Evaluation of the outcomes associated with genetic testing and interventions should also be a component of the process. Ongoing monitoring of access to genetic services and utilization of the range of services is also necessary in order to develop a comprehensive evaluation of genetic health services. Community involvement in ongoing

evaluation is critical. Refer to Appendix A for further elaboration on genetic services.

Population-Based Services

Population-based services are designed to serve the entire community and encompass both health promotion and disease prevention activities. These services are provided generally without regard for ability to pay, with the understanding that they benefit the population as a whole.

Public Education

The general public and key policy makers should be well informed about genetics, its impact on health, and the ethical, legal, and social issues that are important to the provision of genetic services and the use of genetic information.

The state genetics plan should address educational mechanisms for the public as well as for key policy makers. The general public and policy makers will need basic information about genetics and its relationship to creating and maintaining good health. An understanding of medical genetics enhances cultural literacy, engenders understanding of genetic diversity and holds the promise of improved treatment and even prevention of some diseases. An informed public is the best societal protection from possible abuses of genetic technology in the future. In addition, effective planning of genetic health care policy must take into account a community's perceptions of genetic information and technology and a community's values.

Comprehensive Newborn Screening Systems

All newborns should be screened shortly after birth for certain treatable and preventable heritable disorders.

Newborn screening for disorders leading to catastrophic health consequences, including death and mental retardation, has been a concern of public health departments since the development of screening algorithms in the early 1960s [13]. Shortly after it was shown that newborn population screening for inborn errors of metabolism could reduce the frequency of, or prevent, mental retardation and consequently reduce the economic burden to individual families and to society, state-based screening programs spread rapidly across the country. These early studies and the health promotion that followed were funded by federal health dollars (from the Children's Health Bureau, now Maternal and Child Health Bureau), and were critical in establishing public health newborn screening systems that included education, screening, follow-up, diagnosis, man-

agement, and evaluation [13–15]. Over the ensuing years, newborn screening programs have remained state-based with tremendous variations in how they are administered. Despite these variations (not because of them), state public health newborn screening systems have been successful in creating efficient, productive disease detection and service delivery systems for thousands of newborns identified annually with heritable disorders.

Within newborn screening systems, there are certain critical junctures between system components where responsibilities and coordinated activities must be seamless and nonduplicative. Defined beginning and ending points are critical in ensuring that all patients with presumptively identified disorders receive full advantages of the screening system. Partnerships between public and privately funded system components (e.g. laboratories) are possible, but care must be taken to achieve the ultimate goal of fast and effective diagnosis and treatment. Thus, for example, private testing laboratories may augment the screening system effectively only if their information can feed into and out of the public health infrastructure seamlessly and efficiently in order to maintain the full integrity of the system and full service availability. Private systems cannot replace mandated public systems unless they can manage the entire population for whom the services are mandated (e.g. without regard to ability to pay). Care must be taken to avoid destructive competitions between public and private system components through communication, education, evaluation, and carefully defined responsibilities. Ultimately, public health departments must ensure that newborn screening systems function to the benefit of the public's health [15, 16].

Lessons learned from newborn screening implementation and expansion over the years must be utilized as new systems are developed. Current dried blood spot screening programs evolved over a period of almost 40 years from fragmented systems of public and private laboratory services with disjointed follow-up, to their current operational levels quietly and efficiently integrating education, screening, follow-up and ancillary services [15]. Today's improved systems resulted from valuable input from the public and professional communities. Virtually all newborns now receive newborn screening and essentially all who need follow-up testing, diagnosis, and treatment obtain it. Now newborns are receiving expanded dried blood spot screening for even rarer metabolic disorders in many states as a result of new technologies such as tandem mass spectrometry. Often screening is optional (at the discretion of the parent) and is uncoordinated (with the state data and follow-up system) through private screening lab-

oratories. Additionally, almost all states have begun universal or targeted newborn hearing screening programs, in which genetics often plays a critical role. This expanded newborn screening should be embraced by state public health systems in such a way that there is: (1) minimal duplication of data tracking between programs that serve the same population; (2) rapid follow-up coupled with efficient and effective delivery of ancillary medical services; (3) adequate privacy protection, and (4) appropriate outcome data for system evaluation and improvement. Where nationally recognized standards of care exist, public health systems must comply. Ultimately, public health newborn screening systems must address nationally issues of uniformity in: (1) service availability (education, testing, follow-up and care), (2) public health decision-making processes related to testing, and (3) methods of financing.

Summary

While *enabling* and *direct* genetic health services currently exist to varying degrees within the health care system, public health genetics contributes a new dimension for the provision of all health care services. Through a public health genetics perspective, a system can be envisioned that allows determination of the unique biological and environmental factors that cause a given person to express a given condition at a given point in time in his or her developmental history. This approach will necessitate a strong relationship between public health and primary care providers. This approach will also necessitate building a sound public health infrastructure for genetics.

The primary focus of this document is to provide guidance for developing the necessary components of that state-based genetics infrastructure for the public's health. We believe that the genetics infrastructure will serve as the foundation for integrated genetic services and supports population-based, enabling, and direct genetic health services. The MCHB/Genetics pyramid displays dynamic and necessary relationships within and between its levels. Using the pyramid as a planning tool results in a logical approach to systems building in public health genetics and other public health programs.

A discussion of enabling and direct genetic health services is provided in Appendix A, 'Recommended Genetic Services for Local/State/Territorial Public Health Systems'. A more comprehensive presentation is provided in the 1997 edition of *Guidelines for Clinical Genetics Services for the Public's Health* [2], available at the NNSGRC website at <http://genes-r-us.uthscsa.edu>.

Acknowledgments

The assistance of Susan Panny, MD, Maryland Department of Health is acknowledged for her comments in preparation of the materials in Appendix A, and to Cassie Lauver, Maternal and Child Health Bureau, Health Resources and Services Administration and Maricela Aguilar, University of Texas Health Science Center at San Antonio for advice in preparing the final report.

Support for this project came from the Maternal and Child Health Bureau of the US Health Resources and Services Administration (grant No. 5 U93 MC00148-03). The National Newborn Screening and Genetics Resource Center is a cooperative agreement between the Maternal and Child Health Bureau and the Department of Pediatrics, University of Texas Health Science Center at San Antonio.

Appendix A: Recommended Genetic Services for Local, State, or Territorial Health Systems

Preface

The purpose of this guidance document is to provide details for integrating genetic services into the framework of local, state, and territorial public health programs, and community/statewide systems of care. These guidelines are intended to include public health professionals and health care providers, as well as other stakeholders, as primary players within such an integrated system. They build upon previous guidelines [2] developed to assist public health systems in integrating genetics into public health planning.

The role of public health in the organization and provision of genetic services to specific populations has been well recognized since the early 1960s, when newborn screening for PKU and subsequently other disorders was initiated. Never before, however, have we stood on the threshold of an era in which the potential need for genetic information or services is faced by practically every individual, family and population throughout the life cycle.

The recent completion of a draft map of the human genome [17, 18] and other advances in genetics and related technologies have resulted in the need to integrate all aspects of genetic medicine, together with its unique potential for health promotion, into public health. This document clarifies that need, provides definitions, and gives examples of the role of genetic services in health promotion and disease prevention throughout the life cycle. A logistical framework and mechanisms of integration of genetics into public health and health care are proposed that build on detailed descriptions of genetic services, staff, and operational requirements previously outlined [2].

Introduction and Information

The ultimate goal of genetic services is to reduce mortality and morbidity and to alleviate suffering associated with conditions having a genetic component in individuals, families, and populations at risk. This can be achieved through: (1) optimizing the health of the population by employing scientific knowledge about (genetic) disease causation, illness prevention, management and health promotion; (2) early diagnosis, intervention and management, and (3) minimization of disparities among individuals and populations through assurance of equal access to and availability of genetic health services and resources.

Until recently, genetic disorders were considered individually rare, though cumulatively fairly common. It was previously estimated that 20% of the population either had, or had within their families, a disorder with genetic implications. Today, as we discover more about genetic involvement, i.e. gene function and mutation (change), in the most common disorders of adulthood, such as can-

cers, diabetes, cardiovascular disease, neurological impairment, kidney diseases, and others, we realize that instead of 20% with genetic disorders, all families within all populations have at least one genetic disorder. This has resulted in the recognition of a need for genetic services and information for a large proportion of patients, families and populations. It has been reported [2] that each year:

- An estimated 3–5% of all newborns have a birth defect or disorder, recognizable at birth and associated with genetic factors, which requires serious medical or surgical intervention.
- Birth defects comprise the most common cause of death in infants under the age of 12 months.
- Through newborn screening, about 3,000 newborns are identified at birth in the United States with a disorder that requires lifelong management or treatment.
- Children with genetically determined disorders occupy about one-third of pediatric beds in tertiary care centers.
- About 10% of the school-aged population has sensory, developmental, behavioral or emotional difficulties, many of which have genetic components.
- The most common causes of death in adults (not including injuries) are complications of cardiovascular, neoplastic or diabetic diseases. Each of these has a genetic component that is detectable, recognizable and possibly amenable to early intervention.

Clinical disorders with a genetic component affect not only the patient but also their immediate and extended families, and a large proportion of the population. Although several of these disorders (e.g. birth defects, newborns identified through screening, cancers) are already registered, recorded, and monitored by public health systems in some states, it is imperative that all states incorporate information and education about the genetic components of all of these disorders (including the adult onset disorders such as cardiovascular and diabetic diseases) into their programs. Genetic services must, therefore, become an inseparable link in the chain that connects all public health systems with patients, families, and health service providers.

Genetic services cannot be provided without serious consideration of their financial, ethical, legal and social implications. Although it is beyond the scope of the present document to encompass these issues in detail, it must be emphasized that genetic services, perhaps somewhat more than other medical disciplines, may be susceptible to abuse, such as breach of confidentiality, invasion of privacy, disregard for autonomy, and causation of unintended harm.

It must be remembered that the fear of potential discrimination against individuals, families, and/or populations with known or suspected genetic diseases or risk factors by employers, insurers, and others is prevalent among many professionals and consumers familiar with genetics. Even though a few states (Wisconsin was among the first, in 1991) have enacted legislation to prevent such discrimination, it may be up to the federal government to provide assurance that such discrimination becomes illegal. The Health Insurance Portability and Accountability Act (HIPAA) of 1996 is a step in that direction [19]. Its purpose is to protect the privacy and security of health information. After much discussion and review, the HIPAA Privacy Rules became effective on April 14, 2001. An informative summary of these rules, as they affect genetics professionals and their patients, is available in the July/August 2001 issue of *Genetics in Medicine* [20]. The three sentinel requirements of the Privacy Rules are the: (1) control of the use and disclosure of protected health information (PHI); (2) safeguarding of patients' rights; and (3) establishment, documentation and implementation of compliance policies

and procedures. All health care providers must be in compliance with these rules by April 14, 2003. It is strongly recommended that all health care providers and investigators who work with sensitive protected health information become acquainted with HIPAA, as well as with the recommendations from the Secretary's Advisory Committee on Genetic Testing [21].

Since this document describes genetic services, it is important to precede any recommendations for their appropriate utilization with certain definitions and clarifications.

Human genetics is the science of variation and heredity within the human species.

Medical genetics is the study of hereditary factors and variation in human disease. It is a subspecialty that is recognized by the American Board of Medical Subspecialties.

Clinical genetics is a medical discipline whereby comprehensive services are provided to patients, families and populations at all ages who have or are suspected of having hereditary, inherited or congenital disorders. Clinical genetics is conventionally divided into several disciplines, and clinical geneticists frequently have expertise in one or more of the following areas:

- dysmorphology (study and measurement of physical and anatomical features and associated disorders);
- biochemical/metabolic genetics (study of disorders associated with underlying biochemical or molecular abnormalities);
- molecular genetics (study of gene function and mutations and associated disorders);
- cytogenetics (study of chromosomes and their functional/structural abnormalities and associated disorders), now rapidly becoming closer to molecular cytogenetics;
- prenatal genetics (the diagnosis of preconceptional or embryonic/fetal disorders, the recognition and management of which will result in the optimization of pregnancy outcome for fetus and family);
- pharmacogenetics (the study of genetic involvement in individual drug/medication reactions and responses);
- public health genetics (the use of genetic information and molecular technology to promote, protect and thereby improve the public's health and well-being), and
- teratology (study of the causes and biological processes leading to abnormal development and birth defects at the fundamental and clinical level, and appropriate measures for prevention).

Predictive genetic information indicates whether an individual is or is not predisposed, at a higher or lower risk, to developing a specific disease in the future.

Diagnostic genetic information is used to indicate whether or not a person has a particular disorder.

Genetic counseling is a communication process whereby the patient and the family receive applicable information about a disorder or issue, its clinical course, management, treatment and prognosis. Reassurance is also an inseparable component of the counseling process, as is information about all options available to family members. Genetic counseling should be provided in a supportive, compassionate, and nondirective manner. Referral for genetic counseling (by genetic professionals) must be available to all providers who serve patients/families with genetic diseases. This includes providers within genetics units, clinics and centers as well as providers outside genetics units, such as primary care physicians, specialists in cancer clinics, surgeons who care for patients with birth defects, pediatric and adult cardiologists, neurologists, care providers in sensory deficits clinics, and others.

Health Promotion and Disease Prevention Throughout the Human Life Cycle

Clinical genetic services are components of all branches of health care and are applicable to patients, families, and populations at all stages of the life cycle. More and more disorders are recognized in which local, state, and territorial public health programs can assume an active health promotion and disease prevention role. Generally, three levels of health promotion and disease prevention are recognized: primary, secondary and tertiary prevention. *Primary prevention* genetic services are services intended to prevent a birth defect, genetic disorder, or disease before it occurs. Genetic counseling is a form of primary prevention. As described above, genetic counseling provides couples with information about their pregnancy and reproductive risks and pregnancy options. *Secondary prevention* genetic services are services intended to prevent the unfavorable sequelae of an existing disorder or genotype. Newborn screening is a classic example of secondary prevention. *Tertiary prevention* genetic services are services aimed at ameliorating the unfavorable consequences of existing disorders through enabling services (MCHB/Genetics pyramid), such as parent-to-parent support and empowerment. Tertiary intervention includes educational and other comprehensive services to children and adults with special needs, appropriate management of genetic conditions, access to orthodontic and other auxiliary devices, dietary supplements, special occupational and physical therapy, and ongoing support group services. Since family-centered care is at the heart of all genetic services, referral of families to condition-specific support groups and facilitation of contact with similarly affected families is an intrinsic component. This component also includes collaboration with consumers and consumer support and advocacy groups. They provide support, information, and much needed help aimed not only at families and populations with genetic disorders, but also at health and social services professionals, educators, policy makers, and society in general.

In addition to the three traditional levels of health promotion and disease prevention outlined above, a 'quaternary' level of prevention involves ongoing basic research into genetic diseases by scientists worldwide. Prevention also extends to the prevention of discrimination on the basis of genetic disease or testing by employers, insurers, and peers and involves close collaboration with legislators [20, 21].

Below are examples of health promotion and primary and secondary disease prevention activities that can occur at all stages of the life cycle.

Preconceptional and Prenatal Period

Timely and effective preconception interventions, prenatal monitoring, and management of maternal diseases associated with fetal risk result in improved pregnancy outcome. A carefully explored family, medical and obstetric history within the primary health care setting in couples contemplating pregnancy can also identify many risk factors prior to conception, and appropriate management can sometimes prevent an unfavorable pregnancy outcome. Intrauterine exposure to alcohol, smoking, cocaine, and other hazardous agents increases the risk for physical and developmental disabilities. Prevention of prenatal exposure to known teratogenic agents from conception through delivery can prevent the primary occurrence of deleterious effects of alcohol, cocaine, smoking, and other hazardous agents. Well-coordinated public educational efforts by health care professionals and, on a larger scale, by public health programs aimed at women of childbearing age have the potential for saving thousands

of children and hundreds of millions of dollars each year. However, such efforts must be undertaken prior to conception and continue throughout pregnancy if they are to be effective. For example, fetal alcohol syndrome affects an estimated 7,000 infants each year in the US [22]. Yet intrauterine exposure to alcohol is totally preventable. Many state public health programs have educational efforts that aim to reduce the maternal consumption of alcohol during pregnancy.

Pre- and periconceptual folic acid prophylaxis can prevent the primary occurrence of 2,000 (about 50%) neural tube defects per year [23]. An affected infant, if not lost during the prenatal period, requires hundreds of thousands of dollars of treatment, services, and support throughout life. Folic acid prophylaxis can improve the likelihood that an infant will be born free of this birth defect and lead a normal life. Primary prevention includes education aimed at relevant professionals and consumers (in this example, health care providers and women of childbearing age). Monitoring and evaluation of outcomes is an essential part of a successful primary prevention program. Most state public health agencies are already involved in folic acid prophylaxis programs.

Teratogen information services (TIS) play an important role in primary prevention, especially for prenatal monitoring for birth defects. Birth defect surveillance systems in states and territories can obtain baseline information to monitor changes in the incidence or prevalence of specific types of birth defects. TIS should be available in each state and territory to provide information to physicians and their patients who are concerned about the risk that a particular agent will cause an unfavorable pregnancy outcome. In states without TIS, access to a national or regional information source such as Reprotox [24] and the Organization of Teratogen Information Services should be provided. Each TIS should be a part of a national network and comply with national guidelines. Collaboration within the existing network in the US has already resulted in research into the teratogenic effects of several agents, and has led to the primary prevention of adverse pregnancy outcomes. Furthermore, teratology courses have been introduced into most genetic counseling programs in the country [25].

Awareness and appropriate management of maternal diseases and infections, such as PKU, diabetes, rubella, and toxoplasmosis, can result in the primary prevention of birth defects and mental retardation. Collaboration between primary care providers, infectious disease experts, geneticists and physicians specializing in high-risk obstetrics is essential for a successful pregnancy outcome. Well-documented examples include the prevention of: (a) mental retardation in offspring of mothers with PKU; (b) congenital malformations and/or metabolic compromise in offspring of diabetic mothers; (c) complications in offspring of mothers with prenatal infections including HIV and parvovirus, and (d) Rh incompatibility disease, one of the most common and easily preventable conditions. Rh disease prevention occurs through appropriate prenatal screening and diagnosis, antibody detection, and treatment during pregnancy.

Prenatal screening offered through maternal serum markers, fetal ultrasonography, and cytogenetic and DNA analyses can not only identify affected fetuses and provide options about pregnancy outcome, but also can identify those pregnancies in need of: (a) special delivery (e.g. cesarian section to prevent damage to infants with open spine defects); (b) management of metabolic defects, for example, fetuses with methylmalonic acidemia or congenital adrenal hyperplasia; (c) recognition and management of prematurity or intrauterine growth retardation, and (d) potential prenatal intervention, including fetal surgery in rare instances. For example, many state public health

agencies monitor maternal serum multiple markers for the detection of birth defects (some states, such as California, mandate the offering of this screening). Maternal serum marker screening for specific birth defects identifies an estimated 5% of pregnancies at higher risk (approximately 200,000 per year in the US), facilitates their appropriate management, and prevents unnecessary complications of labor and delivery [26].

Prenatal monitoring also includes selected use of fetal ultrasound to identify structural abnormalities. The prenatal identification and diagnosis of infants with special needs, when feasible, spares much time, energy and resources that are frequently spent in unnecessary postnatal work-up and diagnostic testing. In addition, screening for fetal chromosomal abnormalities of an estimated 6% (or 240,000) of pregnant women in the US each year who are 35 years or older, or in those pregnancies in which a hereditary chromosomal abnormality is suspected, enables parents not only to make informed decisions, but also plan for timely and effective intervention for affected infants (estimated 5,000/year) [27]. Health care professionals are aware of the age-related risks and usually refer their patients to centers or clinics with experience in procedures needed to provide optimal care to mothers, infants, and families. Early identification facilitates appropriate management (e.g. the need for an earlier or cesarian delivery or other intervention), combines perhaps both primary and secondary prevention and optimizes pregnancy outcome. Additionally, rapid developments in alternative reproductive techniques (e.g. preimplantation diagnostics and egg donation) are promising an ever-expanding set of specialized approaches to the primary prevention of genetic disorders.

The Perinatal Period

Approximately 3–5% of infants (120,000–200,000 per year) in the US are born with birth defects that have serious medical or surgical implications. Many of these disorders are preventable. Over one-third of all children hospitalized in tertiary care medical centers have genetic disorders. Most public health genetics programs in the perinatal period are aimed at secondary prevention. For example, newborn screening for inherited disorders identifies approximately 3,000 infants each year in the US who are born with diseases such as PKU, hypothyroidism, galactosemia, and sickle cell disease. Rapid detection with the recent addition of new technology such as tandem mass spectrometry leads to appropriate treatment and prevents mental retardation, physical disability, and death. Newborn genetic screening is the most well known paradigm for a successful population-based genetics public health program. All 50 states and all territories screen for PKU and congenital hypothyroidism (CH), providing early identification and treatment for approximately 400 PKU and 1,200 CH infants annually. More than 40 states also mandate universal newborn screening for sickle cell diseases (>1,300 cases detected annually) and some states screen for 30 or more disorders [16]. Newborn hearing screening is a recent addition to many state screening programs. Early detection and proper management of hearing impairment has been shown to produce a language quotient in hearing-impaired children that is comparable to that in normal-hearing children [27]. Guidance for newborn screening systems exists [13, 14] and the AAP Newborn Screening Task Force has adequately defined the role that federal and state agencies should play in partnering with health professional and consumers to further the success of these programs [16].

Secondary prevention programs in the perinatal period are generally designed to ameliorate or reduce the effects of the primary disorder.

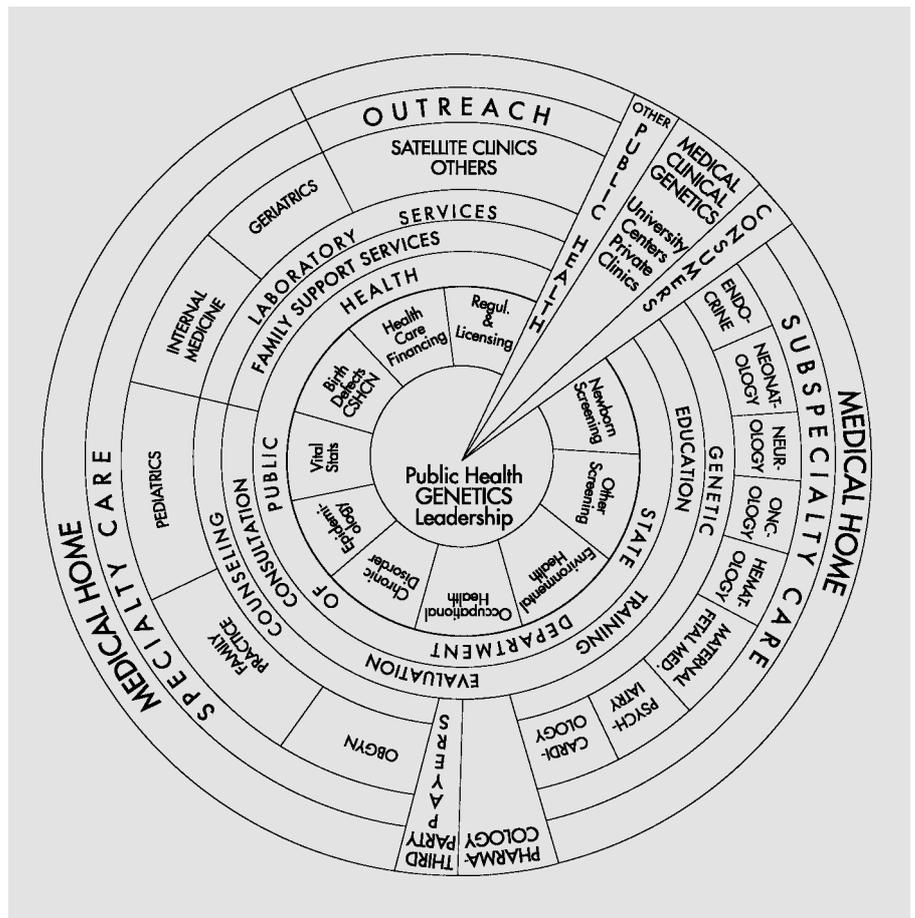


Fig. 4. Structural integration of genetics and public health.

der. For example, special phenylalanine-restricted diets instituted soon after birth prevent irreversible mental retardation in infants with PKU. Similarly, recent studies have demonstrated that penicillin prophylaxis saves the lives of infants with sickle cell disease.

Childhood and Adolescence

Approximately 3% of school-aged children are challenged cognitively and/or have special health care needs. In addition, common genetic disorders such as mental illness, learning disabilities, diabetes, asthma and some metabolic disorders appear in childhood and adolescence. It is estimated that their prevalence ranges from 15–20%. Early provision of organized, well-coordinated, family-focused, culturally competent services for children with special needs prevents certain later complications. Later adolescence is also the time for the identification of individuals at risk for carrier status for some single gene disorders (e.g. Tay-Sachs disease, cystic fibrosis, etc.). Case management within a comprehensive system of care that includes a medical home for these children is an important component of such services. A medical home is defined as a source of ongoing comprehensive care, including mental health, within a person's community. Sensitive transition into adulthood with continuity of care is essential [28].

Screening of individuals for cystic fibrosis and Down syndrome to prevent or reduce many of the disease complications associated

with these conditions is now available. One can envision the screening of children for some adolescent- and/or adult-onset diseases where early identification with appropriate and timely treatment and intervention would lead to the reduction of later morbidity and mortality that may be associated with the disease, e.g. screening for allergies, asthma, cancers, cardiovascular disease, diabetes, and hypercholesterolemia.

Adulthood

In the future, prevention programs utilizing predictive genetic testing may be applied to those who are at risk for adult-onset disorders of genetic etiology. Adulthood is associated with the appearance of common disorders of great personal and public health significance including allergies, asthma, hypertension, heart disease, diabetes, and cancers, many of which have an environmental as well as a genetic component. The variant genes in question might differ from family to family, as might the environmental factors. Most physicians already screen their patients for hypertension, glaucoma, elevated cholesterol and lipid levels, and some early signs of breast, colon, and prostate cancer. The recent identification of genes which, when mutated, increase the risk for breast/ovarian or colorectal cancers offers the possibility in the future of presymptomatic screening for some of these diseases which have a significant cumulative lifetime risk. Presymptomatic identification may result in improved outcome

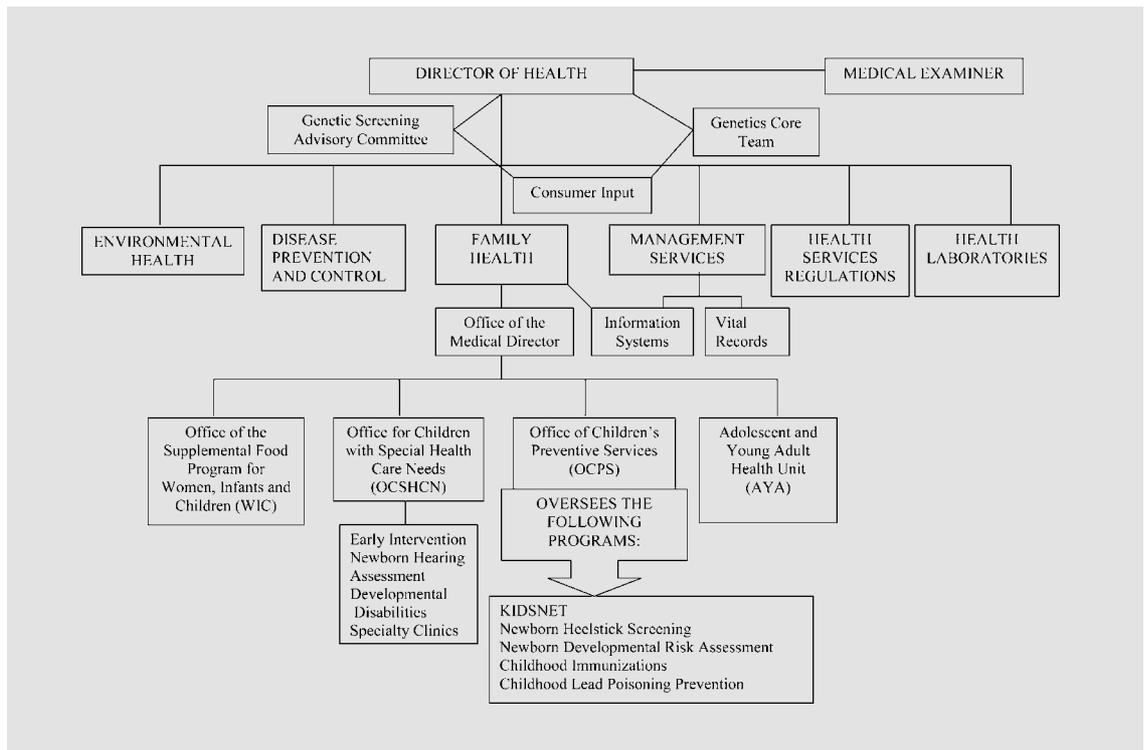


Fig. 5. Rhode Island Department of Health organizational structure.

through, for example, early management and avoidance of potential environmental risk factors.

The education of health professionals and families as well as the development of appropriate surveillance techniques will be essential for many primary disease prevention programs in public health. There will be a need for both the public and health professionals to understand the difference between predictive, diagnostic, and screening genetic testing. For example, DNA-based tests are being developed to detect those at risk for a number of different cancers. Specifically, it is estimated that current methods of screening for specific DNA mutations could detect thousands of women at risk for hereditary breast cancer. The cumulative lifetime risk for breast cancer in all women is approximately 1 in 8 [29]. A mutation in BRCA1 gene is seen in approximately 1 in 800 non-Jewish women with breast cancer and 1 in 107 Jewish women with breast cancer. The identification of a mutated BRCA gene in an individual significantly increases the risk for cancer development in the patient and the family.

In the future genetic testing may aid in evaluating an individual's susceptibility to these diseases. Later onset and degenerative diseases with known genetic etiology include Huntington and Alzheimer diseases. Mental illnesses, including schizophrenia and manic depression, also have genetic components. Research is in progress to identify specific genetic risk factors and interventions for these and other disorders. Hemochromatosis, a common single-gene disorder of iron metabolism that is fatal when untreated, affects about 1 in 400 individuals. Screening and treatment are effective, feasible at the primary care level, and life saving [30]. However, the use of DNA tests for

predictive testing is often complicated by the low penetrance of mutated genes – or in this case, the frequency with which the mutated hemochromatosis gene produces the characteristic effect in those individuals possessing it. The genotype cannot always be directly correlated to phenotype. An accurate diagnosis of hemochromatosis also may be masked by its concomitant complications, such as diabetes, liver cirrhosis, congestive heart failure and arthritis. Thus, physicians must be aware of the existence of this common and easily treatable disease, if its preventive strategies are to succeed.

α_1 -Antitrypsin is an example of a genetic disorder associated with environmental exposure. About 1 in 5,000 individuals is homozygous for the abnormal α_1 -antitrypsin gene, the morbidity of which is severely aggravated by smoking and is partially preventable [30]. Thus, knowledge about abnormal α_1 -antitrypsin status may assist in targeting antismoking behavior. Other genetic issues associated with the adult childbearing years include pregnancy loss, infertility and pregnancies at risk for unfavorable outcome. Appropriately designed population screening to detect carriers of serious genetic disorders such as Tay-Sachs disease enables couples to make informed reproductive decisions.

Structural Integration of Genetics and Public Health

State Examples

A great deal of education, communication, information, and data sharing must occur at all levels of the local/state/territorial public health structure before it is possible to achieve true integration and

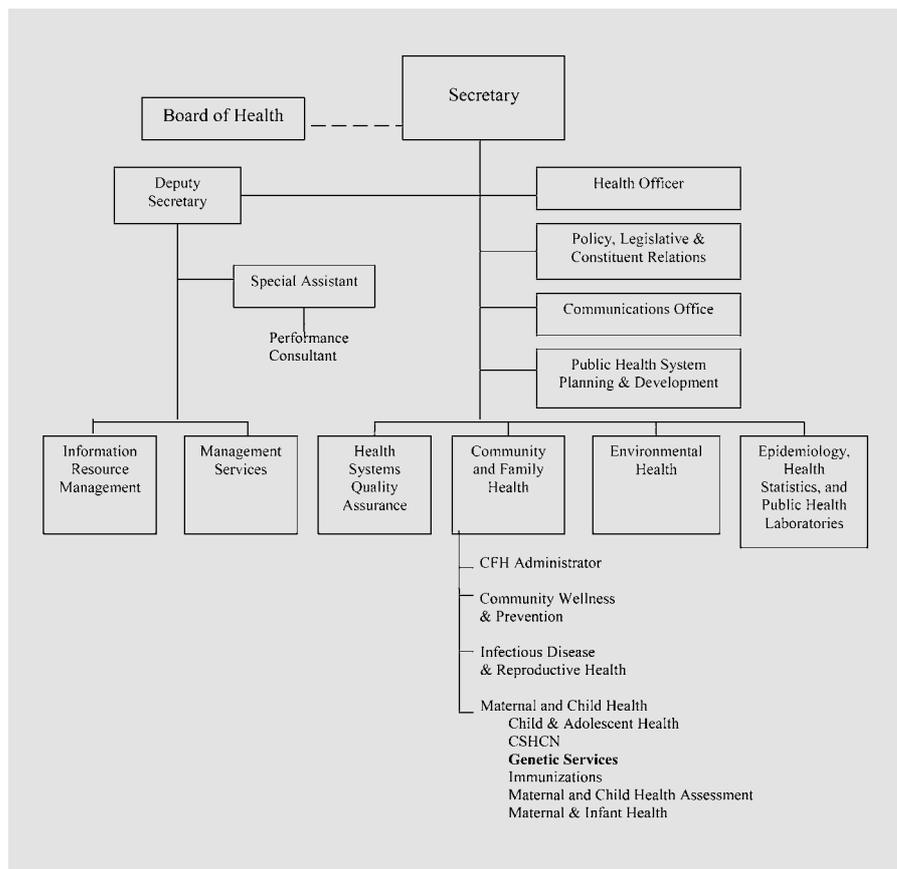


Fig. 6. Washington State Department of Health organizational structure.

collaboration with health professionals. Only then will the three-dimensional, four-level pyramid become a solid, dynamic structure. The dramatic breakthroughs in genetic technology and the resulting expansion of molecular diagnostics make it critically important that we pay attention to cultural differences, quality assurance at all levels, and the active involvement of consumers and families in all genetically related services and deliberations.

The circle diagram in figure 4 emphasizes genetic service interactions and their relationships with the local/state/territorial departments of public health, i.e. the hub (center) of the wheel. The hub, representing the state genetics leadership in the form of the advisory committee, the needs assessment and plan, evaluation, and development of standards, roughly corresponds to the base (level 4) of the pyramid. It is surrounded by 'spokes' or offices within the department of public health, such as vital statistics, newborn screening, chronic diseases, occupational, and environmental health (population-based services – level 3). All clinically related services (level 1) form the outer perimeter of the circle, and counseling and laboratory services are an uninterrupted circle with connections to all. Enabling services (e.g. family support services – level 2), as well as education, training, evaluation, and consultation, are also connected with the perimeter (direct health services), as well as with the center (leadership and population-based services) of the circle. The wedges in the diagram represent those programs or groups that not only interact with all adjacent sections at all levels, but are also directly connected to the state department of public health. They are consumers, who

participate in all components of the system, genetics units, laboratory and other service providers, and public health programs at university, school, county, and city levels who also interact with adjacent programs and the hub's genetics leadership. Pharmacology or pharmacogenetics touches on enabling services and may become part of direct services in the future.

A more specific overview of the structure of genetics and public health within several states/territories reveals major differences in the structure of individual health departments and in the position of the 'Genetics Office' within their hierarchy.

For example, in Rhode Island (fig. 5), two groups, the Genetics Screening Advisory Committee and the Genetics Core Team, each with formally established consumer input, respond to the State Director of Health. The Division of Family Health and other divisions report directly to the State Director of Health. Other offices, all serving children and families (WIC, CSHCN, children's preventive services, adolescents and young adults) are connected and integrated within the system [31].

In Washington State (fig. 6), the office of Maternal & Child Health (MCH), together with two other offices, namely Infectious Diseases and Reproductive Health and Community Wellness and Prevention, responds directly to the director of the Division of Community and Family Health. Genetics is one of the six programs within MCH [32].

These states and others with organizational structures that foster spontaneous, ongoing communication and natural alliances may

integrate genetic services more readily. Consideration of existing organizational structures within local and state departments of public health should be part of both short- and long-term planning.

Recommendations for the Initiation of Integration

State Genetics Coordinator

Each state should identify a full-time state genetics coordinator with a background in genetics, service delivery, and other related public health issues. The coordinator's activities should include responsibility for, awareness of, and monitoring duties in the following areas (includes all four levels of the pyramid):

Advocacy. Advocacy for the role of genetics in public health includes initiation and maintenance of linkages between and among the well-established genetics unit within the division of MCH and other public health programs such as the department of laboratory services, occupational and environmental health, chronic disease, epidemiology and vital statistics. The coordinator also initiates and maintains linkages between providers of genetics services, consumers, and all relevant components of a comprehensive state genetics system.

Collaboration and Communication. Collaboration and communication with other public health programs should occur in order to maximize use of existing data collection programs within the department of public health and other agencies, such as newborn and other screening, vital statistics, cancer and solid tumor registries, immunization registries, birth defects registries and any others that share common data elements. The coordinator should actively participate in any state/territorial genetics advisory committee activities. In addition, the coordinator should collaborate with other state genetics coordinators to share information and partner in developing and implementing cooperative genetics activities.

Identification of Need for Current and Additional Services. The coordinator should initiate or contribute to the service needs assessment within the state/territory and to the identification of needs and resources for additional services as applicable and required.

Coordination of Genetic Services and Awareness of Their Types, Distribution, and Quality. Coordination of genetic services involves familiarity with all aspects of clinical and laboratory components of state/territorial genetic systems including: types and levels of services, personal and population-oriented; distribution of services within the state/territory and promotion of availability and accessibility for those who need them; training and dissemination of information, education programs and materials; mechanisms of reimbursement; recognized professional standards for clinical and laboratory personnel, facilities and genetic services; mechanisms of monitoring contracts related to state funded services, assuring their quality, performance, effectiveness and appropriate evaluation; and recommendations for regulation of state genetic services.

Organization of Consumer Involvement and Monitoring Education and Satisfaction. The coordinator works closely with consumer representatives and interest groups, as well as with other advisory bodies such as the cancer council, the diabetes council, council on aging, vocational rehabilitation, and others. The coordinator monitors consumer education and satisfaction surveys.

Legislative Issues. The coordinator works closely with the state/territorial health department, genetics advisory committee, consumer groups, and other interested parties to advocate that legislative initiatives and regulatory efforts be directed at genetic medical issues, issues of privacy, confidentiality, prevention of discrimination, and quality assurance.

Financing of Genetic Services. The coordinator is aware of types of funding for genetics services within the state/territory and resources available for coverage of underinsured and underserved communities. The coordinator monitors and applies for federal funding opportunities to improve genetics infrastructure and activities at the state level.

Strategic Planning. The coordinator initiates the establishment of a state/territorial genetics advisory committee with representation from both public health and health care providers as well as the community at large. Stakeholders could include participants from the departments of environmental and occupational health, chronic diseases, laboratory services, vital statistics, health care financing, and others within public health programs. In addition, the committee should include those whose activities are essential for successful implementation of the state/territorial plan, e.g. the insurance industry, biotechnology industry, legislators, genetic service providers from the academic, private and public sectors, University Affiliated Programs (UAP), and consumers. This is not to say that participation in the state/territorial genetic advisory committee should be so numerous as to render it incapable of concrete work and goal achievement. Representatives of the above advisory committee, together with the coordinator, initiate the development of a state/territorial genetics plan. An outline for the process of the development of such a plan is provided in table 1. Each state genetics plan will be unique but, as stated in the framework document, it is assumed that a central genetics program will be maintained within departments of public health.

Linking Public Health and the Health Care Delivery System

The schematic diagram in figure 7 demonstrates the anticipated direction of genetic medicine. The current and potentially near future role of the primary health care provider is demonstrated in bold, while the remainder of the diagram currently represents the role of the genetics researcher.

Public health genetics integrates genetics into the public health disciplines such as epidemiology, biostatistics, environmental science, ethics, and social science. Often, this is accomplished by linking the genetics program office within the department of public health with other public health units such as environmental or occupational health, chronic diseases, and vital statistics (hub and spokes of the 'genetics wheel' in fig. 4). In turn, this linkage provides connection between systems for data collection, evaluations and assessments that cut across programs. Routine linkage of data systems should encompass linkage of birth and death certificates, birth defects and tumor registry data, birth defects registry records with vital statistics (births, fetal deaths and deaths), statewide inpatient hospital discharge records with birth certificates, newborn screening records with birth certificates, the statewide clinical genetic services database and birth/fetal death certificates. There should be assurance that systems exist for direct referral from clinical genetics to early intervention services for infants and children [33]. Similar examples of linkages can be envisioned at the adult level (chronic disease) that would enable data collection about adult onset disease such as cancer and cardiovascular disease.

Whatever structures evolve at the state level, the structure of genetic services must take into consideration the milieu of public health departments and health care systems and, most importantly, the people they serve. In addition to a central genetics office within the state public health department, there should be a genetics health care system outside the department. Details of such a system are pre-

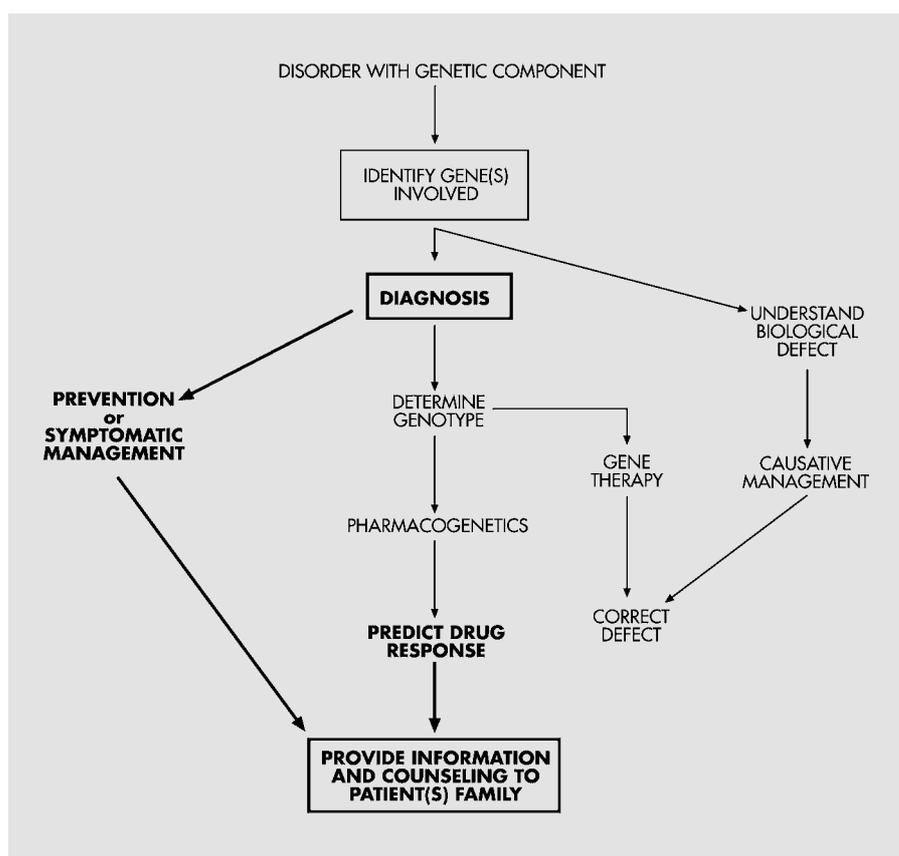


Fig. 7. Schematic diagram of the anticipated direction of genetic medicine. The role of the primary caregiver within this scheme is in bold.

sented in the CORN guidelines [2]. Descriptions are provided for types of services, both direct and population-oriented, laboratory services, facility and operational requirements, staff classification and credentials, details of genetic evaluations, patient records and rights, and quality assurance. At a minimum, every state should have access to medical support services necessary for diagnosis and management of genetic and congenital disorders. Laboratories associated with a genetics unit should participate successfully in available proficiency testing programs. Also, when planning for its system of services, every state must adhere to licensing requirements, published guidelines, standards and regulations such as the 2001 edition of The Joint Commission on Accreditation of Health care Organizations (JCAHO) document and its revisions in support of patient safety and medical/health care error reduction [34].

Workforce Capacity

Every state that is providing genetic services should assure that expert consultation is available (either at its own institutions or by referral to another institution). Genetic health professionals including clinical geneticists, PhD medical geneticists, genetic counselors, cytogenetic, biochemical, and molecular genetic laboratory directors, and cytogenetic technologists should be certified by their own professional organizations, such as the American Board of Medical Genetics (ABMG), the American Board of Genetic Counseling (ABGC), and the National Certification Agency for Medical Laboratory Personnel (NCA). Staff should be familiar with practice guidelines

developed for specific disorders or groups of disorders by the American College of Medical Genetics Subcommittee on Practice Guidelines, the American Academy of Pediatrics, the American College of Obstetrics and Gynecology, the American Academy of Family Physicians, and others.

The CORN guidelines [2] provide a description of the clinical and laboratory personnel that are needed to deliver genetic services. In addition to staff delineated in the CORN guidelines, public health systems also need a public health workforce with competencies in genetic epidemiology, ethics, law, biostatistics, and related fields. Public health agencies have an active role in training and should provide public health trainees with exposure to genetic professionals through rotations within their agency and other programs. Programs in schools of public health should be encouraged to ensure that public health students become aware of the existence of genetic services, genetic epidemiology, and the role of genetics in health promotion and disease prevention in a comprehensive health care system. For example, trained professionals are needed in the areas of quality assurance, epidemiology, data collection and follow-up required in plans for genetic testing, screening, and reporting. It is recommended that genetics courses be offered in all school(s) of public health. States also need health care professionals who are competent to treat specific genetic disorders in the field of genetic medicine. States can encourage curricular changes in schools of medicine, nursing, and other health professions through licensing requirements.

Appendix B: Membership of the NNSGRC Genetics Advisory Committee

(References to organizations do not imply official approval by these organizations)

Celia I. Kaye	Co-chairperson, UTHSCSA School of Medicine
Edward R.B. McCabe	Co-chairperson, UCLA School of Medicine
Michele Lloyd-Puryear	Advisor – MCHB/HRSA
Marie Y. Mann	Advisor – MCHB/HRSA
Bradford L. Therrell	Advisor – NNSGRC Director
Judith Livingston	Support – NNSGRC Staff

Committee Members (alphabetical order): Representing:

Sylvia Au	Coalition of State Genetics Coordinators
Gerard T. Berry	Society for Inherited Metabolic Diseases
Jannine D. Cody	Genetic Alliance
Leslie Cohen	National Society of Genetic Counselors
George C. Cunningham	American College of Medical Genetics
Franklin Desposito	American Academy of Pediatrics
Robert M. Fineman	March of Dimes Birth Defects Foundation
Carol Greene	Genetics Community at Large
Stanley L. Inhorn	Association of Public Health Laboratories
Amy Klein	Association of State and Territorial Health Officials
Kathy Peppe	Association of Maternal and Child Health Programs
Eugene Pergament	Genetics Community at Large

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