

A State Plan for Michigan

Genetics Through the Life Cycle: Improving Health and Preventing Disease

2003

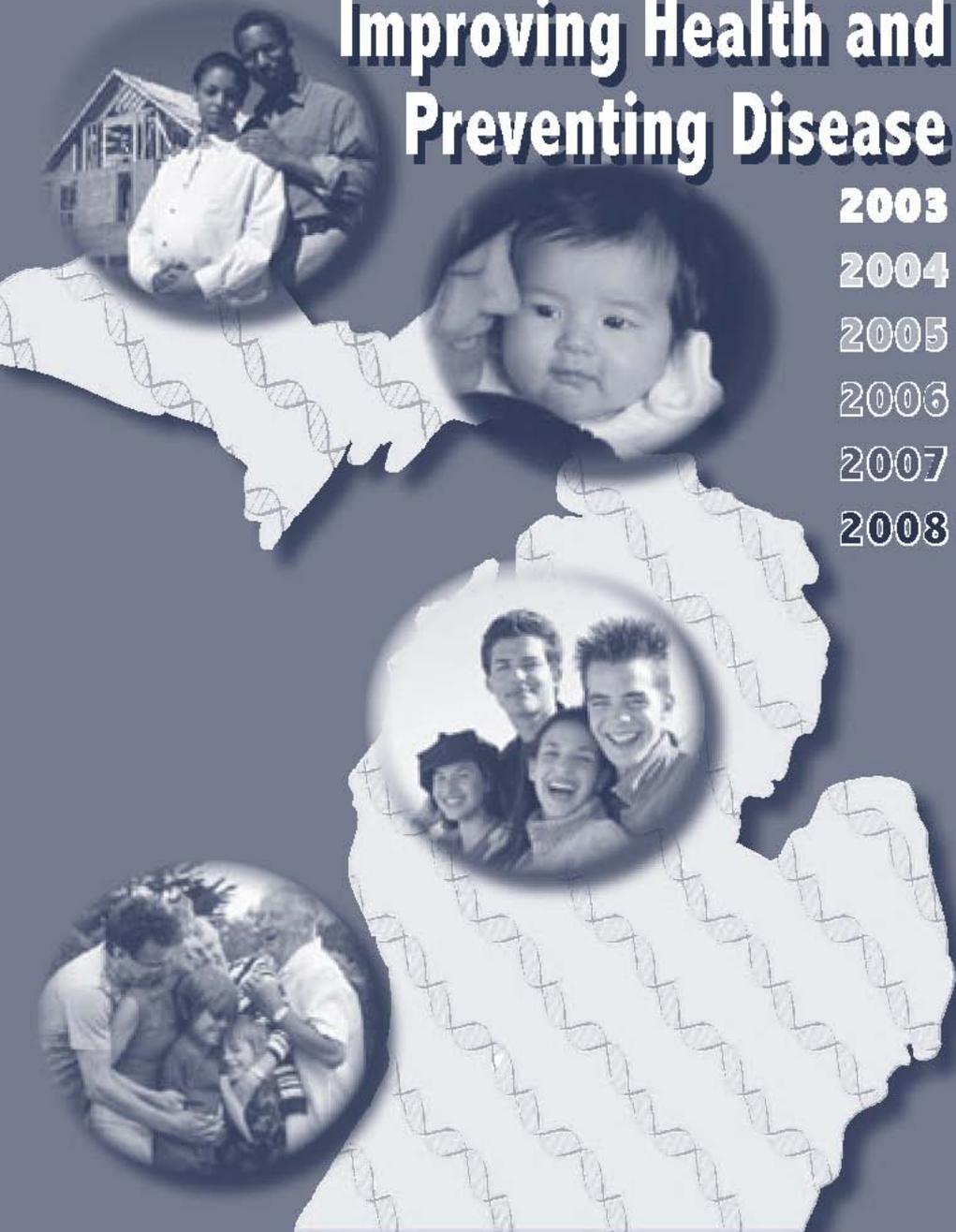
2004

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Michigan Department of Community Health

**A Vision for the Role of
Genetics in Public Health:**
*Improved health outcomes and an
enhanced quality of life
for the people of Michigan
through appropriate use of genetic
information, technology,
and services.*

Introduction

This pamphlet provides an introduction to the five-year genetics action plan developed for the State of Michigan by the Department of Community Health and a wide array of partners and stakeholders. It is based on the findings of a statewide needs assessment conducted from 2000 through 2002.

A two-year grant for Infrastructure Development from the Genetic Services Branch of the Health Resources and Services Administration/Maternal Child Health Bureau (HRSA/MCHB) provided the impetus and funding needed to accomplish this strategic planning project.

The plan consists of six goals that will improve traditional maternal and child public health genetic services, as well as create a more comprehensive agenda spanning common chronic diseases with onset in adult life. The goals encompass the emerging field of "genomics", promote the integration of genetics within existing programs, and emphasize the core functions of public health: assessment, policy development and assurance.

Some Helpful Definitions

Genetics:

The science of heredity; the study of genes and the way they determine traits and characteristics passed from generation to generation. In contrast to genomics, "genetics" refers to a single gene and its effects.

Genome:

All of an organism's genetic material—the DNA contained in the chromosomes and mitochondria of cells.

Genomics:

The study of the entire genome, including the complex interactions among multiple genes as well as between genes and the environment. Applied to public health, genomics offers the potential to better understand the role of genes, environment, and behavior as risk factors for complex, chronic diseases.

Newborn Screening:

A public health program mandated by state law to test newborns for certain rare but treatable disorders.



Needs Assessment

- The Need for a Plan**
- Issues Identified**
- Key Findings**
- Development of the Plan**

Why does Michigan Need a Genetics Plan?

The definition of “genetic disorder” continues to expand. Such conditions are no longer considered rare, but instead are known to affect a large segment of the population. Many developmental disabilities, congenital malformations, metabolic, neurologic and other diseases of childhood, as well as common chronic diseases of adulthood all have a genetic component and constitute a major health burden for Michigan’s citizens. In addition, technology continues to advance, moving from the research setting into health care delivery systems. Medical, public health, and human service professionals face new challenges in helping society uphold appropriate use of genetic information through policies and programs to promote health and prevent disease. “Public Health Genetics” spans a wide array of disciplines and represents an unprecedented opportunity to effectively target biological, behavioral and environmental factors leading to morbidity and mortality, based on new understanding of the human genome. Having a strategic plan helps to focus our efforts and maximize the use of existing resources to better address the most pressing issues.

“Human genetic variation is associated with many, if not all, human diseases and disabilities, including the common chronic diseases of major public health impact”

--Muin Khoury et al ¹

How Were the Issues Identified?

The needs assessment process collected both qualitative and quantitative data using a variety of techniques: a review of literature and other state plans, key informant interviews, focus groups, survey questionnaires, and expert working groups. The purpose of the needs assessment was to define genetic health service priorities for Michigan—as seen by a broad array of stakeholders—for all four stages of the life cycle: prenatal, newborn, childhood and adulthood. The stakeholders included advocacy organizations; consumers; educators; funding sources; general public; genetic service providers; health professional training programs; health care providers; industry; media; mental health and developmental disability program providers and clients; policy makers; and research scientists. Overall, nearly 1,000 people—individual citizens, as well as those representing numerous different public and private organizations and agencies—participated in the consensus process.

¹ Khoury MJ, Burke W, Thomas EJ (eds). *Genetics and Public Health in the 21st Century: Using Genetic Information to Improve Health and Prevent Disease*, New York: Oxford University Press, 2000.

What Were the Key Findings of the Needs Assessment?

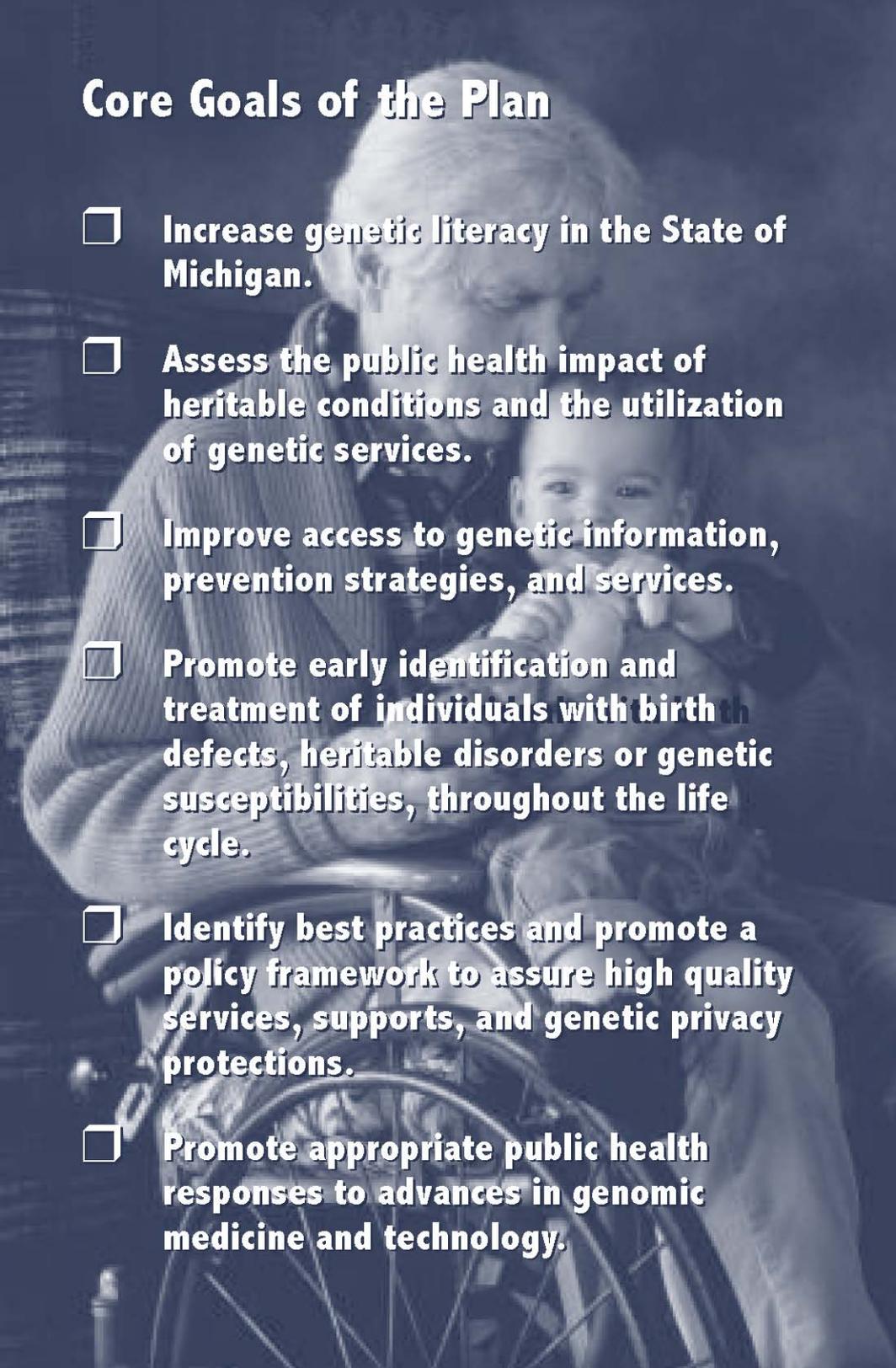
There were many important conclusions, with a great deal of consensus across all stakeholder populations. The key findings were summarized into seven areas²: organization and administration, prevention, available services, research, education, data collection and documentation, and funding. The findings highlight the need to:

- Increase the visibility of the state genetics and newborn screening program;
- promote collaboration with local health departments;
- increase public and professional awareness of prevention strategies across the lifespan;
- increase early identification of disorders that benefit from treatment;
- reduce barriers to utilization of existing genetic services;
- nurture public interest and participation in genetic research;
- utilize existing public health data sets to increase understanding of birth defects and genetic disease;
- reduce the communication gap between gene-environment research and public health;
- educate all sectors of the population about the role of genetics in health and disease;
- increase capacity for assessment, planning and evaluation of genetic health care services;
- find ways to address reimbursement issues and demonstrate the cost-effectiveness of genetic services.

Who Developed the Plan?

Twelve expert working groups were convened and charged with identifying priority objectives using the data collected in the needs assessment, along with their own knowledge and expertise. Common themes among 52 work group objectives were summarized, and formulated into six overarching goals, along with relevant action steps. These recommendations were further reviewed by the Genetics Advisory Committee (GAC), as well as an internal public health Genetics Work Group and Birth Defects Steering Committee. The full plan was drafted, then offered for comment by the GAC and all work group members before administrative review and approval.

² Summary areas based on the Council of Regional Networks for Genetic Services (CORN) document, "Guidelines for Clinical Genetic Services for the Public's Health", 1997.



Core Goals of the Plan

- Increase genetic literacy in the State of Michigan.**
- Assess the public health impact of heritable conditions and the utilization of genetic services.**
- Improve access to genetic information, prevention strategies, and services.**
- Promote early identification and treatment of individuals with birth defects, heritable disorders or genetic susceptibilities, throughout the life cycle.**
- Identify best practices and promote a policy framework to assure high quality services, supports, and genetic privacy protections.**
- Promote appropriate public health responses to advances in genomic medicine and technology.**

What are the Goals?

The six core goals and their related objectives are:

1. Increase genetic literacy in the State of Michigan

A major and overwhelming theme through all components of the needs assessment was the importance of having a more informed, genetically literate public in order to maximize the use of genetic knowledge and technology to improve health and/or quality of life. A large segment of the population (45% of the general public in our survey) was not even aware of the worldwide Human Genome Project, in progress since 1990, let alone possible implications for the future of medicine and health care decision-making. Therefore, a major focus of the statewide genetic services system should be to educate the general public, consumers, and health and human service professionals about the role of genetics in health and disease.



Recommended objectives include:

- Expand public and provider knowledge regarding the impact of genetics on health
 - Integrate human genetics into curricula throughout the educational system
 - Increase awareness of genetic ethical, legal and social issues (ELSI) by educating health care professionals and the public
 - Develop avenues for communication about gene-environment issues between academic, public health, primary care professionals, and the public
- ### 2. Assess the public health impact of heritable conditions and the utilization of genetic services
- In order to determine the effectiveness of public health program initiatives, it is critical to understand the populations in need of services, as well as those currently utilizing available services. A new emphasis on developing, analyzing, and disseminating statistical public health information on birth defects and genetic disease should be pursued. Such data will be of value not only to public health programs but also to service providers and advocacy organizations.

Recommended objectives include:

- Improve the utilization of existing data sources for planning, implementing, and evaluating program activities
- Develop methods to assess the use of reproductive genetic services by individuals of childbearing age
- Develop and maintain systems to improve the accuracy and completeness of newborn screening data; establish linkages with vital records and other children's databases, in order to identify health services needed/received by high risk populations
- Improve the assessment and understanding of birth defects as a public health problem
- Develop methods to assess the public health burden of genetic/familial disease in the adult population
- Conduct public health surveillance and research regarding hereditary cancer in Michigan

3. Improve access to genetic information, prevention strategies, and services

A tremendous amount of genetic information is now available and some effective strategies for primary, secondary and tertiary prevention are known. A major concern identified by consumers was the need for improved access to genetic information, especially in underserved populations including rural and low-income areas. While many genetic disorders cannot be prevented from occurring, it is still important to prevent secondary or tertiary complications or disabilities to the greatest extent possible. Genetic specialty clinic services are currently available at seven medical centers and 10 outreach clinic sites throughout the state. However, public awareness of existing services is still relatively low.

Recommended objectives include:

- Establish a central genetic resource center to make information and resources more readily available to the public and providers
- Provide information to the public and professionals about known causes of birth defects and strategies for prevention
- Improve dissemination of information about resources and services to families of children with or at risk for birth defects and heritable disorders
- Assure availability of comprehensive genetics clinics throughout Michigan
- Explore strategies for financing genetic health care, testing and support services

- Assure availability of DNA testing for children with heritable disorders and their relatives
 - Increase referral of patients affected or at risk for conditions with a genetic component to appropriate genetic services regardless of ability to pay
4. Promote early identification and treatment of individuals with birth defects, heritable disorders or genetic susceptibilities, throughout the life cycle

Many people with or at risk for heritable disorders will benefit from early treatment, even if a cure is not possible. However, affected and susceptible individuals are not always recognized or diagnosed as early as possible, even when screening tests are available or family history suggests an increased risk. Promoting methods of early identification is an important role for the statewide genetic services system. Eighty-four percent of all survey participants (n=710) agreed that available resources should be used to expand screening programs for early identification of persons predisposed to genetic diseases who might benefit from early treatment or other interventions.

Local health departments recognized that chronic disease program areas including diabetes, cancer, cardiovascular, and obesity, would need to incorporate new genetics information over the next 3 to 5 years.



Recommended objectives include:

- Assure that all Michigan infants receive an initial newborn metabolic screen by 24-36 hours of age in accordance with guidelines established by MDCH; and identify all infants with positive screens by 6 days
- Provide appropriate follow-up, diagnosis, and treatment for infants with positive screening tests in accordance with nationally recognized guidelines
- Expand the newborn screening program to reflect current technological advances including tandem mass spectrometry
- Assure early identification, evaluation and genetic counseling/education for all children with birth defects, heritable disorders and developmental delay

- Reduce the public health burden related to preventable chronic diseases with a significant genetic component
- Reduce morbidity and mortality related to hereditary cancer by increasing utilization of appropriate cancer risk assessment services
- Monitor developments in current knowledge about gene-environment interactions of potential public health relevance for the Michigan population

5. Identify best practices and promote a policy framework to assure high quality services, supports, and genetic privacy protections

A variety of policy issues related to promoting best practices and assuring high quality services are addressed through goal five.

Assuring the quality of services available to the public is an important role for public health. Advocacy organizations and consumers reported that proper assessment or optimal care for their genetic condition is not always received during health visits. Potential health risks related to gene-environment interactions are complex

and not easily understood. There is also concern among both the public and providers about the possibility of discrimination occurring on the basis of a genetic predisposition.

Current Michigan “genetic privacy” laws require written informed consent before genetic testing; prevent the use of genetic test results as a basis for health insurance coverage or renewal; and prohibit workplace discrimination based on genetic information unrelated to a person’s ability to perform the duties of a particular job.

Recommended objectives include:

- Promote genetic competencies among health care professionals serving Michigan residents
- Assure quality of genetic laboratory testing in Michigan
- Assure quality and availability of clinical reproductive genetic services statewide and disseminate consensus guidelines for reproductive genetic health care
- Implement the recommendations of the Governor’s Commission on Genetic Privacy regarding retention and storage of newborn screening dried blood spot samples

- Assure privacy protections for reporting newborn screening results to physicians, medical management centers, and others
- Evaluate the effectiveness of the newborn screening program using identified outcome measures
- Develop and test a consensus diagnostic approach for genetic evaluation of developmental delay and mental retardation

6. Promote appropriate public health responses to advances in genomic medicine and technology

It is important for the public health community to stay abreast of scientific discoveries, in order to interpret the significance of new findings for the public and incorporate state-of-the-art knowledge into health promotion and disease prevention activities. Public

The challenge facing public health is to find practical applications for genomics “today” while building knowledge, experience, and capacity to prepare for the breakthroughs of “tomorrow”

health plays an important role in facilitating statewide dialogue and collaboration to assure appropriate integration of genomic medicine and technology throughout public and private health care systems.

Recommended objectives include:

- Promote integration of public health genomics within MDCH and other relevant state and local agencies
- Enhance communications with genetic service providers and promote partnerships with relevant stakeholders
- Identify funding opportunities to increase state and local public health capacity to respond to current and emerging technical and administrative needs relative to a comprehensive statewide genetics and newborn screening program
- Ensure an adequate workforce by promoting awareness of careers in genetics for interested individuals
- Designate a central biochemical laboratory to provide confirmatory testing on infants with positive newborn screens and other biochemical genetic disorders
- Ensure prompt and appropriate state response regarding national recommendations for voluntary population-based genetic screening
- Address the identified public health risks related to gene-environment interactions and assure the public has access to appropriate information and resources

The Next Steps

How can the goals be achieved? This strategic plan, “Genetics Through the Life Cycle: Improving Health and Preventing Disease”, represents an opportunity to better understand the public health impact of gene variants on disease, death, and disability within our own state, as well as to define the role of public health in the genetic health care delivery system. The goals will be achieved by taking action to accomplish specific objectives recommended by the expert working groups and the Genetics Advisory Committee. Much progress can be made with existing resources— through increased dialogue, collaborations with partner organizations, and federal grant initiatives, in addition to simply incorporating a new “public health genetics perspective” into currently funded program activities. To accomplish certain objectives, additional funding is needed and will be identified as new opportunities become available. The plan serves as an important blueprint for mobilizing the resources and partnerships needed to advance a new vision of genetics in public health.

For more information or a copy of the complete needs assessment and plan, call the MDCH state genetics program at (517) 335-8110 or e-mail genetics@michigan.gov

Michigan Department
of Community Health



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