A Needs Assessment and State Plan for Michigan

Genetics Through the Life Cycle: Improving Health and Preventing Disease

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Michigan Department of Community Health

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"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

EXECUTIVE SUMMARY

This report recommends a framework for a genetics action plan for the State of Michigan, 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. Many of the plan's action steps are already being pursued using current resources and collaborations, whereas additional funding will need to be identified in order to fully achieve all of the recommended objectives.

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. Although many Michigan residents have benefited over the last 25 years from available genetic evaluation and counseling services, the number is small compared with the millions potentially at risk for rare disorders and common, complex conditions. 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 designed 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.

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 genetics as well as chronic disease plans, key informant interviews, focus groups, survey questionnaires, and expert working groups. The Council of Regional Networks for Genetic Services (CORN) document, "Guidelines for Clinical Genetic Services for the Public's Health", served as the basis for topics to be explored, including: organization and administration; prevention; available services; research; education; data collection and documentation; and funding. The goal 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. Major sectors of the population thought to influence or be impacted by a state genetics plan were identified, and attempts were made to include those perspectives in the needs assessment process. Such stakeholders include: 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.

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.

Who Developed the Plan?

Twelve expert working groups were convened and charged with developing priority objectives using the data collected in the needs assessment, along with their own knowledge and expertise. Common themes among 52 identified 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 plan was written, then offered for comment by the GAC and all work group members before administrative review and approval.

What Are the Goals?

The six core goals of the genetics plan for Michigan are to:

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

How Can the Goals Be Achieved?

"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, a vision that will lead to improved health outcomes and enhanced quality of life for Michigan families through appropriate use of genetic information, technology and services.

ACKNOWLEDGMENTS

The state genetics plan for Michigan is the result of many months of collaborative effort by nearly 1,000 individuals. A special acknowledgment and heartfelt thank you is extended to everyone - key informant interviewees, focus group participants, survey respondents, work group members, advisory and steering committee members, reviewers, and staff - who gave their time and insight to shape this consensus plan.

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INTRODUCTION

Major advances in the science of medical genetics have occurred at an unprecedented rate over the last two decades. The worldwide **Human Genome Project,** initiated in 1990 to map and sequence the human genome, has been an important catalyst in elucidating the genetic basis of human disease. New molecular diagnostic and treatment technologies, derived as a result of the Human Genome Project and ongoing basic genetic research, are expected to dramatically shift the focus of health care from late stage treatment to prevention and early stage diagnosis over the next several years. Moreover, it may become feasible, even efficacious, to target public health monitoring efforts and preventive measures at populations that share increased susceptibility to certain diseases based on genotype or specific environmental exposures. The plan outlined in this document is intended to help the State of Michigan anticipate changes in health care infrastructure based on an evolving recognition of the role genes play in health and disease.

Rapid technological changes are presently outpacing our ability to educate the public and professionals about the health implications of genetic discoveries. At the same time, our most vulnerable citizens are tempted to believe media reports of the benefits of new tests or treatments for genetic disorders that may be greatly exaggerated. There is an important role for public health in providing accurate information and a balanced view of genetic technology. Former Governor John Engler recognized the significance of these advances and the concomitant need to address ethical, legal, and social issues by appointing an 11- member Commission on Genetic Privacy and Progress in 1997. Many of the commission's recommendations were enacted as genetic privacy legislation in 2000.

State Program History

A mandated newborn screening program, coordinated by the Michigan Department of Community Health (MDCH), has been in place since 1965, when a filter-paper test to screen babies for phenylketonuria (PKU) first became available. Infants are now screened for six additional disorders: congenital hypothyroidism, galactosemia, biotinidase deficiency, maple syrup urine disease (MSUD), hemoglobinopathies, and congenital adrenal hyperplasia (CAH). The program is funded by charging a fee ($42.61 as of October, 2002) for the filter paper card used to screen each infant. A comprehensive approach provides follow-up of positive screens and assures medical management for infants diagnosed with these disorders. Since the program began, more than four million Michigan babies have been screened. The state genetics program was first established by MDCH with federal funding in 1978 as a result of the National Genetic Diseases Act. The program has evolved over the past 25 years and currently consists of a state genetics coordinator, a few staff members funded by federal grants, and contractual agreements with regional genetics centers. During 2001, about 3,000 individuals and their families received genetic clinic services through the regional network supported in part by MDCH. A cooperative agreement with the Centers for Disease Control and Prevention (CDC) since 1999 has allowed development of a program component to promote birth defects prevention and examine ways to assure that infants and toddlers reported to the Michigan Birth Defects Registry (MBDR) are linked with available services. A four-year grant from the Health Resources and Services Administration (HRSA) will provide funding to implement many of this plan's recommendations related to early identification and newborn screening quality assurance. In addition, a new focus on the genetic impact of common adult-onset chronic diseases has been initiated in collaboration with the MDCH Division of Chronic Disease and Injury Control.

Public Health Impact

Genes impact Michigan's public health in significant ways, yet the role of genetics in health and disease is often not appreciated. During 2001, 15 children with PKU (seven requiring diet treatment) were identified through newborn metabolic screening. The program also found one child with MSUD, three with classical galactosemia, eight with biotinidase deficiency, 113 with

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.

congenital hypothyroidism, nine with congenital adrenal hyperplasia, and 93 with sickle cell anemia, for a total of 17.8 per 10,000 births. The exact prevalence of other genetic conditions in the state is not available, but efforts have been initiated to view existing public health data sets through a genetic lens in order to better characterize the public health impact of genetic disease. Initially, this effort will focus on five sentinel conditions: cystic fibrosis, Down syndrome, spina bifida, hereditary hemochromatosis, and hereditary breast cancer.

About 8,000 children from birth to two years of age are reported to the MBDR annually. The largest category of congenital malformations is related to the heart and circulatory system, with 1,884 children reported in 1999 for an incidence rate of 142.6 per 10,000 live births. More than 50 percent of congenital deafness is genetic in origin and 172 infants were identified with confirmed hearing loss through Michigan's community-based voluntary newborn hearing screening program during the year 2000. Other sources of data include the Children's Special Health Care Services Plan for children with chronic illnesses or other special health needs, and **Early On®** (EO), Michigan's early intervention system. During fiscal year 2000-2001, CSHCS covered medical care and treatment expenses for 27,386 beneficiaries under age 21 at a cost exceeding $96 million. EO provided services to 12,998 infants and toddlers with special needs from birth to three years of age during the year 2000.

Common Chronic Diseases

The important role of genes in the etiology of common, usually adult-onset, chronic diseases is now being recognized. As the nation's population demographics shift, diseases of the elderly will become proportionately more significant and costly to the public health care system over the next 20 years. The use of pharmacogenetics to personalize medicine - by reducing adverse drug reactions, for instance - will become an important tool for reducing health care costs. Of the ten leading causes of death in Michigan last year, at least nine are known to have a genetic component. For instance, genetic factors are important in the development of cardiovascular disease. As the leading cause of death in Michigan and the United States, heart disease is estimated to incur annual health care costs of nearly $300 billion nationwide. About 10 percent of all cancers result from an inherited susceptibility - and multiple genetic predisposition syndromes have already been described for breast, ovarian, colorectal, and prostate cancer. Numerous others - including pancreatic, bladder and lung cancers - are currently under investigation. Stroke, a complex condition involving a combination of genetic and environmental factors, is a leading cause of long-term disability today. Respiratory disease is the result of a number of factors: lifestyle choices such as smoking and environmental exposures, along with an underlying genetic susceptibility. Genetic factors account for about 30 percent of the risk for developing diabetes, which can lead to significant disability including blindness, heart disease, kidney failure and amputation. Although more knowledge is still needed in the area of infectious disease, genetically mediated host susceptibility is an important factor in a person's response to infectious organisms. Several genes for Alzheimer's disease, the most common cause of dementia in older individuals, have now been discovered. Finally, genetic diseases such as polycystic kidney disease and Alport's syndrome contribute to illness and deaths from renal failure.

National Initiatives

Numerous national initiatives are now under way to assist states with integrating genetics throughout public health, while continuing to improve existing programs such as newborn screening and birth defects surveillance. The Office of Genomics and Disease Prevention (OGDP) was established in 1998 at the CDC. Three national conferences on genetics and public health have been held since 1998. In 2001, three centers for genomics and public health were established at academic schools of public health, including the University of Michigan, by the CDC through the Association of Schools of Public Health. The Association of State and Territorial Health Officials has developed a framework for genetics in relation to the ten essential public health services, and is in the process of developing a genomics

"Today, the mounting accomplishments of the Human Genome Project demand that we re-think the role of genomics in every condition of public health interest. We must strengthen the effectiveness of public health interventions by more fully incorporating knowledge of internal host-specific factors and their interactions with environmental exposures."
--Beskow, et. al

Genetic Factors Contribute to the Leading Causes of Death* Among Michigan Residents

1. *Heart disease*
2. *Cancer*
3. *Stroke*
4. *Chronic lower respiratory diseases*
5. *Unintentional injuries*
6. *Diabetes mellitus*
7. *Pneumonia/influenza*
8. *Alzheimer's Disease*
9. *Kidney disease*
10. *Septicemia*

* *MDCH Vital Records Data for Year 2000*

resource tool kit for states to use. The Health Resources and Services Administration (HRSA) has allocated funding to states for genetic and newborn screening infrastructure planning and implementation of state genetic plans. The CDC has established eight centers for birth defects research and provides funding for states to improve their birth defects registries and use surveillance data for public health prevention and intervention programs. In January 2002, the Association of State and Territorial Chronic Disease Directors convened a "Genomics and Chronic Disease Summit" in conjunction with the CDC's National Center for Chronic Disease Prevention and Health Promotion and OGDP. Michigan has been fortunate to benefit from such opportunities and will continue to participate in federal initiatives that provide funding and/or programmatic assistance.

Since the inception of the state genetics program more than two decades ago, efforts have been made to establish and maintain relationships with relevant partners, including public and private health and intervention service providers. Input from clinical genetics providers, anecdotal reports from the field, and discussions with other states have served as the basis for program objectives designed primarily to serve families affected by relatively rare genetic disorders and birth defects. As the underlying genetic etiology of virtually all chronic diseases of public health significance comes to light, there is a new recognition that genetics truly affects everyone in the population throughout the life cycle. Moreover, the potential for using genetic knowledge to improve health and prevent disease is largely untapped. A more comprehensive approach to genetic assessment, policy development and assurance is recommended. By focusing on strategic objectives delineated in this state plan, Michigan can begin to address many of the identified needs - including the overwhelming need for increased genetic literacy - among all populations within our state.

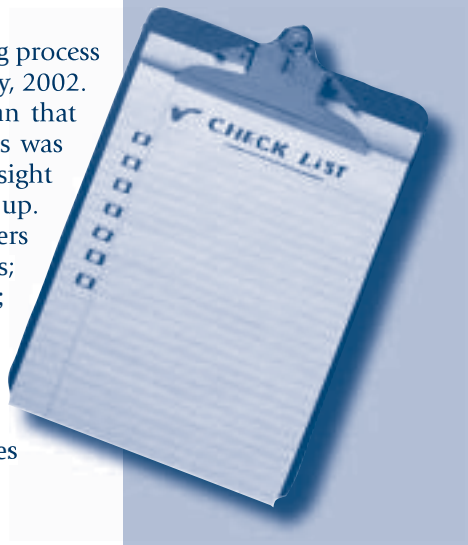
Key milestones impacting genetic health research and service delivery

1908 Archibald Garrod describes rare inborn errors of metabolism
1953 Watson and Crick elucidate the structure of DNA
1956 Tjio and Levan describe the correct number of human chromosomes
1965 Guthrie develops a test for PKU and Michigan initiates newborn screening
1975 Congress enacts Genetic Disease Act
1977 Michigan adds congenital hypothyroidism to NBS Program
1978 Michigan Department of Public Health establishes Genetic Services Program
1984 Michigan adds galactosemia to NBS panel
1987 Michigan adds biotinidase deficiency, MSUD and sickle cell anemia to NBS panel
1987 Legislation establishes the Michigan Birth Defects Registry
1990 Human Genome Project begins
1991 Medical genetics becomes a certified specialty
1992 U.S. Public Health Service recommends folic acid for women of childbearing age to prevent neural defects
1998 CDC establishes Office of Genetics and Disease Prevention
1998 Congress enacts Birth Defects Prevention Act
1998 HHS Secretary establishes Advisory Committee on Genetic Testing
1999 CDC hosts first national conference on genetics and public health
1999 Michigan receives CDC cooperative agreement for birth defects surveillance
1999 Governor Engler appoints Commission on Genetic Privacy and Progress
1999 Congress enacts Children's Health Act
2000 Michigan enacts "Genetic Privacy" laws
2000 Michigan receives HRSA genetics planning grant
2000 Human Genome Project is nearly complete
2000 President Clinton issues executive order prohibiting genetic discrimination in federal employment
2000 CDC establishes National Center on Birth Defects and Developmental Disabilities
2001 University of Michigan establishes Center for Genomics and Public Health
2002 Legislation authorizes addition of MCAD to NBS panel starting April 1, 2003

Today Michigan screens newborns for eight disorders in addition to hearing loss.
Today The Birth Defects Registry contains about 200,000 case reports on 105,000 children.
Today More than 10,000 patients and their families receive genetic services annually

DEVELOPMENT OF THE STATE PLAN

Needs Assessment



The genetics plan for Michigan is based on a statewide needs assessment and planning process (Appendix A) conducted over an 18-month period from August, 2000 through January, 2002. A collaborative approach was used to formulate a comprehensive, yet strategic plan that addresses genetic issues of public health concern through the life cycle. The process was coordinated by a planning grant manager and the state genetics coordinator with oversight by the MDCH Genetics Advisory Committee (GAC) and internal genetics working group. Close to 1,000 Michigan residents representing a wide and diverse range of stakeholders were involved: persons with genetic conditions or birth defects, and their families; primary and specialty care providers; clinical and laboratory genetic service providers; educators; advocacy organizations; public health administrators; and the general public, including union members, church members, and students. Demographic data on the survey participants are included in Appendix B. The mother of a teenager with special needs who also serves as a support group facilitator was hired as a consumer consultant for the project and provided valuable insight through all phases of the planning process.

Planning Process

The planning process included five steps: 1) a review of relevant literature and other state genetic and chronic disease plans; 2) key informant interviews; 3) five focus group meetings; 4) development and dissemination of survey questionnaires to seven different target populations; and 5) convening 12 expert work groups that focused on: birth defects surveillance, cancer genetics, data and evaluation, finance and reimbursement, gene-environment interaction, genetic literacy and education, genetic health services-adult, genetic health services-pediatric, genetic health services-reproductive, laboratory services, newborn screening and policy. Each work group was asked to review available data pertinent to their area of expertise and develop three to five priority recommendations. A total of 52 priority goals and objectives were identified through this process. Recurring themes were summarized by the project team and reduced to six major goals accompanied by objectives and recommended action steps. The goals were reviewed and approved by the GAC. Subsequently, the full plan was submitted in draft form to the GAC as well as to the internal genetics group and MDCH Birth Defects Steering Committee. It was also reviewed by several members of the expert working groups who wished to provide further comment. A second draft incorporated the comments received in the initial review process and was later reviewed by neighboring state genetic coordinators and submitted to the Genetic Services Branch.

Key Findings

Key findings from the needs assessment can be framed in terms of seven core areas outlined in the *Guidelines for Clinical Genetic Services for the Public's Health* by the *Council of Regional Networks for Genetic Services* in 1997. These areas include: a) organization and administration; b) prevention; c) available services; d) research; e) education; f) data collection and documentation; and g) funding. Both quantitative and qualitative data were collected and analyzed in order to identify the major needs summarized below.

- **Organization and Administration:** There is a need to enhance the visibility of the state genetics and newborn screening program, assign additional personnel as funding becomes available, and promote collaboration with local health departments as the role of public health in the genetic health care infrastructure expands.
- **Prevention:** There is a need to increase public and professional awareness of primary prevention strategies including genetic risk assessment related to adult-onset chronic disease. There is a need for earlier identification of heritable disorders in children and adults who could benefit from secondary prevention measures. Utilization of available services and supports by individuals with genetic health needs could be increased as a tertiary prevention strategy.

- **Available Services:** Barriers to utilization of genetic services need to be reduced. The quality and availability of prenatal genetic screening services vary statewide. There is a need to align the newborn screening system with national recommendations and increase in-service training for hospitals and pediatric providers. Specialized cancer risk assessment and genetic counseling services are not currently available in geographically remote regions of the state. There is no biochemical genetics reference laboratory in the state.
- **Research:** There is a need to nurture public interest and participation in genetic research. Currently, existing public health data sets are not being used to increase understanding of the impact of birth defects and genetic disease. There is a wide gap in communication between gene-environment research and public health.
- **Education:** There is a tremendous need to educate all sectors of the population about the role of genetics in health and disease, including related ethical, legal and social issues. A central source is needed as a portal for the public to obtain reliable information about genetic disorders, resources and services.
- **Data Collection and Documentation:** There is a need to increase capacity for assessment, planning and evaluation of genetic health care services based on available data sources.
- **Funding:** Reimbursement for clinical genetic services, as well as clinical research, is an ongoing challenge. Out-of-state reference laboratories frequently do not accept Michigan Medicaid or other insurance plans. Methods of demonstrating the cost-effectiveness of genetic diagnosis, testing and counseling are needed.

A detailed description of the needs assessment process is presented in Appendix A and the results are summarized more fully in Appendix B.

THE STATE GENETICS PLAN: STRATEGIC GOALS AND OBJECTIVES

Goal One

❒ Increase genetic literacy in the State of Michigan

Genetic literacy implies the ability to understand, interpret, and apply genetic information to health and lifestyle decisions and to the ethical, legal, and social issues faced by individuals and society.

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 quality of life. A large segment of the population (45 percent of the general public in our survey) was not even aware of the worldwide Human Genome Project, now in progress for 12 years, 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.

Objectives

1. **Expand public and provider knowledge regarding the impact of genetics on health**

   - Create a genetic literacy campaign targeting the general public to dispel myths and misconceptions, as well as increase recognition of the role of genetics in health and the benefits of genetic services
   - Explore cost-effective methods of providing genetics education to health care providers
   - Make information on underlying genetic causes of common chronic diseases, co-morbidity, and the importance of early detection more readily available to consumers and providers, including those who care for adults with developmental disabilities of genetic origin.

2. **Integrate human genetics into curricula throughout the educational system**

   - Suggest new avenues to increase the teaching of human genetics at all levels.
     - Establish a summer training program for K-12 teachers
     - Collaborate with the Department of Education to examine the current science curriculum
     - Identify or develop a model undergraduate course on general human genetics
   - Foster expansion of professional school curricula
     - Form working groups to identify methods of integrating genetics into medical school and residency training programs; baccalaureate, master's, and doctoral level nursing programs (including nurse midwifery)
     - Assess unmet training needs for allied health care providers, social workers, and other professions such as dentists, psychologists, and the clergy.

"A diverse group of educators, parents, community leaders, and citizens should jointly decide the precise form of genetics education in the school system. This education should include both science issues and the ethical, legal and social implications of genetics research and technology."
-- Recommendation from the Communities of Color Project

"I've had employment discrimination. I didn't run around telling everyone I had a genetic disease, but I didn't exactly hide it either. I got fired from my job."
--A focus group participant

3. **Increase awareness of genetic ethical, legal and social issues (ELSI) by educating health care professionals and the public**

   - Inform citizens about current social issues and their legal rights with regard to genetic privacy and discrimination
   - Provide more information about Michigan's genetic laws to the public, with specific attention to educating professionals about their obligations under the informed consent law
   - Educate practitioners and researchers that identifying information collected on third parties should be limited to assure protection of genetic privacy when medical/genetic information is obtained for clinical or research purposes

4. **Develop avenues for communication about gene-environment issues between academic, public health, primary care professionals, and the public**

   - Identify stakeholders for gene-environment issues such as union health and safety committees, biomonitoring projects and occupational health workers
   - Develop methods of linking stakeholders with sources of specialized information pertaining to genetics and various environmental exposures

Goal Two

❒ 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.

Objectives

1. **Improve the utilization of existing data sources for planning, implementing, and evaluating program activities**

   - Increase interactions with epidemiologists to strengthen infrastructure and capacity for data analysis
   - Use existing databases to assess linkages to care for both children and adults with selected genetic conditions and to evaluate progress with respect to Healthy People 2010 and other national public health objectives

2. **Develop methods to assess the use of reproductive genetic services by individual of childbearing age**

   - Develop methodologies for tracking the use of genetic screening and diagnostic services
   - Assess the needs of medically underrepresented populations
   - Examine the feasibility of tracking birth outcomes associated with assisted reproductive technologies

3. **Develop and maintain systems to improve the accuracy and completeness of newborn screening data and establish linkages with vital records and other children's databases, in order to identify health services needed or received by high risk populations**

- Utilize fully existing public health data systems, including the MDCH data warehouse under development, to assess utilization of public health genetic/newborn screening health services
- Track specific health care services received by high risk populations, such as immunizations for children with sickle cell anemia and WIC eligibility for infants with PKU

4. **Improve the assessment and understanding of birth defects as a public health problem**

- Use the Michigan Birth Defects Registry for epidemiological analyses of selected birth defects including incidence by socioeconomic status, trends over time, a map of selected conditions by county, and recurrence to the same mother
- Explore the feasibility of linking databases containing information on maternal chronic conditions or exposures to birth defects data
- Strengthen local interest and investment in birth defects surveillance, prevention and intervention issues through connections with community health assessment advisory groups and county multi-purpose collaborative boards
- Improve hospital and cytogenetic laboratory understanding of the importance of submitting accurate and timely case reports
- Develop and maintain systems for collecting and reporting accurate data to monitor the prevalence of neural tube defects and preconceptional use of folic acid in conjunction with federal reporting requirements

5. **Develop methods to assess the public health burden of genetic/familial disease in the adult population**

- Design pilot studies to examine mortality related to specific genetic conditions and assess the costs of medical care for selected genetic conditions and related disorders
- Add one or more questions to assess public knowledge and utilization of genetic services to a statewide population-based survey
- Examine issues related to transition from pediatric to adult health care systems for young adults with developmental disabilities, heritable disorders and birth defects, and address barriers to continuity of care for this population

6. **Conduct public health surveillance and research regarding hereditary cancer in Michigan**

- Examine demographic patterns of hereditary cancer
- Test the feasibility of merging existing local cancer genetics registries, in order to determine the value of having a single registry
- Document the current infrastructure and capacity for delivering cancer genetics risk assessment services

Goal Three

❒ Improve access to genetic information, prevention strategies, and services

An overwhelming 95 percent of all survey respondents felt that available resources should be used to assure that anyone who needs genetic evaluation or counseling has access to it.

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.

Objectives

1. Establish a central genetic resource center to make information and resources more readily available to the public and providers

- Assign adequate professional, administrative and support staff to effectively conduct public relations activities and respond to requests for information
- Include information such as existing genetic disease-specific management and health surveillance protocols, support groups, clinical services, websites and laws
- Disseminate a directory of qualified genetic service providers
- Increase the availability of culturally sensitive, educationally appropriate and scientifically accurate information about genetic conditions, risks and services

2. Provide information to the public and professionals about known causes of birth defects and strategies for prevention

- Employ a variety of approaches to increase awareness of birth defect prevention strategies among women of childbearing age in order to reduce the rate of neural tube defects in Michigan
- Create a targeted campaign for high risk groups based on public health survey data
- Increase collaboration with the Michigan Teratogen Information Service in order to assure that information about known risks associated with various prenatal exposures is available to those who need it

3. Improve dissemination of information about resources and services to families of children with or at risk for birth defects and heritable disorders

- Provide uniform information to all families of children with a genetic diagnosis
  - Formulate protocols for information distributed by pediatric genetic centers and newborn screening medical management programs
  - Disseminate information to all pediatricians, family physicians, pediatric and family nurse practitioners, and nurse midwives
- Assess unmet needs and utilization of existing brochures by hospital social workers and neonatal intensive care units
- Identify best practice community referral guidelines based on the findings of a birth defects registry study conducted in various geographic regions
- Assemble best practice guidelines for health surveillance and management of children with or at risk for heritable disorders

4. Assure availability of comprehensive genetics clinics throughout Michigan

- Maintain a network of outreach genetics clinics to underserved geographic regions
- Identify outcome measures to demonstrate the effectiveness of genetic services
- Determine actual costs per patient seen for different types of caseloads at both center-based and outreach clinic sites
  - Compare costs with sources of support through billable activities, and implement appropriate billing practices
  - Assure continued viability of statewide clinical services by providing supplemental financial support as needed

5. Explore strategies for financing genetic health care, testing and support services

- Explore avenues for improving third party coverage and reimbursement
  - Identify liaisons with major third party payers and Medicaid
  - Educate health insurance plans and providers about the value of genetic services
  - Educate genetic and specialty clinic providers about the billing and reimbursement process
  - Evaluate current reimbursement practices for genetic laboratory tests and establish a schedule for periodic review
  - Identify new strategies for public and private funding of genetic services and related needs for individuals and families
- Explore possible funding sources to subsidize services for patients without insurance or ability to pay

6. Assure availability of DNA testing for children with heritable disorders and their relatives

- Examine Medicaid and qualified health plan practices, billing codes and reimbursement for molecular genetic testing
- Explore the feasibility of establishing an agreement with a single reference laboratory to accept Michigan Medicaid for tests not available in-state

7. Increase referral of patients affected or at risk for conditions with a genetic component to appropriate genetic services regardless of ability to pay

- Educate consumers and health care providers to increase recognition of the genetic components of disease and appropriate interventions

"Once I met the doctor who provided me with information about genetic services, things have gone real well for me and my family. It's getting to that point that takes a lot of work…"
--A focus group participant

Goal Four

❒ 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 three to five years.

Objectives

1. **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**

   - Develop a comprehensive program to provide education and in-service training about the newborn screening process to hospitals, physicians and midwives who deliver infants
   - Establish a system to link the newborn screening database with vital records on a continual basis in order to identify unscreened infants in a timely manner

2. **Provide appropriate follow-up, diagnosis and treatment for infants with positive screening tests in accordance with nationally recognized guidelines**

   - Identify core resources needed to comply with national recommendations, including: medical and laboratory personnel; facilities; medical foods, formulas and supplements; medication; and information systems
   - Maintain a network of designated medical management centers of excellence to assure access to treatment services for infants diagnosed through newborn screening
   - Assure that infants with positive screening tests are linked with a medical home for provision of health care

3. **Expand the newborn screening program to reflect current technological advances, including tandem mass spectrometry**

   - Develop general criteria for the inclusion of new neonatal screening tests
   - Consult experts to identify required resources and establish guidelines for an expanded screening program
   - Conduct a pilot study to assess the efficacy of the proposed program in reducing morbidity and mortality
   - Provide the resources needed for all components of the program including follow-up, confirmatory testing and medical management
   - Implement fully an expanded screening program based on the results of the pilot study

4. **Assure early identification, evaluation and genetic counseling or education for all children with birth defects, heritable disorders and developmental delay**

- Identify and promote guidelines for referral through partner programs such as Early On, the Michigan Early Hearing Detection and Intervention Program, CSHCS, and other systems of identification
- Provide genetic counseling to all families of children with PKU, MSUD, biotinidase deficiency, galactosemia and CAH
- Assess utilization of sickle cell trait counseling and barriers to participation

In the year 2001, 132,092 initial screening tests were performed on Michigan newborns.

5. **Reduce the public health burden related to preventable chronic diseases with a significant genetic component**

- Promote the use of family history for genetic risk assessment of common chronic conditions
- Collaborate with national, state and local initiatives sponsored by the CDC, HRSA, chronic disease directors, Michigan Center for Genomics and Public Health, MDCH Chronic Disease Program and others to identify and apply emerging recommendations to public health programs

6. **Reduce morbidity and mortality related to hereditary cancer by increasing utilization of appropriate cancer risk assessment services**

- Develop best practice guidelines for provision of cancer genetic services
- Disseminate standards to primary care providers, payers and the public
- Reduce barriers to access and utilization by increasing knowledge and addressing patients' rights, reimbursement issues, geographic availability, and genetic testing in clinical and research settings

7. **Monitor developments in current knowledge about gene-environment interactions of potential public health relevance for the Michigan population.**

- Define crucial or special factors in Michigan and describe mechanisms for identifying populations at risk based on specific environmental exposures
- Monitor new developments in the collection of biological samples, assay methods and interpretation of knowledge, including ethical considerations and consequences
- Establish a steering committee with designated liaison or staff person to coordinate recommended program activities regarding gene-environment issues
- Increase awareness of the importance of documenting environmental exposures along with family history information

Only 20 percent of primary, specialty, and local public health providers were aware of the Michigan genetics privacy laws passed in 2000.

Goal Five

❒ 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 a state genetics system. 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.

Recommendations

1. **Promote genetic competencies* among health care professionals serving Michigan residents**

   - Identify methods to increase awareness of published genetic competencies for health care professionals
   - Incorporate competencies into public health training and in-service presentations

   * as defined by the National Coalition for Health Professional Education in Genetics and the CDC

2. **Assure quality of genetic laboratory testing in Michigan**

   - Describe current mechanisms for validating clinical genetic tests and document existing types of proficiency tests and inspections
   - Design a pilot study to identify limitations of the system and address gaps through development of supplementary evaluation methods
   - Assess laboratory compliance with existing professional recommendations regarding population-based screening
   - Explore the need for, and ways to enhance, communication among genetic laboratory personnel to increase collaboration and maintain competencies

3. **Assure quality and availability of clinical reproductive genetic services statewide and disseminate consensus guidelines for reproductive genetic health care**

   - Form an expert working group to identify existing protocols and/or develop guidelines related to birth defects prevention, carrier testing and prenatal screening
   - Promote use of the guidelines by primary and specialty health care providers serving women of reproductive age, in order to increase utilization of birth defect prevention strategies and appropriate reproductive genetic screening techniques statewide
   - Designate regional referral centers of excellence to:
     - Identify best practice guidelines for medical management of common genetic conditions and birth defects diagnosed prenatally
     - Assist primary care providers in assuring appropriate follow-up of abnormal prenatal tests
     - Provide genetics training for obstetric/gynecological office staff
     - Disseminate standardized resource materials

4. **Implement the recommendations of the Governor's Commission on Genetic Privacy regarding retention and storage of newborn screening dried blood spot samples**

   - Continue to retain samples in a manner that preserves their integrity for DNA or other types of analysis
   - Restrict access to newborn screening specimens to research approved by institutional review boards and other analyses per MDCH policy
   - Consider development of an appropriate fee structure or handling charge for access to the specimens

5. **Assure privacy protections for reporting newborn screening results to physicians, medical management centers and others**

   - Establish standard protocols to ensure that confidentiality is protected when reporting or storing NBS results

6. **Evaluate the effectiveness of the newborn screening program using identified outcome measures**

   - Identify optimal health and developmental outcomes for each disorder included in the newborn screening panel
   - Assess the benefits of newborn screening to the patient, family and society
   - Implement procedures for ongoing evaluation

7. **Develop and test a consensus diagnostic approach for evaluation of developmental delay and mental retardation**

   - Convene an expert working group, including pediatric geneticists, developmental pediatricians and neurologists, to design a protocol for the initial evaluation of children with developmental delay
   - Pilot implementation and assessment of the protocol's effectiveness at one or more sites
   - Disseminate results and promote widespread implementation if proven to be well received by families and cost effective

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.

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"

Goal Six

❒ 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 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.

Objectives

1. Promote the integration of public health genomics within MDCH and other relevant state and local agencies

- Facilitate activities necessary to achieve the goals of the state genetics plan through collaboration with partner agencies, organizations and programs
- Maintain a state genetics advisory committee and relevant subcommittees
- Establish and maintain partnerships with relevant local, state and national projects
- Increase visibility of the current state genetics/newborn screening unit
  - o Develop a mission statement to define program activities
  - o Identify marketing strategies to create a program image that encompasses the expanding role of genetics in public health

2. Enhance communications with genetic service providers and promote partnerships with relevant stakeholders (disease organizations, local providers, local public health, etc.)

- Examine the role of the regional genetics network and enhance capacity for community partnerships and educational outreach
- Facilitate bi-annual meetings for genetics counselors, an annual statewide genetics symposium, and other work groups or task forces as needed to address specific issues
- Form a new organization of cancer genetics professionals to promote communication, serve as a source of expert information, and participate in the Michigan Cancer Consortium
- Identify panels of experts to provide genetic information on specific chronic diseases such as diabetes to primary and specialty care providers, patients and their families
- Test the feasibility of using regional multidisciplinary coalitions to explore genetics and chronic disease issues in relation to prevention and intervention strategies

3. 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

- Pursue relevant funding opportunities including federal grants and cooperative agreements
- Explore other possible funding sources such as private foundation grants
- Increase collaborative partnerships with state and local agencies and institutions to facilitate successful grant applications

4. **Ensure an adequate workforce by promoting awareness of careers in genetics for interested individuals**

   - Increase collaboration with existing organizations, career counselors, training grants and the Department of Consumer and Industry Services to promote awareness of clinical, laboratory, public health and research careers, and generate support for existing and future training programs
   - Identify ways to increase career opportunities in genetics for underrepresented populations

5. **Designate a central biochemical laboratory to provide confirmatory testing on infants with positive newborn screens and other biochemical genetic disorders**

   - Identify staffing and resources needed to support a biochemical genetics laboratory
   - Facilitate establishment of a central laboratory as a statewide resource for confirmatory testing of positive newborn screens and diagnosis of other metabolic disorders

6. **Insure prompt and appropriate state response regarding national recommendations for voluntary population-based screening**

   - Monitor new and emerging recommendations for population screening
   - Develop methods to address the public health implications of available screening tests

7. **Address the identified public health risks related to gene-environment interactions and assure the public has access to appropriate information and resources**

   - Outline a five-year plan of action to focus efforts and develop an approach to provide accurate and relevant information regarding potential health risks to those who need it

NEXT STEPS: APPROACH TO IMPLEMENTATION AND OPPORTUNITIES FOR COLLABORATION

The mission of Michigan's comprehensive state genetics program is to: Coordinate educational activities that increase genetic literacy; facilitate early identification and treatment of individuals with birth defects, heritable disorders and genetic susceptibilities; and foster collaboration to integrate advances in genomic science throughout public health and other systems of care.

The genetics plan for Michigan provides a blueprint for action to improve health and enhance quality of life through appropriate use of genetic information, technology and services. A wide range of needs have been outlined, as well as numerous strategies to address them. Members of the Genetics Advisory Committee found all of the proposed goals and action steps to be intertwined and equally compelling, and therefore decided against prioritizing one goal over another. However, increasing genetic literacy among all sectors of the population is clearly fundamental to progress in other areas and perhaps the single most urgent priority.

Much progress can be made with existing resources - through collaborations with partner organizations, federal grant initiatives, and by simply incorporating a new public health genetics perspective into currently funded program activities. Continuing partnerships with colleagues in maternal and child health, chronic disease, epidemiology, the laboratory and other public health programs will be of paramount importance to achieving the true integration of genetics needed to attain the goals of the plan. Over the next five years, genetics program staff will focus efforts on the objectives outlined in this document, including enhancement of the newborn screening system. Important roles for MDCH include:

1) providing statewide coordination of services and facilitating communication networks;
2) developing and supporting information systems for data linkages and integration;
3) promoting quality assurance measures, guidelines and best practices;
4) providing leadership on funding issues; and
5) evaluating outcomes.

Many of the objectives delineated in the plan are already being addressed by program staff or through collaborative activities both within and outside the department. For instance, MDCH has already acquired new laboratory equipment needed to test infants for medium-chain acyl-CoA dehydrogenase (MCAD) deficiency and identified changes required in the public health code in order to implement an expanded newborn screening program. Establishment of a centralized genetics resource and information center, along with data integration and development of newborn screening quality assurance measures, are included in a recent grant from the HRSA Genetic Services Branch. To address genetic literacy, available funding may be redirected to emphasize new approaches to outreach education. Steps can also be taken using existing resources to assist genetic centers in exploring possible solutions to difficult funding issues. In order to pursue certain other objectives, new sources of support must first be identified.

Successful implementation of the plan - and fulfillment of the vision for genetics in public health - will depend not only on MDCH but also on the stakeholders who participated in this consensus process, as well as on many new partners at the local, state and national levels. Such key players, in addition to state and local public health programs, include other state and federal agencies, medical care providers and hospitals, consumers, advocacy groups and organizations, educators, industry, media, schools and training programs, and healthcare payers, to name just a few. These partners will each play a vital role in helping to improve health outcomes for the people of Michigan - at all stages of the life cycle - through the wise use of genetic knowledge in medicine and public health.

Key Partners for Achieving State Plan Goals

Key Players	Goals and Objectives
	I. Increase genetic literacy in the State of Michigan
ASTCDD, CDC, Consumer groups, HRSA, LHD, MCGPH, MDCH, Media, Professional organizations, RGC	1. Expand public and provider knowledge regarding the impact of genetics on health
MDCH, MDE, RGC, Schools and teacher groups, Teacher training programs, University training programs	2. Integrate human genetics into curricula throughout the educational system
MCGPH, MDCH, Media, Professional organizations	3. Increase awareness of genetic ethical, legal and social issues (ELSI) by educating health care professionals and the public
LHD, MCGPH, MDCH, Occupational Health and Safety groups & agencies, University researchers	4. Develop avenues for communication about gene-environment issues between academic, public, primary care professionals, and the public
	II. Assess the public health impact of heritable conditions and the utilization of genetic services
MDCH	1. Improve the utilization of existing data sources for planning, implementing, and evaluating program activities
LHD, MDCH, PCP, Reproductive Genetic Centers	2. Develop methods to assess the use of reproductive genetic services by individuals of childbearing age
Hospitals, LHD, MDCH, Community groups	3. Improve the assessment and understanding of birth defects as a public health problem
Managed care plans, MDCH	4. Develop methods to assess the public health burden of genetic/familial disease in the adult population
Cancer Genetics Organization, Cancer Research Centers, MDCH	5. Conduct public health surveillance and research regarding hereditary cancer in Michigan
	III. Improve access to genetic information, prevention strategies, and services
MDCH, RGC	1. Establish a central genetic resource center to make information and resources more readily available to the public and providers
LHD, MDCH, Media, MiTIS, PCP, Reproductive Genetic Centers, Schools, Colleges & Universities	2. Provide information to the public and professionals about known causes of birth defects and strategies for prevention
EO, Hospitals, MDCH, NBS Medical Management Centers, PCP, Pediatric Genetic Centers, Schools	3. Improve dissemination of information about resources and services to families of children with birth defects and genetic disorders
MDCH, RGC, local communities	4. Assure availability of comprehensive genetics clinics throughout Michigan
MDCH, Reimbursement experts, RGC, Third party payers & Managed Care	5. Explore strategies for financing genetic health care, testing, and support services
MDCH, Reference laboratories, Reimbursement experts, RGC, Qualified Health Plans & Third party payers, Specialty clinics	6. Assure availability of DNA testing for children with genetic disorders and their relatives
HCP, LHD, MDCH, RGC	7. Increase referral of patients affected or at risk for conditions with genetic components to appropriate genetic services regardless of ability to pay
	IV. Promote early identification and treatment of individuals with birth defects, heritable disorders or genetic susceptibilities, throughout the life cycle
Hospitals, MDCH, Midwives, Parents, Pediatric PCP	1. Assure that all Michigan newborns receive an initial screen by 24-36 hours of age in accordance with guidelines established by MDCH; and identify all infants with positive screens by 6 days
GAC-Newborn Screening Subcommittee, MDCH, NBS-Medical Management Centers, Parents, Pediatric PCP	2. Provide appropriate follow-up, diagnosis, and treatment for infants with positive screening tests in accordance with nationally recognized guidelines
GAC-Newborn Screening Subcommittee, Legislators, MDCH, Other state programs	3. Expand the newborn screening program to reflect current technological advances
EO, LHD, MDCH, MEHDI,	4. Assure early identification, evaluation and genetic counseling/education for all children

Parents, Pediatric PCP, Pediatric Genetic Centers, Schools	with genetically-based disorders, birth defects and developmental delay
General Public, HCP, LHD, MCGPH, MDCH	5. Reduce the public health burden related to preventable chronic diseases with a significant genetic component
HCP, MCC member organizations, MDCH, Medicaid, Third party payers	6. Reduce morbidity and mortality related to hereditary cancer by increasing utilization of appropriate cancer risk assessment services
MCGPH, MDCH, MDEQ, University researchers	7. Monitor developments in current knowledge about gene-environment interactions of potential public health relevance for the Michigan population.
	V. Identify best practices and promote a policy framework to assure high quality services, supports and genetic privacy protections
MDCH, Professional organizations, RGC	1. Promote genetic competencies (as defined by the National Coalition for Health Professional Education in Genetics) among health care professionals servicing Michigan residents
MCGPH, MDCH, MDEQ, Media, Occupational Health and Safety agencies	2. Address the identified public health risks related to gene-environment interactions and assure the public has access to appropriate information and resources
Genetic laboratory directors, MDCH, SACGT guidelines	3. Assure quality of genetic laboratory testing in Michigan
LHD, MDCH, PCP, Reproductive Genetic Centers	4. Disseminate consensus guidelines for reproductive genetic health care
MDCH	5. Implement fully the recommendation of the Governor's Commission on Genetic Privacy regarding retention and storage of newborn screening samples
MDCH, NBS Medical Management Centers, Pediatric PCP	6. Assure privacy protections for reporting newborn screening results to private physicians, medical management centers, and others
GAC-NBS Subcommittee, MDCH, National experts and published data, NBS Medical Management Centers	7. Evaluate the effectiveness of the newborn screening program using identified outcome measures
EO, Pediatric genetic centers, PCP & subspecialists, Special education	8. Develop and test a consensus diagnostic approach for evaluation of developmental delay and mental retardation
MCGPH, MDCH, RGC	9. Insure prompt and appropriate state response regarding national recommendations for voluntary population-based screening
	VI. Promote Appropriate Public Health Responses to Advances in Genomic Medicine and Technology
Federal agencies, GAC, LHD, MCGPH, MDCH	1. Promote the integration of public health genomics within MDCH and other relevant state and local agencies
Advocacy groups and disease organizations, MCC, MDCH, MPHI/Cancer Genetics Consultant, RGC, University Cancer Centers	2. Enhance communications with genetic service providers and promote partnerships with relevant stakeholders
MDCH, Reimbursement specialists, RGC	3. 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
Consumer and Industry Services, Educational System & Universities, MDCD, MDCH, Professional Associations	4. Ensure an adequate workforce by promoting awareness of careers in genetics for interested individuals
Family Practice & OB/GYN Professional organizations, LHD, MDCH, Medical School and Residency Training Programs, PCP, Reproductive Genetic Centers	5. Assure quality and availability of clinical reproductive genetic services statewide
MDCH, NBS-Medical Management Centers	6. Develop and monitor systems to improve the accuracy and completeness of newborn screening data by establishing linkages with vital records and other children's databases, in order to identify health services needed/received by high risk populations
GAC-Newborn Screening Committee, MDCH, University Health Systems	7. Establish a central biochemical laboratory to provide confirmatory testing on infants with positive newborn screens and other biochemical genetic disorders

Key to partner acronyms

ASTCDD: Association of State and Territorial Chronic Disease Directors
CDC: Centers for Disease Control and Prevention
GAC: Genetics Advisory Committee
HCP: Health Care Providers
HRSA: Health Resources and Services Administration
LHD: Local Health Departments
MCC: Michigan Cancer Consortium
MCGPH: Michigan Center for Genomics and Public Health
MDCD: Michigan Department of Career Development
MDCH: Michigan Department of Community Health
MDE: Michigan Department of Education
MDEQ: Michigan Department of Environmental Quality
MiTIS: Michigan Teratogen Information Service
MPHI: Michigan Public Health Institute
NBS: Newborn Screening
PCP: Primary Care Providers
RGC: Regional Genetic Centers
SACGT: Secretary's Advisory Committee on Genetic Testing

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APPENDIX A:

THE NEEDS ASSESSMENT PROCESS

The framework for the needs assessment was based on seven core areas outlined in the *Guidelines for Clinical Genetic Services for the Public's Health* by the *Council of Regional Networks for Genetic Services* in 1997. These areas include: 1) organization and administration; 2) prevention; 3) available services; 4) research; 5) education; 6) data collection and documentation; and 7) funding. The needs assessment process was designed to collect both quantitative and qualitative data that would allow development of a comprehensive state plan for public health genetic services while incorporating the core public health functions of assessment, policy development and assurance using a collaborative approach. The goal was to define the priorities of patients and their families, communities, the general public, health and human service providers, and educators for all four stages of the life cycle (prenatal, newborn, childhood and adult). Specific objectives of the needs assessment included: 1) identifying available resources including the strengths, weaknesses and gaps in the current genetic service system, and 2) identifying data sources that could be integrated to enhance infrastructure and provide methods of program monitoring and evaluation with emphasis on genetic case-finding and early intervention.

Benefits

It was anticipated that data collected through the needs assessment process would be useful not only in delineating goals for the strategic plan, but also for guiding implementation of programs and special projects over the next several years. In particular, quantitative as well as qualitative data may be used to:

- Develop methods to promote early identification of both children and adults with genetic disorders;
- Assure access to community-based, family centered and culturally appropriate intervention services;
- Identify available resources (data and infrastructure, personnel, legislation);
- Develop methods of measuring the population that is receiving or in need of genetic services;
- Define clinical and research genetic service systems;
- Describe prevention (primary, secondary and tertiary) initiatives related to genetics;
- Assess genetic literacy of professionals, consumers and the public related to the understanding of basic genetic concepts, awareness and utilization of genetic services, and knowledge about ethical, legal and social implications;
- Assess funding sources and reimbursement for genetic services.

Methods

Data Collection

A variety of data collection techniques were used to explore the issues from a wide range of perspectives. Informal discussions were held with professional colleagues and personal acquaintances of the project staff. Meetings and conferences were attended to glean an understanding of ongoing genetic-related initiatives outside the traditional scope of public health. Key informant interviews and focus groups were conducted to obtain a general understanding of the target population's perspective on the need for and utilization of genetic services, level of awareness and interest in medical or public health genetics, and barriers to accessing the state's system. The information collected from this process was then used to develop seven questionnaires in order to solicit more widespread input from populations representing the most critical stakeholders in the state genetics plan: advocacy groups and support organizations; consumers; educators; general public; genetic service providers; health care providers (primary care and specialty); and local health departments. Finally, 12 expert working groups were convened to further identify existing resources and unmet needs, and recommend priorities for the plan.

Survey Populations and Response Rates

Survey Population	Sampling Frame	Surveys Mailed	Surveys Returned	Response Rate
Advocacy groups & organizations	MI Support Group/ Org, 1999-2000 Directory	271	86	32%
Consumers	Advocacy group and genetic service provider distribution to their clients	413	101	24%
Educators	Biology, Life Management, & Health teachers from middle/high schools in the MI Education Directory, 2000	432	168	39%
Clinical & Laboratory Genetic Service Providers	Mailing list of Michigan Genetic Center Staff	93	54	58%
Health Care Providers	Oakland County Nurse Training; MI State Medical Society and MI Osteopathic Association mailing lists	473	140	30%
Local Health Departments	Med. Directors, Health Officers, Env. Health Directors and other health specialists from MDCH Local Health Services Section, June 2001 Directory	245	105	43%
General Public	United Auto Workers; 1999 Michigan Ethnic Directory	360	94	26%
TOTAL		**2287**	**748**	**33%**

Target Populations

The project team recognized the cross-cutting and interdisciplinary nature of public health genetics. Therefore, special care was taken to include individuals representing all four life cycle stages as well as geographic and ethnic diversity. The fact that societal roles are not mutually exclusive, and that a wide array of factors affect knowledge of genetics and utilization of services, was also acknowledged. Major sectors of the state's population were divided into 13 groups that have the potential to either influence or be impacted in some way by implementation of a state genetics plan: advocacy organizations; consumers; educators; funding sources; general public; genetic service (clinical and laboratory) providers; health professional training programs; health care providers; industry; media; mental health and developmental disability program providers and clients; policy makers; and research scientists.

Population Sampling Methods

Relevant directories for each of the survey populations were identified and used to disseminate self-administered questionnaires to a segment of the population stratified by county location, population, and ethnic or religious affiliation. Surveys were distributed over a five month period to assure the best representation possible from the stratified survey populations. Some population groups such as educators, health care providers and the general public were over-sampled in order to decrease the standard error in sampling. In general, all respondents had the opportunity to comment on their level of satisfaction with genetic services and information, and identify priorities for public health initiatives surrounding medical genetics. Where clarity was needed for terminology such as birth defect, genetic condition, predisposition, and genetic counselor, general definitions were provided to maintain content validity. All surveys included seven questions that solicited respondent input about the use of available resources for: 1) public awareness and education about genetic disease; 2) the need for specialists who provide genetic services; 3) screening programs for early identification; 4) research studies to understand the impact of genetic disease on health; 5) assurance that access to services is available; 6) reduction in secondary disabilities; and 7) education of health care professionals and educators. Also included in each survey was a series of closed-ended and open-ended questions to support the construct validity of the survey instrument around the above topic areas.

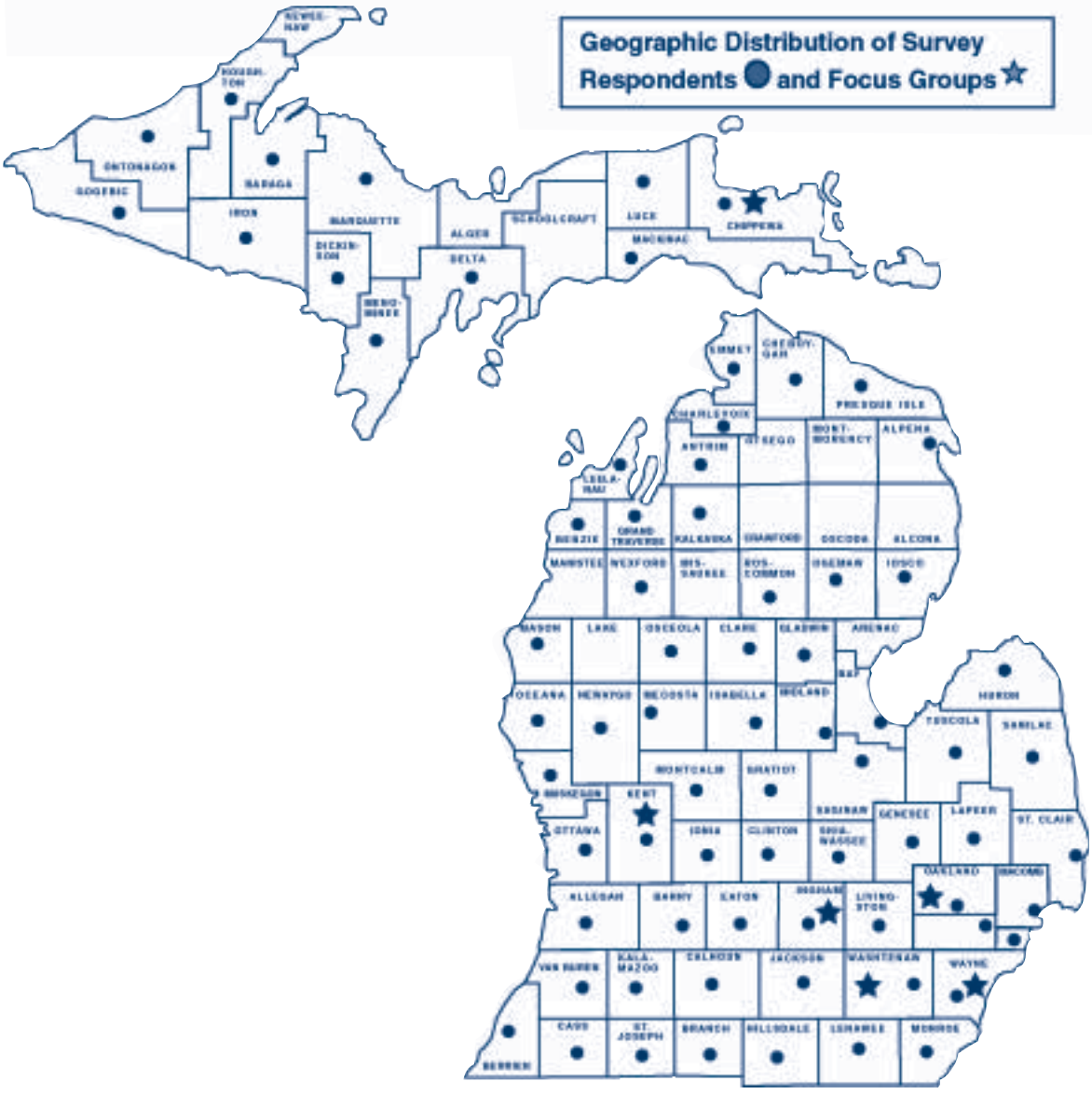
Survey Demographics

Demographic data were provided by 655 individuals who completed the survey questionnaires, with advocacy organization respondents excluded from the demographic profile. Just over one-third of the participants were male, while almost two-thirds were female. The majority were Caucasian, but a total of 114 individuals were of other ethnic origins. The largest minority were African Americans, who represented 6.5% of all survey respondents. Respondents ranged in age from 23 to 80 years, with nine parents also representing their children who ranged in age from 3 to 22 years. Geographic coverage included representation from 69 of Michigan's 83 counties.

Race/Ethnicity of Survey Respondents	
American Indian or Alaska Native	13 (1.9%)
Arab or Middle Eastern American	9 (1.3%)
Asian/Southeast Asian or Pacific Islander	18 (2.7%)
Black or African American	43 (6.5%)
Latino or Hispanic	13 (1.9%)
White or Caucasian	538 (82.5%)
Multiracial American Indian/Spanish	13 (1.9%)
Other	5 (.7%)
TOTAL	652

Gender of Survey Respondents
Female 414 (63.2%)
Male 241 (36.7%)
Total 655

Birth Year Ranges of Survey Respondents	
1920-29	8 (1.2%)
1930-39	37 (5.7%)
1940-49	172 (26.5%)
1950-59	230 (35.4%)
1960-69	118 (18.1%)
1970-79	75 (11.5%)
1980-89 Consumers who answered for their child	5 (.7%)
1990-99 Consumers who answered for their child	4 (.6%)
TOTAL	649



Population Groups

(1) Advocacy Groups and Organizations

This population included support groups and advocacy organizations (such as local Down syndrome support groups, Spina Bifida Association of Southeast Michigan, the American Cancer Society, March of Dimes, etc.) listed in the 1999-2000 edition of the "Guide to Michigan Support Groups and Organizations Serving Families with Inherited Disorders, Birth Defects and Related Conditions" compiled by the Hereditary Disorders Program. Six key informant interviews assessed knowledge about the genetic services system and what barriers constituents might have in using these services. Informants were also asked to comment on their preference in communicating information about genetic services. A 34-item questionnaire was developed and distributed to 271 organizations with a 32% response rate. The surveys asked for general constituent information that could provide additional insight about individuals who use genetic services. The advocacy groups and organizations were also asked to provide further assistance in the data collection process by distributing consumer questionnaires to their members. Organizations were asked which genetics clinics or other state programs they used for referrals. They were also asked to indicate which MDCH data and information systems they utilized or might find useful to their organization in the future. Finally, members of advocacy organizations were included in the expert working groups on pediatric and adult genetic health services, as well as birth defects surveillance.

"I too was ignorant. With muscular dystrophy I thought, that's what it was, muscular dystrophy. Then when I found out I had it, I learned there are different kinds. I've been in the diagnostic process for about three years now. It makes a difference, just knowing what I have and what to expect."
--A focus group participant

(2) Consumers

The needs assessment would not have been complete without input from those served by the genetic service system - individuals and families affected by genetic conditions and birth defects. In order to solicit the consumer voice, four key informant interviews were held with parents of children with special health care needs. In addition, focus groups were held with 1) members of the sickle cell anemia parent support group in Detroit; 2) parents of children with birth defects in Sault Ste. Marie (Upper Peninsula); and 3) adults with genetic disorders in Grand Rapids. Participants were asked to comment on their perception of the word genetics, identify barriers to finding or using genetic information, and how they wished to receive information about genetic services. They were also asked to identify core elements of a state genetic services program and what changes they would implement with respect to genetic education, diagnosis, treatment, and patient support services. As a result, a 58-item questionnaire was developed for adults, or parents representing their children, who have a birth defect, genetic condition, or predisposition. Respondents were asked how soon and what type of information they received about the diagnosis, as well as who provided that information and how satisfied they were with the scope of services they received. Patients who had been seen in a genetics clinic were also asked about their satisfaction with the genetic diagnosis and counseling service. Four hundred thirteen questionnaires were distributed to consumers through genetics clinics and support group organizations, and 101 were returned for a 24% response rate. Consumers, including the mothers of children with PKU, spina bifida, Down syndrome, and a rare metabolic disease, as well as two individuals with adult-onset disorders, also participated in the newborn screening, birth defects, pediatric, and adult expert work groups, respectively. The project's consumer consultant also attended the majority of work group meetings.

(3) Educators

In order to determine unmet needs regarding genetic literacy, and to discern how educating health care providers and the general public might impact utilization of genetic services or health behaviors, the current educational system was examined with respect to elementary, secondary, undergraduate, and professional training as well as special education. A series of five key informant interviews were conducted with special education teachers, secondary educators, and a university professor. Interviewees were asked general questions such as: 1) what did they think of when they heard the word genetics; 2) how they felt genetics would change what they were teaching; 3) what type of role MDCH should play in educating the public; and 4) what educational methods should be used. Special education teachers were asked similar questions but were also asked about genetic diagnosis and counseling clinics, and whether the families of their students ever inquired about genetic services. A 31-item questionnaire was then developed, primarily targeting biology, life management and health education teachers. A total of 432 questionnaires were mailed in May

of 2001 and again in early September to middle and high school educators listed in the 2000 edition of the Michigan Education Directory. Respondents were given the option of filling out the questionnaire online through a web-based version but this was only used by two teachers. In all, 168 responses (39%) were received. Teachers were asked if they were aware of the Human Genome Project, how they perceived their role in providing information to students about genetics, and their level of comfort teaching genetic-related subject matter. They were also asked about their knowledge of available genetic services and who could make the biggest impact on increasing genetic literacy among the general public. To provide additional insight surrounding genetics education, a secondary biology teacher, nursing school administrator, university continuing medical education director, MDCH school health consultant, medical student genetics course director, genetic counseling training program director and several genetic counselors participated in the expert working group on genetic education and literacy.

(4) Financing

Questions about finance and reimbursement for genetic services were included on most of the questionnaires. Finance-related issues were also brought up in several of the expert work groups. Attempts to invite third party payers to participate in the needs assessment met with little success. However, three key informant interviews were conducted with a member of the Michigan Association of Health Plans and two administrators from Upper Peninsula health plans. Those willing to participate were asked to comment on the costs associated with providing genetic services and which services were covered by their plan. Responses to ethical, legal, and social implications surrounding genetics for the health insurance industry were solicited. Finally, participants were asked to describe the role of a public health genetics program in assuring quality and availability of services. Information on MDCH funding for newborn screening and genetic services was gathered. Key members of the working group on finance and reimbursement included a genetic center and laboratory director, genetic counselors and a medical consultant from the Medicaid system.

(5) General Public

Informal discussions with acquaintances as well as community groups were used to capture a snapshot of the general public's perspective on genetics and genetic services. These groups included church members in Ann Arbor and United Auto Worker union members in Ann Arbor, Detroit, Muskegon, Taylor and West Branch. A focus group was also conducted with a university Native American student group. Foreseeing the importance of potential gene-environment interactions, five key informant interviews focused on whether individuals perceived they were at a higher risk of disease from working with certain chemicals in their workplace. Interviewees were also asked to explain possible benefits or problems from knowing this information. A reoccurring theme was the need for educational opportunities about the advancing genetic technology and how this technology contributes to improved medical diagnosis and treatment. Another theme expressed within the groups was a concern about confidentiality and discrimination.

About 89% of the general public surveyed would want to know if they carried genes that could increase the risk of disease when exposed to certain environmental factors. About 5% were unsure, and only 5% did not want to know.

General findings from the qualitative data were used to create a 52-item questionnaire in order to further quantify the awareness and general opinions of Michigan residents about genetics. Survey participants were asked to identify which resources they used to obtain health information and where in particular they find answers to their questions about genetics. There were also questions about the use of genetic services, participation in research studies and how available resources should be used. Respondents were asked whether they would want to know if they carried genes that could increase the risk of getting a disease when exposed to certain environmental factors, and how important it was to them to have access to information about potential gene-environment interactions. Questionnaires were distributed through the 1999 Michigan Ethnic Directory to 360 residents with a 26 percent response rate.

Findings from the Communities of Color and Genetics Policy Project were also reviewed. The report includes recommendations based on a series of community dialogues hosted by 15 African American and Latino community-based organizations in Michigan and Alabama. It addresses topics such as: access to genetic testing and services; education; playing God - perfect children (human cloning and genetic engineering); the right to genetic privacy; genetic research; genetic testing; and trust and distrust.

(6) Genetic Service Providers

a) *Clinical:* A focus group session was held during a meeting of the Michigan Association of Genetic Counselors to elicit the perspectives of genetic service providers working in reproductive, pediatric and adult genetics clinic settings. Participants were asked to explore the role of public health in supporting the clinical genetics infrastructure. They were also asked to identify gaps and strengths in the state's genetic service delivery system, and what changes they would implement with respect to genetics education, diagnosis, treatment, patient support services and research within a five year period. Genetic counselors, clinical geneticists and genetic center directors participated in nearly all of the expert work groups, including those addressing the areas of reproductive, pediatric, and adult genetic health care services, as well as cancer genetics. A questionnaire was developed and mailed to 93 genetic service providers on the mailing list maintained by the Hereditary Disorders Program. Respondents had the opportunity to identify barriers for patients seeking their services. They were also asked to estimate how they spent their time, the percentage of patients seen for various reasons and the number of patients seen with certain diagnoses. Genetic center directors were also asked to identify the level of staffing, types of databases used and level of staff activity.

b) *Laboratory:* Questions for genetic laboratory directors were included on the survey, such as the number of tests performed, the types of tests available and levels of reimbursement received from third party payers. In addition, laboratory directors participating in a laboratory work group represented the cytogenetic, molecular (DNA), maternal serum screening and state public health laboratory perspectives.

Overall, 54 genetic service providers returned questionnaires for a 58 percent response rate.

(7) Health Professional Training Programs

Information about this sector was gathered primarily from participants in the education work group representing genetic counselor training programs, nursing and medical education. The need for trained laboratory personnel was also highlighted by the laboratory work group. In addition, personal contacts and knowledge of national initiatives, including the recent development of genetic competencies for various types of health care providers, were used to identify possible needs with respect to workforce training.

(8) Health Care Providers

a) *Primary and Specialty Care:* The providers in this population included primary care and specialty physicians and nurses. Two key informant interviews were conducted with Ingham Regional Medical Center nurses to gather preliminary information regarding the role of health care providers in relation to the public health genetics system. They were asked to identify gaps in the system and barriers to using genetic screening, clinical or laboratory services. A Southeast Michigan March of Dimes "Genetics and Your Practice" conference for primary care providers was attended. An 85-item questionnaire was then developed and disseminated to nurses attending an in-service conference in Oakland County. In addition, 473 questionnaires were sent to primary care and specialty care providers using mailing lists obtained from the Michigan State Medical Society and the Michigan Osteopathic Association. A total of 140 (30 percent) questionnaires were returned by primary care providers such as pediatricians, obstetricians, internal medicine and family practice; and by those providing specialty services–such as hematology, oncology and allergy. Practicing physicians including a neonatologist, neurologist, obstetrician/gynecologist, pediatric hematologist, pediatric cardiologist, internist, oncologist and adult cardiologist participated in the birth defects, newborn screening, reproductive, pediatric, cancer and adult genetics work groups, respectively.

b) Local Health Departments: Key informant interviews were held with two local health officers as well as the MDCH liaison to local public health. Informants were asked how they view the current role of local public health with respect to providing genetic information and services. They were also asked how they perceived the role of MDCH in supporting delivery of public health genetic services at the local level, e.g. training; quality assurance; capacity building; reimbursement; resource and referral information; and public education. As a result of the qualitative data collected, a 46-item questionnaire was developed and distributed to 245 health officers and medical directors, environmental health directors, health education and planning directors, and public health nurses throughout the state, with a 42 percent response rate. Respondents were asked where they found information about advances occurring in medical and public health genetics, as well as their opinions on which chronic disease program areas should incorporate new genetics information over the next three to five years. A local Children's Special Health Care Services coordinator from Ingham County participated in the pediatric genetics work group. Other local public health representatives were invited to participate in several of the relevant expert working groups but were unable to attend.

(9) Industry and Commercial Companies

The role of industry was explored primarily by attending the BioMed Expo and the University of Michigan School of Business Health Care Forum, through informal interviews with conference attendees. In addition, members of the gene-environment work group shed light on the ways that pharmaceutical companies are using knowledge of the human genome in their product development.

(10) Policy Makers

A meeting of the MDCH adult genetics work group was used as a focus group session to explore the perspectives of state program administrators. Informal discussions with newborn screening and genetic program staff also provided insight into potential needs relating to the policy arena. Michigan's responses to the 1999 CSTE survey were reviewed. Existing genetic privacy legislation was also reviewed. A policy work group identified potential ethical, legal and social issues. Although 74 percent of health care providers and local health departments were unaware of the Michigan genetic privacy legislation, they reported that they felt that their role included informing the public about current social issues and their legal rights. Approximately 75 percent of consumers and the general public noted the importance of obtaining information regarding laws protecting genetic privacy.

(11) Media

Insight into the possible roles of the media (print, television, radio) in relation to implementing a statewide genetics plan was gained by attending a workshop on "Genetics and the Media" held during the 2000 National Conference on Genetics and Public Health in Ann Arbor. Resources available for developing public relations campaigns were discussed with the MDCH Director of Health Promotions and Publications. Questions were included on several questionnaires to further define the role of the media in increasing awareness and access to genetic information.

(12) Mental Health and Developmental Disabilities

Members of the mental health and developmental disability community were included in the survey process through advocacy organizations and consumers. In addition, two staff members from Community Mental Health for Central Michigan participated in the Adult Genetic Health Services work group. Findings from a survey conducted by the statewide Prader-Willi Syndrome consultant were reviewed. Interviews with program staff provided additional insight into the genetic information needs of providers caring for individuals with developmental disabilities.

(13) Research Scientists

Information about existing clinical and public health genetics research in Michigan was requested from the major universities or found on their websites. Discussions were held with MDCH Bureau of Epidemiology staff. In addition, research scientists participated in the work groups on education, gene-environment interaction, adult genetic services and cancer genetics. The need for translating genomic research from the academic centers to public health has been highlighted by the CDC Office of Genomics and Disease Prevention, and many of these documents were reviewed by project staff. During the course of the needs assessment process,

MDCH staff collaborated with public health researchers at the University of Michigan School of Public Health to develop a Center for Genomics and Public Health, funded by the CDC.

Expert Work Groups

In addition to the approaches already described, 12 expert working groups were convened and asked to review specific topic areas critical to a comprehensive state plan. The groups were initially asked to identify key issues and barriers, suggest possible action steps, and ultimately to recommend priority goals and objectives relating to their area of expertise. Most groups consisted of six to 12 members including experts identified through partner organizations, consumers and relevant MDCH personnel. The meetings were held in mid and southeast Michigan and all except two of the groups met twice to complete their recommendations. Relevant survey data were presented to support the goals and objectives formulated by each group.

Birth Defects Surveillance

Discussions focused on improving the use of current data sources, such as the Birth Defects Registry, for producing statistics on incidence rates, client populations, cluster investigations, epidemiological research and evaluation of linkage with services. The need to examine ways of linking environmental and teratogenic exposures with birth outcomes was identified. The group also identified priorities related to preventing birth defects, especially neural tube defects, and assuring that children with birth defects receive intervention services.

Cancer Genetics

The group explored the meaning of public health genetics, as well as the role of public health in supporting the infrastructure for cancer genetics services and research. Members were aware of many ongoing clinical genetics research projects within the state, notably at institutions such as the University of Michigan Cancer Center, Karmanos Cancer Institute, Henry Ford Hospital and Michigan State University. Problems related to such research include a lack of funding, lack of knowledge among possible participants and lack of large enough patient numbers at any one center. The group felt there was a need to better understand existing practices with respect to cancer risk assessment and educate practitioners about best practice models, including how to obtain good family histories and triage referrals. The usefulness of establishing an ongoing cancer genetics advisory group with an outcome-based approach to education, patient care and research was also highlighted.

Data and Evaluation

A central finding from this work group was the need for quality data for use in genetics program planning and evaluation, as well as the challenge of extrapolating data for programmatic needs when knowledge of various existing databases is incomplete. MDCH data sources were discussed, including vital records, the newborn screening database, Birth Defects Registry, CSHCS, and other maternal and child health program databases. The potential usefulness of the department's new data warehouse was also examined, although there may be unforeseen challenges to access and limitations related to data confidentiality.

Finance and Reimbursement

Potential funding sources for genetic and newborn screening services were discussed. The primary source of revenue for public health genetic services is the newborn screening fee. Third party reimbursement for clinical services is usually in the range of 50-70 percent and there are no other sources of funding to subsidize direct genetic health care to patients. The group felt that continued public health funding for genetic counseling services was critical and that it would be useful to track expenditures based on the number of families served per year. The cost-effectiveness of genetic services needs to be assessed by examining the potential for improved health outcomes and promoting the preventive aspects of genetic health care.

Gene-Environment Interaction

The group described national research studies as well as new and emerging technology that will increase awareness of the effects of environmental factors in relation to genetic disease or predispositions. The potential value of the newborn screening cards in relation to gene-environment research was underscored. Different types of gene-environment interactions were

identified as those related to: lifestyle; nutrition; occupational exposures; general environment; and medicine (pharmacogenetics). Adolescents and the elderly were identified as especially vulnerable populations and the potential for reducing the problem of adverse drug reactions in the population through individual genotyping was highlighted. Society needs to be educated in preparation for new applications of gene-environment science, including a better understanding of risk concepts. The connections between occupational health and public health should be strengthened, and establishment of an ongoing advisory committee on gene-environment interaction was recommended.

Genetic Education and Literacy

The need for increased genetic literacy has been recognized nationally, and the situation is no different in Michigan. Genetics education was a common theme among almost all of the expert work groups. This group reviewed past and current genetic education initiatives in the state and explored the need for improved integration of genetics within curricula throughout the educational system, from elementary through high school, to undergraduate and professional school training. Barriers as well as possible strategies for pre-service and in-service continuing education were discussed, as was the need to ensure an adequate workforce by promoting awareness of careers related to human genetics.

Genetic Health Services – Adult

The group explored a definition of the target population for "adult" genetic services and agreed that all adults are included, noting that some individuals have special genetic risk factors. A distinction was made between adults with conditions identified at birth or in childhood, often of Mendelian or chromosomal etiology, and those with later onset conditions that are often of a polygenic or complex nature. The needs of these two populations may be very different. From a public health standpoint, it would be important to differentiate health management strategies for disorders with a possible prevention component verses chronic genetic disorders with service needs. Transition of medical care between age groups was identified as a major concern. The work group acknowledged that access to specialized resource information is nearly impossible for some residents who most need it, and strongly recommended development of a central portal for genetic resources and research that would assist the general public as well as health care providers in finding information about services available within the state.

Genetic Health Services – Pediatric

The group defined the population for pediatric genetic health services as newborns with positive genetic screening tests, children with known genetic conditions, children identified through MBDR, and children with developmental delay. Fears related to receiving a genetic diagnosis for their child, and the range of medical, legal and financial issues facing families were explored. Potential sources for case finding of children who would benefit from genetic evaluation were discussed. The work group also discussed possible implications for disease management related to a shortage of qualified medical personnel, lack of training on genetic disorders, medical and social service needs of children and families, timely coordination of care, and equitable access to diagnostic services, particularly molecular genetic testing in out-of-state laboratories. The group identified gaps in the evaluation of children with developmental delay and suggested development of standardized protocols.

Genetic Health Services – Reproductive

A key issue identified by this work group was the need to assure quality and availability of clinical reproductive genetic services statewide. Systems are needed to monitor recurrent pregnancy loss as a sentinel condition in addition to the long term implications of assisted reproductive technologies. Patients need to be better informed about maternal and prenatal screening and consumption of folic acid to prevent certain birth defects. A need for standardized educational materials to be available at low or no cost, and ways to assist practitioners in providing accurate information were highlighted. Medical management of unusual or high-risk cases, and a lack of options for women faced with diagnosis of a fetal anomaly, including the need for bereavement services, were also discussed. The group suggested designating reproductive genetic centers of excellence to develop consensus guidelines for reproductive genetic care and provide training statewide.

Laboratory Services

Existing clinical facilities include university, hospital-based and commercial laboratories providing molecular, cytogenetic and clinical chemistry tests for maternal serum screening, as well as the newborn screening and other public health laboratories. University research laboratories are another potential source of genetic testing for specific disorders. There is currently no full-service biochemical genetics laboratory in the state. This was identified as a major deficiency in the provision of appropriate laboratory services. Work group members pointed out a need to evaluate reimbursement for genetic laboratory tests, which varies by health plan and type of billing (hospital vs. direct), and assure the quality of laboratory services. Also of concern was the effect of gene patenting on access to testing, as well as an inadequate genetic laboratory workforce. Educating physicians about the appropriate use of genetic testing, including informed consent, was another important issue for this group.

Newborn Screening (NBS)

This group focused on the need to develop standard criteria and a process for integrating new tests into the NBS screening panel based on the medical facts, available technology, budgetary considerations, and timelines for legislative approval. Methods of evaluating the current NBS system and assuring ongoing quality of medical management services, in compliance with national guidelines, need to be developed. The importance of an ongoing NBS advisory body was recognized and this work group has been designated as a subcommittee of the Genetics Advisory Committee.

Policy: Ethical, Legal, and Social Issues (ELSI)

The group discussed ethical, legal, and social issues (ELSI) of potential relevance to a statewide genetics system, and was given an opportunity to review recommendations by the Governor's Commission on Genetic Privacy and Progress as well as the Michigan genetic privacy laws passed in 2000. The project team also presented policy-related issues from the other work groups for consideration by the members. The need for a public that is more informed about ELSI, including informed consent and legal rights, was highlighted. The group recommended exploring potential barriers and implementing policies to assure access to genetic services statewide. They also suggested that privacy protections be assured for reporting of NBS results, and that a framework be established to insure appropriate state response as national recommendations on population-based screening emerge.

Genetics Advisory Committee

As part of the needs assessment and planning process, the MDCH Genetics Advisory Committee was refocused to better address the future direction of genetics throughout public health. New members represent a broader range of stakeholder perspectives including consumers, advocacy organizations, clinical and laboratory genetic providers, local public health, secondary educators, medical and public health training. The committee will advise the department on an ongoing basis. Its mission is to:

- Provide expertise and recommend appropriate ways for the department to integrate genetics into public health programs and activities addressing all stages of the life cycle;
- Review and evaluate current laboratory technology, follow-up, and medical management protocols for newborn and other population genetic screening programs;
- Assess public health implications of new genetic screening, diagnostic and treatment technologies.

A standing subcommittee to address newborn screening has been established and a subcommittee on birth defects surveillance and prevention is planned. Ad hoc subcommittees will be developed as needed to address specific issues.

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APPENDIX B:
KEY FINDINGS FROM THE NEEDS ASSESSMENT

Key findings from the needs assessment have been summarized below according to the framework delineated by the Council of Regional Networks for Genetic Services. Where findings from the survey questionnaires are presented, "n" represents the number of persons who responded to the questions addressing the particular issue under discussion. In order to assess the level of consensus regarding use of available resources to address genetic concerns, seven of the same or very similar questions were included on questionnaires for all survey target groups. For most questions, there was agreement or strong agreement and minimal disagreement across all populations, as noted in Table 1. The top two priorities were educating health care providers about advances in genetics and assuring access to genetic evaluation and counseling services, followed closely by a desire to increase public awareness of genetic factors in health and disease.

A. Organization and Administration

Current Status & Existing Resources

An organizational structure to administer genetics and newborn screening programs exists at the state level. Currently housed in the MDCH Bureau of Epidemiology, staffing for the Genetics and Newborn Screening Unit includes a state genetics coordinator, newborn screening director, newborn screening nurse consultant, and adult genetics consultant, as well as a birth defects coordinator funded through a CDC cooperative agreement. Public health genetics activity at the local level consists primarily of case identification and referral through programs such as Children's Special Health Care Services and WIC, and birth defect prevention education through programs such as Maternal and Infant Support Services and Women's and Reproductive Health Services.

Identified Needs

There is a need to enhance the visibility of the state genetics and newborn screening program,increase the number of genetics and newborn screening personnel as funding becomes available, and increase collaboration with local public health departments as an avenue for providing genetic education and expanding genetic health care infrastructure at the community level.

- About 31 percent (n=134) of health care providers were not familiar with the newborn screening program and only 74 percent of genetic service providers (n=46) correctly identified sickle cell anemia as a newborn screening test.
- More than 78 percent of health care, genetic service and local public health providers agreed they should know more about the state public health genetics program.
- In the general public survey, 82 percent of participants (n=92) agreed there should be a central state office to help people find genetic information and services, and 50 percent of health care providers (n=119) thought MDCH could better support their efforts by maintaining a central resource and referral line for providers or patients to call.
    - More than half (54 percent, n=279) of the health care, genetic service and local public health providers surveyed felt that having additional genetics personnel at the state level would help to facilitate utilization of existing genetic service programs by the public and health professionals.
- There is a need for more collaboration with local health departments as the role of public health in the genetics health system infrastructure continues to expand.
- As illustrated in Figure 1, MDCH could better support local public health and health care providers by:
    - Developing and disseminating statistical information on birth defects and genetic disease

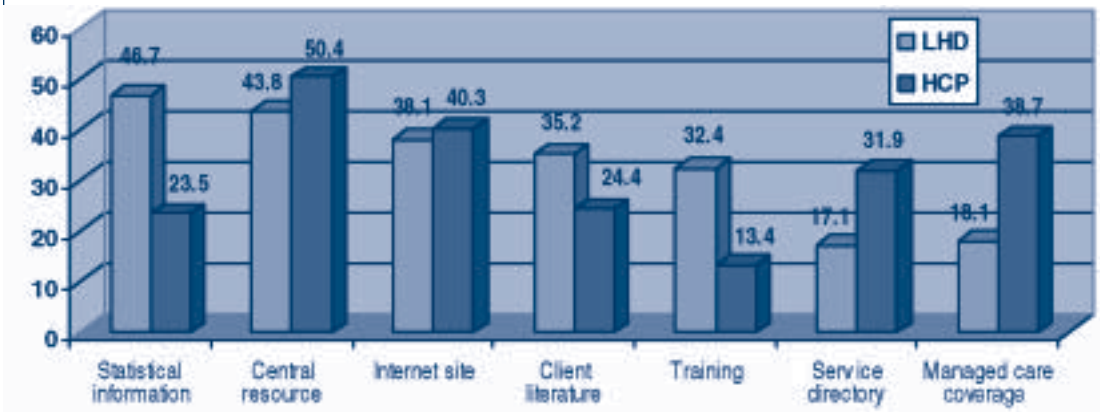
- Maintaining a central resource and referral line for providers and clients
- Maintaining an Internet site with links to clinical genetic databases and patient support organizations
- Providing client and patient literature for their use
- Providing in-service training opportunities and conferences
- Working with managed care plans to assure coverage for genetic services
- Providing a service directory of genetic specialists and clinical research studies within the state

Table 1. Comparison of Responses on Use of Available Resources Across All Survey Populations
(Percent indicates those who strongly agreed or agreed, combined.)

	ADV	CON	EDU	PUB	GSP	HCP	LHD	GRAND TOTAL
1. Increase public awareness and education about genetic factors in health and disease	96.2% n=79	97.9% n=95	93.3% n=164	94.6% n=93	97.8% n=47	89.6% n=125	95.2% n=104	**94.3%** n=707
2. Increase the number of specialists available to provide genetic diagnosis and counseling services	80.6% n=77	91.5% n=94	77.3% n=163	82.8% n=93	89.3% n=47	66.6% n=126	58.4% n=101	**76.4%** n=701
3. Expand screening programs for early identification of people predisposed to genetic diseases who might benefit from early treatment or other interventions	89.9% n=79	96.9% n=96	87.2 n=165	92.6% n=94	91.4% n=46	85.8% n=127	86.4% n=103	**89.4%** n=710
4. Fund research studies to better understand the impact of genetic disease on the health of Michigan's citizens	84.6% n=78	90.4% n=94	84.2% n=164	92.6% n=94	82.9% n=47	81.6% n=125	77.4% n=102	**84.6%** n=704
5. Assure that anyone who needs genetic evaluation or counseling has access to it	92.3% n=78	100% n=96	92.1% n=165	97.9% n=94	100% n=47	95% n=133	92.3% n=103	**95.5%** n=716
6. Help to prevent or reduce secondary disabilities in people with existing genetic diseases	90.8% n=76	98.9% n=95	87.8% n=164	97.9% n=94	91.1% n=45	94.7% n=132	83.4% n=102	**91.8%** n=708
7. Educate physicians and other health care providers (and science/ health educators) about advancements in medical genetics and birth defects	93.7% n=79	99% n=96	93.9% n=164	94.7% n=94	100% n=47	96.9% n=131	91.3% n=103	**95.2%** n=714

ADV: Advocacy Groups and Organizations CON: Consumers EDU: Educators PUB: General Public
GSP: Genetic Service Providers (clinical and laboratory) HCP: Health Care Providers (primary care and specialty)
LHD: Local Health Departments

Figure 1
Priorities for MDCH: Local Health Department (LHD) vs. Health Care Provider (HCP) Needs
(percent of responses in agreement when participants selected top 3 priorities)



B. Prevention

Current Status & Existing Resources: Primary Prevention[1]

Numerous birth defect prevention initiatives exist within the state and were summarized in the 1999 document *Birth Defects in Michigan*, produced as part of the 1999-2002 CDC cooperative agreement on birth defects surveillance. A new CDC cooperative agreement for 2002-2005 will provide continuation funding for the birth defects follow-up coordinator, a birth defects epidemiologist, and a part-time folic acid coordinator. The March of Dimes has been a major partner in leading a folic acid campaign nationally and within Michigan, but is now shifting its focus to the issue of prematurity. As a result, the lead responsibility for folic acid education statewide will need to be assumed by MDCH. Fortunately, 84 percent of local health department personnel surveyed (n=105) view their role in educating the public about birth defect prevention strategies as important or very important. Another important resource for disseminating prevention information is the Michigan Teratogen Information Service (MiTIS), a statewide teratogen information system located at the Detroit Medical Center. MDCH and partner advocacy groups such as the American Heart Association, American Cancer Society, and many other similar organizations, play an important role in promoting healthy lifestyle choices that can help prevent adult-onset chronic diseases. The importance of family history and underlying role of genetic predisposition has heretofore not been a major emphasis of health education campaigns, but is now starting to receive more attention.

More than 89 percent of the total survey population agreed that available resources should be used to expand screening programs for early identification of people predisposed to genetic diseases who might benefit from early treatment or other interventions.

Identified Needs: Primary Prevention

- According to a recent analysis of data from the Pregnancy Risk Assessment Monitoring System, awareness of folic acid among women giving birth is leveling off or even decreasing among certain population groups. With the March of Dimes reducing its emphasis on folic acid, new leadership is needed to maintain basic folic acid awareness and target educational efforts at high risk populations.
- There is a need to increase awareness and utilization of the MiTIS, and identify a stable funding source to assure continued availability of this statewide resource for the childbearing population.
- There is a need to improve awareness and patient compliance with recommended guidelines for preconception management of maternal medical conditions (e.g. diabetes, PKU) known to affect pregnancy outcomes.
- There is a need and apparent desire among the public for better genetic risk assessment related to adult disorders such as hereditary breast cancer where preventive measures may be available.

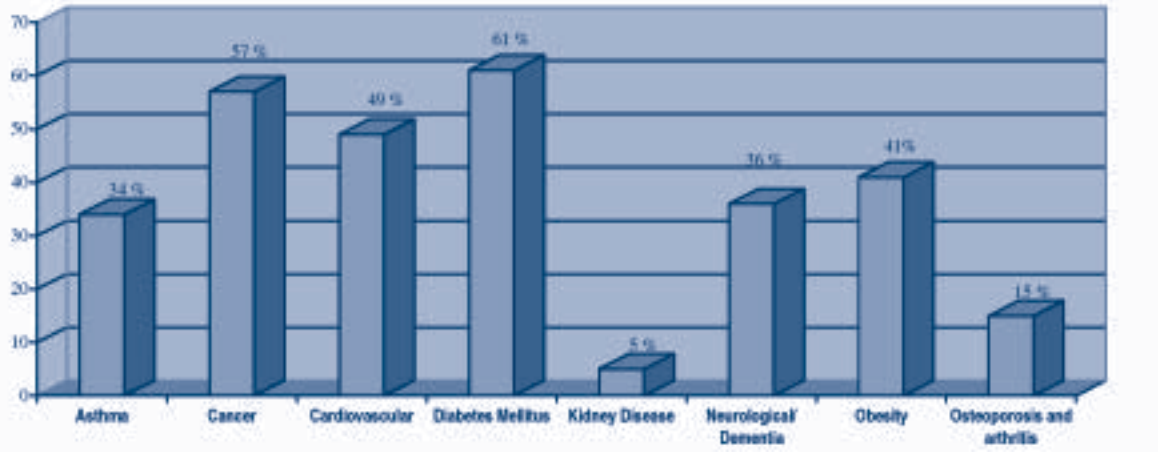
[1] *Primary prevention* refers to preventing the occurrence of a birth defect, genetic condition or disease. Examples include preventing neural tube defects by maternal folic acid consumption, preventing Fetal Alcohol Syndrome by maternal abstinence from alcohol use during pregnancy, or preventing cardiovascular disease by lifestyle modifications.

More than 90 percent of the general public would want to know if they were going to get a genetic disease running in their family if there was a treatment to prevent or slow the disease.

Current Status & Existing Resources: Secondary Prevention[2]

The appropriate use of prenatal genetic diagnosis is an important tool for identifying high risk infants with a wide range of structural anomalies or metabolic conditions who might benefit from special perinatal management or intervention such as fetal surgery. Seven reproductive genetic centers are available in Michigan to provide state-of-the-art prenatal diagnosis of birth defects or genetic disorders using techniques such as detailed ultrasonography, chorionic villus sampling, and amniocentesis. MDCH also plays a major role in fostering secondary prevention by assuring follow-up and medical management of infants identified through the newborn screening system. The success of the program depends heavily on the participation of hospitals and primary care pediatric providers, as well as the parents of newborns with positive screening tests. Seven disorders are currently included in the newborn screening panel. Primary care and specialty providers, managed care plans and a large number of disease-specific non-profit organizations are important partners in promoting general awareness of secondary prevention as well as specific recommendations for persons with identified genetic disorders.

Figure 2
Chronic disease program areas needing new genetics information in the next three to five years.
Responses of local health department personnel asked to select the top three priorities.
(Data shown in percentages, n= 100)



Identified Needs: Secondary Prevention

- There is anecdotal evidence of variable quality across the state in the medical management provided for fetal anomalies diagnosed prenatally.
- Advancing technology now enables detection of additional early childhood disorders in which prognosis is improved by early treatment. There is a need to add these disorders to the newborn screening panel.
- Many individuals with or at increased genetic risk for insidious chronic diseases such as diabetes, celiac disease, cardiovascular disease, cancer, hereditary hemochromatosis, and many others are not being identified as early as possible to receive maximum benefit from secondary prevention measures. As noted in Figure 2, local health department personnel identified diabetes, cancer and cardiovascular program areas as those most in need of genetic information over the next few years.

Current Status & Existing Resources: Tertiary Prevention[3]

Numerous services and organizations are available in Michigan to assist families with birth defects and genetic conditions. The Children's Special Health Care Services plan provides medical care and treatment for children with chronic conditions up to age 21 years. Early On coordinates systems to provide early intervention for children from birth to three years and their families. Special education is available through the public school system for students with special needs to age 26. Community mental health provides mental health and support

2 *Secondary prevention* refers to preventing unfavorable consequences from existing genetic conditions or predispositions. A good example is the prevention of mental retardation in children with PKU by early detection and treatment with a special low-protein diet. Other examples include preventing organ damage in persons with hereditary hemochromatosis (an iron-overload disease) by regular phlebotomy or preventing medical complications of celiac disease through a gluten-free diet.

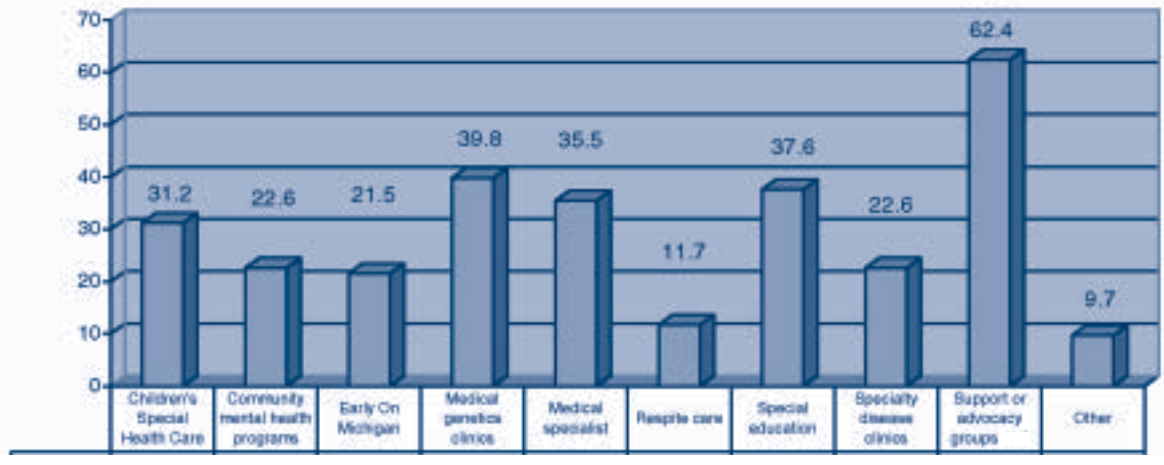
services for children, adolescents and adults with developmental disabilities. The Family Support Subsidy assists families caring for severely mentally or multiply impaired and autistic impaired children. Medicaid, managed health plans and third party payers provide insurance coverage for many of the diagnostic tests, treatments, medications and surgeries needed by individuals with genetic conditions. A wide range of medical subspecialties are available through major university medical centers and private health systems. A statewide Family Support Network links families of children with special needs, and numerous disease-specific support groups and organizations also exist for adults. The MDCH Hereditary Disorders Program has maintained a directory of such support groups for more than 10 years. Figure 3 highlights the services used by Michigan consumers participating in the needs assessment survey.

92 percent of all survey participants agreed that available resources should be used to help prevent or reduce secondary disabilities in people with existing genetic diseases

Identified Needs: Tertiary Prevention

- While a number of services and supports are available to individuals with birth defects and genetic disorders, and their families, they are not being utilized to the greatest extent possible.
- Support or advocacy groups and organizations were the single service used most often by the consumers in our survey, and yet these groups often struggle to survive. There was anecdotal evidence of a need to help maintain local community support groups organized and run by volunteers.
- Factors limiting access include uneven geographic distribution, a shortage of available qualified providers in some areas, and a lack of awareness of services or eligibility guidelines on the part of families and referring providers. Less than one-half (48 percent) of health care providers (n=132) viewed their own role in referring patients to community support services and programs as being very important.
- Many genetic disorders are rare, meaning that practitioners and other support service providers may have only limited experience managing or treating the condition and its complications.
- Patients and their families need information about their condition and options for management or treatment. More than one-quarter (28 percent) of consumers (n=91) reported that they did not have all the information they needed.
- Appropriate medical care is not always received by individuals with genetic conditions: about 54 percent of advocacy organizations (n=77) reported that, in general, they did not think their constituents were getting the medical care and treatment they need for their genetic condition. More than 20 percent of consumers (n=93) reported dissatisfaction with their outpatient care.

Figure 3
Services used for special health needs reported by Michigan consumers
(Data shown by percentages, n=93)



3 Tertiary prevention refers to ameliorating unfavorable consequences of existing birth defects or genetic disorders. Examples include providing educational, dietary, occupational/physical therapy and other support services for individuals with special needs; providing appropriate medical management for genetic conditions; and helping to sustain family support groups and parent-to-parent networks.

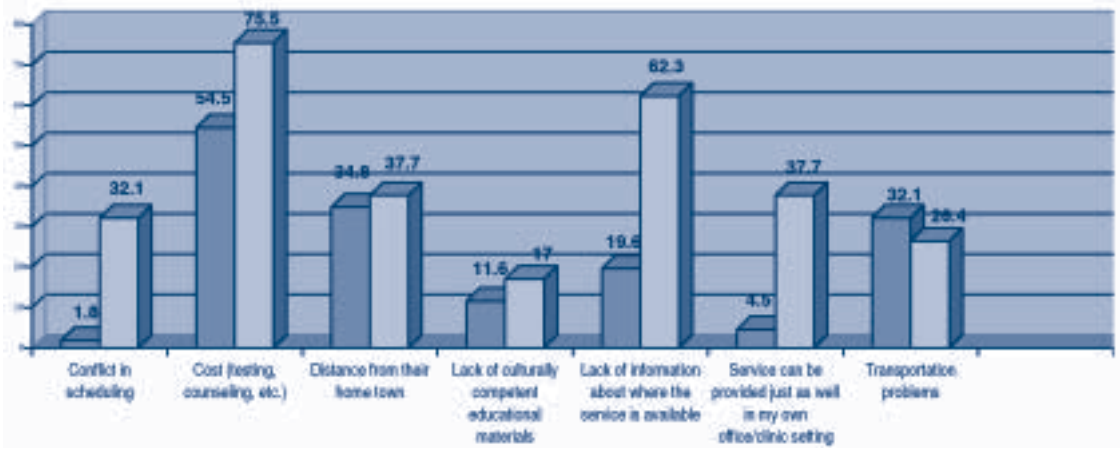
"When we found out, my son was only 3 months old. We got all this bad information. We called the hospital and they let us use the medical library. We went over that stuff for three days before we found a support group from a doctor who knew about it. Until then we were just devastated. You know, we thought we were the only people with this condition on earth."
--A focus group participant

C. Available Services

Current Status & Existing Resources: Family-Focused Services

The existing network of clinical genetic services includes seven hospital or university-based centers located in mid- and southeast Michigan as well as ten outreach sites. Clinic locations and contact information are included in Appendix C. The types of clinics include reproductive, pediatric and adult services including specialty clinics for neurogenetics, cancer and inherited retinal or macular degenerations, although these genetic subspecialties are not available throughout the state. There are approximately 20 practicing board-certified clinical and Ph.D. medical geneticists in the state and 35 certified or board-eligible practicing genetic counselors. Based on 2000 census data the ratio of pediatric geneticists to newborns is about 1: 17,006 infants, while the ratio of geneticists per total population is about 1: 496,222 Michigan residents. More than 10,000 individuals and their families receive genetic diagnostic and counseling services each year. Approximately 8,359 new outpatient visits were reported by 12 genetic clinic directors in the year 2000; 3,084 return outpatient visits and 2,184 inpatient consults were also reported. Advocacy groups and organizations are an important source of information about genetic services for their members and constituents. More than half refer members or clients to genetic counseling services, and 25 percent reported an increase in the number of inquiries received regarding genetic research, testing or treatment compared to the previous year.

Figure 4.
Barriers to referral or utilization of genetic clinic services reported by Michigan health care and genetic service providers
(Data shown in percentages, HCP n=112; GSP n=53)



Identified Needs: Family-Focused Services

- Cancer risk assessment and genetic counseling services are not uniformly available statewide and there is a need to assure the availability of such services in geographically remote regions of the state.
- The leading perceived barriers to utilization of genetic services identified by health care providers and genetic service providers are depicted in Figure 4. These include: cost, lack of public knowledge, patient refusal, and lack of insurance coverage. More than one-third of genetic service providers (n=53) also identified fear of knowing results as a factor, while 41percent identified concerns about confidentiality of results as a barrier to seeking their services. There is a need to make genetic specialty services geographically accessible. About 78% of consumers and the general public (n=187) thought it was very important to be able to see a genetic specialist within their own county. However, 82 percent of the general public (n=94) would be willing to travel up to four hours to get information from a specialist if there was a rare genetic disease in their family. About 19 percent of consumers (n=57) who have already received genetic services had to travel more than 100 miles.
- Acceptable, cost-effective methods of increasing accessibility need to be explored. While the use of telemedicine might improve service availability in geographically remote regions, patients may not be receptive to this modality. More than 4 percent

of consumers (n=53) disagreed that information they received in their genetics clinic visit could effectively be provided this way.

- There is anecdotal evidence that approaches used to evaluate the etiology of developmental delay in young children are neither uniform nor cost-effective.
- There is a need to improve monitoring of the quality of genetic clinic services. Sixty-one percent of 13 clinic directors reported always monitoring patient satisfaction annually, while 23 percent reported this occurring sometimes and 15 percent, "rarely". While more than 70 percent of consumers reported being satisfied or very satisfied with staff sensitivity to their needs and concerns, more than one-third were dissatisfied with referrals to community services and 20 percent were dissatisfied with the follow-up they received after the appointment.
- While most consumers reported overall satisfaction with the services and information they had been given, some dissatisfaction was expressed.

Current Status & Existing Resources: Clinical Laboratory Services

Clinical molecular genetic, cytogenetic, and maternal serum screening studies are currently available in six major medical centers, as well as through several national commercial laboratories. Cytogenetic services are available at approximately two additional hospitals, and several hospital-based clinical chemistry laboratories also offer maternal serum screening tests. It is often necessary to send specimens out of state to specialized reference or research laboratories for diagnostic or carrier tests on rare disorders.

Identified Needs: Clinical Laboratory Services

- There is currently no biochemical genetics reference laboratory in the State of Michigan under the direction of a board-certified clinical geneticist. This represents a significant deficiency in the state's capacity to provide timely confirmatory diagnosis of inborn errors of metabolism.
- Genetic laboratory directors report a shortage of qualified technologists, likely related in part to a relatively low rate of reimbursement for time-consuming genetic laboratory tests.
- There is a need to monitor laboratory quality assurance measures and applications of emerging genetic technology.

Current Status & Existing Resources: Population-Based Screening Services

Prenatal maternal serum screening for neural tube defects and certain chromosome abnormalities is now a part of standard obstetrical care. The American College of Obstetrics and Gynecology has also recently issued a recommendation for cystic fibrosis carrier screening in pregnant women and their partners or those contemplating pregnancy. Carrier screening for certain populations is recommended because of possible increased risk for conditions such as sickle cell anemia, Tay-Sachs disease, and other serious, often degenerative disorders as a result of higher gene frequencies among some ethnic groups. Michigan's Newborn Screening (NBS) Program currently tests infants for seven disorders that benefit from treatment soon after birth. A comprehensive system of follow-up and medical management is included as part of the program. The feasibility of adding other diseases to the screening panel is now being explored but new staff and equipment will be needed in order to implement changes. The Michigan Early Hearing Detection and Intervention (MEHDI) Program promotes community-based newborn hearing screening in hospitals. Efforts are underway to integrate genetic referral and evaluation of infants with confirmed permanent hearing loss into the MEHDI program. Adult screening initiatives have focused primarily on early detection of chronic diseases such as diabetes, hypertension, cardiovascular disease, glaucoma, osteoporosis, and selected cancers. Screening for these conditions includes a variety of methods and approaches but has not generally included genetic testing or an emphasis on inherited factors.

Identified Needs: Population-Based Screening Services

- The quality and availability of prenatal screening appears to vary across the state and there is a need to assist primary care providers in assuring patient access to uniform information and screening services.

More than two-thirds of the general public agreed that the impact of human genetic technology should be examined by state agencies every five years. Nearly half thought genetic technology and testing should be regulated by a state agency while 29 percent were unsure.

Eighty-two percent of the general public felt it was important to have information available about the interaction between the environment (diet, lifestyle, medications, etc) and their genetic makeup as well as workplace exposures that could lead to a genetic disease

- There is a need to expand the NBS program based on the availability of new technology and emerging recognition of potential treatments for rare metabolic disorders in children.
- All aspects of the NBS program need to be aligned with the national taskforce recommendations published in 2000 to assure a high quality program with ongoing evaluation of outcome measures.
- Parents, primary care providers, and hospitals need ongoing in-service education regarding newborn screening procedures and the benefits and limitations of testing. About 60 percent of health care providers (n=119) reported their experience interacting with the NBS system as excellent, while 25 percent reported a poor experience.
- There is a need to identify the role of genetics and further promote integration with existing population screening programs such as MEHDI.
- There is a need to incorporate the use of family history as a tool for eliciting genetic risk factors in screening programs for adult-onset conditions.

D. Research

Current Status & Existing Resources

Numerous clinical and basic genetic research studies are being conducted at the major universities in Michigan and across the nation. Within the state, recent projects range from the molecular genetics of obsessive compulsive disorder to familial psoriasis to macular degeneration. The genetics of smoking and nicotine dependence is being investigated, as is a susceptibility gene for Crohn's disease. However, there is typically a considerable lag time between new discoveries and the development of widespread public health applications. A Center for Genomics and Public Health has recently been established at the University of Michigan School of Public Health through a grant from the Association of Schools of Public Health funded by the Centers for Disease Control and Prevention. The mission of the center is to contribute to the public health genetics knowledge base, primarily in the area of cardiovascular disease (Coronary Artery Calcification and Long QT Syndrome) and provide technical assistance to state and local public health agencies to hasten the integration of genomics into existing programs. Detailed statewide epidemiological assessment of the impact of genetic conditions or service utilization has not been occurring in Michigan, although this would now be possible to some extent based on available public health data collection and management. Given adequate staff resources, existing databases could now be mined to maximize use of available data for program planning, monitoring, and evaluation. Development of genetic health indicators to monitor the population over time would be useful.

Identified Needs

- There is a need to nurture interest in genetic research among the general population and provide access to information about existing research studies. Researchers reported concerns about their ability to recruit subjects for scientific research in light of confidentiality restrictions. More than 16 percent of consumers reported dissatisfaction with the information- (or lack thereof)- they had received about genetic research. About 37 percent of the general public (n= 92) felt they would consider participating in a research study to help society better understand genetically based diseases.
- Existing registries (e.g. cancer, birth defects) are not currently being used to their maximum potential for population research or to identify individuals for clinical research studies.
- Public health data sets are underutilized for analyzing the impact of birth defects and genetic disease in Michigan.
- Scientific knowledge related to gene-environment interaction is growing rapidly, yet there is a large gap in communication between researchers in this field and public health personnel, let alone the general public. There is a great need to explore ways of increasing emphasis on this facet of public health genetics that can lead to potentially cost effective strategies for disease reduction.

E. Education

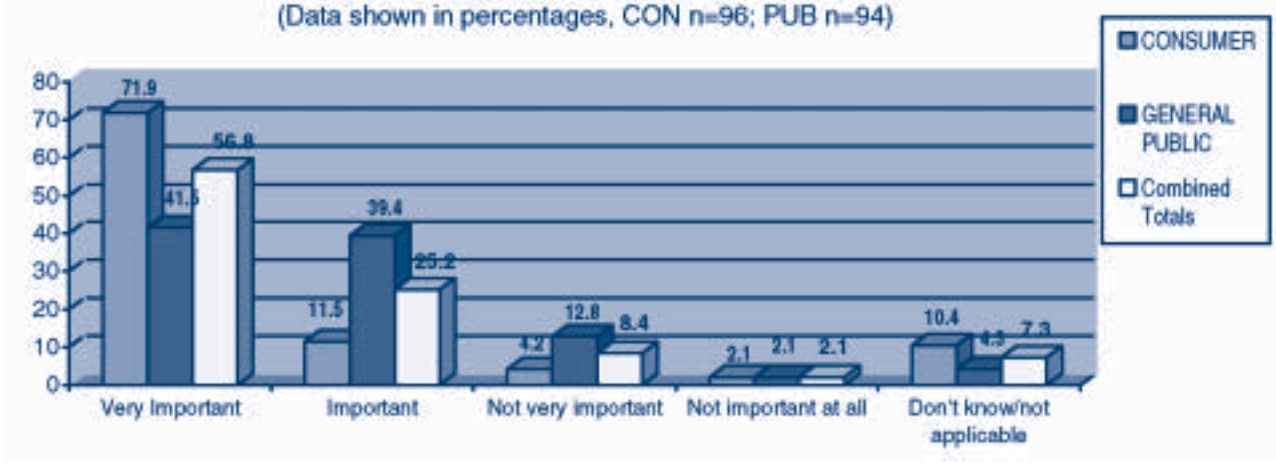
Current Status & Existing Resources

Numerous efforts have been underway in Michigan for more than 20 years to provide genetics education to students, the health care workforce, affected families and other sectors of society. Participants in this process include primary and secondary teachers, post-secondary professors, genetics providers, public health personnel, the Michigan Department of Education and local school districts, the media, and many others. Federal agencies, non-profit organizations, and the commercial sector also play a major role in developing teaching materials, textbooks, curricula, and professional competencies. Despite all this, the pace of discovery continues faster than our ability to educate the public about the implications of genetics for their decisions regarding health and disease.

Identified Needs

- There continues to be a tremendous need, as well as support, for educating all sectors of the population about genetics and the role genes play in health and disease. More than two-thirds (68 percent, n= 227) of local health departments and health care providers wish that their clients and patients had more background knowledge about genetics.
- Major avenues for increasing the level of genetic literacy include: the media and classroom teachers. Educating physicians and other health care providers about advances in medical genetics and birth defects would also provide a positive impact.
- About 75 percent of teachers (n=165) need to learn more about implications of the Human Genome Project for the subject area they teach. Of those (n=96) who indicated they were uncomfortable teaching topics related to human genetics in health and disease prevention, 36 percent said they had too little knowledge about the subject
- Local public health departments believe they play an important role in educating the public about possible gene-environmental interactions that might affect a person's health status (68 percent, n= 105), and therefore need better access to current state-of-the-art information to support this role.
- Physicians and other health care providers are the leading source of health information for consumers, followed by support groups or organizations, the Internet, media, and family members as well as self help reference books or brochures. This was generally true for the public as well, except the media played a slightly more important role and support groups were not a major source.
- Health care and local public health providers obtain information about new advances primarily from continuing education seminars and conferences, medical journals, the media, and health professional colleagues.

Figure 5
Importance of having a central source that provides information about genetic diseases and services for families
(Data shown in percentages, CON n=96; PUB n=94)



- Culturally sensitive educational materials are needed to promote genetic messages. Two-thirds of the general public and consumers (n=190) felt that having information about genetics in their primary language was very important.
- There appears to be a workforce shortage of professionals adequately trained in human genetics and therefore a need to promote careers in this field.
- A central source is needed to serve as a portal for information about genetic diseases and services within the state. This is especially important to individuals and families affected with birth defects and genetic conditions but also supported by the majority of the general public as illustrated in Figure 5.
- While substantial clinical genetics expertise is available in the state, these professionals generally have little time to write educational articles or participate in training and outreach education activities unrelated to their clinical work with patients. Of the genetic service providers who responded to the survey, about one-half spend roughly 25 percent of their time teaching or lecturing and 41 percent spend no time at all on such educational activities.
- There is a lack of knowledge among the public and health care providers about Michigan's genetic privacy legislation and requirement for informed consent before genetic testing, even though nearly 75 percent of consumers and the general public (n=186) felt it was important or very important to be able to find out about existing laws, and two-thirds of the general public respondents (n=93) were concerned that genetic information might be used to discriminate against some people.

F. Data Collection and Documentation

Current Status & Existing Resources

Improvement of the public health infrastructure for data collection, management and analysis is needed and already in progress through a major MDCH initiative to develop a data warehouse. The warehouse will significantly increase current capacity to link existing data sets such as birth and death records, newborn screening, Medicaid, Children's Special Health Care Services, WIC, and others. Such linkages between public health data sources will be beneficial for improving the planning, assessment and assurance functions related to the genetics and newborn screening program. In addition, a new NBS database is currently under development that will enhance all aspects of record-keeping, reporting and tracking newborn screening specimens and results. An epidemiologist has been hired to analyze data in the Birth Defects Registry. Most genetics clinics maintain a database of patients seen, although such demographic data are not submitted to MDCH. Pediatric genetics clinics participate in birth defects reporting to MBDR, and most of the reproductive genetics clinics submit non-identifiable data on birth defects diagnosed prenatally.

Identified Needs

- There is a need to develop more capacity for genetic assessment. Approximately 64 percent of genetic service providers, health care providers and local health departments (n= 276) indicated that a better system of data infrastructure at the state level would help to facilitate planning and evaluation of genetic health care needs.
- Advocacy organizations also felt they could benefit from access to statistical data. About 23 percent of respondents (n=86) currently use MDCH data and information systems while 40 percent felt their organization could benefit from such data and 20 percent wanted to know more. Specific areas of interest that were expressed included: diagnosis rates of attention deficit and hyperactivity disorders; treatments, surgeries, and complications associated with celiac disease; cerebral palsy trend data; counseling services for individuals with dyslexia; Rett syndrome support groups; connective tissue disease data including scleroderma and lupus; primary immune deficiency qualified providers and access to services; data and information systems for developmental disabilities; and deaf and blindness issues.

G. Funding

Current Status & Existing Resources

Clinical genetic and laboratory services are financed primarily through fee-for-service charges and reimbursement by third party payers, including the Medicaid system. The reimbursement is low compared with the time-intensive nature of most genetic evaluations. Although master's degree level genetic counselors are a cost-effective adjunct to physician services, national billing codes for genetic counseling do not currently exist. Thus, funding for genetic services continues to be a major issue in assuring availability of center-based services. The network of 10 outreach clinic sites has been supported to a large extent by Children's Special Health Care Services and the Hereditary Disorders Program since its inception. The NBS program is directly supported by the fee charged to screen each newborn.

Identified Needs

- Adequate reimbursement mechanisms for genetic counselors do not currently exist and funding for direct patient services is an ongoing problem, especially in the pediatric setting.
- It is often difficult for scientists to find funding sources for *clinical* (as opposed to *basic)* genetics research
- Out of state reference laboratories frequently will not accept Michigan Medicaid or insurances - this is a major and growing problem for many clinics and patients.
- Existing funding mechanisms need to be systematically examined, and ways sought to maximize available reimbursement through third party payers.
- Methods of demonstrating the cost-effectiveness of genetic diagnosis, testing, and counseling services are needed. Studies are needed to assess improved health outcomes from prenatal diagnosis that can lead to life-saving procedures, to examine the relationship between the number of children identified with particular genetic disorders and the financial costs associated with providing appropriate care, or to demonstrate costs saved through preventive care for adult-onset disorders.

More than 60 percent of the general public and consumers said it was very important to them to have genetic testing and counseling covered by their insurance. More than half of health care providers felt that a lack of insurance coverage and associated costs would keep them from referring a patient to a genetics clinic.

APPENDIX C:
GENETIC RESOURCES IN MICHIGAN

(as of 2002)

Clinical Genetic Centers

- Henry Ford Hospital *(Detroit)*
  - Department of Medical Genetics- Pediatric, Reproductive, Adult, Cancer and Neurogenetics (313) 916-3188
- Michigan State University *(East Lansing)*
  - Pediatric, Reproductive and Adult Genetics (517) 353-2030
- Oakwood Hospital *(Dearborn)*
  - Clinical Cytogenetics- Reproductive and Cancer Genetics (313) 593-8483
- Spectrum Health *(Grand Rapids)*
  - Pediatric, Reproductive, Adult, and Cancer Genetics (616) 391-2700
- St. Joseph Mercy Health System *(Ypsilanti)*
  - Reproductive Genetics (734) 712-7903
- University of Michigan Health System *(Ann Arbor)*
  - Breast and Ovarian Cancer Risk Evaluation Program (734) 764-2248
  - Medical Genetics Clinic- Adult and Cancer (734) 763-2532
  - Inherited Retinal and Macular Degenerations (734) 763-5906
  - Neurogenetic Disorders Clinic (734) 936-8173
  - Pediatric Genetics Clinic- includes Biochemical Genetics (734) 764-0579
  - Pediatric Neurology Metabolic Clinic (734) 763-4697
  - Perinatal Assessment Center (734) 764-6834

- Wayne State University/Detroit Medical Center *(Detroit)*
  - Karmanos Cancer Institute- Cancer Genetic Counseling Service (313) 966 -7780
  - Children's Hospital- Pediatric Genetics Clinic (313) 745-4513
  - Harper Hospital- Neurogenetics Clinic(313) 577-8317
  - Hutzel Hospital- Reproductive Genetics Clinic (313) 745-7067

- William Beaumont Hospital *(Royal Oak)*
  - Pediatric Genetics Clinic (248) 551-0847
  - Reproductive Genetics Clinic (248) 551-0395

Clinical Genetic Specialty Providers

- ~20 Clinical Geneticists (MD/DO)
- 4 Medical Geneticists (Ph.D.)
- ~35 Genetic Counselors (M.S.)
- ~6 Genetic Nurse Specialists (R.N.)

Newborn Screening

- Newborn Metabolic Screening Program (517) 335-9205
- Michigan Early Hearing Detection and Intervention Program (517) 335-8878

Public Health Genetics

- Hereditary Disorders Program (517) 335-8887
  - Genomics and Adult Genetics; Birth defects prevention and follow-up
- Children's Special Health Care Services (517) 241-7186
- Division of Chronic Disease and Injury Control (517) 335-8368
- Division of Family and Community Health (517) 335-8928
- Bureau of Epidemiology (517) 335-8900
  - Maternal and child health, chronic disease, environmental, and infectious disease epidemiology
- Bureau of Laboratories (517) 335-8063

Surveillance Systems

- Behavioral Risk Factor Surveillance System
- Michigan Birth Defects Registry (517) 335-8678
- Michigan Cancer Registry (517) 335-8678

Training Programs

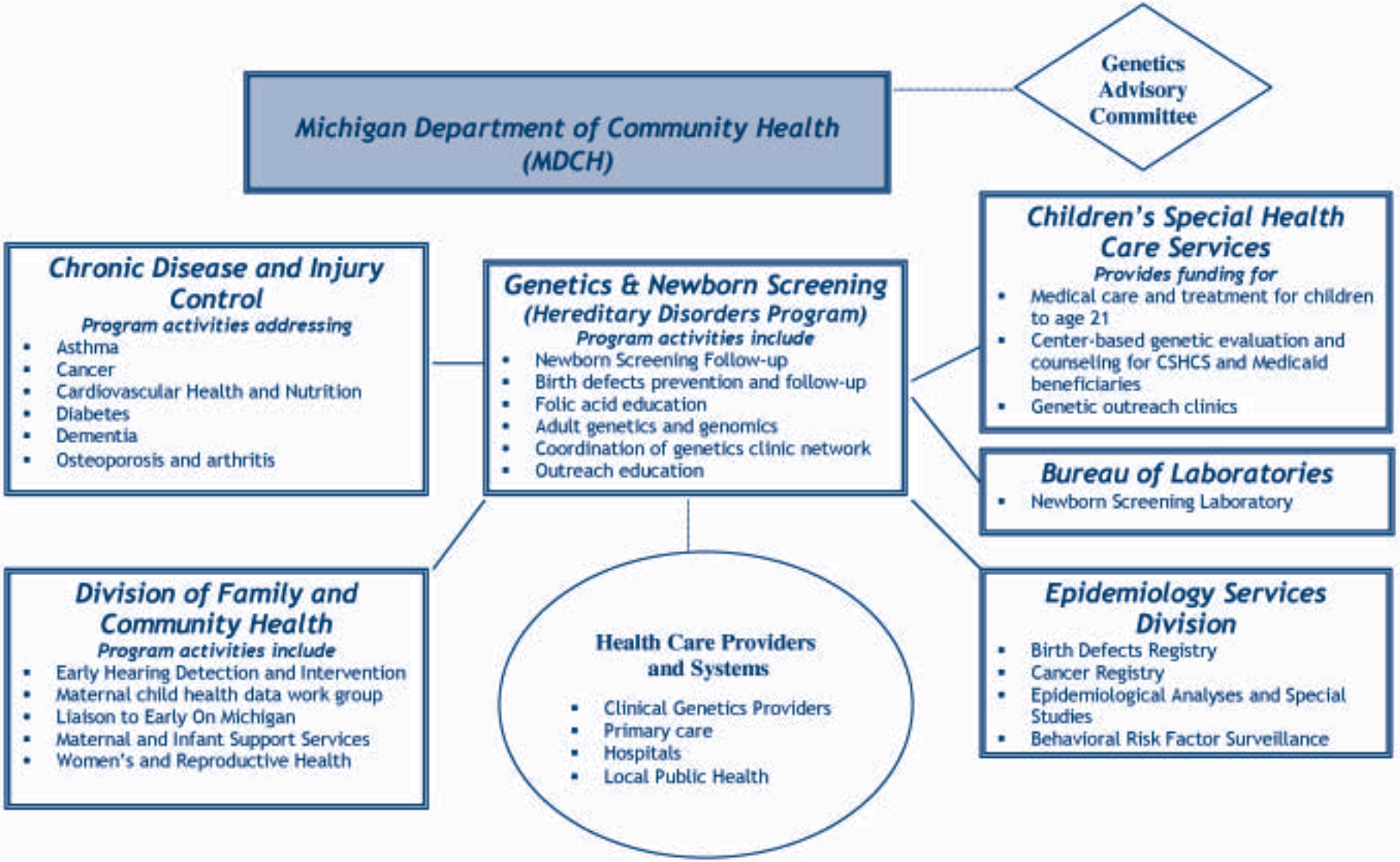
- Three Genetic Residency Programs (University of Michigan, Wayne State University, Henry Ford Health System)
- Two Genetic Counselor Training Programs (University of Michigan, Wayne State University)

APPENDIX D: KEY MICHIGAN LEGISLATION

Act 368 of 1978 (Public Health Code)	Requires the public health department to establish a chronic disease prevention and control program including arthritis, cancer, dental disease, diabetes, *genetic disease*, heart disease, hypertension, renal disease, and any other disease the department designates as chronic… Also requires the department to cooperate with the department of mental health in establishment of a statewide program for genetic screening and counseling in the area of mental disabilities.
Act 14 of 1987	Expands the newborn screening program to six disorders, designates a central laboratory for testing, and authorizes collection of a fee to support program costs.
Act 236 of 1988 (H.B. 4007)	Amends the public health code to require reporting of each diagnosed incidence of a birth defect, congenital or structural malformation, or a biochemical or genetic disease, to the state public health department.
Act 26 of 2000 (S.B. 589)	Prevents health care corporations from requiring members or their dependents, or applicants for coverage to: undergo genetic testing before issuing, renewing or continuing a health care corporation certificate; or to disclose whether genetic testing has been conducted or the results of genetic testing or genetic information.
Act 27 of 2000 (S.B. 590)	Prevents hospital, medical or surgical policies from requiring an insured or his or her dependents, or asymptomatic applicants for insurance to: undergo genetic testing before issuing, renewing or continuing a policy or certificate; or to disclose whether genetic testing has been conducted or the results of genetic testing or genetic information.
Act 28 (S.B. 591)	Prevents a health maintenance organization from requiring an enrollee or his or her dependent or an asymptomatic applicant to: undergo genetic testing before issuing, renewing or continuing a health maintenance organization contract; or to disclose whether genetic testing has been conducted or the results of genetic testing or genetic information.
Act 29 of 2000 (S.B. 593)	Amends the public health code to require a physician or his/her delegate to obtain written, informed consent of the test subject before ordering a presymptomatic or predictive genetic test.
Act 30 of 2000 (S.B. 594)	Regulates disposal of DNA samples and identification profile records in criminal investigations.
Act 31 of 2000 (S.B. 595)	Regulates the confidentiality of genetic testing material used for paternity determinations.
Act 32 of 2000 (S.B. 815)	Prevents employers from failing or refusing to hire, recruit, or promote an individual because of a disability or genetic information that is unrelated to the individual's ability to perform the duties of a particular job or position; and from discharging or otherwise discriminating against an individual because of a disability or genetic information.
Act 33 of 2000 (S.B. 807)	Amends the public health code regarding newborn screening to allow health professionals and hospitals to offer to draw an additional blood specimen from an infant to be retained by the parent for future identification purposes.
Act 691 of 2002 (H.B. 5998)	Amends the public health code regarding newborn screening to increase the fee and add testing for medium chain acyl-coenzyme A dehydrogenase deficiency (MCAD)

APPENDIX E:
KEY PLAYERS IN STATE PUBLIC HEALTH GENETICS INITIATIVES

(as of 2002)



APPENDIX F: FEDERAL INITIATIVES in PUBLIC HEALTH GENETICS

(as of 2002)

Department of Health & Human Services (HHS)

Secretary's Advisory Committee on Genetic Testing

http://www4.od.nih.gov/oba/sacgt.htm

Advises HSS on the medical, scientific, ethical, legal, and social issues raised by the development and use of genetic tests

National Institutes of Health (NIH)

Numerous institutes conduct basic scientific health research pertaining to genetics, including:

National Human Genome Research Institute
http://www.nhgri.nih.gov/
Lead agency for the publicly funded Human Genome Project, including ethical, legal, and social implications.

National Cancer Institute,
Genes & the Environment Initiative
http://www3.cancer.gov/initiatives/grp-cancer.html
Purpose: To discover genetic, environmental, and lifestyle factors and their interactions that define cancer risk and inform strategies for cancer control.

National Institute for Environmental Health Sciences, *Environmental Genome Project*
http://www.niehs.nih.gov/
includes National Center for Toxicogenomics

National Institute of General Medical Science
Pharmacogenetics Research Network
http://www.nigms.nih.gov/pharmacogenetics

Centers for Disease Control & Prevention (CDC)

Office of Genomics and Disease Prevention
www.cdc.gov/genomics/ Seeks to integrate advances in human genetics into public health research, policy, and programs. Provides a weekly electronic newsletter and. funds Centers for Genomics & Public Health at the University of Michigan, School of Public Health; the University of North Carolina and the University of Washington

National Center for Birth Defects and Developmental Disabilities
www.cdc.gov/ncbddd/ provides national leadership for preventing birth defects and developmental disabilities and improving the health and wellness of people with disabilities national Center

Health Resources & Services Administration (HRSA)

Maternal & Child Health Bureau, Genetic Services Branch
http://mchb.hrsa.gov/programs/
Supports newborn screening and funds projects to increase professional and public knowledge of how genetic diseases affect health:

- State Genetics Planning & Implementation Grants
- National Newborn Screening & Genetics Resource Center
  http://genes-r-us.uthscsa.edu/
- Gene Tests: Medical Genetics Knowledge Base
  http://www.genetests.org/
- Genetics in Primary Care: A Faculty Development Initiative
  http://bhpr.hrsa.gov/dm/genpc.html

Some National Organizations With Public Health Genetics Activities:

Association of State & Territorial Health Officials — www.astho.org/phiip/genetics.html

National Association of Chronic Disease Directors — www.chronicdisease.org/

National Coalition for Health Professional Education in Genetics — www.nchpeg.org

Coalition of State Genetics Coordinators — www.stategeneticscoordinators.org

National Society of Genetic Counselors — www.nsgc.org

Notes

Notes

F*or more information, call the MDCH state genetics program at (517) 335-8887 or e-mail genetics@michigan.gov*

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