

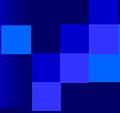
An Overview:

Genomics and Birth Defects Programs



Janice Bach, Genomics Unit Manager

Division of Genomics, Perinatal Health
& Chronic Disease Epidemiology
genetics@michigan.gov



From 20th to 21st Century: **A paradigm shift**

■ **“Medical model” addressing rare disorders**

- Newborn Screening
- Maternal and Child Health
- Clinical Genetic Services

■ **“Public health model” addressing common diseases**

- Many people affected
- Gene-environment-behavioral interactions
- Major impact on public health
- Potential for prevention
- Focus on core public health functions & 10 essential services

From "Genetics" to "Genomics"

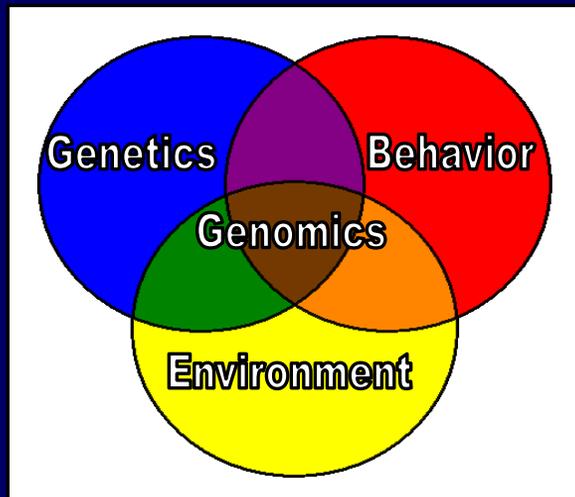
Genetics

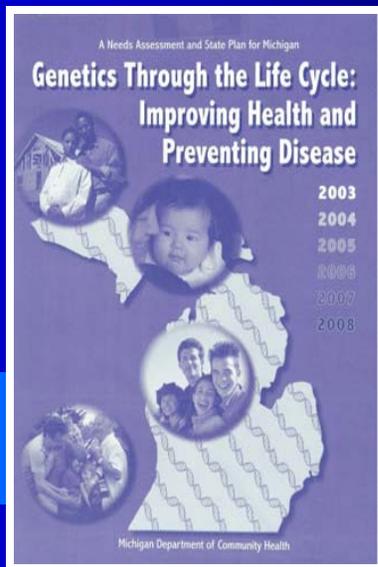
The science of heredity; refers to a single gene and its effects



Genomics

The study of the entire genome including the complex interactions among multiple genes as well as between genes and the environment





A Vision for the Role of Genetics in Public Health

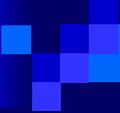
Michigan Genetics Plan 2003-2008

Improved health outcomes and an enhanced quality of life for the people of Michigan through appropriate use of genetic information, technology, and services.

MDCH Public Health Genomics Program Mission

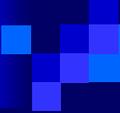


- Coordinate educational activities that increase genetic literacy;
- Facilitate early identification and treatment of individuals with birth defects, heritable disorders and susceptibilities; and
- Foster collaboration to integrate advances in genomic science throughout public health and other systems of care



MDCH Public Health Genomics Program Components

- Newborn Screening Unit
- Genomics Unit
 - Birth Defects Prevention and Follow-up
 - Genomics and Chronic Disease
 - Clinical Genetic Services
 - Genetics Education/Resource Center



MDCH GENE TEAM (2007)

Janice Bach

State Genetics Coordinator

Joan Ehrhardt

Birth Defects Program
Coordinator

Jane Simmermon

Birth Defects Follow-up Nurse

Birth Defects Epidemiologist

Valerie Ewald

Administrative Assistant

Deb Duquette

Genomics Coordinator

Mary Teachout

Genomics Educator

Ann Annis-Emeott

Genomics Epidemiologist

John Gehring

Gene-Environment Specialist

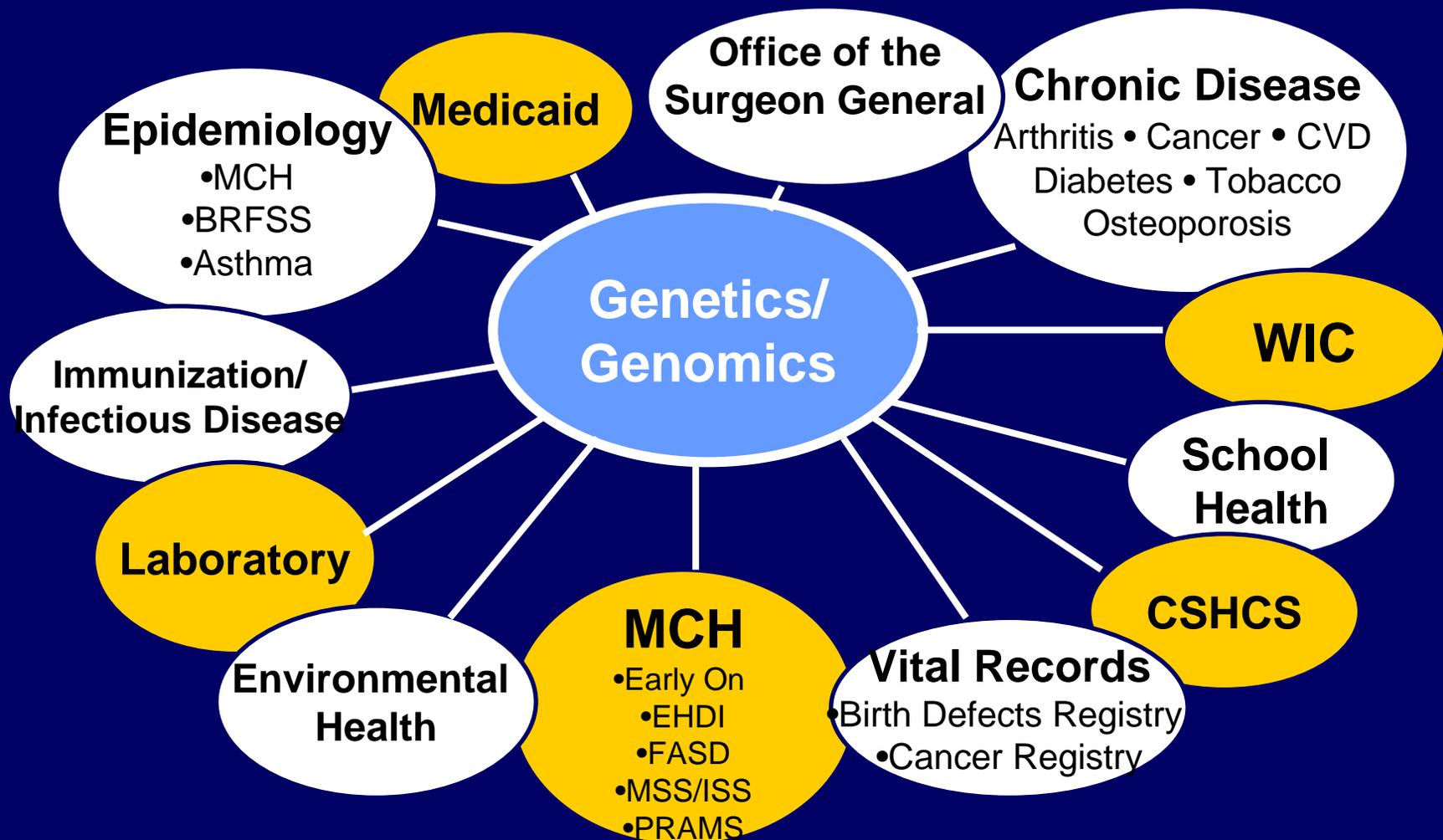
Carrie Langbo

Clinical Services Liaison

Champa Bhatia

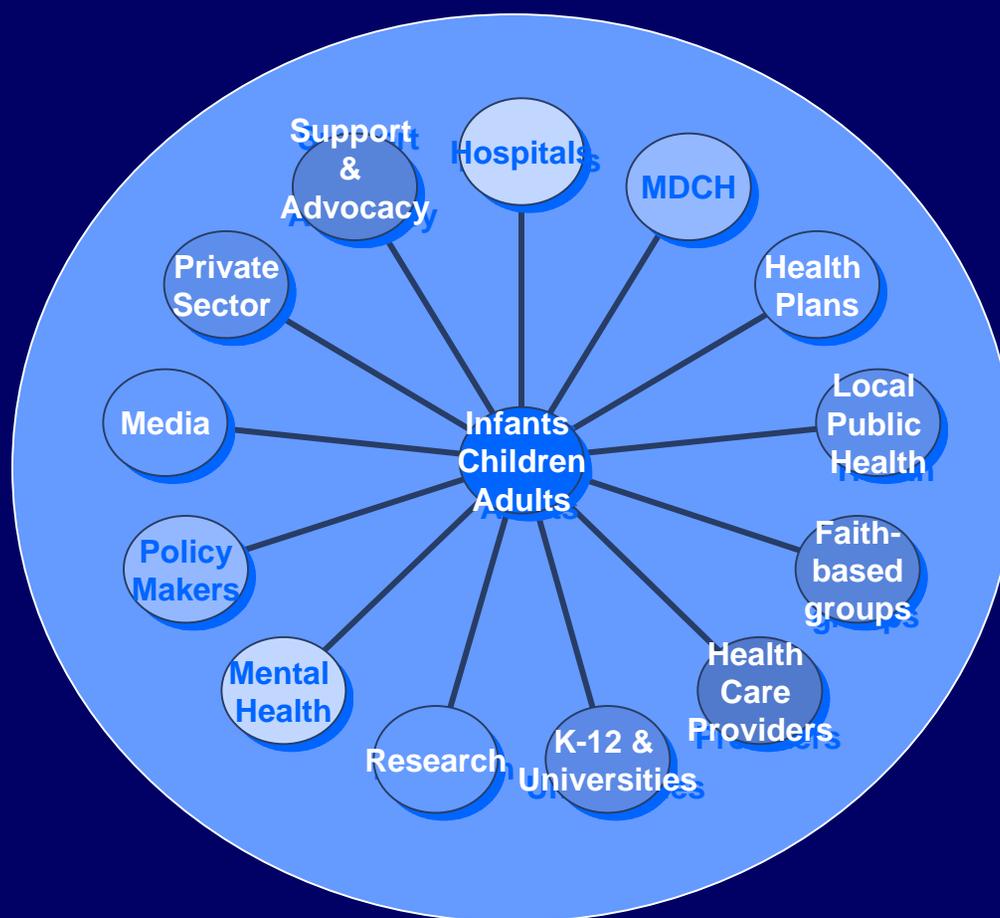
Reimbursement Specialist

MDCH Genomics Program relies on partnerships and collaborations



External Partnerships

A Genetics Plan for the Life Cycle





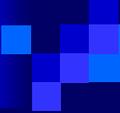
Region4
Genetics Collaborative

- **HRSA: Enhance and support the genetics and newborn screening capacity of States across the nation**
 - Promote translation of genetic medicine into public health and health care services
 - Address maldistribution of genetic resources
 - Ensure that individuals with heritable disorders have access to quality care and appropriate genetic expertise in the context of a medical home

- **Region 4 Coordinating Center: MPHI** (Cindy Cameron, project director)
 - Illinois, Indiana, Kentucky, Michigan, Minnesota, Ohio and Wisconsin
 - Six goals related to improving access to genetic expertise and care coordination for children with heritable disorders in the context of a medical home
 - Special projects related to use of tandem mass spec. technology and long term follow-up of children with metabolic diseases



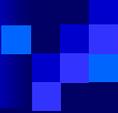
Region 4



CDC Cooperative Agreement for Population-based Birth Defects Surveillance Programs and the Utilization of Surveillance Data by Public Health Programs, 1999- 2010

Goals:

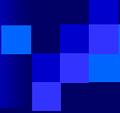
- Improve, expand and evaluate Michigan's population-based birth defects surveillance system
- Implement and evaluate a population-based birth defects prevention program
- Expand and evaluate the effectiveness of activities to improve access to health services and early intervention programs for children with birth defects and their families



CDC Cooperative Agreement for Chronic Disease Prevention and Health Promotion: Genomics Component, 2003-2008

Goals:

- Enhance state and local leadership capacity for integrating genomics into chronic disease prevention and health promotion programs
- Strengthen MDCH organizational capacity for developing and implementing population-based assessments; and utilize existing surveillance and data systems for ...the integration of genomics into public health programs
- Educate the health workforce, policymakers, and general public about the role of family history and genetic risk factors in chronic disease...
- Expand the use of genomics in core public health programmatic activities ...
- Plan and coordinate the use and evaluation of targeted risk assessment strategies based on genomics and/or family history tools...



1. Increase genetic literacy in the State of Michigan

■ **General Public**

Family History and Your Health newsletters

Genetics to Genomics for Teachers

Genealogy groups

■ **Public Health Workforce**

MDCH Cancer Section seminar series

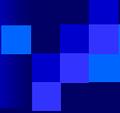
Local public health in-service trainings

■ **Health Professionals**

Conference planning and presentations: Healthy Mothers

Healthy Babies; Family History in Primary Care; Diet and DNA

Diabetes ED1 and ED2 modules

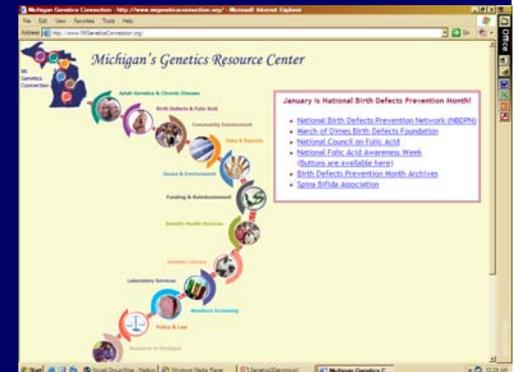


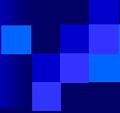
2. Assess the public health impact of heritable conditions and utilization of genetic services

- **Birth Defects Registry**
- **Cancer Registry**
 - Early Onset Deaths; Family History Collection
- **Sudden Cardiac Death of the Young Mortality Review System**
- **Population Surveys**
 - PRAMS
 - BRFS 2004, 2005, 2006, 2007:
 - Provider practice/family history
 - Colorectal Cancer Family History
 - Breast Cancer Family History
 - Direct-to-consumer genetic testing
 - Sudden Cardiac Death

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

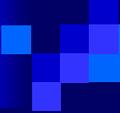
- www.MiGeneticsConnection.org
- Call toll-free 1-866-852-1247 or e-mail genetics@michigan.gov
- Family-to-Family Health Information and Education Center
- Genetics Clinic Network
- Genetic Services Payment Program
- NTD follow-up
- Brochures and publications
- Birth defects resource kit
- Conference exhibits and workshops
- Press releases; governor's proclamations
- Birth Defects Prevention Month & Folic acid awareness campaign
- Multi-vitamin distribution to low income women in high risk counties (March of Dimes grant)
- NTD family history study





4. Promote early identification and treatment of individuals with ...heritable disorders or genetic susceptibilities

- Genetics Education for Early On[®] providers
- Cancer risk assessment guidelines
- WISEWOMAN family history education
- Healthy Homes University
- Partnerships with EHDI, CSHCS, FIMR, Mental Health (Autism Spectrum Disorders), FASD Task Force

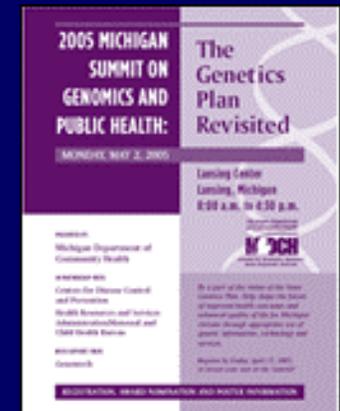


5. Identify best practices and promote a policy framework to assure high quality services, supports and genetic privacy protections

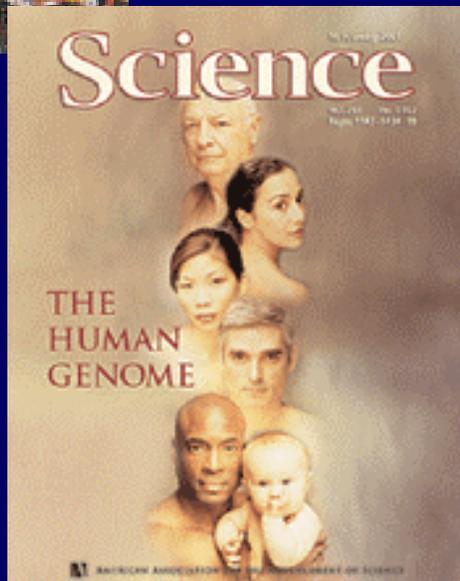
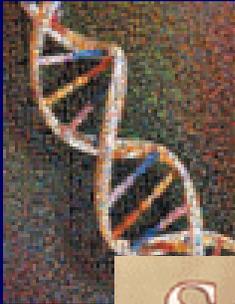
- **Informed consent for genetic testing**
- **Collection and use of family history in primary care**
 - Health plans and systems
- **Hospital best practice guidelines for birth defects referral**
- **Collaboration with CSHCS**
 - Medical home
 - Genetic services payment program & reimbursement for genetic counseling

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

- 2005 Summit on Genomics and Public Health
- Michigan Cancer Genetics Alliance
- CSHCS/Medicaid reimbursement for new genetic tests
- Genomics Work Group
- Birth Defects and Cancer Genetics Listservs
- Integration of genomics in chronic disease programs
- Gene-environment Work Group
- Development of Neonatal BioTrust



The Promise of Genomics for Public Health



- Early identification of at-risk populations
- Targeted interventions to high-risk populations
- Improved responses to acute public health events