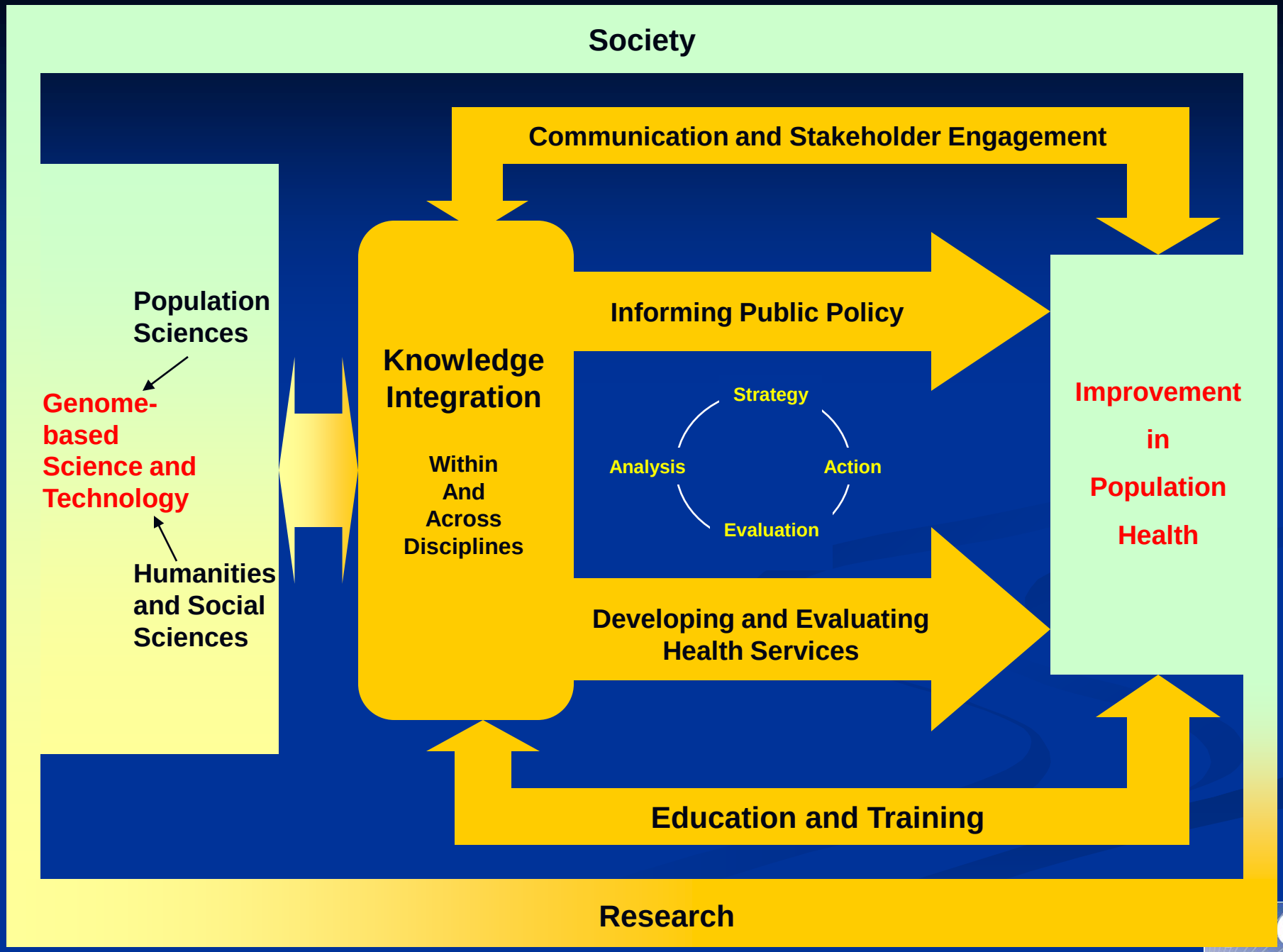
Public Health Core Functions and Essential Services in Integrating Genomics into Practice

Muin J. Khoury, MD, Ph.D.

CDC National Office of Public Health Genomics

Acknowledgements

- Debbie Klein Walker
- Jon Kerner
- Scott Grosse
- Laura Beskow
- Marta Gwinn
- Katrina Goddard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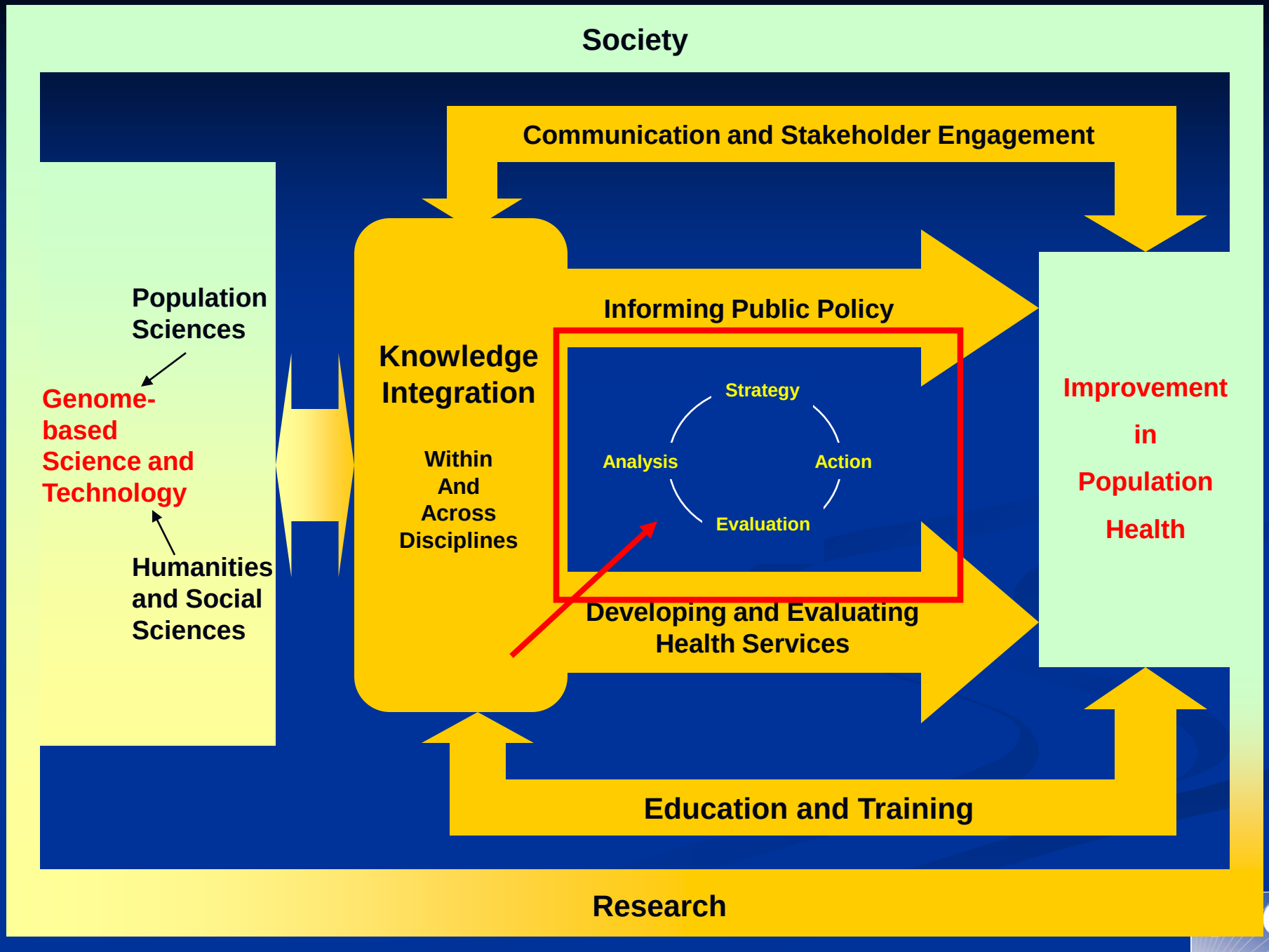
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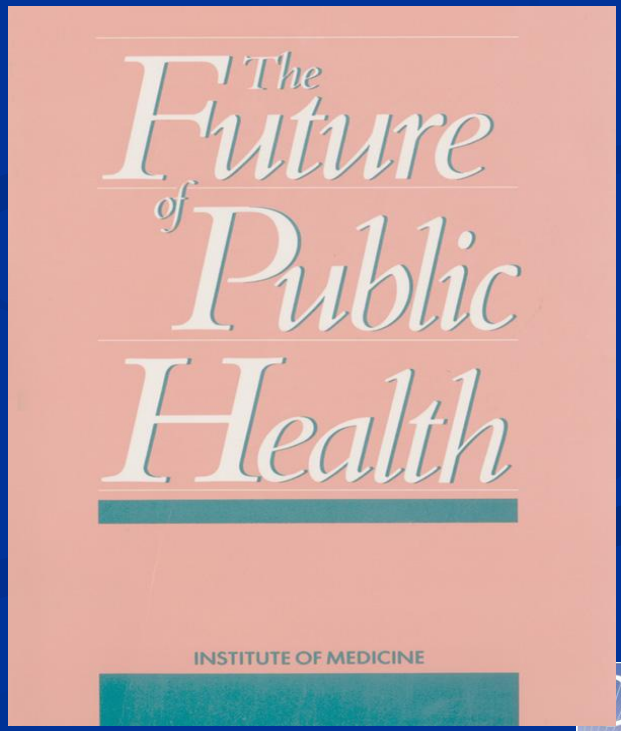
Outline

- What is Public health?
- Public Health and the Paradigm Shift from Traditional Genetics to Genomic Medicine
- Core Public Health Functions and Essential Services
- Examples: from Newborn Screening to DTC Genetic Testing

Definition of Public Health

"Public health is what we do collectively to fulfill society's interest in assuring the conditions in which people can be healthy."

Institute of Medicine, 1988



Who Will Keep the Public Healthy? (IOM, 2002)

"Actors in the Public Health System"

Characteristics of the US Health Care System

- Escalating costs and unequal access
- Considerable variation in
  - primary care and specialists per population
  - coverage for vulnerable populations
  - coverage of mental health, substance abuse treatment and dental health
- Expanding technologies and treatments
- Financial pressures to control costs
- Increased push for quality and electronic health records

Source: D Klein-Walker, President APHA, 2007

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Health Coverage Today in the US

- Single payer system for 65 years plus
- Medicaid is insurer for poor and vulnerable in US today
- Many uninsured Americans (~ 50 million)
- Attempted universal coverage several times in past
- Children and youth are closest to universal coverage in many states
- State experiments are evolving solutions
- Health care viewed as a right

Source: D Klein-Walker, President APHA, 2007

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"System" Differences for BRCA testing: US vs. UK

US (Individual)

- Access to findings restricted for fear of discrimination
- Level of testing based on ability to pay
- Counseling not required
- Physicians facilitators to increase demand
- Individuals empowered to make own decisions but also assumed to be informed consumers of testing

UK (Population)

- Universal access to health care mitigates concerns
- Level of testing restricted by fixed allotments
- Counseling provided
- Physicians gatekeepers to manage demand
- Individuals represent patient groups (low, moderate & high risk) whose access to testing should be managed by health professionals

From J Kerner

Parthasathy S. *Social Studies of Science* 35/1 (2005): 5-40.

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"Public Health Practice" in the United States

- Federal
  - HHS: CDC/HRSA/FDA/AHRQ/CMS/etc..
  - Other Agencies
- State/Local
  - State health departments
  - Local health departments

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The Challenge of Moving from Genetics to Genomics

Genetic Disorders	Genetic Information
Mendelian Disorders	**All Diseases**
Disease burden: 5%	**Disease Burden: 95%**
Mutations/One Gene	**Variants/MultiGenes**
High Disease Risk	**Low Disease Risk**
Environment +/-	**Environment ++**

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Delivery System for Traditional Genetics

- Key for individuals and families with conditions
- Most people not directly affected
- Relatively rare and, therefore, small role in health care (and in society)
- Care supplied primarily by medical geneticists and genetic counselors with occasional involvement of other medical specialists and primary care providers

D Klein-Walker, APHA president 2007

Delivery System for Genomic Medicine

- Care will be supplied primarily by primary care and other healthcare providers with occasional involvement of medical specialists and genetic counselors
- The role of family history will be extremely important in determining appropriate behavior and care

D Klein-Walker, APHA president 2007

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What Role of Public Health in Genetics and Vice Versa?

Holtzman NA, Comm Genet 2006

- "The only genetic service for which a public health role is paramount is newborn screening"
- "There is little need for further integration of genetic services and education into public health especially in countries in which public and private health services are dichotomized."

The Challenge of Moving from Genetics to Genomics

- Genetic Disorders
- **Mendelian Disorders**
- **Disease burden: 5%**
- **Mutations/One Gene**
- **High Disease Risk**
- **Environment +/-**

- **Role of Public Health Agencies**
- **Newborn screening programs**
- **Delivery of genetic services**

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The Challenge of Moving from Genetics to Genomics

- **Role of Public Health Agencies?**
- **Explore the "wheel"**

- Genetic Information
- **All Diseases**
- **Disease Burden: 95%**
- **Variants/MultiGenes**
- **Low Disease Risk**
- **Environment ++**

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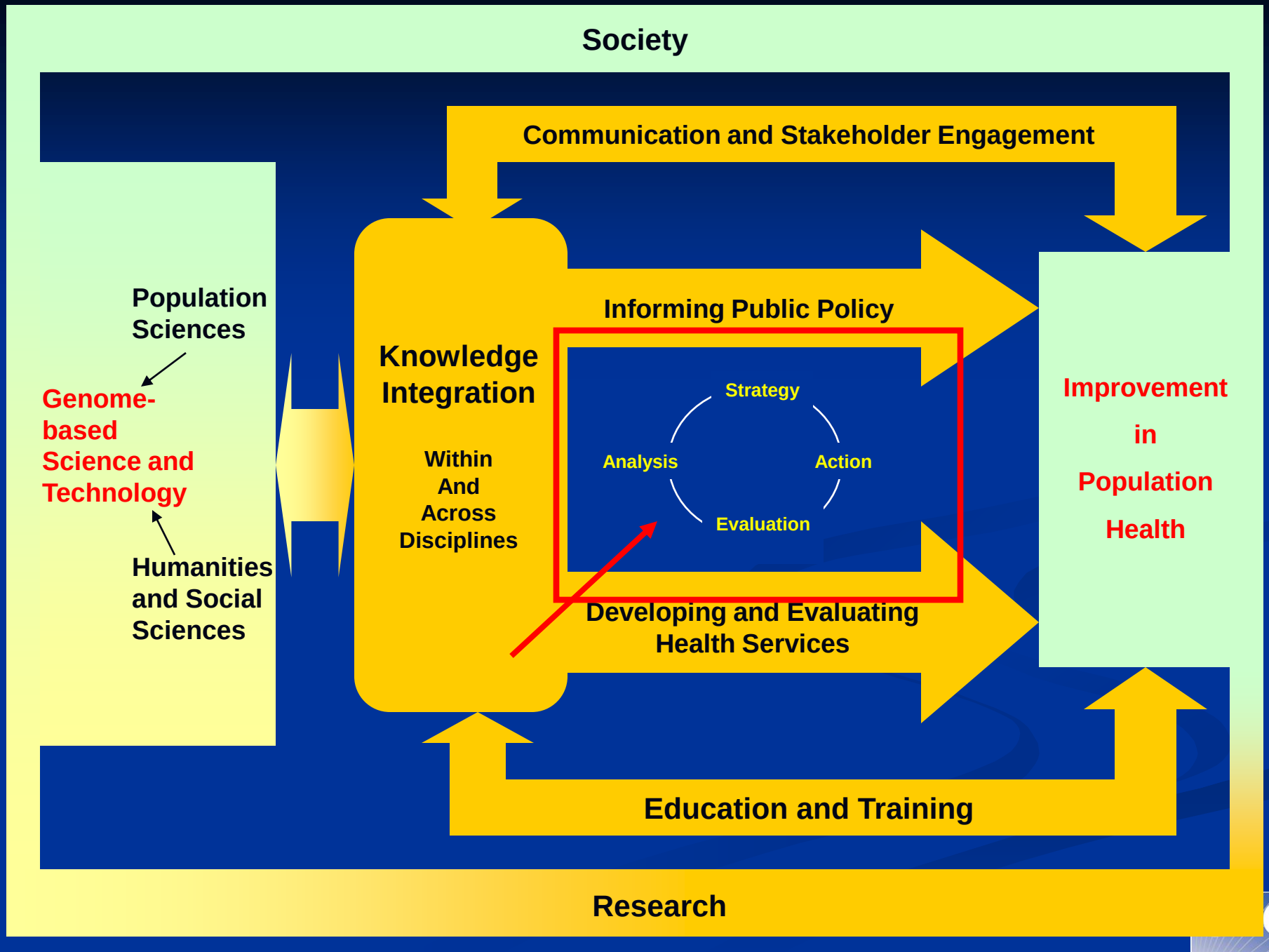
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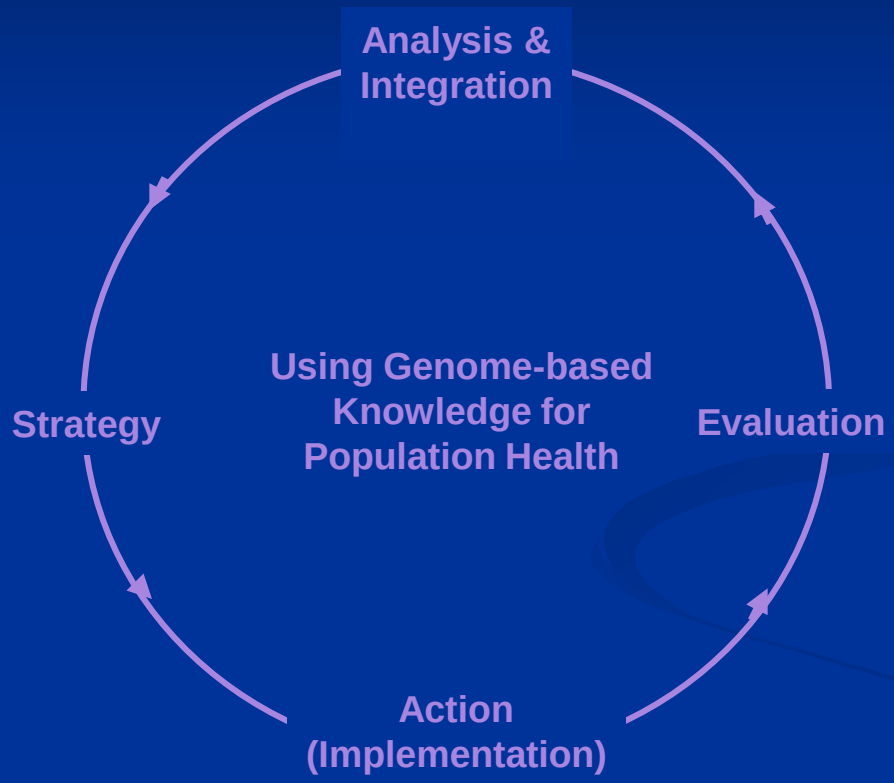
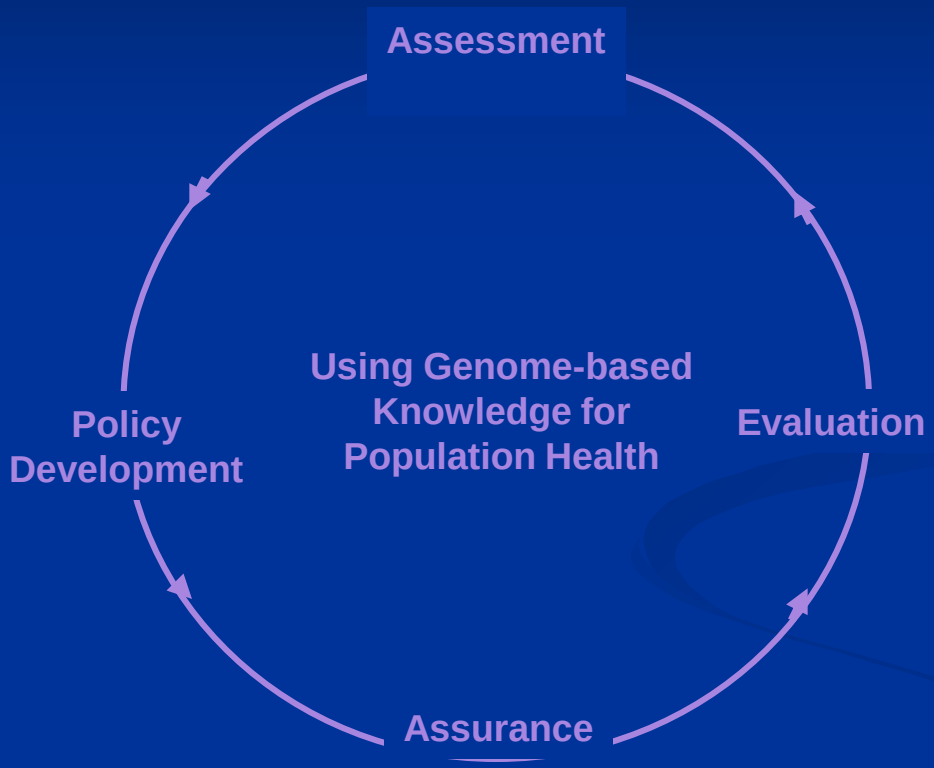
The Cycle of Public Health Practice: The Bellagio Report

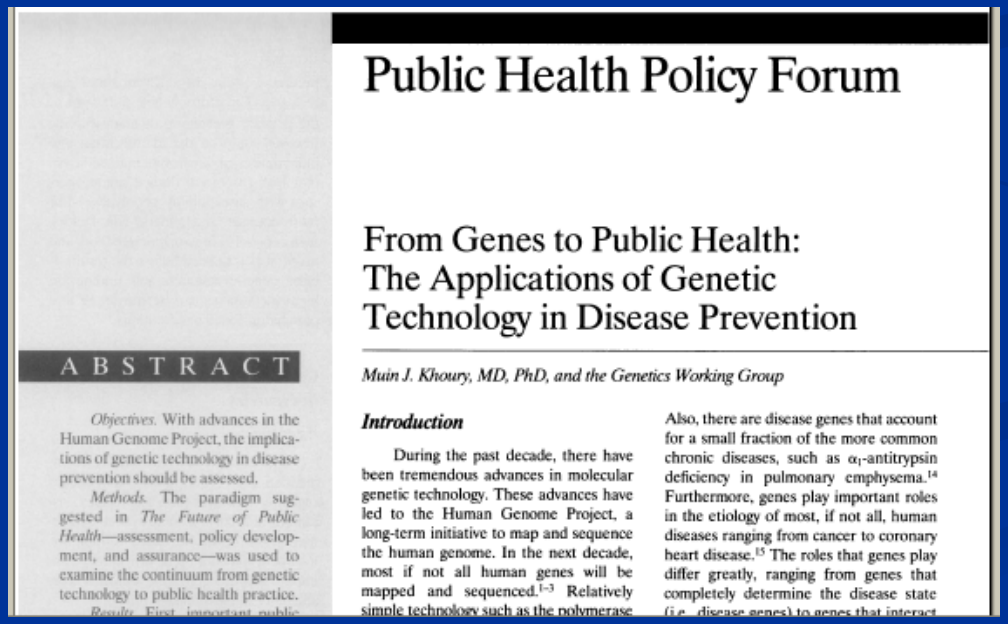


The Cycle of Public Health Practice In the USA, IOM Report 1988



Genetics and Public Health
(Khoury et al, 1996)

- Public Health Functions
  - Assessment
  - Policy Development
  - Assurance



Public Health Policy Forum

From Genes to Public Health: The Applications of Genetic Technology in Disease Prevention

Muin J. Khoury, MD, PhD, and the Genetics Working Group

ABSTRACT

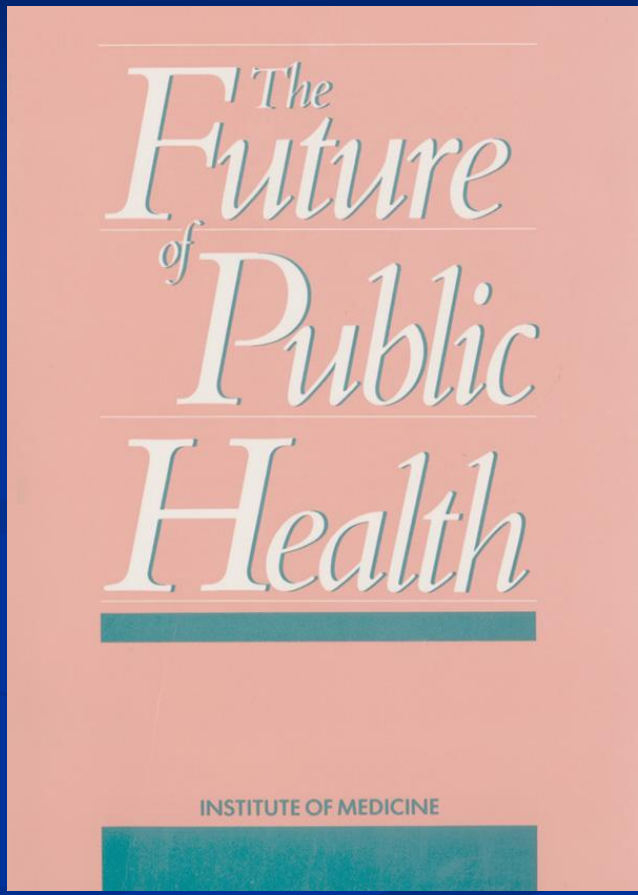
Objectives. With advances in the Human Genome Project, the implications of genetic technology in disease prevention should be assessed.

Methods. The paradigm suggested in *The Future of Public Health*—assessment, policy development, and assurance—was used to examine the continuum from genetic technology to public health practice.

Introduction

During the past decade, there have been tremendous advances in molecular genetic technology. These advances have led to the Human Genome Project, a long-term initiative to map and sequence the human genome. In the next decade, most if not all human genes will be mapped and sequenced.[1–3] Relatively simple technology such as the polymerase

Also, there are disease genes that account for a small fraction of the more common chronic diseases, such as α_1-antitrypsin deficiency in pulmonary emphysema.[14] Furthermore, genes play important roles in the etiology of most, if not all, human diseases ranging from cancer to coronary heart disease.[15] The roles that genes play differ greatly, ranging from genes that completely determine the disease state



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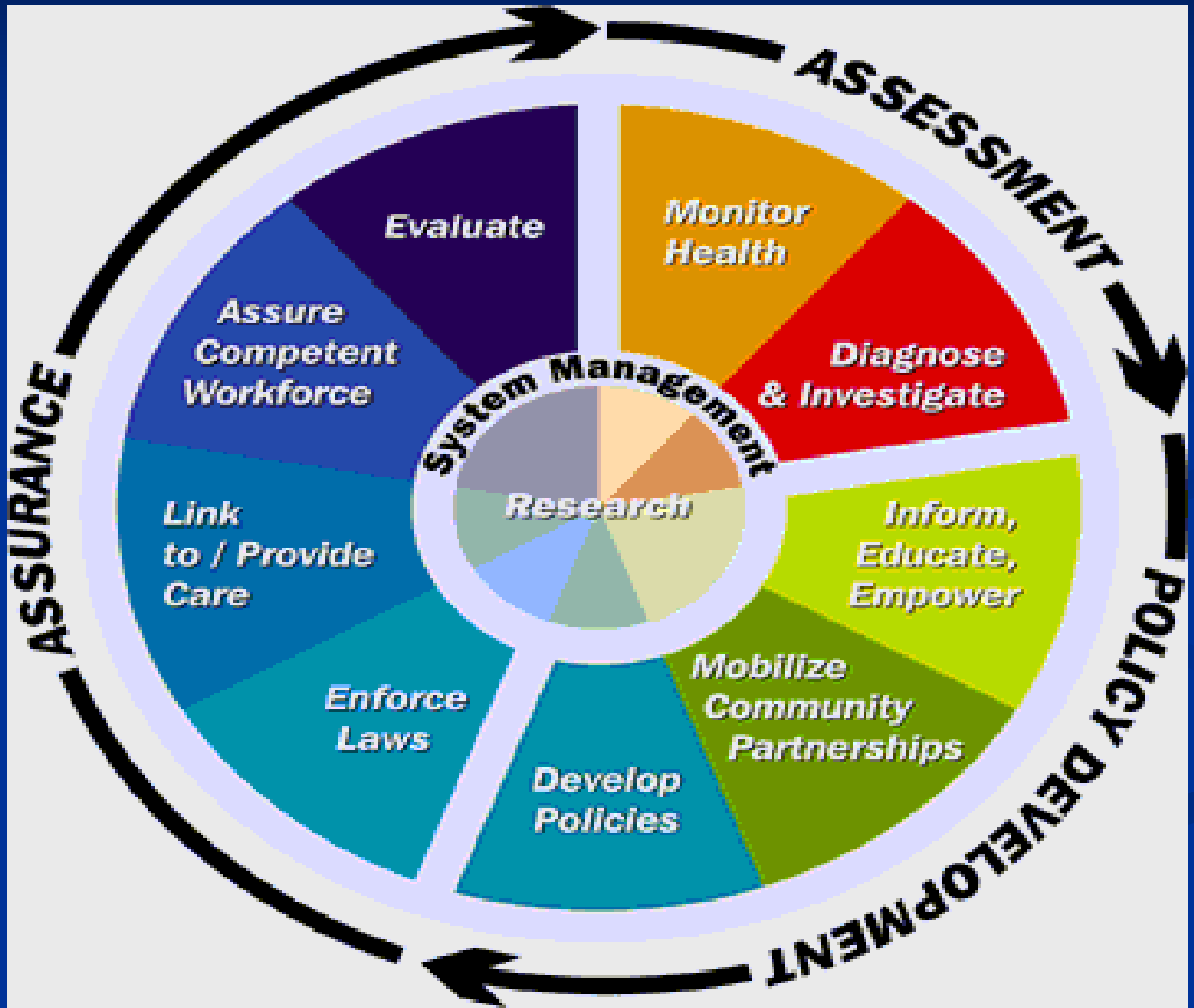
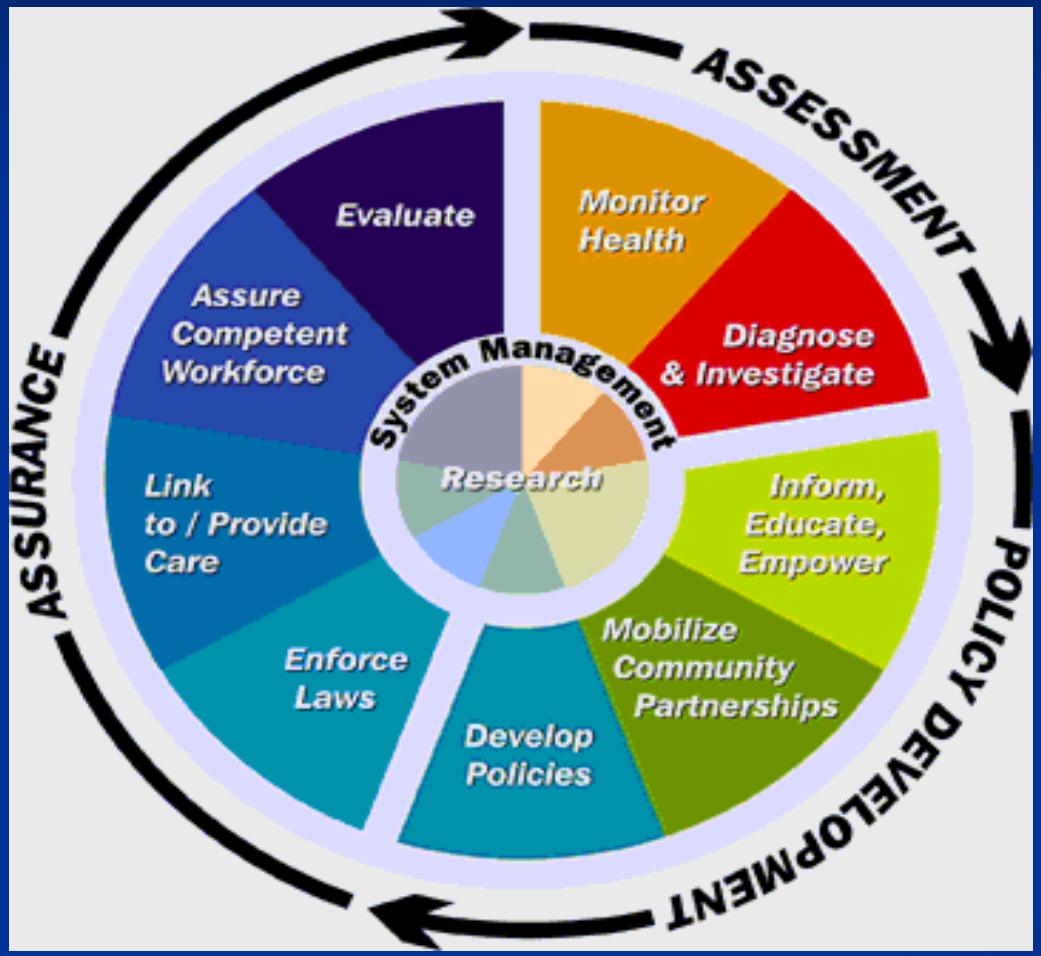
The 10 Essential Public Health Services



The Integration of Genomics into Public Health: Core Functions

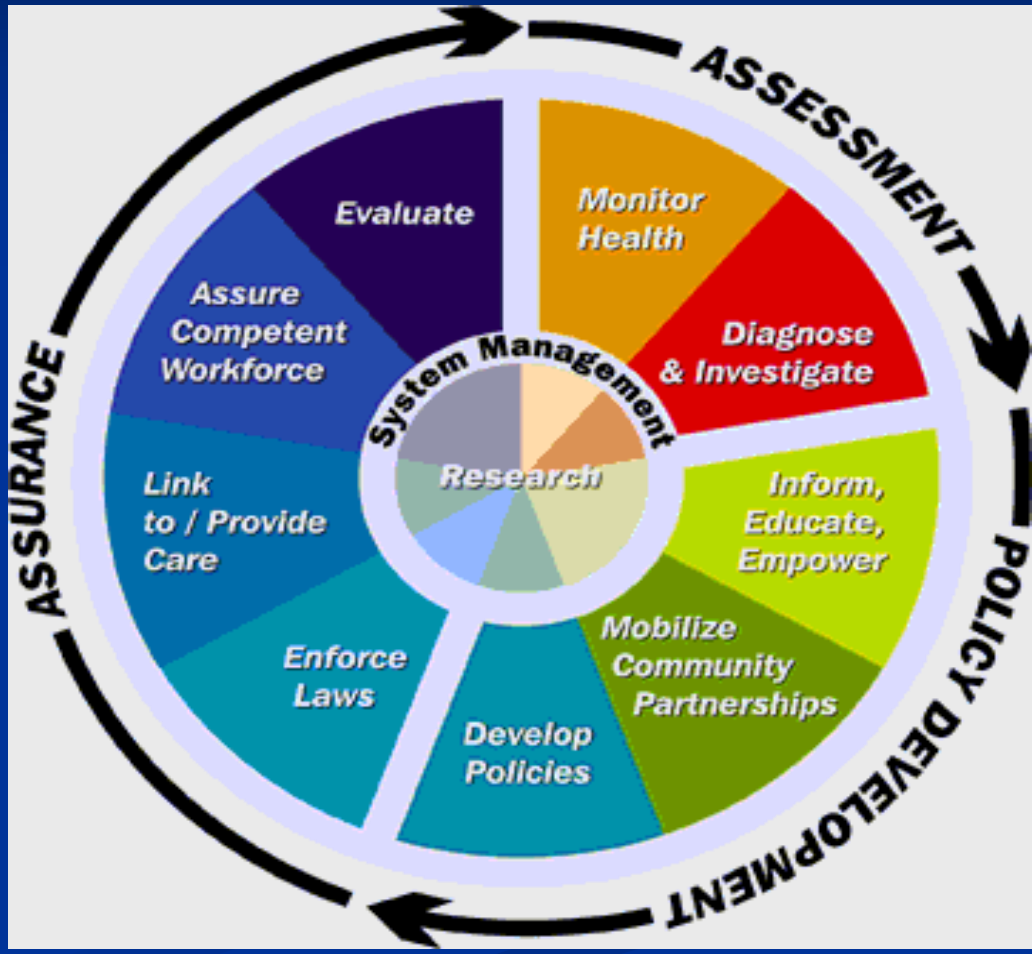


Beskow et al. Comm Genetics 2001

- Assessment: The regular systematic collection, assembly, analysis, and dissemination of information, including genetic epidemiologic information, on the health of the community.

The Integration of Genomics into Public Health: Core Functions

- **Policy Development**
  Formulation of standards and guidelines, in collaboration with stakeholders, which promote the appropriate use of genetic information and the effectiveness, accessibility, and quality of genetic tests and services



Beskow et al. Comm Genetics 2001

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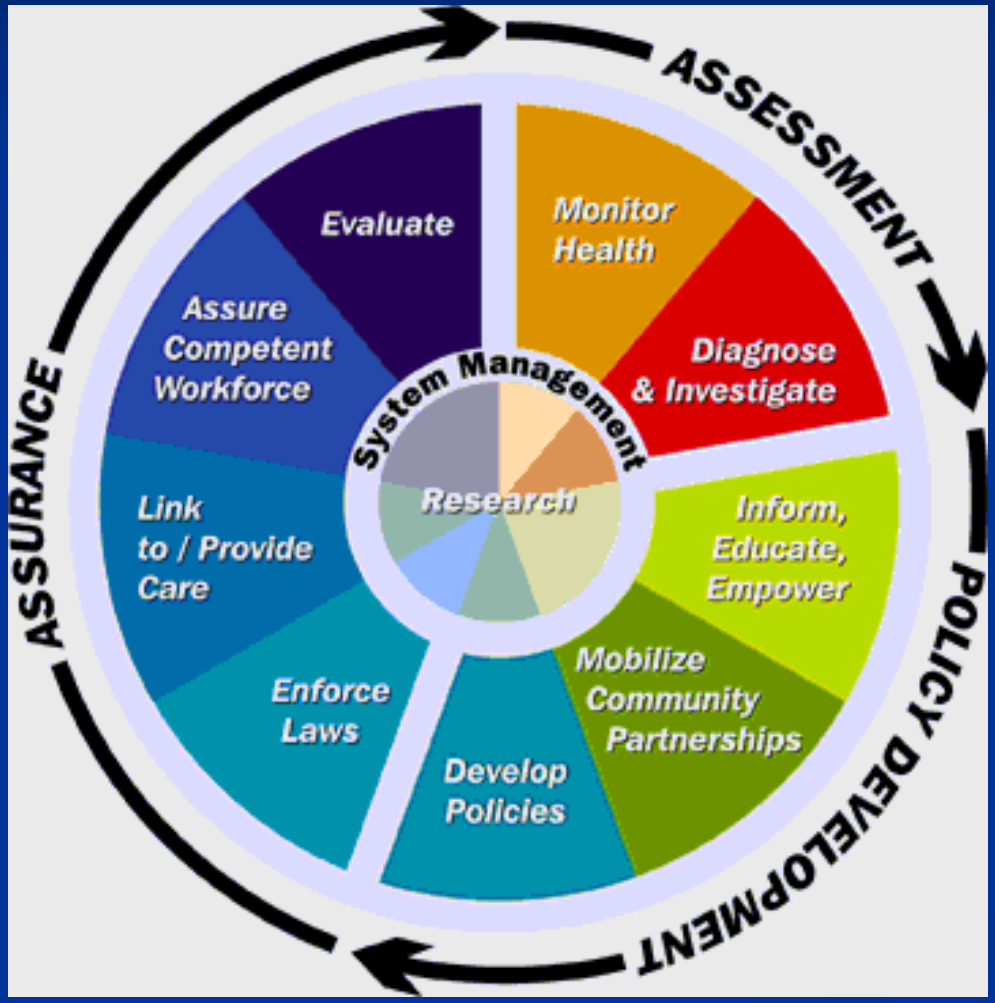
The Integration of Genomics into Public Health: Core Functions

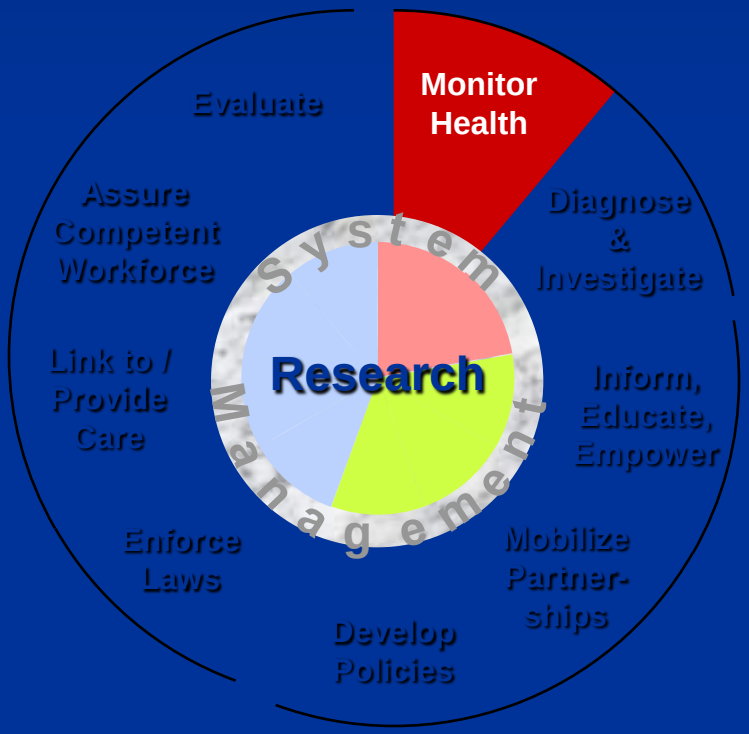


Beskow et al. Comm Genetics 2001

- Assurance

  The assurance to constituents that genetic information is used appropriately and that genetic tests and services meet agreed upon goals for effectiveness, accessibility, and quality.

Essential Services: Assessment



- **Monitor Health:** Monitor health status, including genetic factors, to identify health problems within the community.

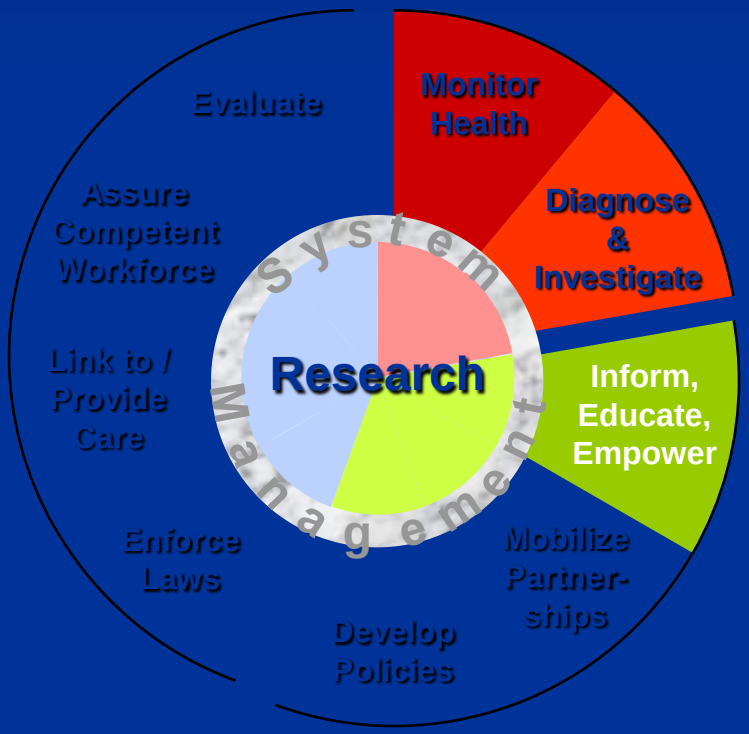
Beskow et al. Comm Genetics 2001

Essential Services: Assessment

- **Monitor Health:** Monitor health status, including genetic factors, to identify health problems within the community.
- **Diagnose & Investigate:** Investigate the distribution of genetic and modifiable risk factors within the community to determine their contribution to identified health problems and to improve health outcomes.

Beskow et al. Comm Genetics 2001

Essential Services: Policy



- **Inform, Educate, Empower:** Facilitate communication and education about the integration of genetic information into health promotion and disease prevention programs.

Beskow et al. Comm Genetics 2001

Essential Services: Policy

- **Inform, Educate, Empower:** Facilitate communication and education about the integration of genetic information into health promotion and disease prevention programs.
- **Mobilize Partnerships**: Foster collaboration among public and private agencies and constituent groups to promote effective and efficient policy making about genetics.

Beskow et al. Comm Genetics 2001

Essential Services: Policy

Beskow et al. Comm Genetics 2001

- **Inform, Educate, Empower:** Facilitate communication and education about the integration of genetic information into health promotion and disease prevention programs.
- **Mobilize Partnerships**: Foster collaboration among public and private agencies and constituent groups to promote effective and efficient policy making about genetics.
- **Develop Policies**: Establish policies and guidelines for when and how genetic information should be applied to promote health and prevent disease.

Essential Services: Assurance

- **Enforce Laws:** Promote the enforcement of policies and standards enacted to ensure the appropriate use of genetic information, and the effectiveness, accessibility, and quality of genetic tests and services.

Beskow et al. Comm Genetics 2001

Essential Services: Assurance

- **Enforce Laws:** Promote the enforcement of policies and standards enacted to ensure the appropriate use of genetic information, and the effectiveness, accessibility, and quality of genetic tests and services.
- **Link to / Provide Care**: Ensure the availability and accessibility of intervention strategies that incorporate genetic information to improve health and prevent disease.

Beskow et al. Comm Genetics 2001

Essential Services: Assurance

- **Assure Competent Workforce**: Ensure that present and future health professionals have training and skills in the appropriate use of genetic information to promote health and prevent disease.

Beskow et al. Comm Genetics 2001

Essential Services: Assurance

- **Assure Competent Workforce:** Ensure that present and future health professionals have training and skills in the appropriate use of genetic information to promote health and prevent disease.
- **Evaluate**: Evaluate the impact of genetic information, and the effectiveness, accessibility, and quality of genetic tests and services.

Beskow et al. Comm Genetics 2001

The Integration of Genomics into Public Health

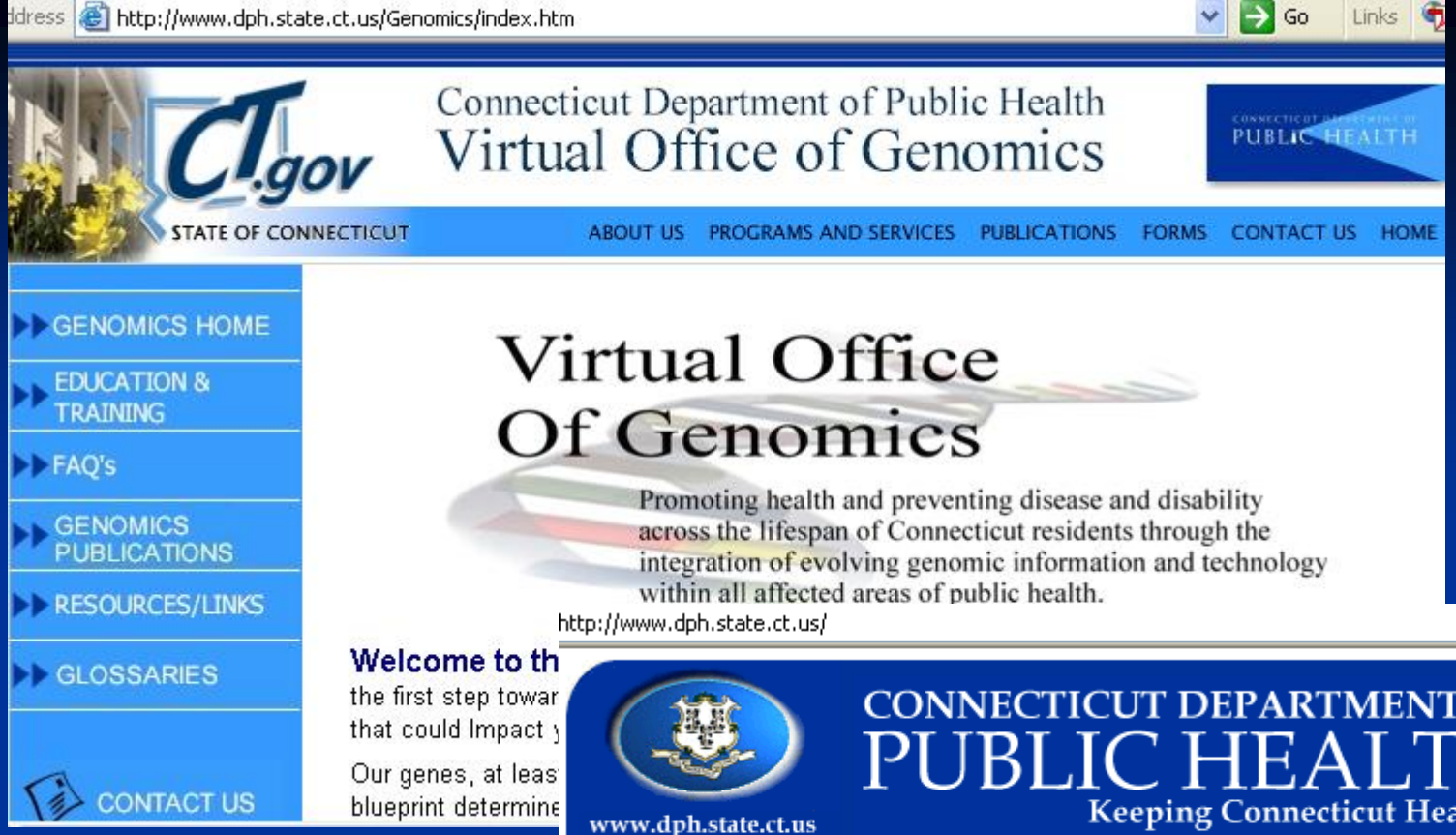
- **System Management**: Building and maintaining the capacity of the public health infrastructure to integrate genetic information into public health research and practice.

Beskow et al. Comm Genetics 2001

The Integration of Genomics into Public Health

- **Research**: A systematic investigation designed to develop or contribute to generalizable knowledge of the impact of human genetic variation on health and disease.

Beskow et al. Comm Genetics 2001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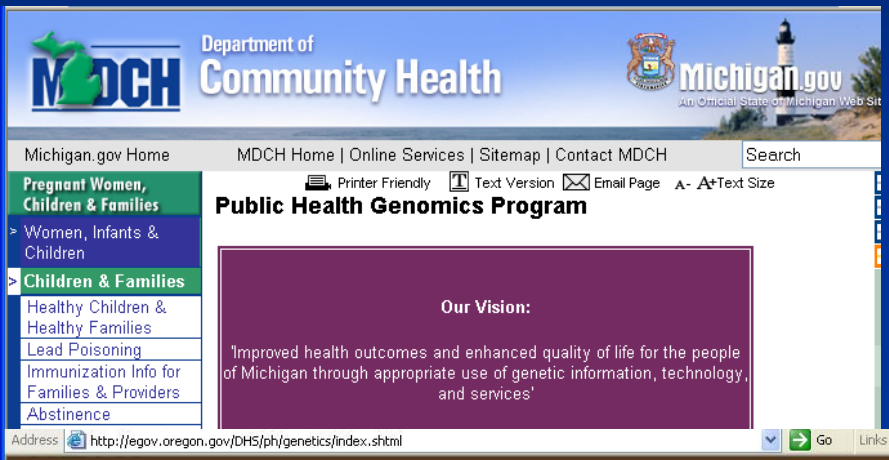
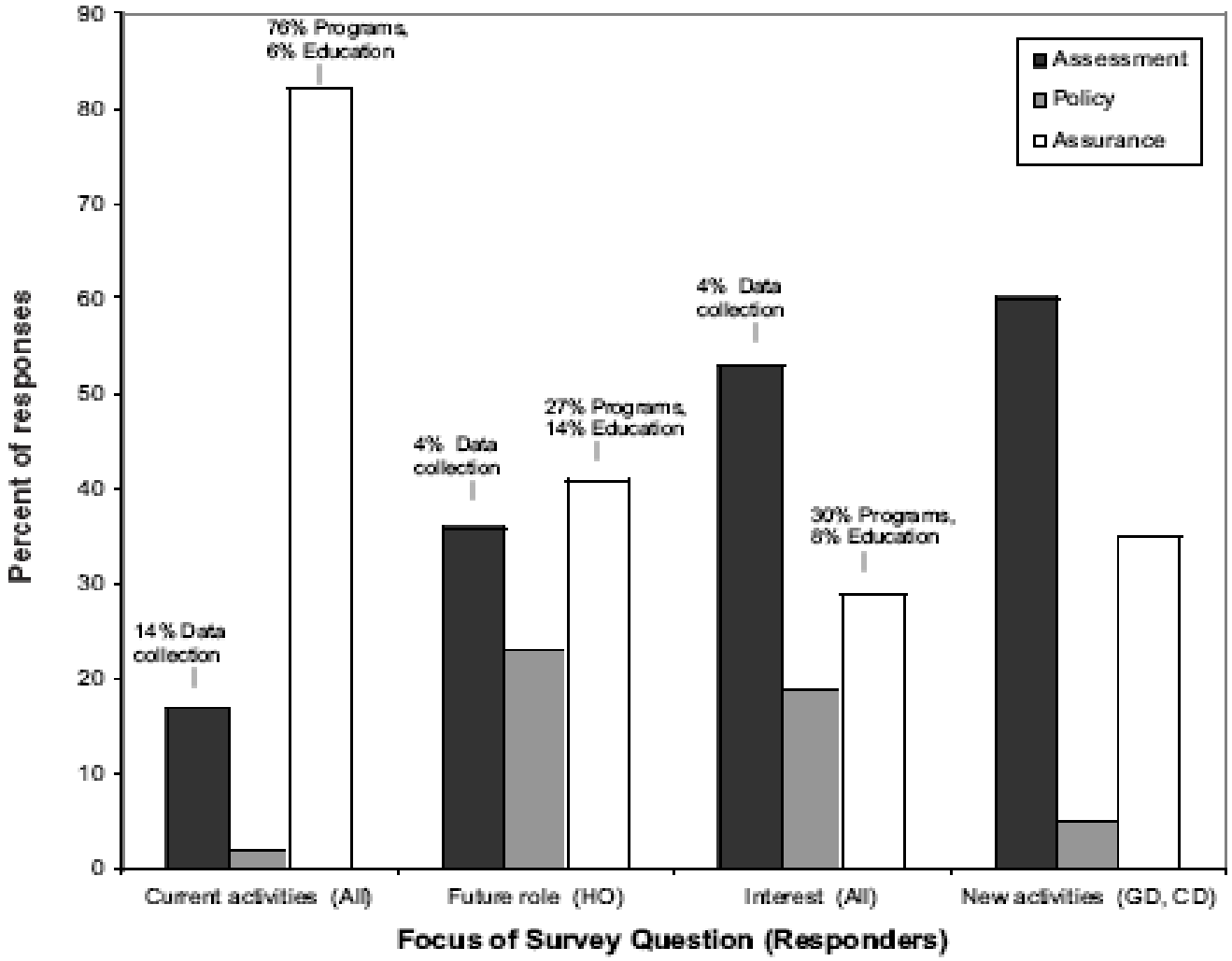
Health Care Quality
Physician Profile
Press Releases
Public Health Preparedness & Response
Department of Public Health Launches Web-based Training for Newborn Screening
Private Well Water Concerns
P&W Investigation of Possible Brain Cancer Cluster - Information concerning the Pratt & Whitney investigation
Emergency Credentialing Program - to pre-qualify the credentials of health care practitioners who are willing to assist in the event of a bioterrorist...

Integrating Genomics into State Public Health Programs



The Role of State Public Health Agencies in Genetics and Disease Prevention: Results of a National Survey

Piper MA et a. Publ Health Rep 2001



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Newborn Screening Public Health Genetics Program

- Almost all infants in US are screened for at least 4 types of disorders
  - Phenylketonuria (PKU), 1 in 20,000 births
  - Congenital hypothyroidism, 1 in 2,500 births
  - Galactosemia, 1 in 67,000 births
  - Sickle cell disease (SCD) 1 in 2,600 births
- Largest genetic testing program in US
  - Mostly biochemical, phenotypic screening tests
  - DNA tests can be used for confirmatory or diagnostic purposes

From S. Grosse

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Newborn Screening As a System

- Authority – states mandate screening panels
- Activities
  - Birthing centers collect bloodspots
  - Screening by public health or designated lab
  - Reporting and follow-up
  - Referral and diagnostic testing
  - Treatment
  - Monitoring and evaluation
- Funding
  - State fees and taxes
  - Federal funding – Title V (MCH Block Grant)
  - Other

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History of Newborn Screening

- Began in early 1960s with PKU screening test
  - Why PKU?
    - Mental retardation preventable if treated < 3 months
    - Pushed by test developer and parent advocates
  - State mandates widely adopted, beginning 1963 in MA
    - Public health emergency rationale
    - Cost saving rationale
- Other conditions added by states
  - Galactosemia began in 1964, spread more slowly
  - Congenital hypothyroidism initiated 1975, spread quickly
    - Very common disorder
    - Inexpensive test and treatment

From S. Grosse

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Tandem Mass Spectrometry (MS/MS) Screening

- Tandem mass spectrometry (MS/MS) new technology that can be used to screen for >30 metabolic disorders at once
  - Organic acid disorders
  - Fatty acid oxidation disorders
- Proprietary technology developed in 1990s
  - Commercial impetus behind expansion and advocacy
  - Public-private rivalry or partnership
- Can detect dozens of analytes simultaneously
- Adopted by majority of states since 1997
  - Focus of advocacy
  - Some states use MS/MS to screen for limited set of disorders

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Recent Developments in U.S. Newborn Screening Policy

- In 2002, HRSA contracted with the American College of Medical Genetics (ACMG) to
  - Develop new screening criteria and
  - Recommend uniform core screening panel
  - Report issued March 2005
    - Primary screening panel of 29 disorders
- Creation of Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children (SACHDGDNC)
  - Authorized by Children's Health Act of 2000
  - Established in 2004
  - Has recommended that all states adopt a new uniform screening panel proposed by ACMG expert group
- Most states appear moving to adopt core panel

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NBS for MCAD Deficiency

- MCADD is ‘poster child’ for expanded NBS
  - Most common fatty acid oxidation disorder, 1:15,000
  - Treatment is simple
    - Regular feeding
    - Emergency medical attention
- MCADD outcomes (Grosse et al., *Genet Med*, 2006; Wilcken et al., *Lancet*, 2007; Grosse & Dezateux, *Lancet,* 2007)
  - Metabolic crisis common, perhaps 50-75%
  - Risk of death 10% to 20%, mostly preventable
  - About 10% of survivors have MR according to many studies, but no MR in Australian study
  - Outcomes may be better with clinical awareness

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Evidence for Expanded NBS: Other Disorders

- Challenges
  - Other disorders detected by MS/MS are much less common
  - Reliance on clinical judgment and expert opinion
- Lack of population-based outcomes data
  - NBS data don't indicate outcomes without screening
  - No large scale study of population-based stored blood spots using MS/MS
  - Virginia study of 793 postmortem blood spots for unexplained deaths before 3 years found 8 probable cases of disorders detected by MS/MS, or 1% (Dodd et al., *AJMG*, 2006)
    - Including 2 cases of MCADD

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Cystic Fibrosis (CF) NBS

- CF is second-most common lethal single-gene disorder in US children
  - 1 in 4000 births (1 in 2800 European-origin)
  - Life expectancy 32 years
- Screening introduced in Colorado in 1982
- Slow spread until 1997
  - Wisconsin trial showed nutritional benefits
  - 1997 CDC workshop recommended pilot screening
  - 2003 CDC workshop concluded that routine screening for CF is justified

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Summary of Evidence on Health Outcomes with CF NBS

- Moderate impact on growth – 0.3 Z-score difference in height-for-age
- Moderate impact on cognition – overall difference of 5-6 IQ points in WI study
- Reduction in CF-related mortality
  - Consistently reported in studies from outside US (5-10%)
  - No CF-related deaths < 10 years in Wisconsin trial
  - US data suggest mortality differential of 1.5%-2.0%
- Reduction in hospitalization and cost is likely but not proven consistent
- No consistent improvement in pulmonary outcomes and some risk of harm without adequate infection control

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Balancing CF NBS Benefits and Risks: CDC Summary (2003)

- Certain psychosocial risks…are associated with newborn screening.
- Exposure of young children to infectious agents … is a potential cause of harm from early detection.
- Involving specialists in CF care and infection control, genetic counseling, and communication can minimize these potential harms.
- …on the basis of evidence of moderate benefits and low risk of harm, CDC believes that newborn screening for CF is justified.

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Towards Principles for Population Screening in the Age of Genomic Medicine:

Wilson and Jungner Revisited

(Khoury, McCabe and McCabe, NEJM, 2003)

- Public Health Assessment
- Evaluation of Tests and Interventions
- Policy Development and Screening Implementation

Towards Principles for Population Screening in the Age of Genomic Medicine
Public Health Assessment

- 1. Disease or health condition an important burden to target population (morbidity, disability, mortality)
- 2. Information known on prevalence of genetic trait in the target population and the burden of disease attributable to it
- 3. Natural history of the condition, from susceptibility to latent to overt disease adequately understood

Towards Principles for Population Screening in the Age of Genomic Medicine
Evaluation of Tests and Interventions

- 4. Data available on test PPV and NPV for future disease/health condition in target population
- 5. Safety and effectiveness of the test and accompanying interventions established

Towards Principles for Population Screening in the Age of Genomic Medicine
Policy Development and Screening Implementation

- 6. Consensus achieved using scientific evidence
- 7. Screening acceptable to target population
- 8. Facilities available for surveillance, prevention, treatment, education, counseling and social support
- 9. Screening as a continuous process including pilot programs, assurance of laboratory quality and health services, evaluation of impact and provision for changes based on new evidence

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Towards Principles for Population Screening in the Age of Genomic Medicine
Policy Development and Screening Implementation

- 10. Cost-effectiveness of screening established
- 11. Access of screening and interventions
- 12. Safeguards for informed consent and privacy, avoidance of coercion or manipulation, and protection from stigmatization and discrimination

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Public Health Investigations

- Public health agencies worldwide
- Diseases
  - infectious disease outbreaks
  - cancer or birth defect clusters
- Exposures
  - environmental or occupational
  - bioterrorism
- Adverse response to interventions
  - vaccines
  - antibiotic prophylaxis

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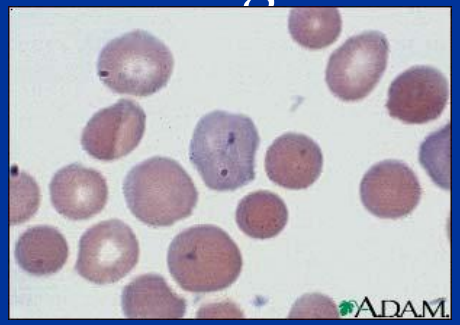
"The Epidemiology Triangle" Revisited

Host

Malaria

Agent

Environment



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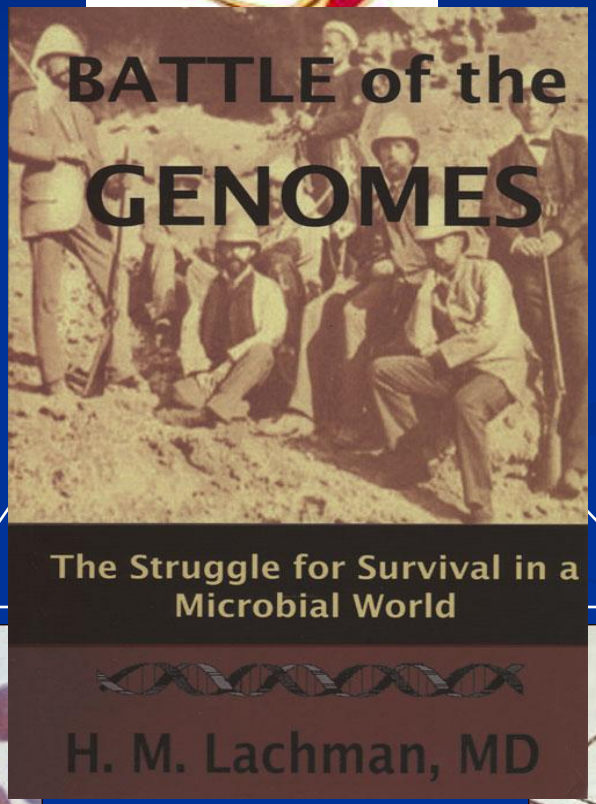
"The Epidemiology Triangle" Revisited



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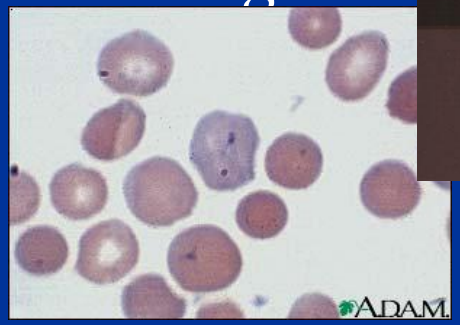

Environment

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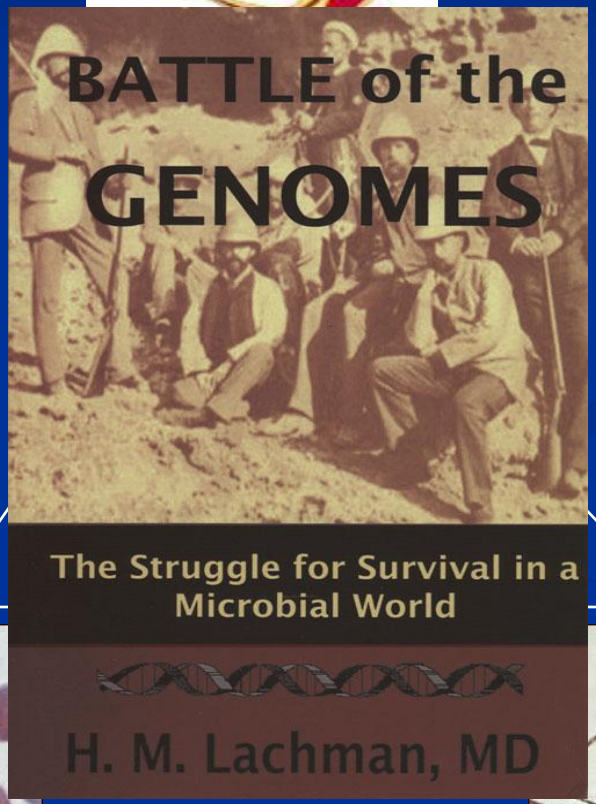
"The Epidemiology Triangle" Revisited



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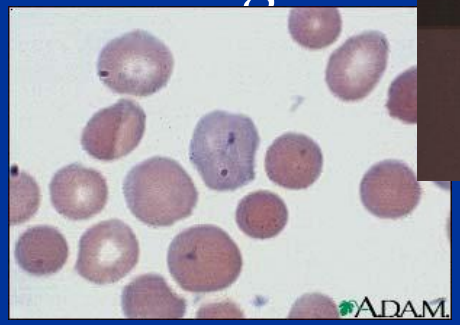
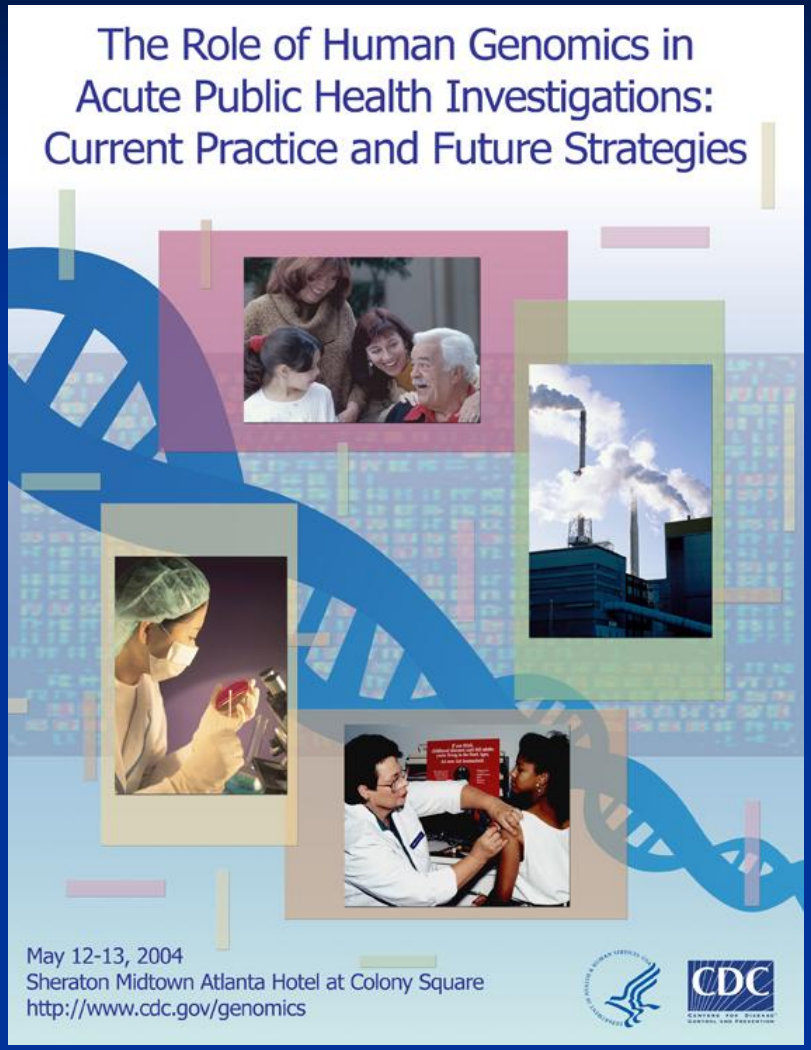


Environment

Transmission
Severity
Complications
Fatality
Persistence
Chronic Dis
Treatment
Vaccines



CDC 2004 Workshop

Opportunities

- diseases occurring only in epidemic settings (SARS)
- rare threats (anthrax)
- mass exposures (toxins)

Challenges

- rapid response
- priority on control
- ethical, legal issues
- resources and logistics

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1998 Springfield Ironhorse Triathlon Leptospirosis Outbreak

- 876 triathletes; 12% reported illness
- Serum from 474; 52 positive for leptospirosis

Genetic studies: TNF-a, HLA-DRB, HLA-DQB

- HLA-DQ6 positive triathletes (compared to DQ6 negatives) were
  - more likely be seropositive for leptospirosis (OR=2.8, p=0.04)
  - especially for those who reported swallowing lake water (OR=8.5, p=0.001)

Lingappa J. et al., Genes & Immunity 2004)

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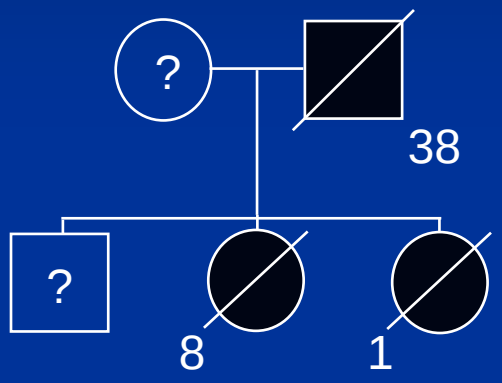
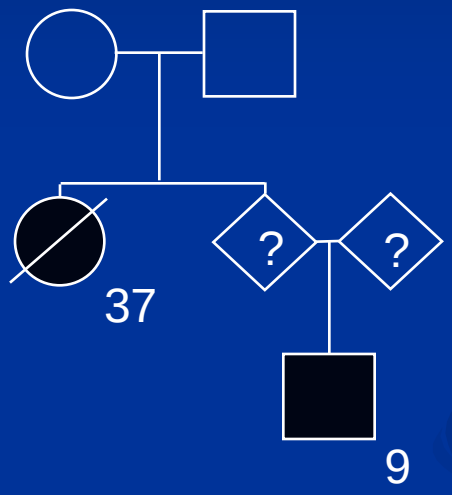
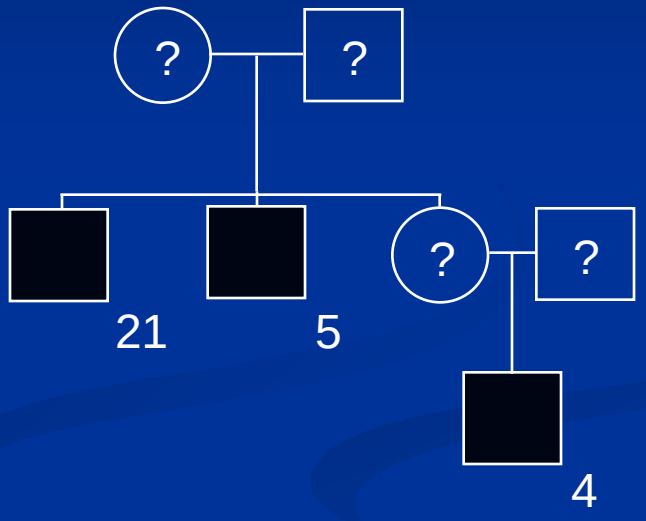
Familial Clusters of H5N1 Influenza



8 household or neighbor contacts

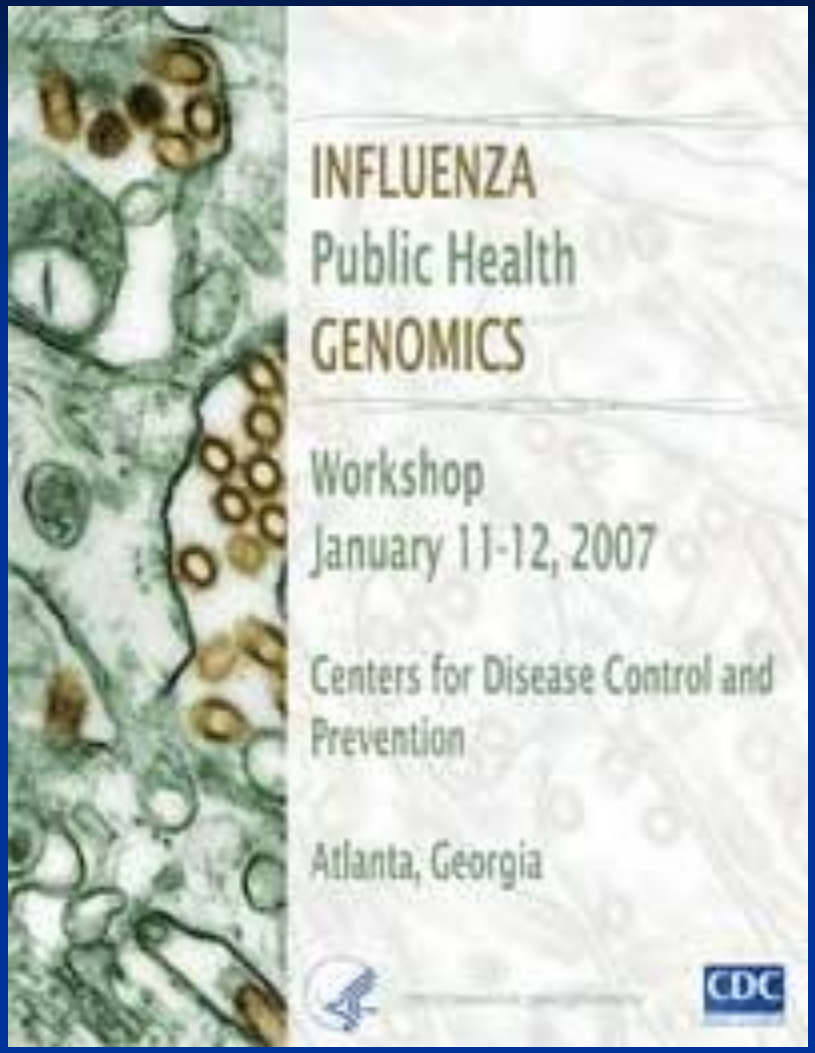


76 household or neighbor contacts



33 household or neighbor contacts

Kandun et al., *New Engl J Med*, 2006



CDC International Workshop

State of the science

- Interdisciplinary
- Interagency and international

Potential opportunities

- outbreak investigations
- "biobanks" in managed care

Issues

- "off-the-shelf" protocols
- privacy, confidentiality
- collaborations

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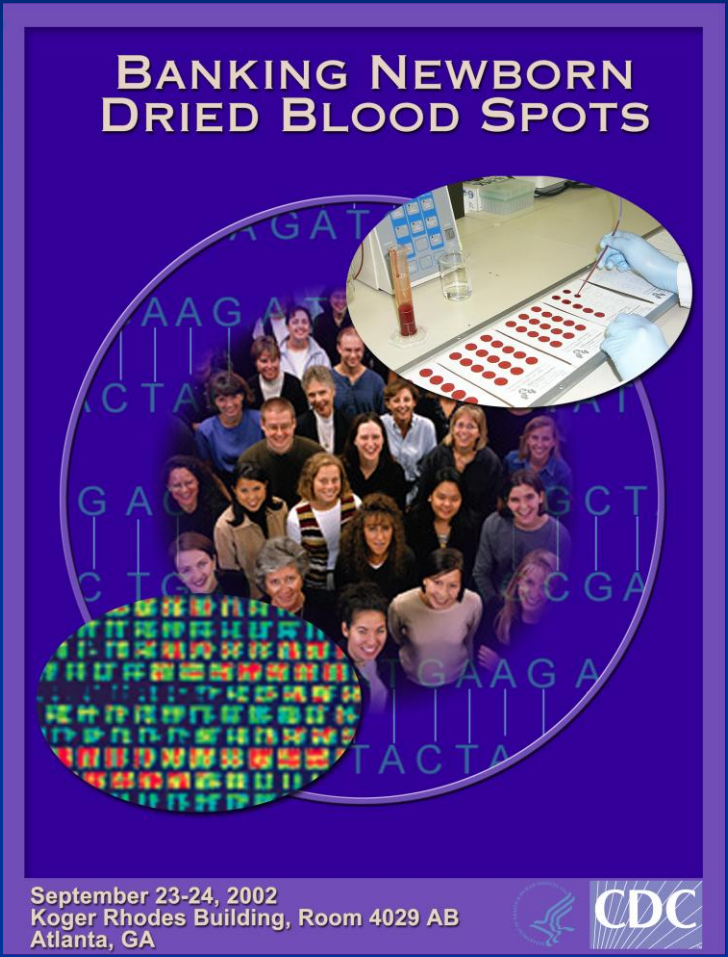
Newborn Bloodspot Workshop



Developing a Strategic Plan to Assess the Feasibility, Utility, and Practical Implementation of Establishing a National/Multi-state Bank of Leftover Newborn DBS

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Outline

- What is Public health?
- Public Health and the Paradigm Shift from Traditional Genetics to Genomic Medicine
- Core Public Health Functions and Essential Services
- Examples
  - Newborn Screening
  - Public Health Investigations
  - DTC Genetic Testing

Public Health Surveillance of DTC Genetic Tests

- The goals of public health surveillance are:
  - To inform about public demand and interest in Health-related Genetic Tests
  - To gauge provider knowledge & experience with DTC tests
  - To assess the impact of new policies or educational efforts aimed towards the public or providers
  - Over time, to describe the evolution of the availability and demand for DTC tests

From K. Goddard (2007)

Background

- Direct-to-consumer genetic tests have recently exploded onto the market
  - **Non-health related examples**: DNA banking, paternity testing, identity testing, genealogical genetics, recreational genetics
  - **Health-related examples**: nutrigenomics, fetal gender, caffeine metabolism, HIV progression, clinical testing
  - The Internet gives *everybody* immediate access

From K. Goddard (2007)

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Concerns Raised in GAO Report, 2006

- Misleading, unsubstantiated, and ambiguous predictions
- Recommended costly dietary supplements
- Supplements may be harmful for some people
- Recommendations not based on unique genetic profile

United States Government Accountability Office

GAO

Testimony

Before the Special Committee on Aging, U.S. Senate

For Release on Delivery
Expected at 10:00 a.m. EST
Thursday, July 27, 2006

NUTRIGENETIC TESTING

Tests Purchased from Four Web Sites Mislead Consumers

The New York Times
Get Home Delivery

Media & Advertising

ADVERTISING

A Genetic Test That Very Few Need, Marketed to the Masses

By ANDREW POLLACK
Published: September 11, 2007

SIGN IN TO E-MAI

BRAC*Analysis*®

A genetic test for hereditary breast and ovarian cancer

BRAC*Analysis*® assesses a woman's risk of developing breast or ovarian cancer based on detection of mutations in the *BRCA1* and *BRCA2* genes. This test has become the standard of care in identification of individuals with hereditary breast and ovarian cancer and is reimbursed by insurance.

Please visit the myriadtests.com website to learn more about inherited breast and ovarian cancer.

"Less than 5% of people at-risk for hereditary cancer know their genetic test results. Myriad provides the answers."

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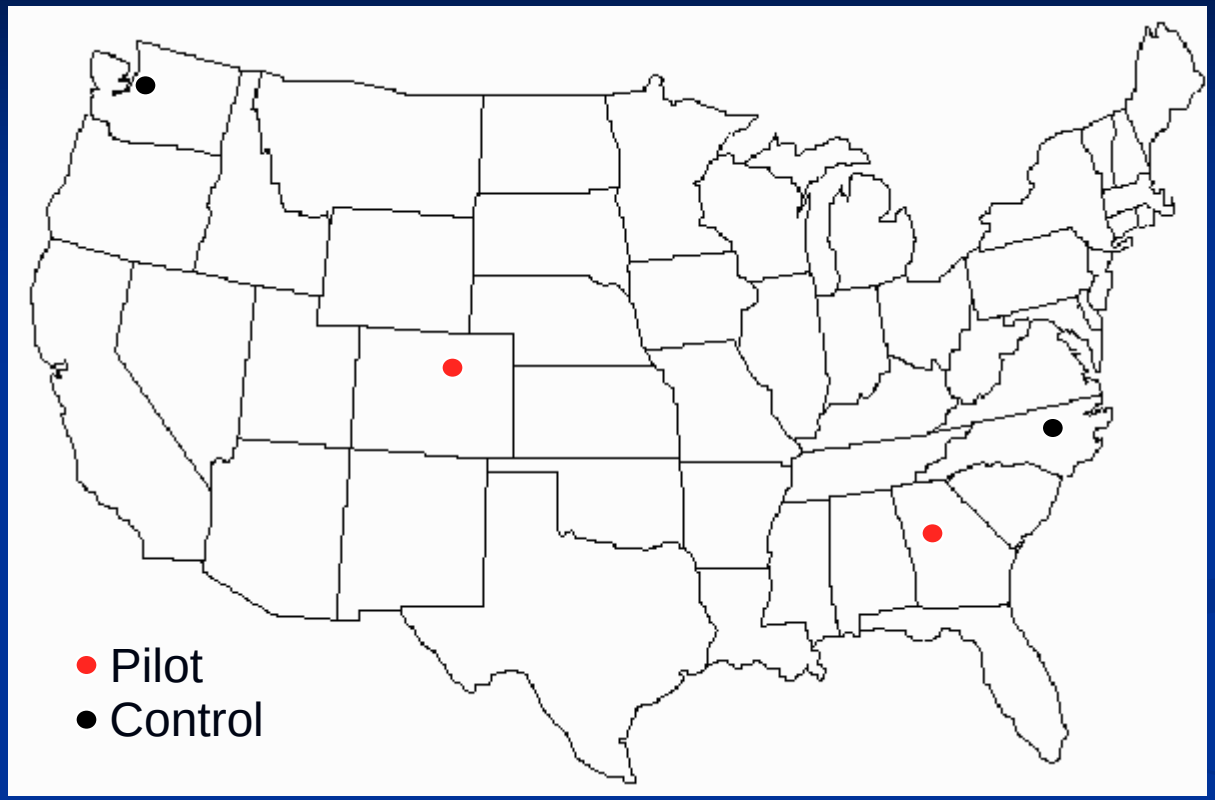
Public Health Impact of DTC Advertising Campaign for BRCA1/2



Advertisement: September, 2002 – February, 2003
Survey: April-May, 2003

From K. Goddard (2007)

Impact of DTCA for BRCA on Consumers

- **DTCA increased consumer awareness (22% vs. 42%)**
- **DTCA increased questions, requests, referrals and orders for BRCAnalysis® approximately 2-fold**
- **Interest in genetic test was not disproportionately increased in women with a + family hx (20% vs. 17%)**

Myers MM et al. Genetics in Medicine 2006

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DTC Genetic Tests: National Surveys

- **HealthStyles, 2006:** National marketing survey conducted by Synovate, Inc. with 5250 respondents
  - Recruited from a consumer mail panel of ~450,000 potential respondents
  - Response Rate: 53% (66% phase I, 80% phase 2)
- **DocStyles**, 2006: Online survey of primary care physicians, internists, and pediatricians with 1250 respondents
  - Recruited from an opt-in, verified panel of 142,000 physicians
  - Response Rate: 61% of eligible, invited physicians

From K. Goddard (2007)

DTC Genetic Tests: State Surveys

- **CDC-funded State Programs in Public Health Genomics**
  - Oregon
  - Michigan
  - Utah
- **Behavioral Risk Factor Surveillance System (BRFSS)**
  - CDC-sponsored health survey system in all 50 states
  - Recruitment of adults through Random Digit Dialing
  - Response Rates: 51.7%, 52.4%, 63.4%

From K. Goddard (2007)

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Results of Surveys

	Oregon	Michigan	Utah	National
Total Sample Size	1867	5499	2441	5250
Awareness of DTC Tests (% Yes, CI)	24.4% [22.2%,26.7%]	7.6% [6.8%,8.4%]	19.7% [17.7%,22%]	14% [12.7%,14.6%]
Use of DTC Tests (% Yes, CI)	0.3%	0.9%	-	0.6% [0.4% - 0.8%]

Goddard K et al. Genetics in Medicine 2006

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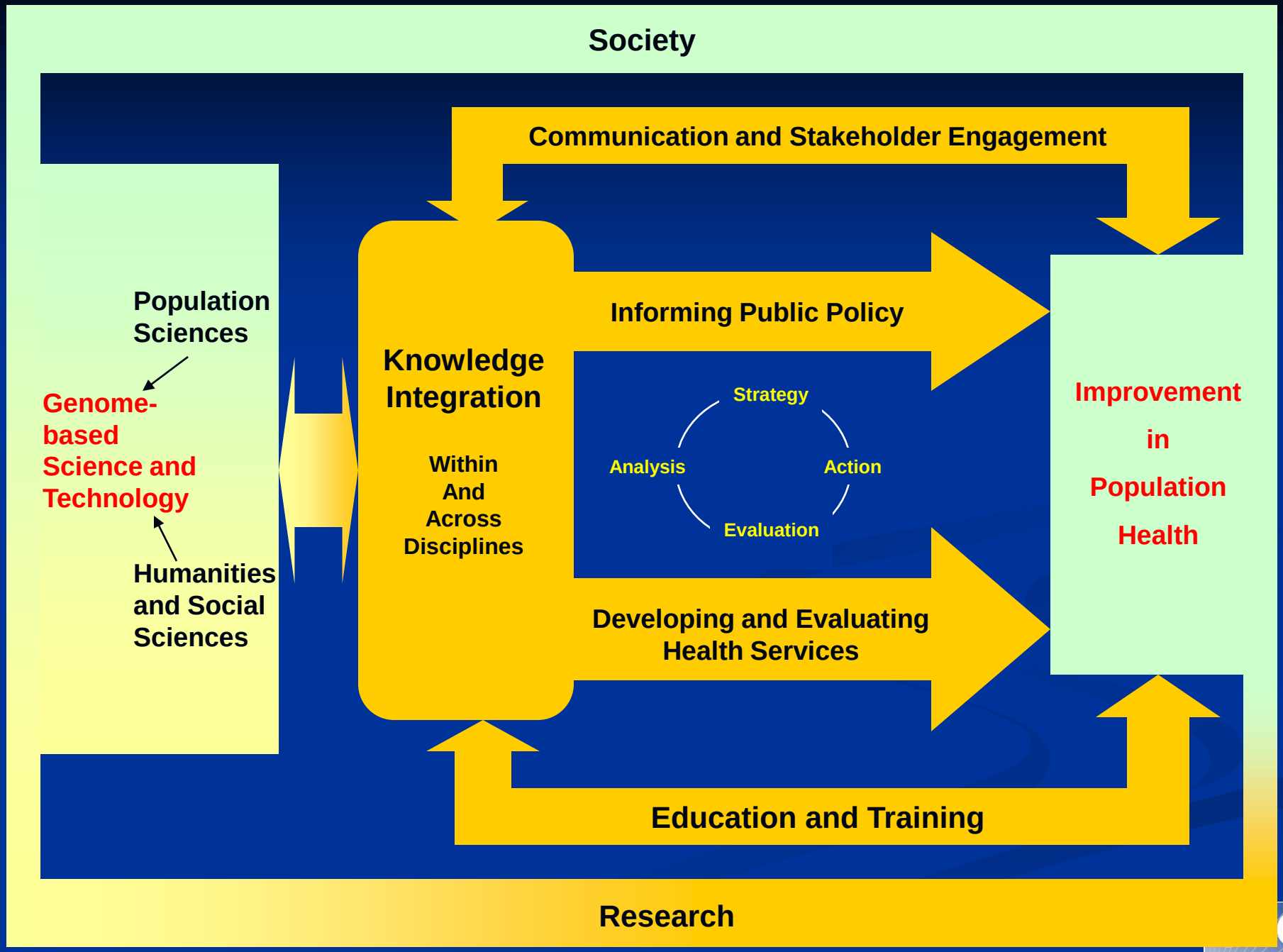
Society
Communication and Stakeholder Engagement
Population Sciences
Genome-based Science and Technology
Humanities and Social Sciences
Knowledge Integration
Within And Across Disciplines
Informing Public Policy
Strategy
Analysis
Action
Evaluation
Developing and Evaluating Health Services
Improvement in Population Health
Education and Training
Research