What role for public health in the era of genomic medicine?

M Gwinn, MD, MPH
National Office of Public Health Genomics, CDC, USA

European Health Forum Gastein
Bad Gastein, Austria
October 4, 2007

What role for public health in genetics and vice versa?

Holtzman NA, Community Genetics, 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."

Genomics and Health, United States, 2007

Medicine	Public Health
• Research	• Programs?
• Diagnostics	• Screening?
• Drugs	• Intervention?
• Promising…	• *Irrelevant?*

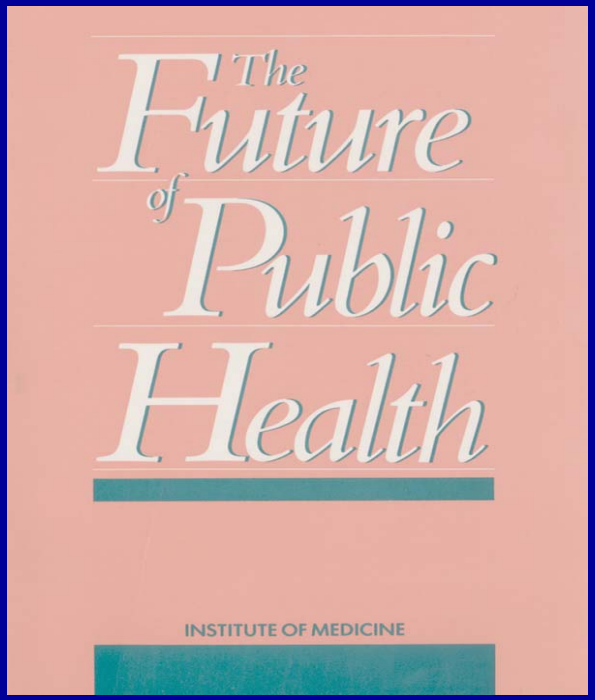
Outline

- What is public health?
- Public health and the paradigm shift from traditional genetics to genomic medicine
- Public health genomics and translation research
- Examples illustrating public health core functions

What is public health?

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

The Future of Public Health
Institute of Medicine, 1988



Who Will Keep the Public Healthy?

Institute of Medicine, 2002

Outline

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From Public Health Genetics…

Genetics

- Mendelian disorders
- Disease burden: 5%
- Mutations in single genes
- High risk
- Environment +/-

Role of Public Health

- Newborn screening programs
- Delivery of genetic services

…to Public Health Genomics

Genomic Information

- All diseases
- Disease burden: 95%
- Multiple common variants
- Small risks
- Environment ++

Role of Public Health?

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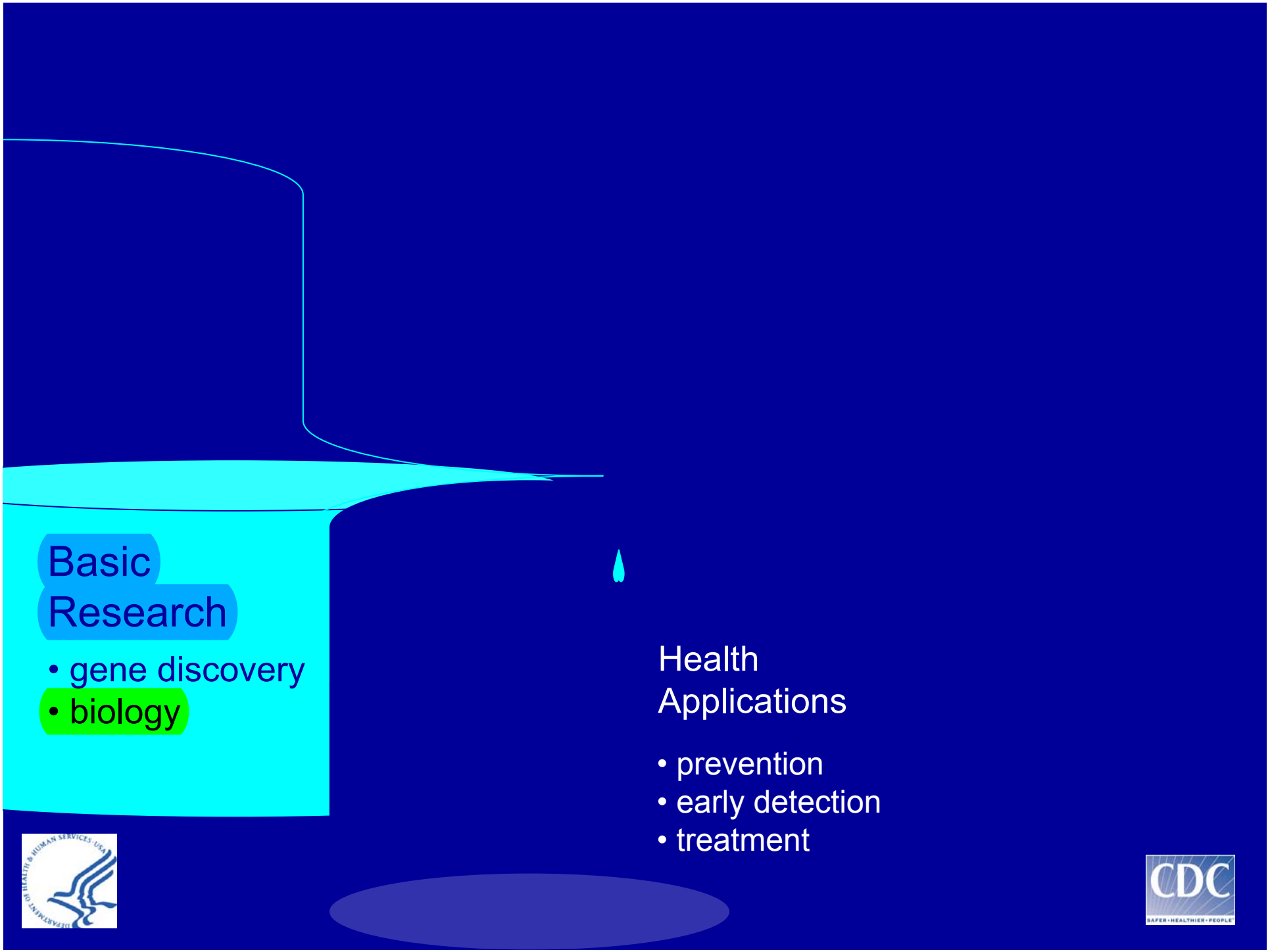
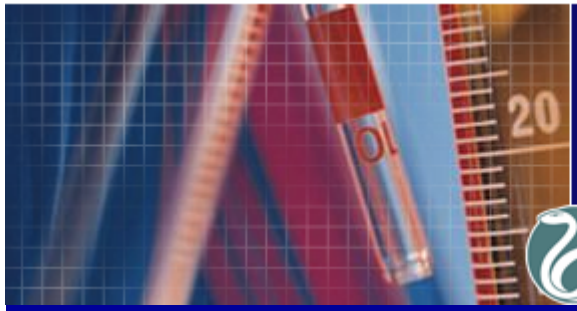

Basic Research
• gene discovery
• biology
Health Applications
• prevention
• early detection
• treatment
CDC
SAFER • HEALTHIER • PEOPLE



INSTITUTE OF MEDICINE
OF THE NATIONAL ACADEMIES

Knowing is not enough, we must apply.
Willing is not enough, we must do.

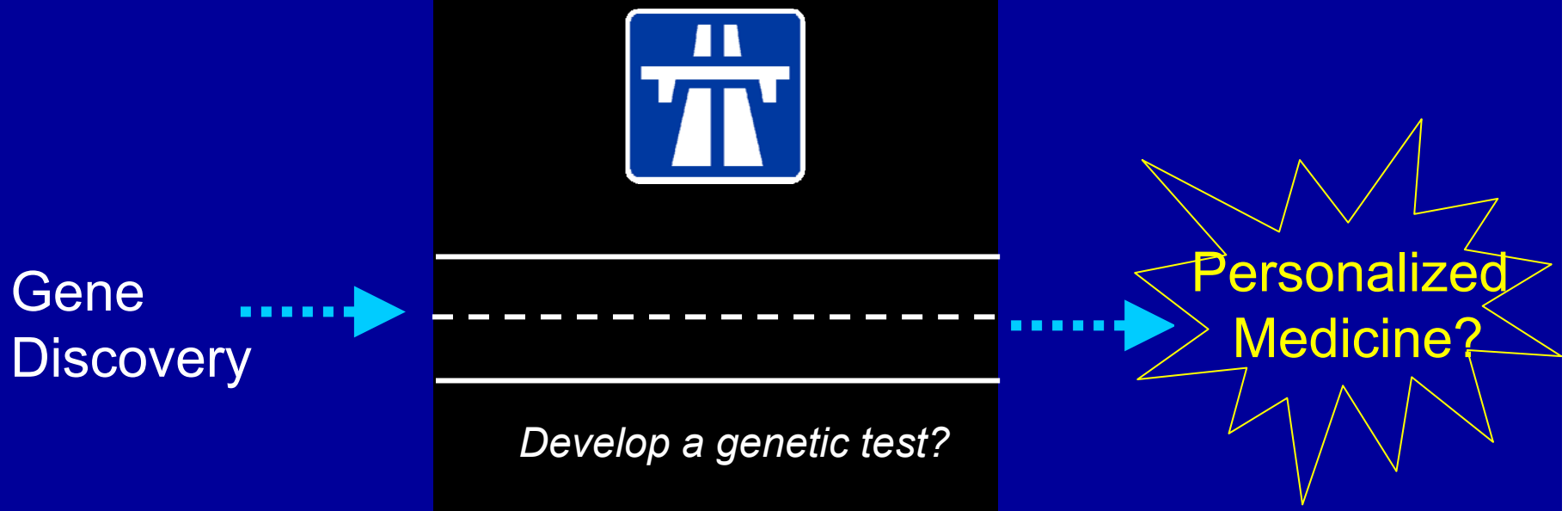
Es ist nicht genug zu wissen, man muss auch anwenden;
es ist nicht genug zu wollen, man muss auch tun.

-- Johann Wolfgang von Goethe

http://www.iom.edu/

Public Health Genomics: Closing the Gap Between Gene Discovery and Population Health

Public Health Genomics: Closing the Gap Between Gene Discovery and Population Health



Public Health Genomics: Closing the Gap Between Gene Discovery and Population Health

Gene Discovery

Prevention
Early detection
Treatment

Individual vs. Population Studies

develop

<u>Population</u>

- establish distribution of risk factors
- study risk factor – disease associations
- assess interactions among risk factors
- develop and test interventions

<u>Individual</u>

- monitor effects of interventions

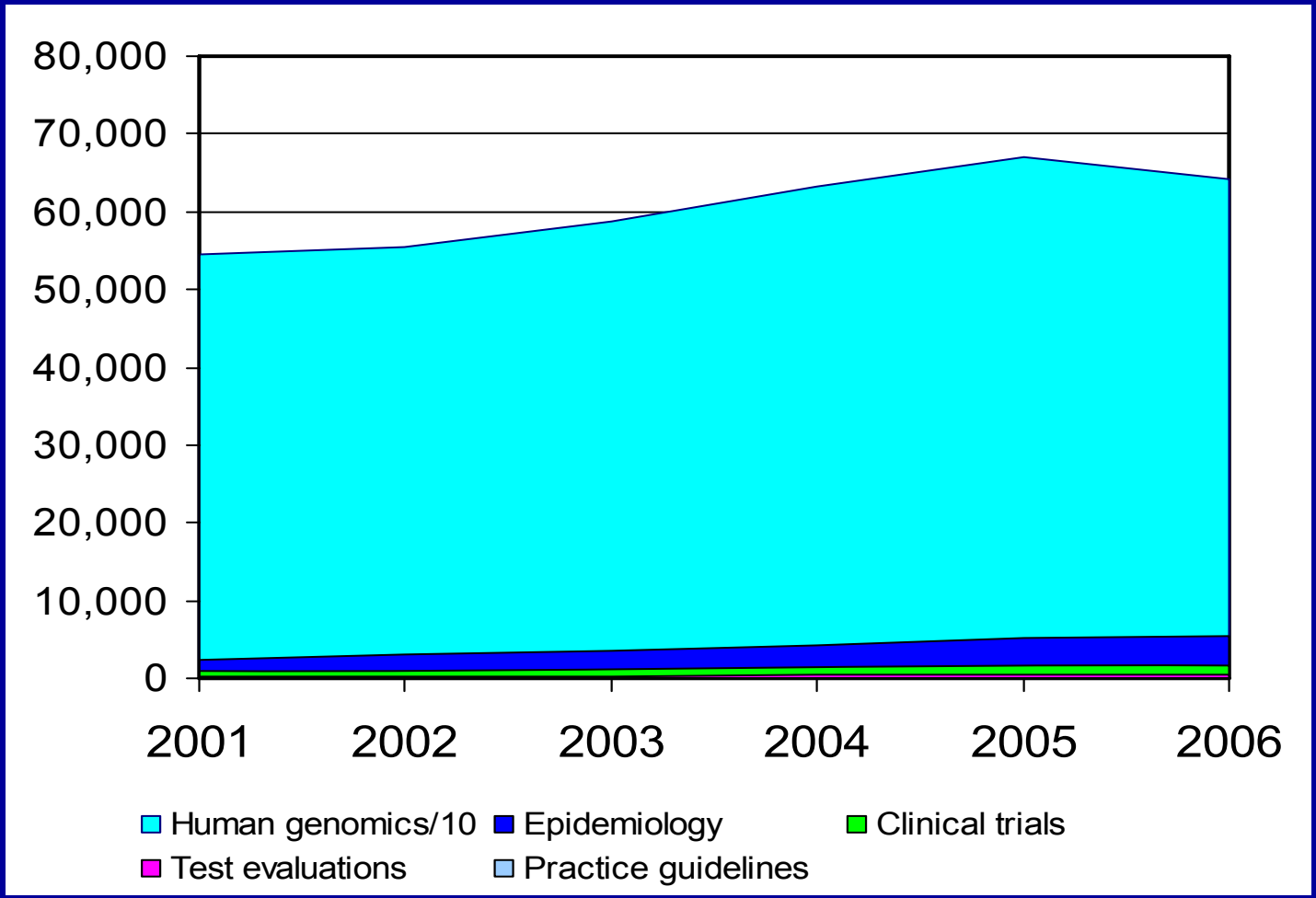
evaluate

Public Health Genomics: Closing the Gap Between Gene Discovery and Population Health

Population-level questions are important for developing genomic applications

- How many people have this genetic variant?
- Is prevalence different in subgroups of the population?
- How much of the population burden of disease does it explain?
- Does the variant interact with other genes and modifiable risk factors?
- Can we construct a risk profile related to genetic and environmental factors?

Numbers of publications related to human genetics and genomics, observational studies, clinical trials, practice guidelines and research on genetic tests, 2001-2006*



*based on PubMed, HuGE Navigator

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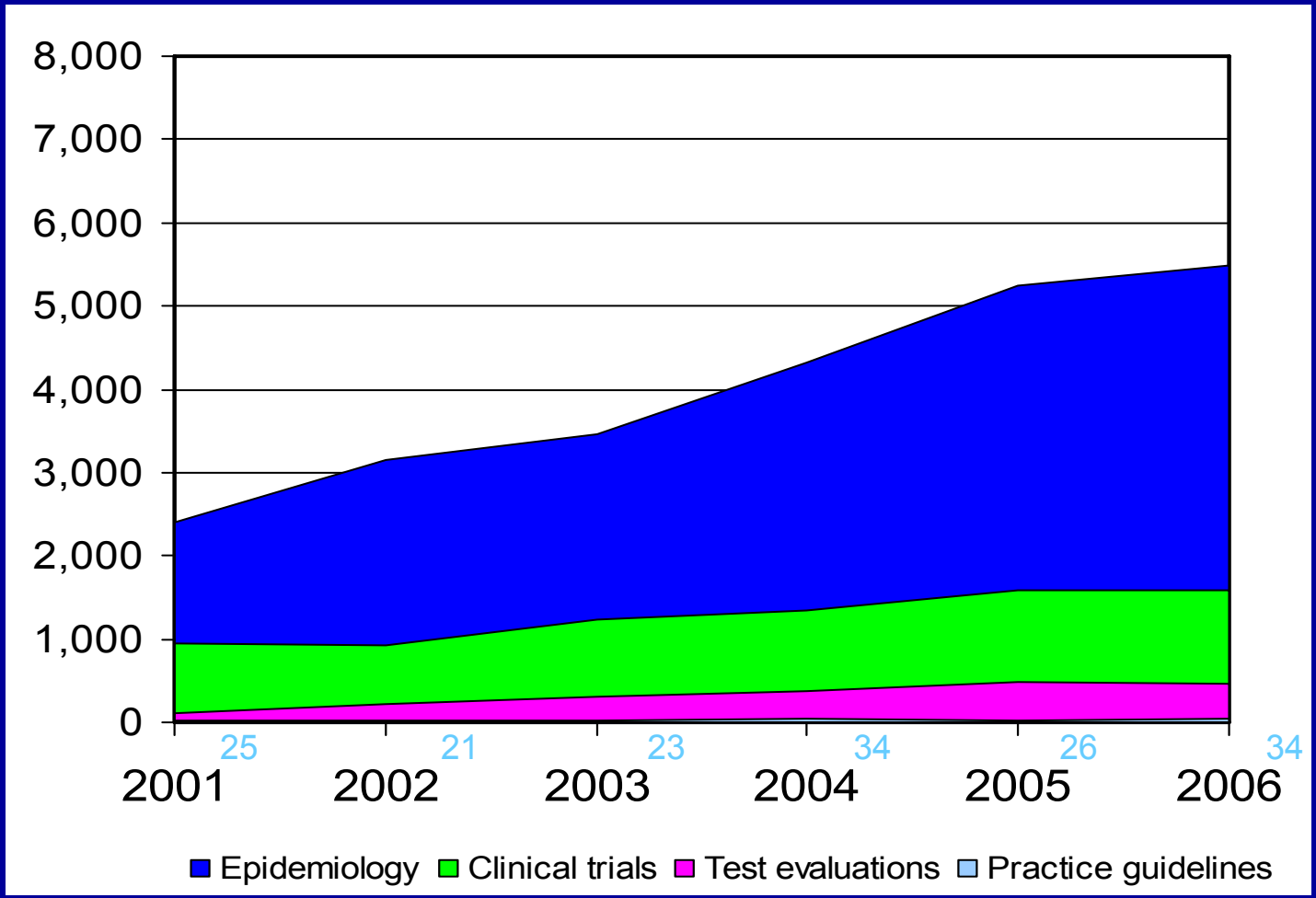
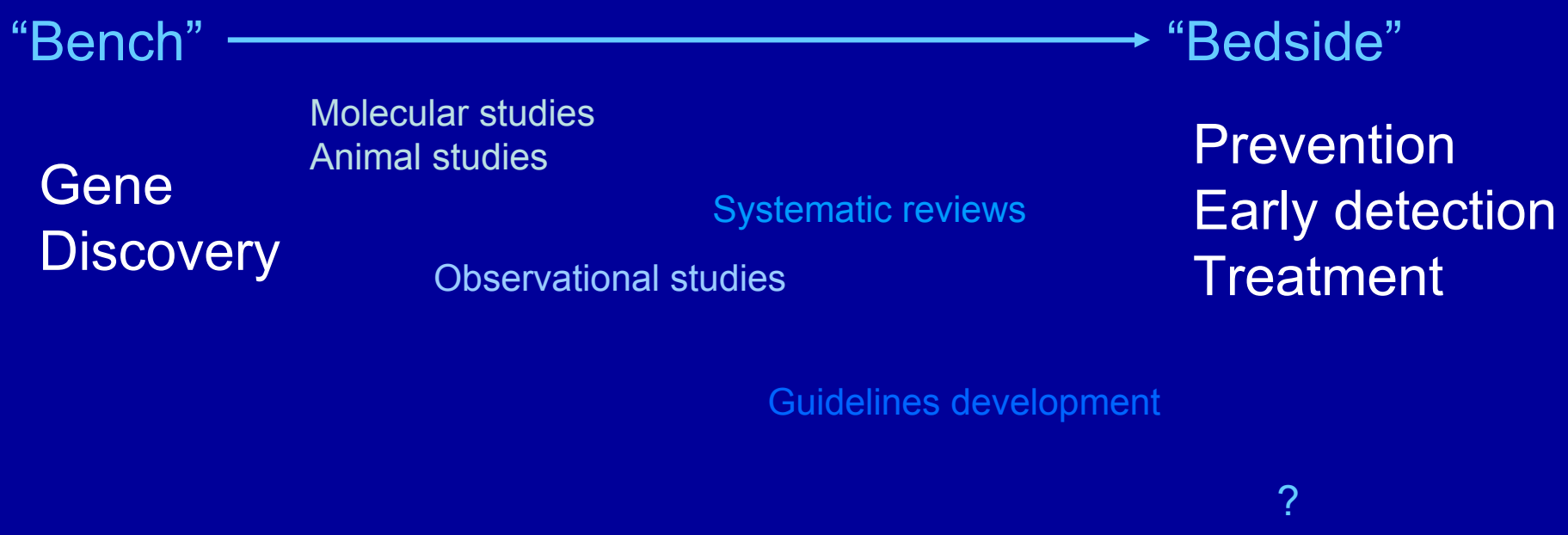


*based on PubMed, HuGE Navigator

From Bench to Bedside?



A Third Phase of Translation

Practice-based research--"Blue Highways" on the NIH roadmap, Westfall et al, JAMA, 2007

- Knowledge synthesis
- Guidelines development

- Dissemination research
- Implementation research

The Translation Continuum from Gene Discovery to Population Health

T1	T2	T3	T4
From Gene Discovery to Health Application	From Health Application to Evidence-based Guideline	From Guideline to Health Practice	From Practice to Health Impact

Khoury MJ et al. Genetics in Medicine 2007 (in press)

The Translation Continuum from Gene Discovery to Population Health

Less than 3% of published genomics research is T2 and beyond

T1	T2	T3	T4
From Gene Discovery to Health Application	From Health Application to Evidence-based Guideline	From Guideline to Health Practice	From Practice to Health Impact

Khoury MJ et al. Genetics in Medicine 2007 (in press)

The Translation Continuum from Gene Discovery to Population Health

T1	T2	T3	T4
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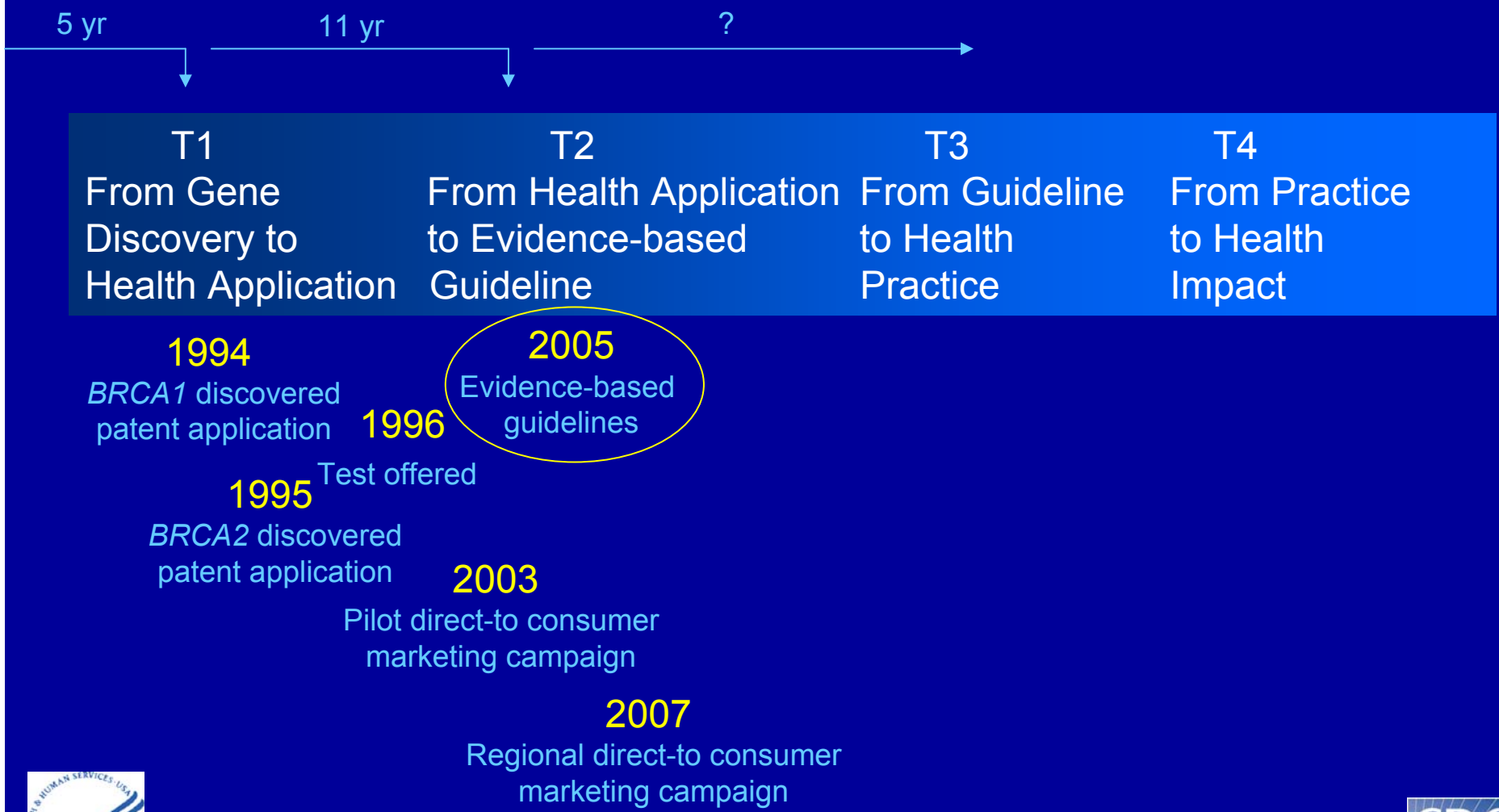
Only 2 USPSTF evidence-based guidelines (*BRCA1/2* and *HFE*)

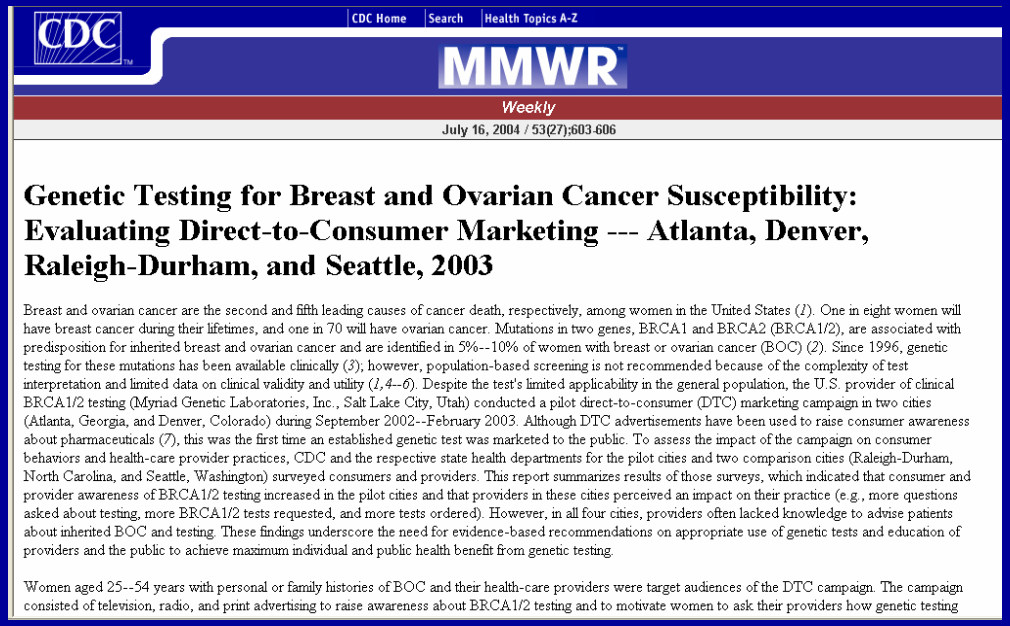
Khoury MJ et al. Genetics in Medicine 2007 (in press)

The Translation Continuum

Case study: BRCA1/2 and Breast Cancer



Public Health Assessment, 2003



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MMWR

Weekly

July 16, 2004 / 53(27);603-606

Genetic Testing for Breast and Ovarian Cancer Susceptibility: Evaluating Direct-to-Consumer Marketing --- Atlanta, Denver, Raleigh-Durham, and Seattle, 2003

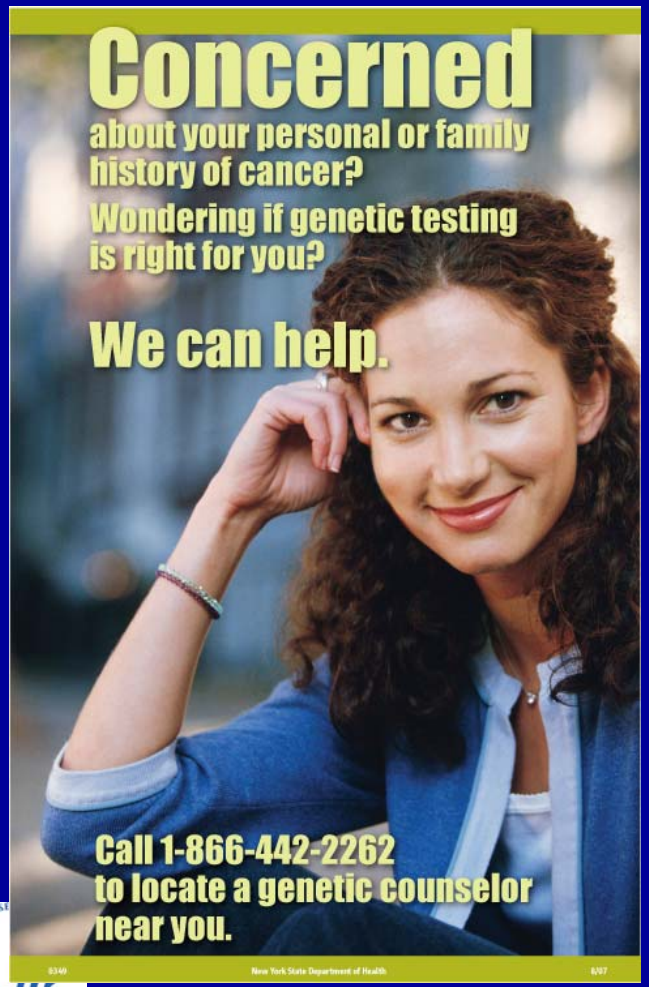
Breast and ovarian cancer are the second and fifth leading causes of cancer death, respectively, among women in the United States (*1*). One in eight women will have breast cancer during their lifetimes, and one in 70 will have ovarian cancer. Mutations in two genes, BRCA1 and BRCA2 (BRCA1/2), are associated with predisposition for inherited breast and ovarian cancer and are identified in 5%--10% of women with breast or ovarian cancer (BOC) (*2*). Since 1996, genetic testing for these mutations has been available clinically (*3*); however, population-based screening is not recommended because of the complexity of test interpretation and limited data on clinical validity and utility (*1,4--6*). Despite the test's limited applicability in the general population, the U.S. provider of clinical BRCA1/2 testing (Myriad Genetic Laboratories, Inc., Salt Lake City, Utah) conducted a pilot direct-to-consumer (DTC) marketing campaign in two cities (Atlanta, Georgia, and Denver, Colorado) during September 2002--February 2003. Although DTC advertisements have been used to raise consumer awareness about pharmaceuticals (*7*), this was the first time an established genetic test was marketed to the public. To assess the impact of the campaign on consumer behaviors and health-care provider practices, CDC and the respective state health departments for the pilot cities and two comparison cities (Raleigh-Durham, North Carolina, and Seattle, Washington) surveyed consumers and providers. This report summarizes results of those surveys, which indicated that consumer and provider awareness of BRCA1/2 testing increased in the pilot cities and that providers in these cities perceived an impact on their practice (e.g., more questions asked about testing, more BRCA1/2 tests requested, and more tests ordered). However, in all four cities, providers often lacked knowledge to advise patients about inherited BOC and testing. These findings underscore the need for evidence-based recommendations on appropriate use of genetic tests and education of providers and the public to achieve maximum individual and public health benefit from genetic testing.

Women aged 25--54 years with personal or family histories of BOC and their health-care providers were target audiences of the DTC campaign. The campaign consisted of television, radio, and print advertising to raise awareness about BRCA1/2 testing and to motivate women to ask their providers how genetic testing

Surveys of target population and providers:

- 2 pilot states vs 2 “control” states
- ↑ awareness of genetic test
- ↔ knowledge of breast cancer genetics among providers

Public Health Response, 2007



NEW YORK STATE
Department of Health

Mailing to 20,000 primary care providers:

- letter from Director of Community Health
- poster with referral information
- Q&A on BRCA1/2 and breast cancer

http://www.health.state.ny.us/

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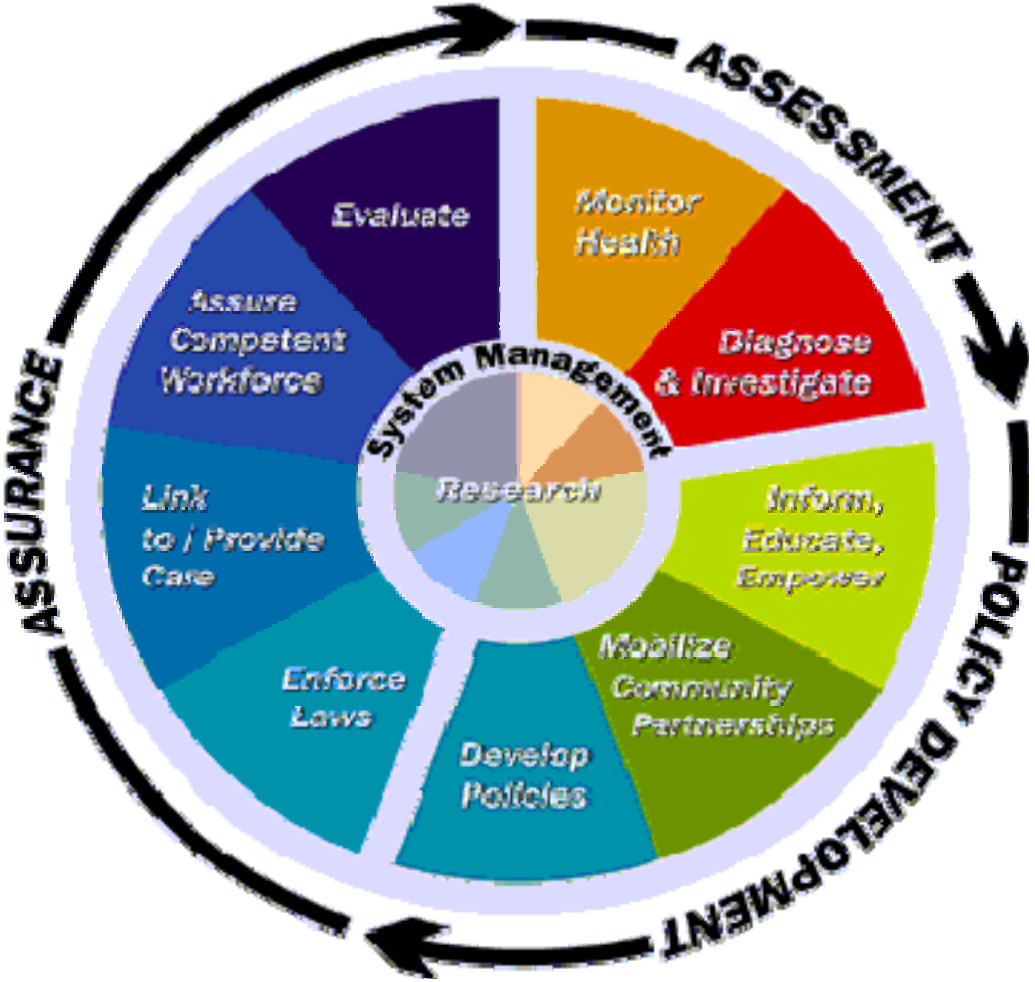
Core public health functions and essential services

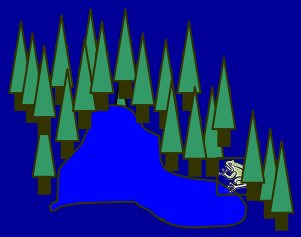


Core Public Health Functions Steering Committee, 1994

Beskow et al, 2001

Assessment

- Monitor health
- Diagnose and investigate



Environment

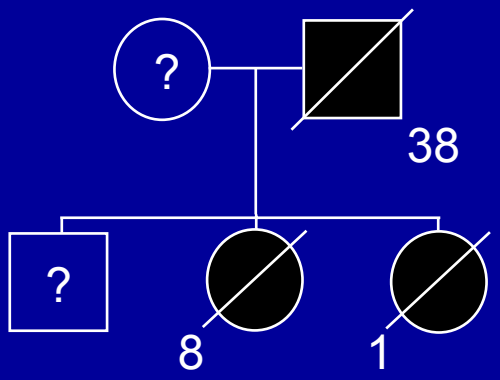
human genome

Host ⟷ Pathogen

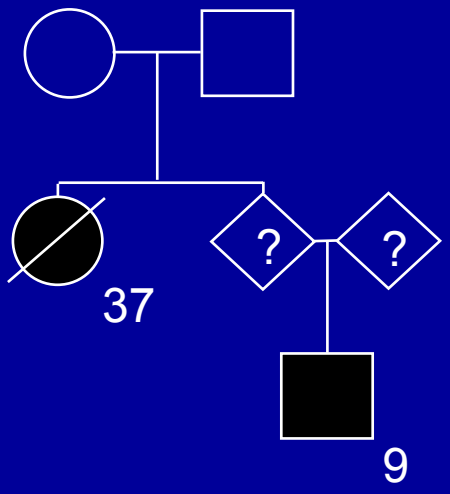
pathogen genome

"Epidemiologic triangle" for public health investigations of infectious diseases

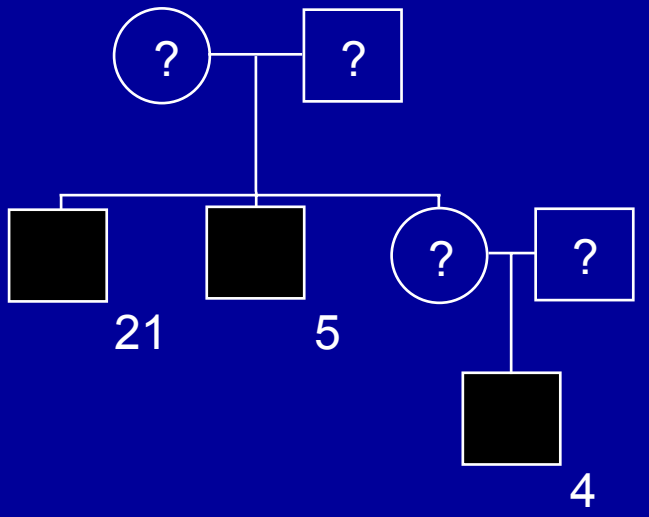
Familial Clusters of H5N1 Influenza



8 household or neighbor contacts

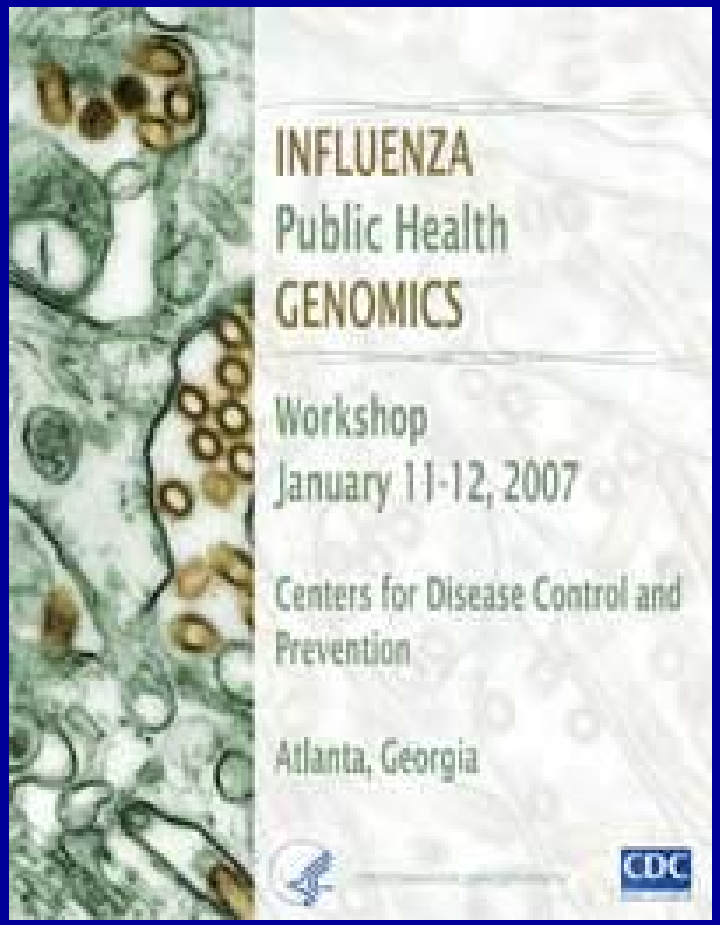


76 household or neighbor contacts



33 household or neighbor contacts

Kandun et al., New Engl J Med, 2006



- Severe influenza
- Vaccine response / reactions
- Translation for public health
- Research ethics, biobanking

Policy development

- Inform, educate
- Mobilize community partnerships
- Develop policies, plans

Policy development http://www.oregon.gov/DHS/ph/genetics/

OREGON.gov

Enter search term(s) Find

Text Size: A+ A- A Text Only Site Accessibility

Oregon Genetics Program

Department of Human Services

Department

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- Provider Resources
- Genetic Services
- Family Health History
- Genetic Privacy
- Data & Surveillance
- State Genetics Plan
- CDC Genomics Grant
- Publications & Talks
- Web Links
- Genetics in the News

Empowering people to make informed decisions about genetics and health

The mission of the Genetics Program is to promote the health and well-being of individuals and families who are impacted by inherited conditions or birth defects through public health assessment, policy development, assurance, and collaboration. The goals of the program are to reduce morbidity and mortality from inherited conditions and birth defects, to improve the quality of life for individuals and families impacted by inherited conditions and birth defects, and to empower people to make informed decisions about genetics and health.

Oregon's Strategic Plan for Genetics and Public Health

Education & Outreach

Family History

Information about collecting and using family history to identify and prevent disease.

Family History

Spanish Resources

Hoja Informativa Para Usuarios de AtenciÃ³n MÃ©dica Privacidad GenÃ©tica e InvestigaciÃ³n y NotificaciÃ³n de su derecho a negarse a participar

For Providers

Oregon Genetic Privacy Law

Since 1995, Oregon has had a law to protect the privacy of personal genetic information and prevent misuse of genetic information in clinical, research, employment, and insurance settings.

Privacy and Research

The Oregon Genetics Program Weblog

Empowering people to make informed decisions

DHS Quick Links

Public Health Division

- A-Z Topics
- News and Advisories
- Programs
- Staff Directory

Internet 100%

Assurance

- Enforce laws
- Link to provide care
- Assure competent workforce
- Evaluate

United States Government Accountability Office (GAO) Report, 2006

United States Government Accountability Office

GAO

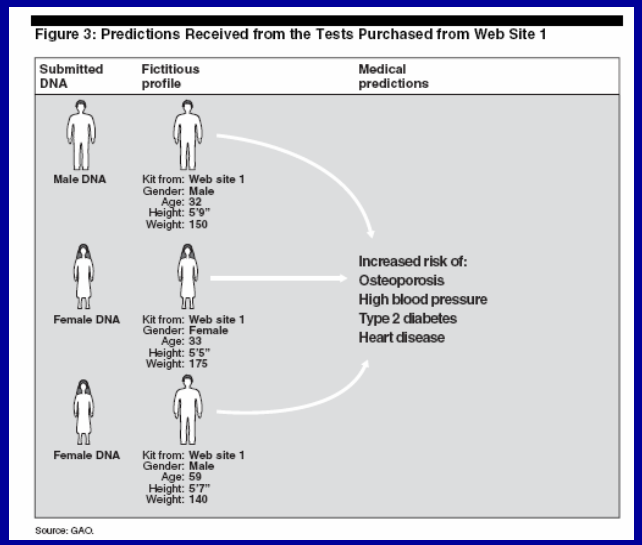
Testimony

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

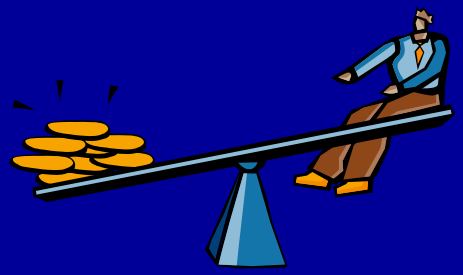


- Recommendations not based on unique genetic profile
- Unsubstantiated, misleading, and ambiguous predictions
- Recommended costly dietary supplements
- Supplements may be harmful for some people

http://www.gao.gov/new.items/d06977t.pdf

United States Health Care System, 2007

- Expanding technologies and treatments
- Increased push for quality and electronic health records
- Considerable variation in coverage
- Escalating costs and unequal access
- Financial pressures to control costs



Source: D Klein-Walker, President APHA, 2007

United States Public Health Functions, 2007

- Expanding technologies and treatments
- Increased push for quality and electronic health records

Assessment

- Considerable variation in coverage

- Escalating costs and unequal access
- Financial pressures to control costs

Policy

Assurance

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HuGENet

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HuGENet Coordinating Centers

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Bad Gastein, Austria
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