



## Genomics and Public Health: Where Are We Now and Where Are We Going?

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- School of Public Health, University at Albany
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- NYS Community Health Partnership

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- NYS Association of County Health Officials
- NYS Nurses Association

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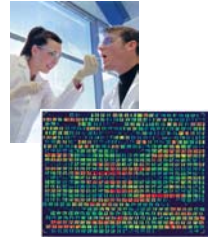
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## Program Objectives

- Present an Overview of Genomics in Public Health
- Discuss Current Initiatives in Genomics and Public Health
- Present Challenges and Action Steps for the Future of Genomics in Public Health

## “Genomics is to the 21st Century What Infectious Disease Was to the 20th Century...”

“...Genomics should be considered in every facet of public health: infectious disease, chronic disease, occupational health, environmental health, in addition to maternal and child health.”



Gerard S. Hayes, M, Rothstein M.J. Law Med Ethics. 2002

## Genetics vs. Genomics

- Genetics is the study of single genes and their effects.
- Genomics is the study of the functions and interactions of all the genes in the genome, including their interactions with environmental factors.

Guttmacher, AE and Collins FS. Genomic Medicine—A Primer. New England Journal of Medicine. 2002 November; 347(19): 1512.

## From Genetics to Genomics

Genetics → Genomics

- |                      |                        |
|----------------------|------------------------|
| • Disease            | • Information          |
| • Single Gene Ds     | • All Diseases         |
| • Mutations/One Gene | • Variation/Many Genes |
| • Rare               | • Common               |
| • “Genetic Services” | • Health care          |

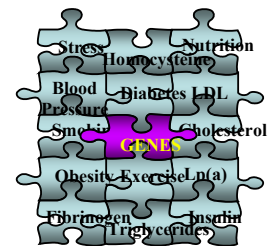
## Examples of Genetic Disorders and Coronary Heart Disease

Apolipoprotein(a) excess	Heparin cofactor II deficiency
Apolipoprotein AI deficiency	Homocystinuria/homoysteinemia
Autosomal recessive hypercholesterolemia	Niemann-Pick disease, type E
Cerebrotendinous xanthomatosis	Progeria
Fabry disease	Protein C deficiency
Familial combined hyperlipidemia	Pseudoxanthoma elasticum
Familial defective apoB	Sitosterolemia
<b>Familial hypercholesterolemia</b>	Spontaneous coronary dissection
Familial partial lipodystrophy	Tangier disease
Familial pseudo hyperkalemia due to RBC leak	Type III hyperlipoproteinemia
	Werner syndrome
	Williams syndrome

Source: Dr Maren Scheuner

## Most Coronary Heart Disease is Due to Gene-Environment Interactions

“Some vegetarians with acceptable cholesterol levels suffer myocardial infarction in the 30’s. Other individuals...seem to live forever despite personal stress, smoking, obesity, and poor adherence to a Heart Association-approved diet”

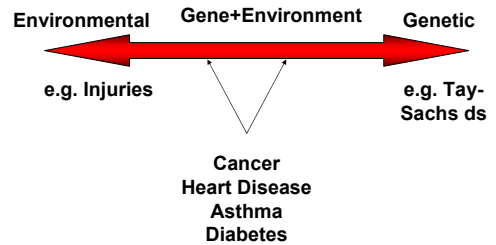


R.A. Hegele (1992)

## Genetic Susceptibility is NOT Health Destiny



## Almost all Diseases are Caused by Gene-Environment Interaction



## Top 10 Causes of Death in the U.S.

9 of 10 Have a Genetic Component

- Heart Disease
- Cancer
- Cerebrovascular Disease
- Chronic Lower Respiratory Disease
- Accidents/Unintentional Injuries (?)
- Diabetes
- Pneumonia/Influenza
- Alzheimer's Disease
- Kidney Disease
- Septicemia

National Vital Statistics Report, Vol 50, No. 16, September 16, 2002

## Using Genomics to Prevent Disease and Promote Health Across the Lifespan

### Infants

- Newborn screening prevents morbidity and disability in thousands of children annually.



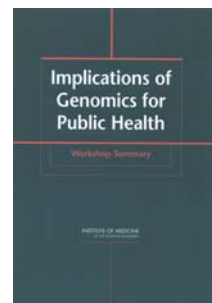
## Using Genomics to Prevent Disease and Promote Health Across the Lifespan

- **Children**
  - Genomics might explain why some children die from influenza infection.
- **Adolescents**
  - Understanding gene-drug interactions could help reduce asthma morbidity and drug side effects.
- **Adults and Older Adults**
  - Promoting screening for persons with family history could double the number of prevented colorectal cases.

## Public Health Genomics

- “An emerging field that assesses the impact of genes and their interaction with behavior, diet and the environment on the population's health”

(IOM, 2005)



## Current Priorities in “Public Health Genomics”

- Develop a coherent understanding of the scientific literature in genetics and its applications to public health and health care
- Identify gaps in knowledge and conduct public health genomics research

Cont.

(IOM, 2005)

## Current Priorities in “Public Health Genomics”

Cont.

- Develop a new health services paradigm to stratify the population into different risk groups
- Inform public policy
- Determine and implement education needs of workforce

(IOM, 2005)

## CDC’s Public Health Genomics Initiatives

- Family History Public Health Initiative
- Integrating Genomics into Public Health Surveys and Investigations
- Centers for Excellence in Genomics and Public Health
- Integrating Genomics into State Public Health Programs

## Family History

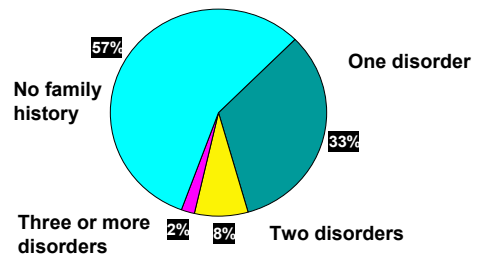
“Family history of disease reflects the interactions of genetic susceptibility, shared environment, and common behaviors.”



## Family History is a Risk Factor for Most Common Diseases

	<u>Relative Risk</u>
Heart disease	2.0 – 5.4
Breast cancer	2.1 – 3.9
Colorectal cancer	1.7 – 4.9
Prostate cancer	3.2 – 11.0
Melanoma	2.7 – 4.3
Type II diabetes	2.4 – 4.0
Osteoporosis	2.0 – 2.4
Asthma	3.0 – 7.0

## Family History of Common Diseases is Common in the Population



Scheuner et al. Am J Med Genet 1997;71:315-324.

## Family History Can Help Achieve Population Health Goals

- 14% of families account for almost half of the burden of heart attacks in Utah (Hunt 2003)
- Targeting colorectal cancer screening to close relatives can double the number of prevented colorectal cancer cases (Tyagi and Morris, 2003)
- More than 70% of adults with diabetes have a family history of diabetes (Hariri et al, 2005)

## CDC's Family History Public Health Initiative

- Developing and validating family history tool for use by practitioners and the public for six common diseases
- Diabetes, stroke, heart disease, breast, ovarian and colorectal cancer
- Improve targeting of screening and interventions for health impact

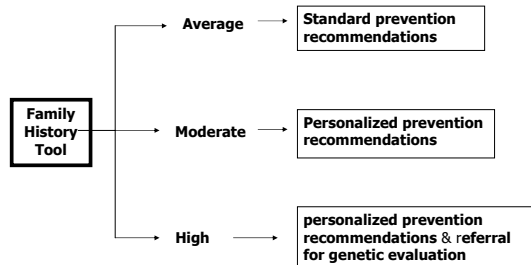
## CDC's Family History Web site for the Public



<http://www.cdc.gov/genomics/public/famhistMain.htm>

## Family history Risk Stratification Concept

Assessment Risk stratification Intervention



## U.S. Surgeon General's Family History Initiative



<http://www.hhs.gov/familyhistory/>

## Integrating Genomics into Public Health Surveys and Investigations

- Measure prevalence of variants of genes in USA and population subgroups
- Develop tools and methods for understanding population susceptibility to infectious and environmental exposures and response to intervention

## 1998 Springfield Ironhorse Triathlon Leptospirosis Outbreak

- 876 athletes; 12% reported illness
- Drinking infested lake water was the common risk factor
- HLA-DQ6 positive athletes were more likely to be seropositive for leptospirosis especially if they reported swallowing lake water

Lingappa J. et al., Genes & Immunity 2004

## Challenges in Public Health Genomics

- Lack of population-based data
- Rapid commercialization of genetic tests
- Ensuring quality of laboratory testing
- Availability of and access to interventions
- Potential discrimination/stigmatization
- Public and professional education

## IOM Report, 2002 *Who Will Keep the Public Healthy?*

- Critical areas for public health education in the 21<sup>st</sup> century
  - Informatics
  - **Genomics**
  - Communication
  - Cultural competence
  - Community-based research
  - Global health
  - Policy and law
  - Public health ethics

## Centers for Excellence in Genomics and Public Health

- 3 Centers (*Michigan, Washington, North Carolina*)

### Examples of Activities

- Technical assistance to state public health departments
- Training public health professionals
- Development of genomics knowledge-base

## Integrating Genomics into State Public Health Programs

- 4 states (Utah, Oregon, Michigan, Minnesota)

### Examples of Activities

- Workforce development
- Integration of genomics into surveys (e.g. BRFSS)
- Use of Family History

## Action Steps for Public Health

- Educate ourselves and our constituents
- Support the development and enactment of policies related to ethical and effective use of genome-based knowledge for population health
- Develop partnerships with stakeholders

## Action Steps for Public Health

- Use existing population surveys and investigations to help determine genetic contributions to disease and gene-environment interaction
- Identify populations at high and moderate genetic and familial risk who could benefit the most from medical, behavioral and environmental interventions

## Summary Points

- Genomics will affect public health practice beyond the traditional domain of genetic diseases.
- Scientific gaps exist in translating gene discoveries into population health benefits.
- We are seeing the emergence of “public health genomics” as a multidisciplinary field for the 21st Century.
- Know your Family History: It Could Save Your Life

## Learn More about Genomics

### Genomics for Public Health Practitioners

A screenshot of a CDC webpage titled "What Does Genomics Have to do with Public Health?". The page features a header with the title and a sub-header "Genomics for Public Health Practitioners". Below the header is a main image showing a group of people, and a sidebar with a list of topics including "Genetics in Public Health", "Genetics in Action", and "Genetics in the Future". The URL [http://www.cdc.gov/genomics/training/GPH/menu\\_content.html](http://www.cdc.gov/genomics/training/GPH/menu_content.html) is visible at the bottom.

### Six Weeks To Genomic Awareness

A screenshot of a CDC webpage titled "Want to Learn More about Genomics?". The page features a header with the title and a sub-header "Six Weeks To Genomic Awareness". Below the header is a main image showing a group of people, and a sidebar with a list of topics including "Genetics in Public Health", "Genetics in Action", and "Genetics in the Future". The URL <http://www.genomicawareness.org/index.htm> is visible at the bottom.

## Genomic Resources

### Centers for Genomics and Public Health

- University of Michigan:  
[www.sph.umich.edu/genomics](http://www.sph.umich.edu/genomics)
- University of Washington:  
<http://depts.washington.edu/cqph>
- University of North Carolina:  
[www.sph.unc.edu/nccqph](http://www.sph.unc.edu/nccqph)

## Genomic Resources

- CDC's Office of Genomics and Disease Prevention  
[www.cdc.gov/genomics](http://www.cdc.gov/genomics)
- National Newborn Screening and Genetics Resource Center  
<http://genes-r-us.uthscsa.edu>
- Association of State and Territorial Health Officials (ASTHO)  
[www.astho.org/phiip/genetics.html](http://www.astho.org/phiip/genetics.html)



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