But How Do We Translate Advances in Genomics into Population Health?

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CDC National Office of Public Health Genomics





What is "Public Health Genomics?"

 A multidisciplinary field concerned with the effective and responsible translation of genomebased knowledge and technologies to improve population health

Focus:

- Populations
- Gene-environment Interaction
- Disease prevention
- Evidence-based applications
- Health disparities

July 2006 - Vol. 8 - No. 7 commentary

The path from genome-based research to population health: Development of an international public health genomics network

Wylie Burke, MD, PhD¹, Muin J. Khoury, MD, PhD², Alison Stewart, PhD³, and Ronald L. Zimmern, MA, FFPHM⁴ for the Bellagio Group⁵

The health benefits of the Human Genome Project have been widely anticipated. Experts predict a new era of individualized disease prevention based on testing for genetic susceptibilities, and safer, more effective use of drugs based on Which vision of the future should the prudent clinician believe: A cornucopia of healthcare innovations based on genomic research, or a stream of genetically-based interventions that fail to deliver value to the public? We argue that both visions are

Genome-based Research and Population Health



Report of an expert workshop held at the Rockefeller Foundation Study and Conference Centre Bellagio, Italy, 14–20 April 2005









The Public Health Genomics Enterprise

Genomebased Science and Technology



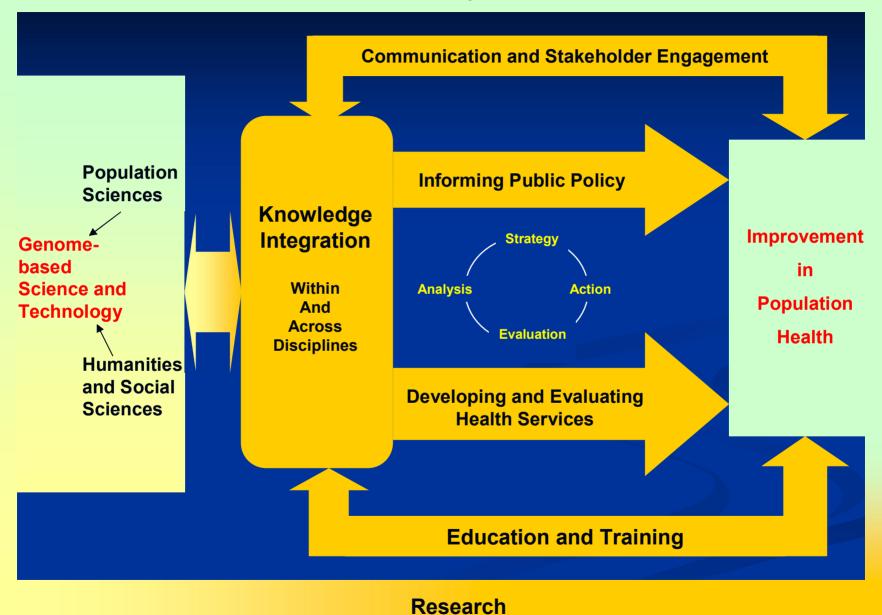
in
Population
Health

Closing the Gap Between Gene Human Genome Discoveries and Population Health

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

"Public Health" or "Population Health"?





SAFER-HEALTHIER-PEOPLE

Society Communication and Stakeholder Engagement Population Sciences ment Genome-In January, Dr Stephen Channock based from NCI gave us a primer Science and tion **Technology** on genome-based science th **Humanities** and technology and Social **Sciences Education and Training**

Research

AFER · HEALTHIER · PEOPLE

Communication and Stakeholder Engagement

Population Sciences

Genomebased Science and Technology

Humanities and Social Sciences

In February, we explored with Drs Bob
Hoover and Teri Manolio the role of
population sciences
with a focus on epidemiology

nt

Education and Training

Research

Communication and Stakeholder Engagement

Population Sciences

Genomebased Science and Technology

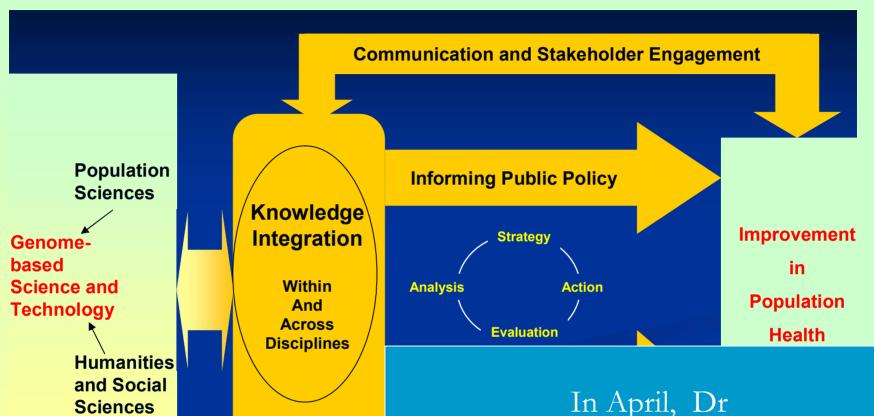
Humanities and Social Sciences

In March, we explored with Drs Colleen
McBride and David Abrams
the role of behavioral and social sciences

∍nt

Education and Training

Research



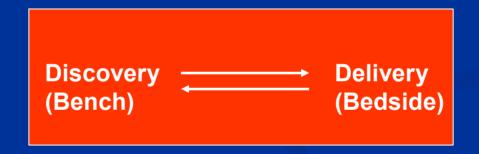
In April, Dr
Nat Rothman and I explored
the role of knowledge
integration within
the discipline of epidemiology



Society **Communication and Stakeholder Engagement Population Informing Public Policy Sciences Knowledge Improvement** Integration Strategy Genomebased in Science and Within **Analysis Action Population** And **Technology Across Evaluation** Health **Disciplines Humanities** and Social In May, Dr Al Berg and I **Sciences** explored the role of knowledge integration across disciplines, with a focus on evaluation of genetic tests

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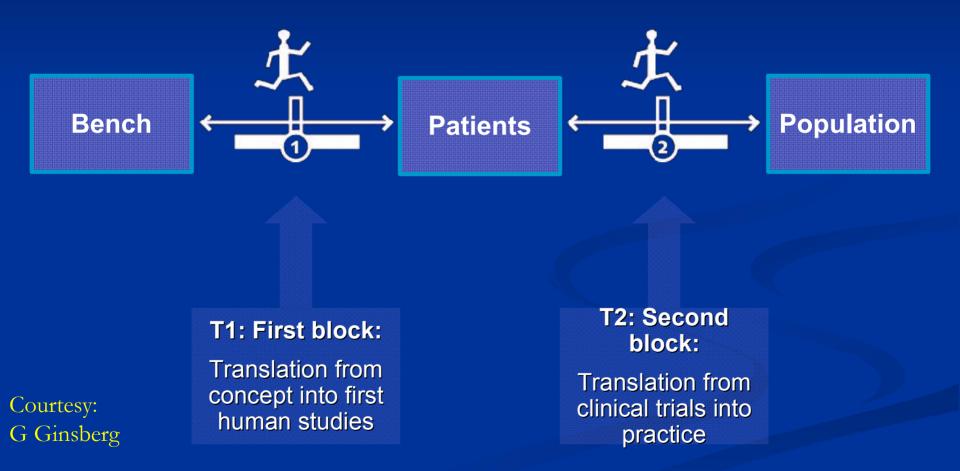
"Translational and Clinical Science— Time for a New Vision" E. Zerhouni NEJM 2005;353:15



It takes an estimated average of 17 years for 14% of new scientific discoveries to reach day to day clinical practice JM Westfall JAMA 2007;297:403



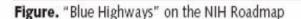
Translational Blocks Apply to Genomics in Medicine





Practice-Based Research-Blue Highways on the NIH Road Map

JM Westfall et al JAMA 2007;297:403.



BENCH

Basic Science Research

Preclinical Studies Animal Research T1

Case Series
Phase 1 and 2
Clinical Trials

TRANSLATION TO HUMANS BEDSIDE

Human Clinical Research

Controlled Observational Studies Phase 3 Clinical Trials PRACTICE

Clinical Practice

Delivery of Recommended Care to the Right Patient at the Right Time Identification of New Clinical Questions and Gaps in Care

T2

Guideline Development Meta-analyses Systematic Reviews

> TRANSLATION TO PATIENTS

Practice-Based Research

Phase 3 and 4 Clinical Trials Observational Studies Survey Research T3

Dissemination Research Implementation Research

TRANSLATION TO PRACTICE

The Genomics Translation Highway: 2001-2006

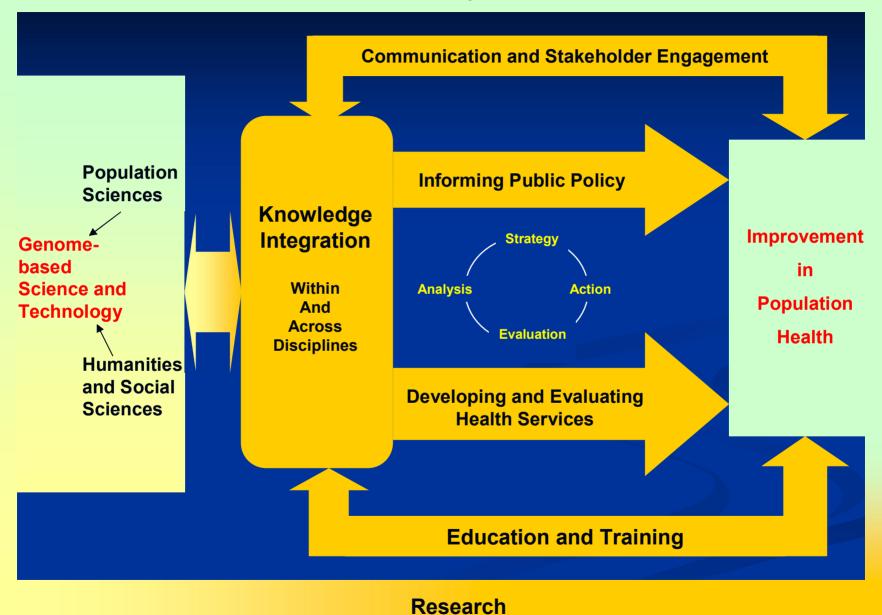
- More than 350,000 published human genetics/genomics articles
 - Only 2% deal with translation research T2 and Beyond
 - Only 2 USPSTF evidence-based recommendations
 - BRCA1
 - HFE



USPSTF 2005 Recommendations on BRCA Testing

- Recommended "against routine referral for genetic counseling or routine breast cancer susceptibility gene (BRCA) testing for women whose family history is not associated with an increased risk for deleterious mutations in breast cancer susceptibility gene 1 (BRCA1) or breast cancer susceptibility gene 2 (BRCA2)."
- Recommended that "women whose family history is associated with an increased risk for deleterious mutations in BRCA1 or BRCA2 genes be referred for genetic counseling and evaluation for BRCA testing."
- USPSTF spelled out clearly what family history criteria warranted the referral for counseling and possible testing.





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