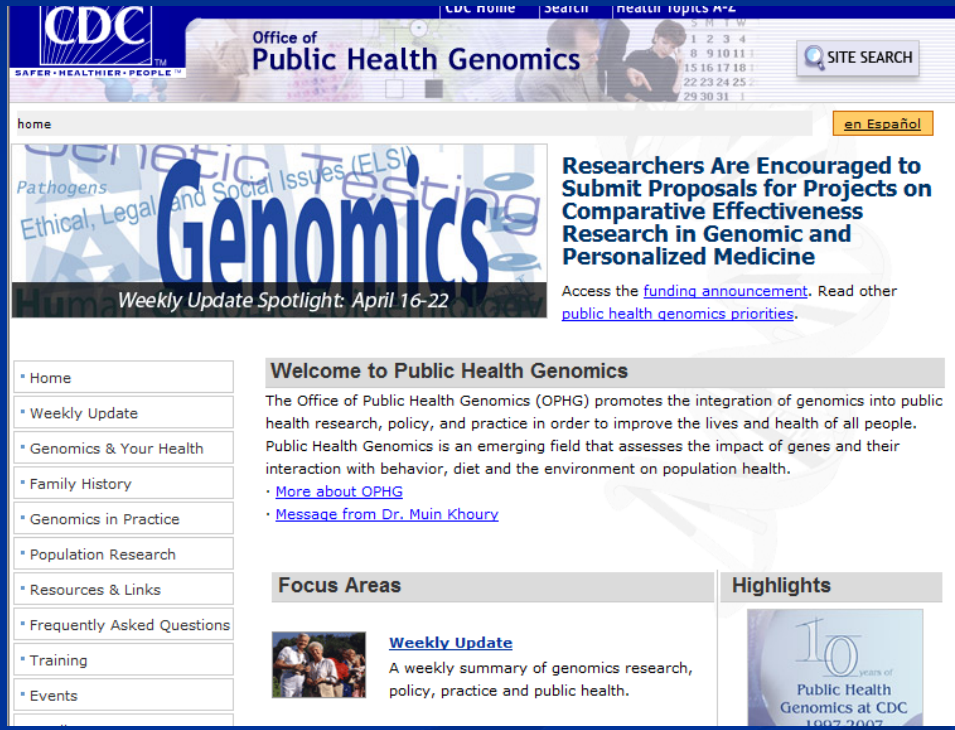
CDC Update
SACGHS, Feb 2010

Muin J. Khoury MD, PhD

Director, Office of Public Health Genomics

Centers for Disease Control and Prevention



Outline

- Translation Gap between Gene Discovery and Population Health Impact: The Need for Data to Drive Policy and Practice
- CDC Focus on Public Health Genomics
- Integration of Genomics Objectives into Healthy People 2020: What Gets Measured Gets Done

What Do You Do with Genes When You Find Them?

Predictive Health

Personalized Medicine

Prevention

"I predict that comprehensive, genomics-based health care will become the norm with individualized preventive medicine and early detection of illnesses" (E. Zerhouni, 2006)

"We are on the leading edge of a true revolution in medicine, one that promises to transform the traditional "one size fits all" approach into a much more powerful strategy that considers each individual as unique and as having special characteristics that should guide an approach to staying healthy" (F. Collins, 2010)

From Genomic Discovery to Health Impact: The Translation Gap!

Health Affairs Dec 2008

The Evidence Dilemma In Genomic Medicine

We need a roadmap for the appropriate integration of genomic discoveries into clinical practice.

by Muin J. Khoury, Al Berg, Ralph Coates, James Evans, Steven M. Teutsch, and Linda A. Bradley

ABSTRACT: An ongoing dilemma in genomic medicine is balancing the need for scientific innovation with appropriate evidence thresholds for moving technology into practice. The current low threshold allows unsubstantiated technologies to enter into practice, with the potential to overwhelm the health system. Alternatively, establishing an excessively high threshold for evidence could slow the integration of genomics into practice and present disincentives for investing in research and development. Also, variable coverage and reimbursement policies can lead to differential access to technology, exacerbating health disparities. There is an urgent need for a collaborative process for appropriate transition of genomic discoveries from research to practice. [*Health Affairs* 27, no. 6 (2008): 1600–1611; 10.1377/hlthaff.27.6.1600]

COMMENTARY

JAMA Dec 3, 2008

Closing the Evidence Gap in the Use of Emerging Testing Technologies in Clinical Practice

Kathryn A. Phillips, PhD

NEW TESTING TECHNOLOGIES—INCREASINGLY BASED on genomic information—are essential in the shift toward personalized medicine and molecular targeted therapies. Considering the rapid proliferation of new tests, health care insurers and policy makers are

There is no consensus about optimal testing methods. Guidelines recommend using either immunohistochemistry, with indeterminate results confirmed by fluorescence in situ hybridization (FISH), or FISH to determine HER2 status.[1] Although FISH is a better predictor of response to treatment, immunohistochemistry costs substantially less and is more easily performed in community laboratories.[1]

Despite the clinical success of trastuzumab, there are con-

PERSPECTIVE

The Human Genome And Translational Research: How Much Evidence Is Enough?

Given the lack of a robust translational infrastructure, conflict between those developing new technologies and those who must u or pay for them seems inevitable.

by Janet Woodcock

ABSTRACT: Multiple new genomic diagnostic tests are currently under developm Given the lack of an efficient translational infrastructure, it is not clear how, or whether, ro-bust evidence for their clinical value will be generated. [*Health Affairs* 27, no. 6 (2008): 1616–1618; 10.1377/hlthaff.27.6.1616]

PERSPECTIVE

The Health Benefits Of Genomics: Out With The Old, In With The New

We must dispense with old models of research support and regulatory guidance designed for the pre–Human Genome Project world.

by Kathy Hudson

Genomics Translation Gap: From Bench to Population Health Impact

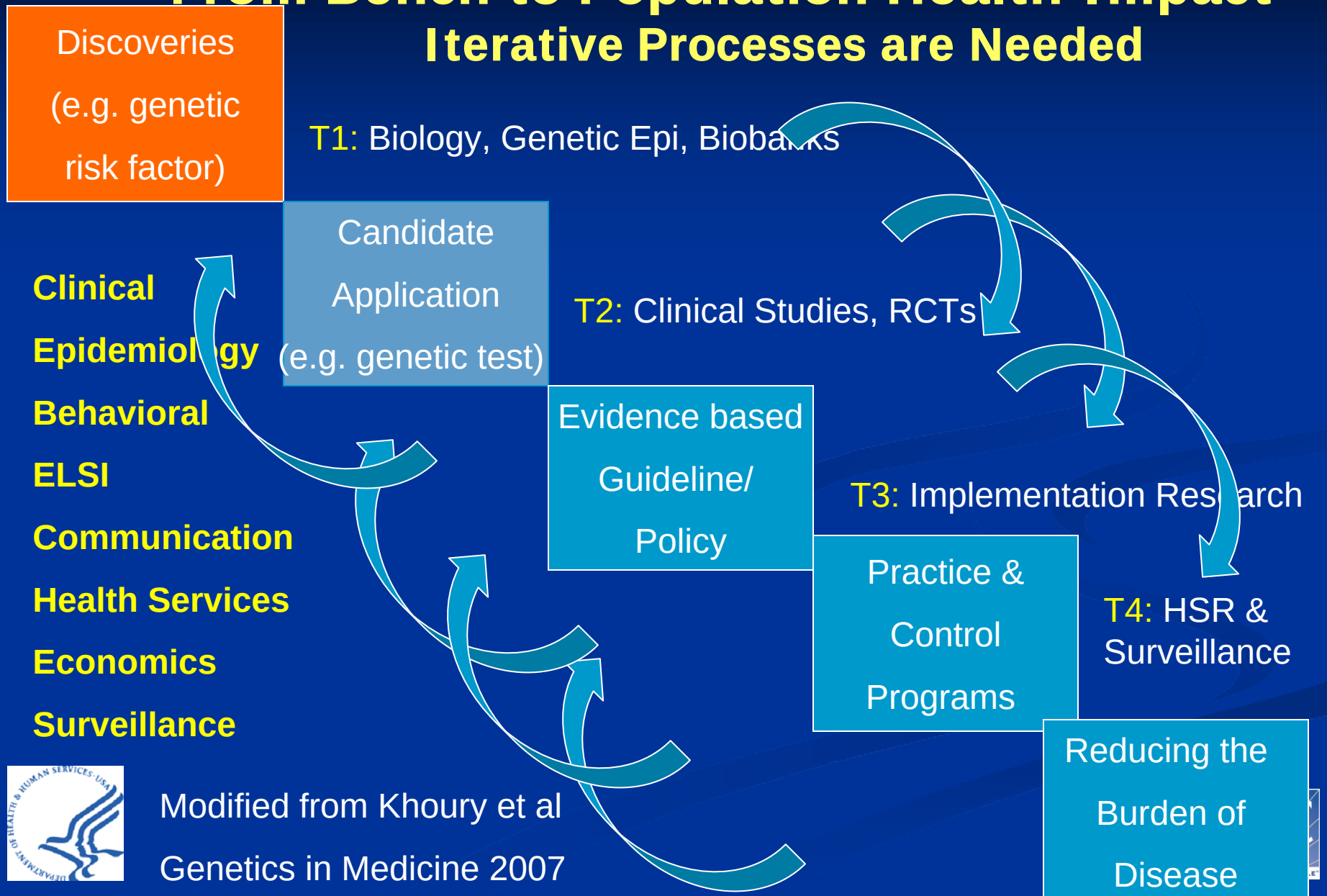
Discoveries (e.g. genetic risk factor)

Modified from Khoury et al Genetics in Medicine 2007

Reducing the Burden of Disease

Genomics Translation Gap: From Bench to Population Health Impact

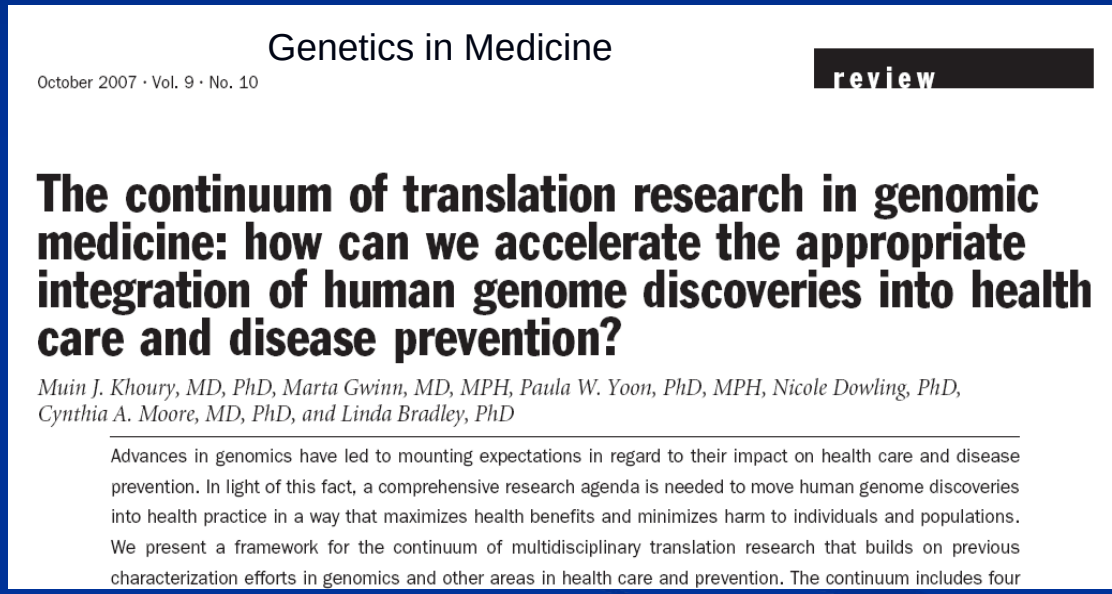
Iterative Processes are Needed



Modified from Khoury et al Genetics in Medicine 2007

The Genomics Translation Research Continuum: 2001-2006

- **More than 350,000 published human genetics/genomics articles**
  - **Almost all discovery**
  - **~ 2% Translation Research T2 +**
  - **Only 2 evidence-based recommendations**
    - *BRCA1* (11 years post gene discovery)
    - *HFE* (10 years post gene discovery)



Genetics in Medicine
October 2007 · Vol. 9 · No. 10

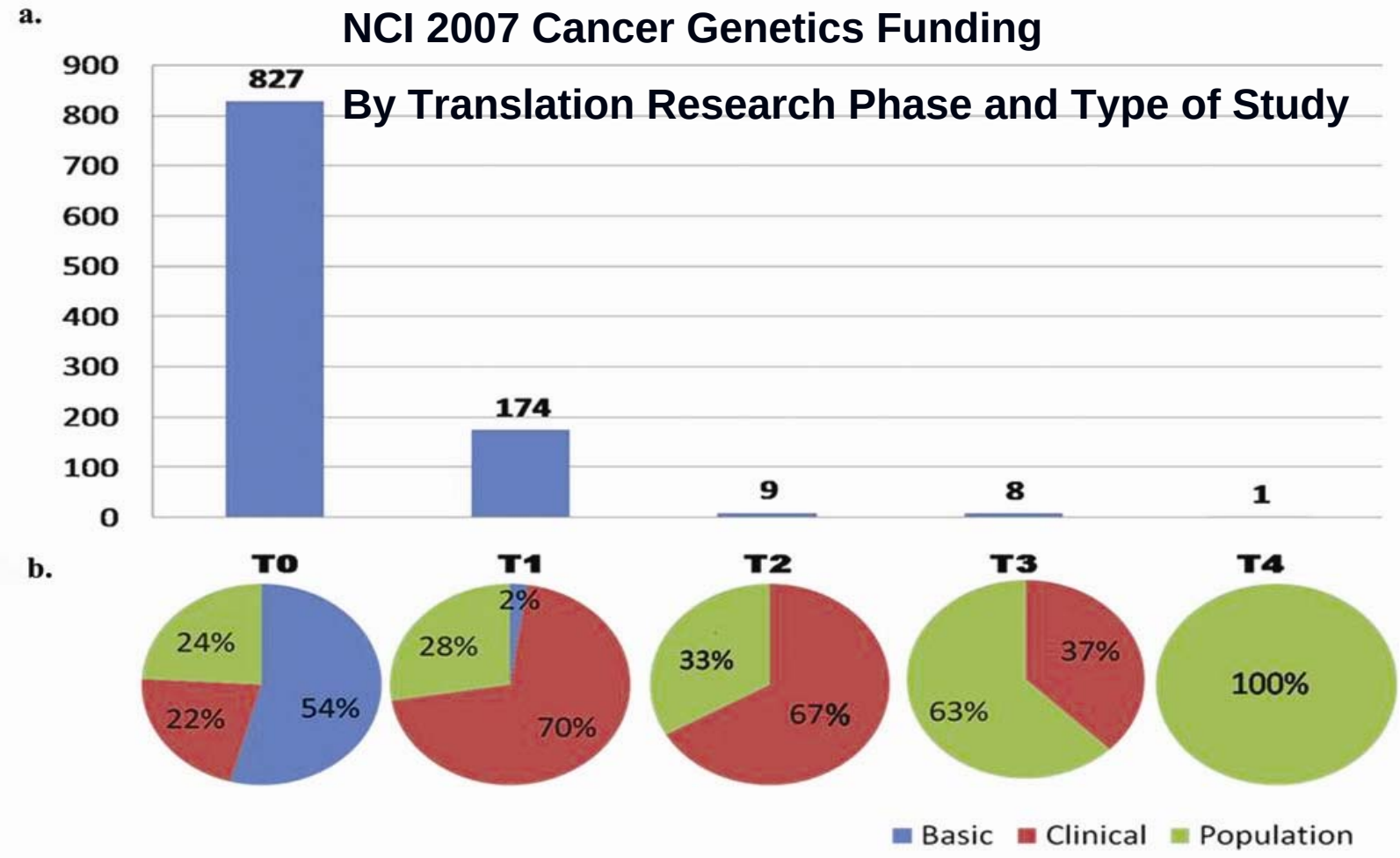
review

The continuum of translation research in genomic medicine: how can we accelerate the appropriate integration of human genome discoveries into health care and disease prevention?

Muin J. Khoury, MD, PhD, Marta Gwinn, MD, MPH, Paula W. Yoon, PhD, MPH, Nicole Dowling, PhD, Cynthia A. Moore, MD, PhD, and Linda Bradley, PhD

Advances in genomics have led to mounting expectations in regard to their impact on health care and disease prevention. In light of this fact, a comprehensive research agenda is needed to move human genome discoveries into health practice in a way that maximizes health benefits and minimizes harm to individuals and populations. We present a framework for the continuum of multidisciplinary translation research that builds on previous characterization efforts in genomics and other areas in health care and prevention. The continuum includes four

We Do not Make Adequate Investments in Genomics Translation Research



Schully S et al. Public Health Genomics 2010 (in press)

Outline

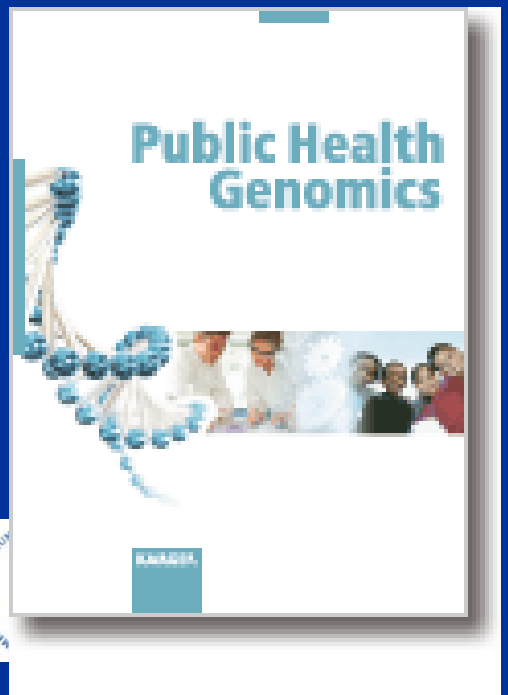
Translation Gap between Gene Discovery and Population Health Impact: The Need for Data to Drive Policy and Practice

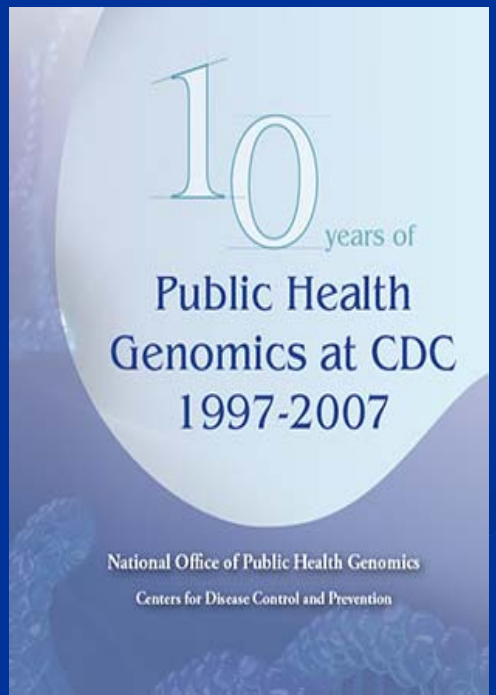
CDC Focus on Public Health Genomics

Integration of Genomics Objectives into Healthy People 2020: What Gets Measured Gets Done

Emergence of "Public Health Genomics"

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





PHG emphasizes

1. Prevention
2. Population approach
3. Health disparities
4. ELSI
5. Evidence-based translation

Data for Action

"The single most important thing that Public Health can do is to increase the degree to which decisions are made using good data"

Dr Tom Frieden , CDC Director , Jan 1, 2010

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 Studies & Surveys

Gene Discovery ⇢ **Closing the Gap** ⇢ **Population Health**

What Does Genetic Information Mean for Population Health & Disease Burden?

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

How can we Integrate the Evidence For the Value of Genomic Applications and Develop Recommendations For Practice and Prevention

Gene Discovery → **Closing the Gap** → **Population Health**

EGAPP

Evaluation of Genomic Applications in Practice & Prevention

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

Translation Research & Programs

GAPPNet

Workforce Issues

Gene Discovery

Closing the Gap

Population Health

How Can We Move Validated Genetic Information From Research to Practice and Document Health Impact in Populations? How can Prepare the Workforce and the Public?

Outline

Translation Gap between Gene Discovery and Population Health Impact: The Need for Data to Drive Policy and Practice

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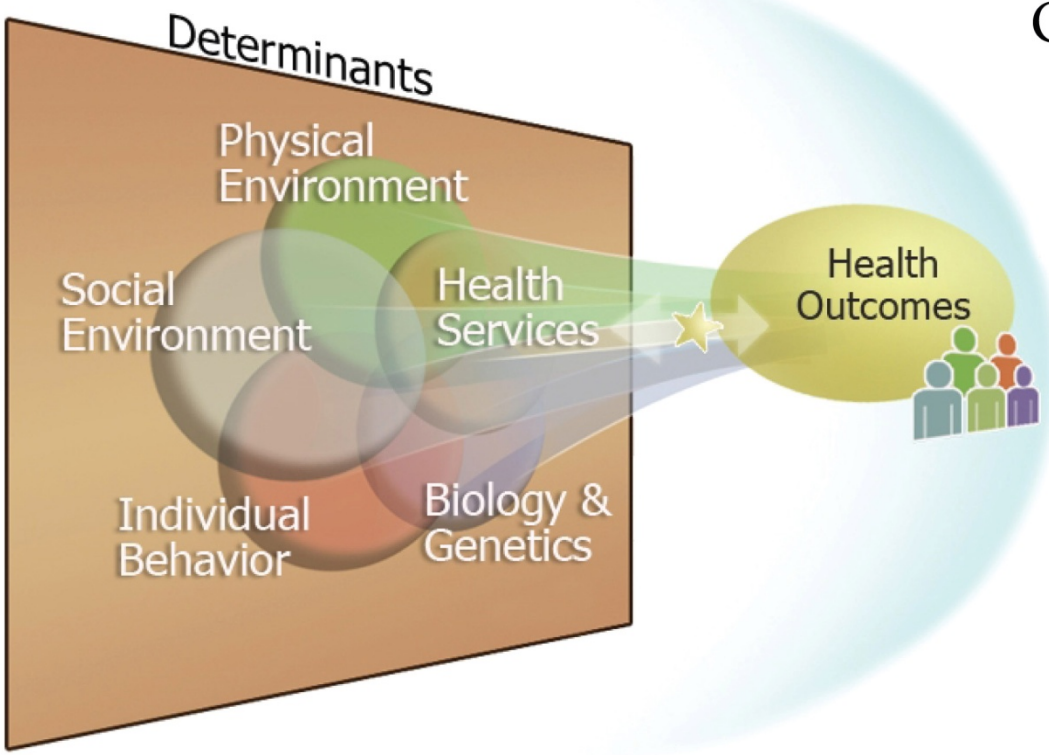
Integration of Genomics Objectives into Healthy People 2020: What Gets Measured Gets Done

Healthy People 2020

A society in which all people live long, healthy lives



Overarching Goals:

- Attain high quality, longer lives free of preventable disease, disability, injury, and premature death.
- Achieve health equity, eliminate disparities, and improve the health of all groups
- Create social and physical environments that promote good health for all.
- Promote quality of life, healthy development and healthy behaviors across all life stages.

Healthy People 2020 Federal Interagency Workgroup

- **Trans-federal membership**
- **55 members, representing 24 HHS agencies/offices**
- **Expanded to include non-HHS Federal partners**
  - ED, EPA, DOI, DOJ, HUD, USDA, VA, and others to come

Draft HP 2020 Organizing Framework

- **Vision: A society in which all people live long, healthy lives.**
- **Mission: Healthy People 2020 strives to:**
  - Identify nationwide health improvement priorities;
  - Increase public awareness and understanding of the determinants of health, disease, and disability and the opportunities for progress;
  - Provide measurable objectives and goals that are applicable at the national, state, and local levels;
  - Engage multiple sectors to take actions to strengthen policies and improve practices that are driven by the best available evidence and knowledge;
  - Identify critical research, evaluation, and data collection needs.

Developing Healthy People 2020 Objectives

- **Eight criteria for proposed objectives:**
  - Important and understandable to a broad audience
  - Prevention-oriented and achievable through various interventions
  - Drive action
  - Useful and reflect issues of national importance
  - Measurable and address a range of issues
  - Build on past iterations of Healthy People
  - Supported with best available scientific evidence
  - Address population disparities

Developing Healthy People 2020 Objectives

- **Data Expectations: Each objective should have:**
  - A valid, reliable, nationally-representative data source (or potential source identified)
  - Baseline data
  - Assurance of at least one additional data point throughout the decade
- **Targets**
  - Each objective will have its own target
  - Target-setting policies on methods to be used are being discussed
- **Approval**
  - Each objective will be approved by the Federal Interagency Workgroup

Healthy People 2010

- Two overarching goals
- 28 focus areas
- 467 specific objectives

No genomics focus area/objectives

Some passing references to genomics in narrative

Genomics Topic Area Proposal for HP 2020

As genetics is recognized by Healthy People 2020 as a major determinant of health outcomes, it could be useful for the Healthy People 2020 initiative to develop a workgroup and objectives to help assure that rapidly advancing knowledge in genetics and genomics is translated into personal and clinical practices that maximize benefits and minimize harms to populations and to individuals.

HP 2020 Genomics Workgroup

Co-lead: Katherine Kolor, CDC
Co-lead: Gurvaneet Randhawa, AHRQ

Janice Bach, MDCH
Coleen Boyle, CDC
Jean Chabut, MDCH
Frederick Chen, UW
Ralph Coates, CDC
Rosaly Correa-de-Araujo, HHS OD
W. David Dotson, CDC
Greg Downing, HHS OS
Debra Duquette, MDCH
Greg Feero, NHGRI
Bob German, CDC
Alaina Harris, HRSA
Sandra Howard, HHS ASPE
Mildred Hunter, HHS OMH
Susan Schneider, CDC
Ira Lubin, CDC
Marie Mann, HRSA
Elizabeth Mansfield, FDA
Geraldine McQuillan, CDC
Laurence Meyer, VA
Debra Nichols, HHS ODPHP
Nakki Price, CDC
Michelle Puryear, HRSA
Jeffrey Roche, CMS
Susan Snyder, CDC

Four Proposed Genomics Objectives: Promoting evidence-based practice

1. **Increasing the knowledge base to support evidence-based practices for genomic applications**
    1. Translation research studies
    2. Evidence-based recommendations
2. **Increasing the implementation of evidence-based practices for genomic applications**
    1. EGAPP Working Group Lynch recommendation
    2. USPSTF BRCA recommendation

Two Approved Genomics Objectives

1. Increase the proportion of persons with newly diagnosed colorectal cancer who receive genetic testing to identify Lynch syndrome (or familial colorectal cancer syndromes)

2. Increase the proportion of women with a family history of breast and/or ovarian cancer who receive genetic counseling.

Public Comment Summary: Genomics Topic Area

1. Topic Area Comments (n=6)
2. Objectives Comments (n= 5)
3. New Objectives Comments (n=9)

Preliminary disposition: No changes to proposed objectives, but some comments will be incorporated in the narrative of the genomics topic area.

Next Steps

1. Final disposition of public comments
2. Identify targets for two objectives
3. Draft narrative section of genomics topic area