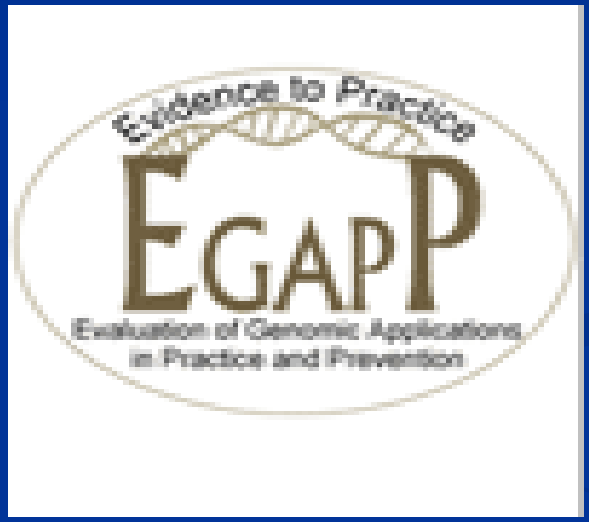
Evaluation of Genomic Applications in Practice and Prevention Initiative: An Update, SACGHS June 2005



Muin J. Khoury MD, PhD

CDC Office of Genomics & Disease Prevention

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Pharmacogenomic Tests: a Public Health Issue

- Can potentially affect a lot of people
- Potential for targeting prevention efforts
- Need for evidence-based transition from research to practice
- Implementation and access
- Provider and public education
- Monitoring impact on population health
- Potential for early applications of genomics to population health

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

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article

Public health impact of genetic tests at the end of the 20th century

Paula W. Yoon, ScD, MPH[1], Bin Chen, PhD, FACMG[2], Andrew Faucett, MS, CGC[2], Mindy Clyne, MHS, CGC[1], Marta Gwinn, MD, MPH[1], Ira M. Lubin, PhD[2], Wylie Burke, MD, PhD[3], and Muin J. Khoury, MD, PhD[1]

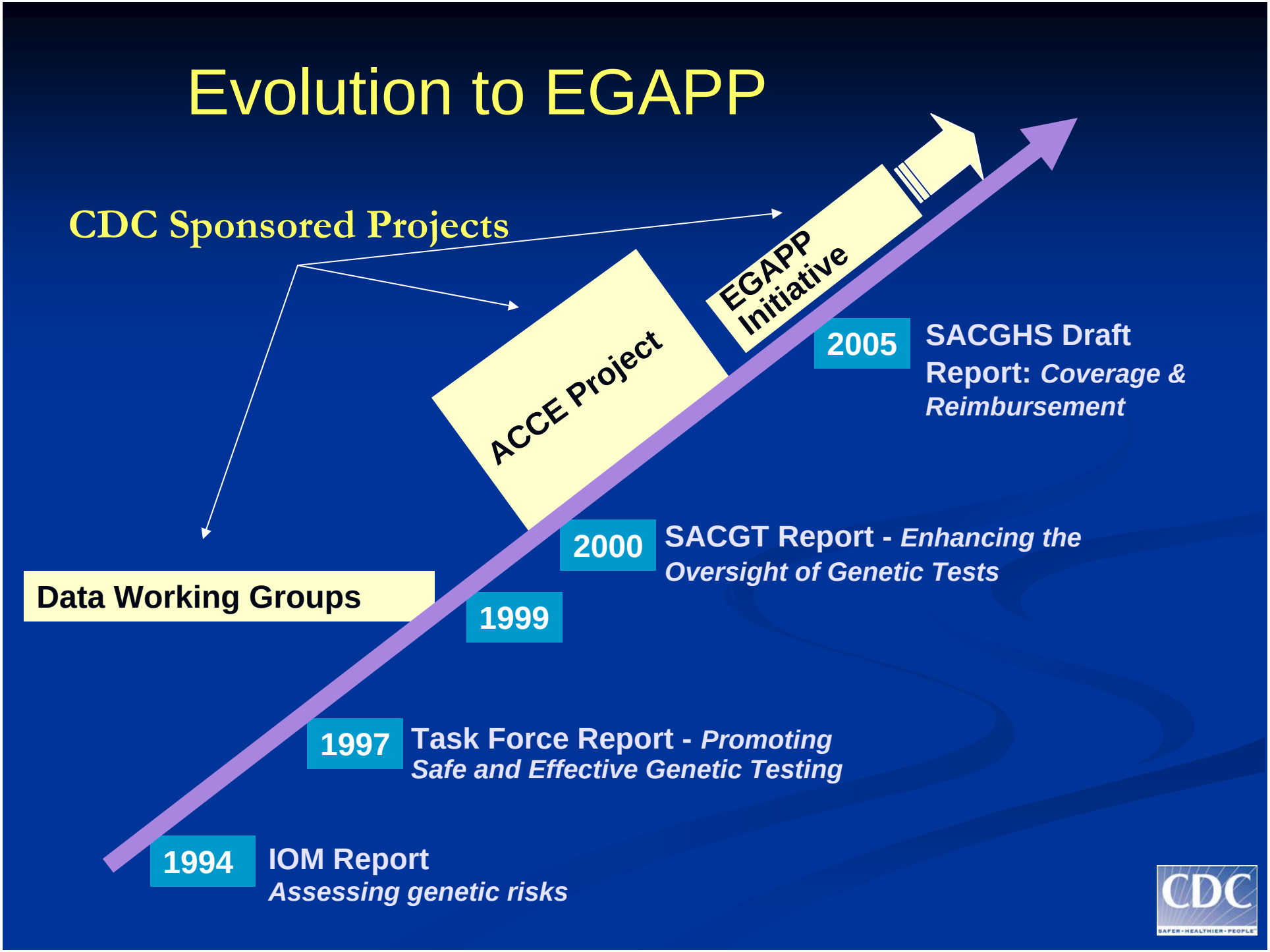
Purpose: To evaluate genetics tests available for clinical, research, and public health purposes in terms of their public health impact as measured by the number of people who could potentially be tested. **Methods:** Genetic tests for the 751 inherited diseases or conditions listed in the GeneTests database as of November 2000, were classified on the basis of their use for population-based testing and the prevalence of the disease or condition being tested. The GeneTests database divides the tests into two groups: those offered for clinical use and those available for research only. **Results:** Of the 423 clinical tests, 51 had potentially greater impact on public health because of their use in statewide newborn screening programs, other population screening programs, or testing for common diseases with a prevalence over 1 in 2,000 people. Among the 328 tests performed for research purposes only, 18 met the criteria for potentially greater public health impact. **Conclusions:** Our classification scheme indicated that fewer than 10% of the genetic tests listed in the GeneTests database at the end of 2000 are highly relevant to public health. The majority of genetic tests are used in diagnosis and/or genetic counseling for rare, single-gene disorders in a limited number of people. However, as more tests are being considered for

Public Health Impact of Genetic Tests at the End of the 20th Century

Yoon et al. Genet Med 2001;3:405-10.

- Assessment of GeneTests database (Nov 2000)
- 751 Conditions, 423 for clinical use
- 51 with significant public health impact
  - 19 newborn screening
  - 9 other population screening
  - 23 conditions with prevalence > 1/2000

Evolution to EGAPP

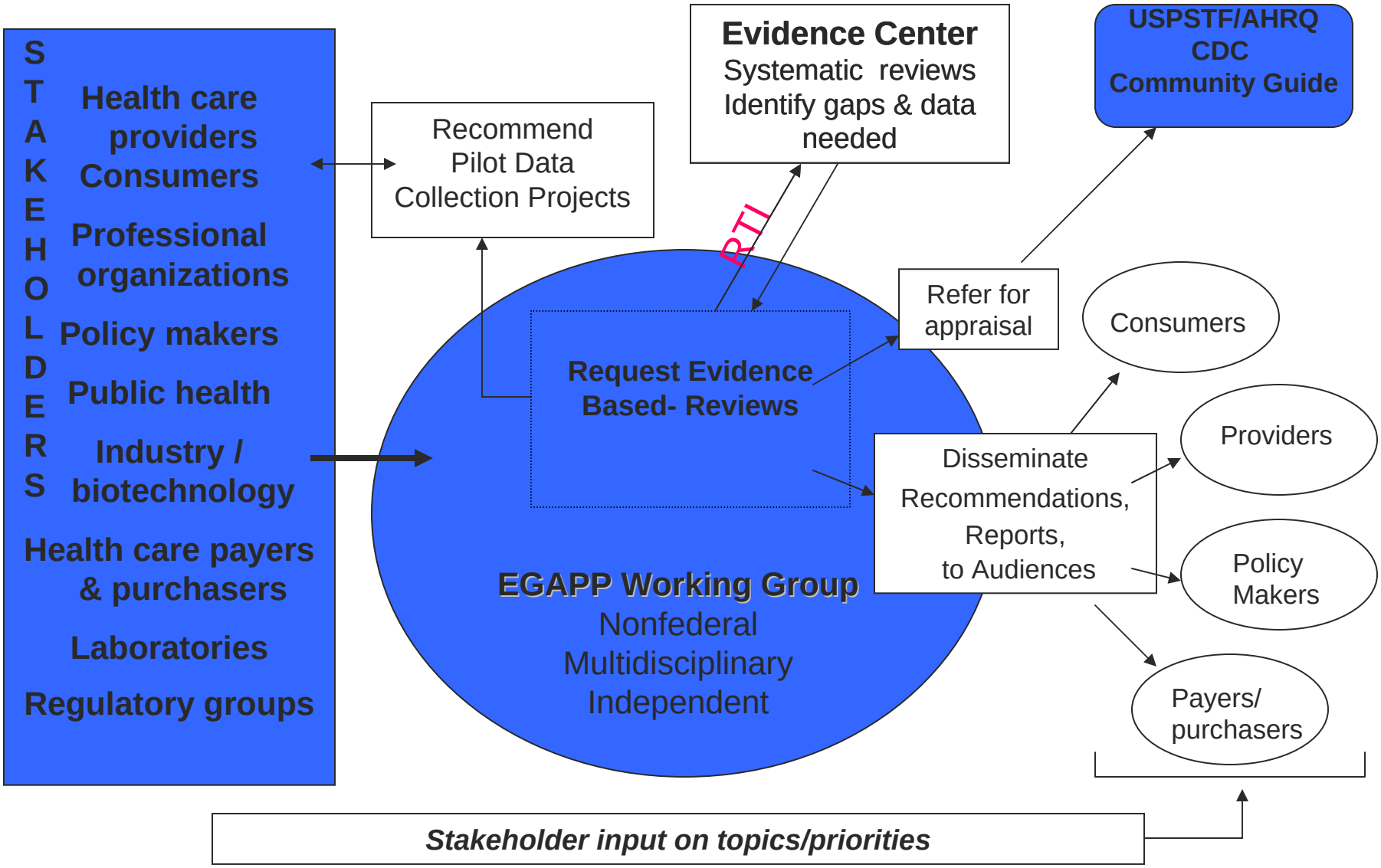


Evaluation of Genomic Applications in Practice and Prevention (EGAPP): A Three-Year Model Project

- Goal

  To establish and evaluate a sustainable systematic, evidence-based process for assessing genetic tests or other applications of genomic technology in transition from research to practice.

EGAPP Overview



EGAPP Planning Objectives

- Work to integrate
  - Previous recommendations for action
  - Knowledge gained from ACCE model project
  - Existing processes for evaluation and appraisal
  - International experience
- Create a transparent process
  - Announce and report on the process
  - Develop and publish methods
  - Provide clear linkage between evidence and conclusions/recommendations

EGAPP Planning Objectives

- Develop and disseminate information that is
  - useful to health care providers, consumers, policy makers, health care payers/purchasers for decision making
  - in appropriate and practical formats
- a key objective is to develop a sustainable process

EGAPP Activities

- ✓ *Expert Meeting on Evidence-Based Review of Genomic Applications*
  - Held January 24-25, 2005
  - 21 invited participants
    - from evidence-based medicine/HTA, health care, genomics, epidemiology, ethics, health economics
    - in US, Canada, UK
    - representing public health, academia, US Task Forces, clinical & lab practice, industry, regulation
  - considered existing and potential methods for systematic evaluation of genetic tests and other genomic applications

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EGAPP Activities

- ✓ Established the Working Group
  - Broad solicitation of nominations in February
    - Good response from individuals and professional organizations
  - Selection by inter-agency (AHRQ, CDC, CMS, FDA, HRSA, NHGRI) EGAPP Steering Committee after full review
  - Completed late March

EGAPP Working Group

Al Berg, MD, MPH - Chair
University of Washington

Katrina Armstrong, MD, MSCE
Univ of Pennsylvania School of Med

Jeffrey Botkin, MD, MPH
University of Utah

Ned Calonge, MD, MPH
Colorado Department of Public Health and Environment

James Haddow, MD
Women & Infants' Hospital, Brown University

Maxine Hayes, MD, MPH
Washington State Dept of Health

Celia Kaye, MD, PhD
Univ of Texas Health Science Center – San Antonio

Kathryn Phillips, PhD
Univ of California – San Francisco

Margaret Piper, PhD, MPH
Blue Cross/Blue Shield Association Technology Evaluation Center

Sue Richards, PhD, FACMG
Oregon Health & Science Univ.

Joan Scott, MD, CGC
Genetics & Public Policy Center

Steven Teutsch, MD, MPH
Merck & Co, Inc

http://www.cdc.gov/genomics/gtesting/egapp.htm#wgroup

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EGAPP Activities

- ✓ Established the Working Group
  - First meeting May 18-19, 2005 – Atlanta
  - Three subcommittees working on
    - Potential topics
    - Design of analytic frameworks
    - Outcomes to be considered
      - Health outcomes
      - Patient, family related outcomes
  - Second meeting July 18-19, 2005 - Atlanta

EGAPP Topic Selection

- ✓ Begin with applications recognized as common and important
  - Screening tests
  - Tests used in a clinical scenario to guide intervention (diagnostic workup, treatment, prevention including pharmacogenomic tests)
- ➢ Tests with potential public health impact
- ➢ Move focus toward prevention
- Less likely candidates
  - Newborn screening - existing processes to address
  - Single gene tests for rare disorders - CDC/NIH initiative

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EGAPP Activities

- Conduct evidence-based reviews on topics selected by the Working Group
  - Plan to select first topic at July meeting
  - Begin EBR process in August – September

EGAPP Activities

- Engage stakeholders
  - Emphasis on health care providers, consumers, policy makers, health care purchasers/payers
  - Approaches
    - ✓ Preliminary survey & research
    - ✓ Stakeholder list – developed & continue to update
    - ✓ Feedback – web & newsletter updates
      - First newsletter May 6
    - Active solicitation – years 2-3 (surveys, focus groups)
    - Partnerships

EGAPP Activities

- Conduct pilot data collection studies
  - Retrospective look at available data
- Develop and implement a comprehensive evaluation plan
  - Process
  - Products
  - Impact/value to stakeholders

EGAPP Products

- From Working Group
  - Published methods
  - Criteria and prioritized list of topics
  - Approved EBRs & conclusions/recommendations
  - Lessons learned and recommendations for a sustained process
- From project overall
  - Dissemination of Working Group products and targeted informational messages
  - Information from stakeholders on value/impact
  - Data from pilot studies
  - Lessons learned

EGAPP Overview