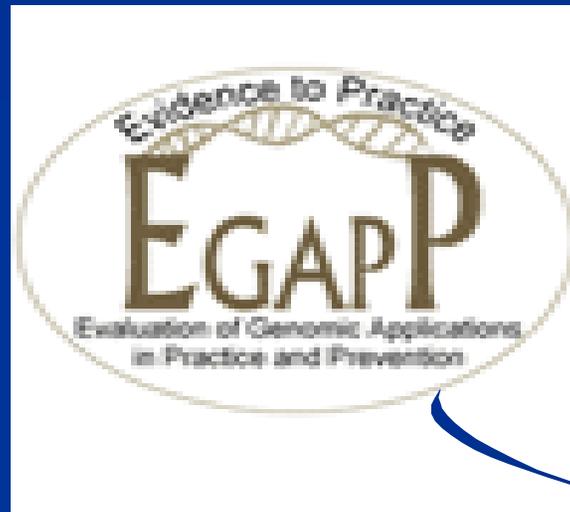


# *Evaluation of Genomic Applications in Practice and Prevention Initiative: An Update, SACGHS June 2005*



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





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article

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

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**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

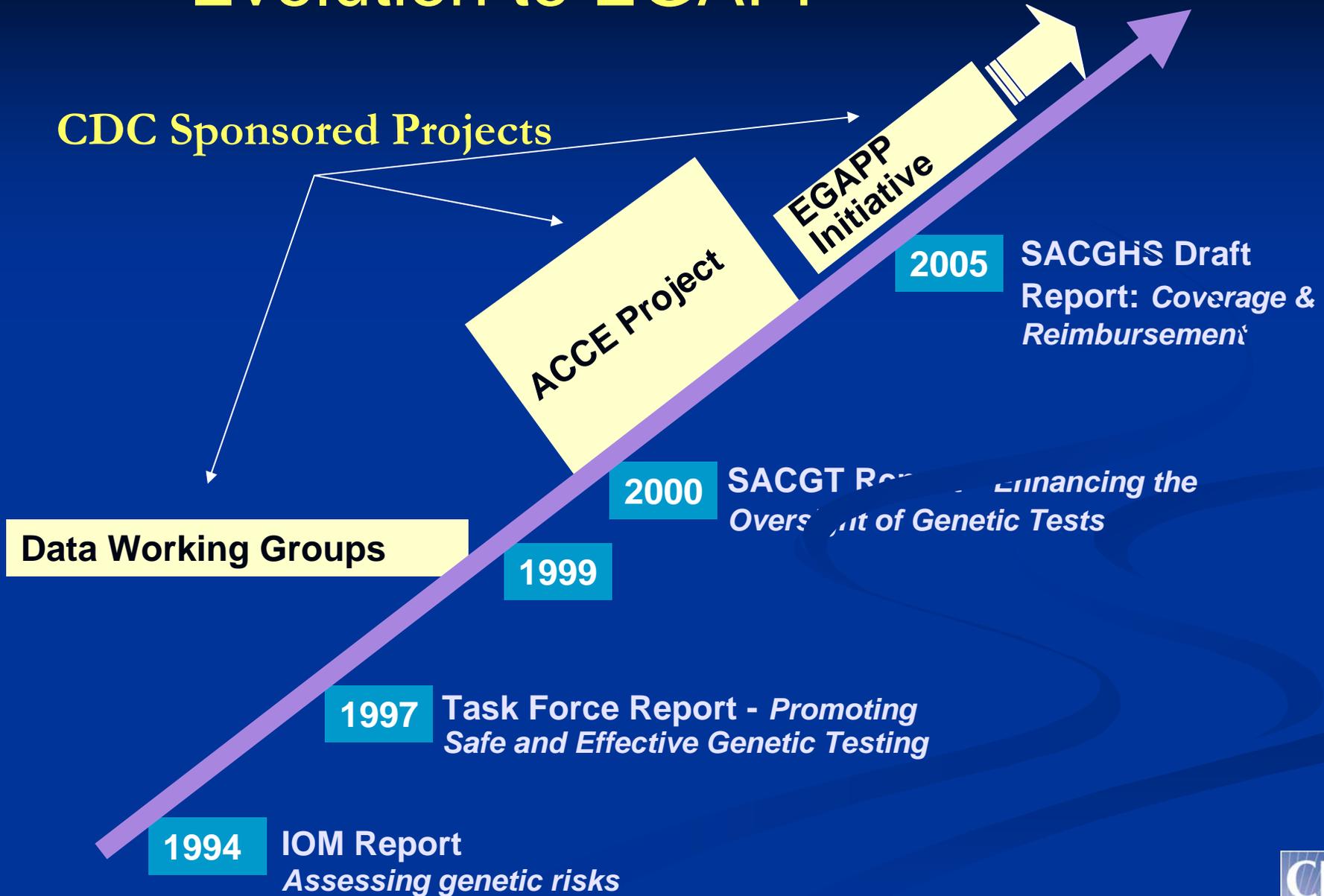
# *Public Health Impact of Genetic Tests at the End of the 20<sup>th</sup> Century*

*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

## CDC Sponsored Projects

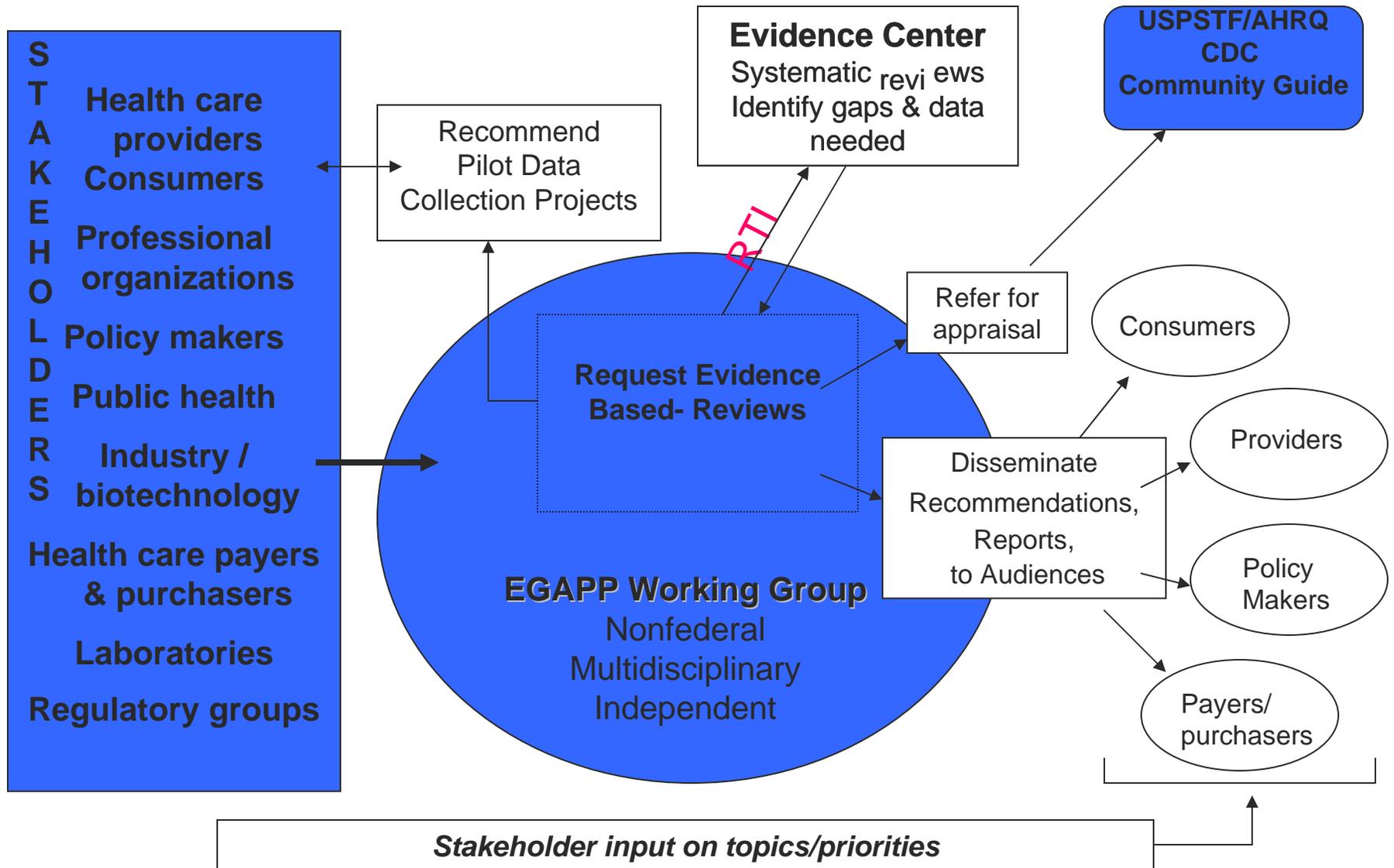


# *Evaluation of Genomic Applications in Practice and Prevention (EGAPP): A Three-Year Model Project*

- 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

# *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>



# *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

# *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

