Outcomes of an NIH-CDC Workshop on Personal Genomics (Dec. 2008)

W. Gregory Feero, M.D., Ph.D.
Senior Advisor to the Director
for Genomic Medicine
NHGRI, NIH

Personal Genomics:
Establishing the Scientific Foundation

Context:

- Personal genome-wide scans have become very inexpensive, and directly available to the public.
- Research discoveries from genome-wide association studies are being leveraged to provide consumers with interpretations of their genotypes within days of publication.

Personal Genomics:
Establishing the Scientific Foundation (cont.)

Context:

- There is vigorous debate about how (and when) to translate research discoveries from genome-wide association studies to health applications.


Analytic Validity?
Clinical Validity?
Harms?
Disparities?
Education?
Intellectual property?
Intellectual property?
Clinical Utility?
Benefits?
Informed consent?

The Spectrum of Genetic Testing

Accepted

Dubious

Rare disorders:
Huntington's disease

Prenatal screening:
Cystic fibrosis

Expression profiling:
Breast cancer

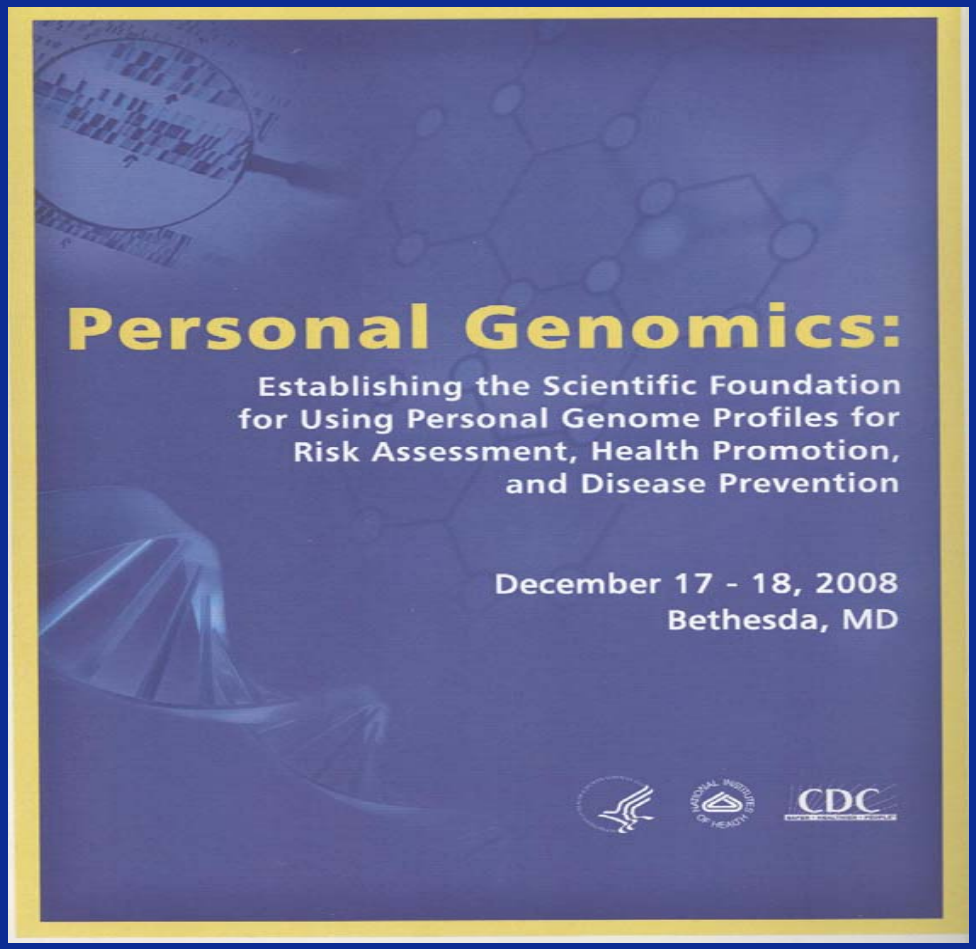
Genome scans:
Complex disease risk

Cancer syndromes:
BRCA1

Pgx:
Abacavir hypersensitivity

Pgx:
Warfarin metabolism

Treatment selection:
EGFR/breast cancer



Sponsors:

NIH – National Cancer Institute, National Heart, Lung and Blood Institute, National Human Genome Research Institute

CDC – National Office of Public Health Genomics

Personal Genomics:
Establishing the Scientific Foundation (cont.)

Meeting:

- 2 day event – approximately 100 attendees
- About 40 speakers/panelists
- Diverse perspectives – government, academia, industry
- Presentations and mediated “discussion”

Personal Genomics:
Establishing the Scientific Foundation (cont.)

Day 1

I. Genome Profiles, Risk Assessment, and Personalized Health: The Basics (Downing, HHS)

II. The Scientific Foundation For Which Genetic Variants Should be Included in Genome Profiles: The Credibility of Genetic Associations (Manolio, NHGRI)

Personal Genomics:
Establishing the Scientific Foundation (cont.)

Day 1 (con't)

III/ IV. The Scientific Foundation for Establishing Clinical Validity and Utility of Genome Profiles (Wanke, OBSSR; Greene, NCI)

Personal Genomics:
Establishing the Scientific Foundation (cont.)

Day 2

V. Case Studies and General Discussion of Clinical Validity and Utility (Lauer, NHLBI)

VI. Models for Conduction Translational Research on Genome Profiles (Feero, NHGRI)

VII. Panel Discussions and Next Steps for Research and Practice Agenda

Next Steps:

Consensus paper in preparation with key recommendations:

1. *Develop and implement industry-wide scientific standards for personal genomics*

   More later from A. Miller.

Next Steps:

2. *Develop and implement a multidisciplinary research agenda*

A variety of perspectives/expertise including genetic epi., clinical, behavioral, public health must be brought to bear to study complex research, clinical, and public health issues. Consider novel public/private partnerships (e.g. GappNet)

Next Steps:

3. *Enhance credible knowledge synthesis and dissemination of information to providers and consumers*

Public, health care providers, and policy makers require unbiased sources of information that are updated and made fully accessible (cost, literacy).

Next Steps:

4. Link scientific research on validity and utility to evidence-based recommendations for use of personal genomic tests

Public, health care providers, and policy makers need unbiased recommendations on when and how to act on proposed health applications of genomics (EGAPP).

Next Steps:

5. Consider the value of personal utility

Personal genomic information may have value to individuals beyond reducing morbidity and mortality. Objective measures are needed to incorporate into research to better understand personal utility.

Next Steps:

Agenda and slides are available at:

http://cancercontrol.cancer.gov/od/phg/workshop.html#agenda