



# Throughput of DNA Sequencing Technologies

## Capillary Array Electrophoresis

96 channels x 24 runs/day x 800 bp per run  $\approx$  1.8 Mb/day

6x coverage of 3 Gb genome takes 26 years with 1 machine

~ 3 months with 100 machines

## Sequencing by synthesis on array (projected, end of 2010)

200-300 Gb/run, ~ 7-14 days/run

30x coverage of 3 Gb genome takes ~ 0.5 run

~ 1 week with one machine ( ~ 2 genomes)

## Nanosensor

1 msec per base

10x coverage of 6 Gb genome takes

~ 2 years with single nanopore

**< 1 day with 1000 nanopore array**

[1] 2 Next »

## The \$30 Genome?

A startup is developing a new and potentially much cheaper sequencing technology based on microfluidics.

By Emily Singer

MONDAY, JUNE 07, 2010

[E-mail](#) [Audio](#) [Print](#) [Favorite](#) [Share](#) [T](#) [T](#)

LOG IN

Username

Password

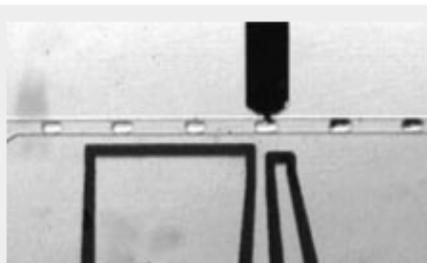
Submit



[Forgot your password?](#)

[Register »](#)

At a time when the longtime goal of a \$1,000 genome is still just out of reach, a Harvard University physicist is promising an even cheaper price--the ability to sequence a human genome for just \$30. [David Weitz](#) and his team are adapting [microfluidics](#) technology that uses tiny droplets, a strategy developed in his lab, to DNA sequencing. While the researchers have not yet sequenced DNA, they have successfully demonstrated parts of the process and formed a startup, GnuBio, to commercialize the technology. Weitz presented the findings at the Consumer Genomics Conference in Boston last week.



Drop by drop: Droplets moving

Weitz's team had previously developed a way to create picoliter droplets of water, which act as tiny test tubes. The droplets can be precisely moved around on a microfluidics chip, injected with chemicals and sorted based on color. (The technology has been commercialized by [RainDance Technologies](#), which Weitz cofounded in 2004. The company markets the droplet technology to amplify select regions of DNA.)

**Analytic Validity?**

**Clinical Validity?**

**Harms?**

**Disparities?**

**Education?**

**Intellectual property?**

**Clinical Utility?**

**Benefits?**

**Informed consent?**

# Outline

- Clinical utility... or “I know it when I see it.”
- Getting to utility.

Criteria for the evaluation of genetic tests: analytic validity, clinical validity, and clinical utility.

National Institutes of Health–  
Department of Energy Task Force on  
Genetic Testing, 1997

<http://www.genome.gov/10001733>

Clinical utility, “the balance of benefits to risks..... Before a genetic test can be generally accepted in clinical practice, data must be collected to demonstrate the benefits and risks that accrue from both positive and negative results.”

# Clinical utility continued...

## Benefits

- Decreased anxiety
- Decreased monitoring
- Life planning
- Reproductive planning
- Informing relatives
- Improved survival, cost savings, etc, etc

## Risks

- Lack of treatments
- Poor predictive ability
- Discrimination
- Psychological harm
- Physical harms, cost, etc, etc

# The Primary Care Perspective on Health Care



# Solutions – primary care style...

- **33%** of diabetics are undiagnosed --- blood glucose. (Almanac, 2008)
- **24%** of hypertensives undiagnosed --- blood pressure measurements. (Almanac, 2008)
- **37%** of high cholesterol undiagnosed --- fasting lipid panel. (Almanac, 2008)
- Universal health care, better screening programs, smoking cessation, exercise programs, improved nutrition etc...

# The Primary Care Perspective on Relative Importance of Genomics to Health Care



# Waiting for the genetic revolution

Will 2008 be the year that genomics delivers on its promises?

The sequencing of the human genome was completed in 2003. Since then we've been told that we're living in the "genomic era"—the biggest in human health since antibiotics, some say, and the beginning of scientific, personalised medicine.

In the United States we've spent about \$4bn (£2bn; 2.8bn) since 2000 to fund the National Human Genome Research Institute, so it seems fair to ask what we've got for our money.

Certainly there have been improvements in the clinic. DNA sequencing and other technologies. Polymerase chain reaction and other amplified techniques have made what was tedious and painstaking commonplace. Linkage analysis, that geneticist's bread and butter, and genomics can be found in the surgery. Human papilloma virus testing, rapid tests for some infectious diseases by polymerase chain reaction, HIV analyses, and diagnostic laboratory tests have taken their way into general practice.

Genomic tools have been used to develop some drugs that specialists use, and more are being evaluated all the time. But most that I've heard of are the provincial oncologists

What about the common, everyday diagnoses—heart disease, diabetes, and other "lifestyle diseases"? Hope

if you're a doctor, you'll be able to

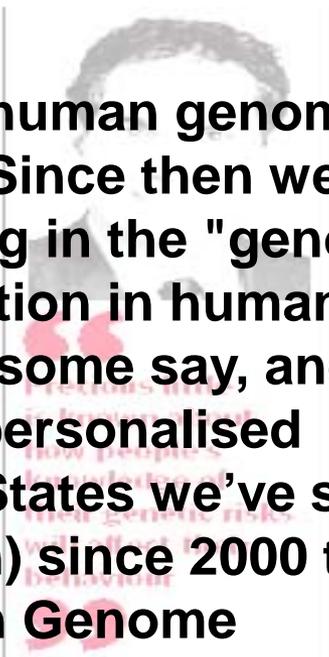
It's wrong to say that we're living in the "genomic era" with heraldic letters for testing. You're going to have a

family history of heart disease, you're going to have a

genetic test, how do you know that you're going to have a

genetic test, how do you know that you're going to have a

These "personal genomic services" allow you to "unlock the secrets of your own DNA." They can tell you your risk of developing a



behaviours. And even less is known about how people's knowledge of their genetic risks will affect them. The US Centers for Disease Control and Prevention convened a panel of experts in 2004 to assess genetic tests and technologies for their appropriateness in practice. It was a couple of years of work setting up a systematic, evidence based process they have just issued in their first recommendation. They recommend pharmacogenomic testing for cyclosporine P450 in depressed patients to predict how well selective serotonin reuptake inhibitors will work. Their conclusion: the evidence is insufficient to assess the value of such testing in this situation (Genetics in Medicine 9:125).

And what about all the legal and ethical challenges involved in genetic testing, especially the broad genetic testing? It's probably that an accident that has even now websites steer clear of conventional medical care. What will happen if for whatever reason companies get hold of our genetic profiles? Legislation that would prohibit discrimination on the basis of genetic risks has been pending at the US Congress for a number of years but never seems to pass. It is no surprise that the US National Human Genome Research Institute has

# Evidence-Based Medicine

"the conscientious, explicit and judicious use of current best evidence in making decisions about the care of the individual patient. It means integrating individual clinical expertise with the best available external clinical evidence from systematic research." Sackett D, 1996

# SORT Criteria AFP, June 2004

<i>Strength of recommendation</i>	<i>Definition</i>
A	Recommendation based on consistent and good-quality patient-oriented evidence.*
B	Recommendation based on inconsistent or limited-quality patient-oriented evidence.*
C	Recommendation based on consensus, usual practice, opinion, disease-oriented evidence,* or case series for studies of diagnosis, treatment, prevention, or screening.

Use the following table to determine whether a study measuring patient-oriented outcomes is of good or limited quality, and whether the results are consistent or inconsistent between studies.

<i>Study quality</i>	<i>Diagnosis</i>	<i>Treatment/prevention/screening</i>	<i>Prognosis</i>
Level 1—good-quality patient-oriented evidence	Validated clinical decision rule SR/meta-analysis of high-quality studies High-quality diagnostic cohort study†	SR/meta-analysis of RCTs with consistent findings High-quality individual RCT‡ All-or-none study§	SR/meta-analysis of good-quality cohort studies Prospective cohort study with good follow-up
Level 2—limited-quality patient-oriented evidence	Unvalidated clinical decision rule SR/meta-analysis of lower-quality studies or studies with inconsistent findings Lower-quality diagnostic cohort study or diagnostic case-control study§	SR/meta-analysis of lower-quality clinical trials or of studies with inconsistent findings Lower-quality clinical trial‡ Cohort study Case-control study	SR/meta-analysis of lower-quality cohort studies or with inconsistent results Retrospective cohort study or prospective cohort study with poor follow-up Case-control study Case series
Level 3—other evidence	Consensus guidelines, extrapolations from bench research, usual practice, opinion, disease-oriented evidence (intermediate or physiologic outcomes only), or case series for studies of diagnosis, treatment, prevention, or screening		

## *Consistency across studies*

Consistent	Most studies found similar or at least coherent conclusions (coherence means that differences are explainable) or If high-quality and up-to-date systematic reviews or meta-analyses exist, they support the recommendation
Inconsistent	Considerable variation among study findings and lack of coherence or If high-quality and up-to-date systematic reviews or meta-analyses exist, they do not find consistent evidence in favor of the recommendation

# Patient-Oriented Outcomes

These are outcomes that matter to patients and help them live longer or better lives, including reduced morbidity, reduced mortality, symptom improvement, improved quality of life, or lower cost.

AFP, February 2004

EBM

---

Genomics

# Morbidity and mortality

---

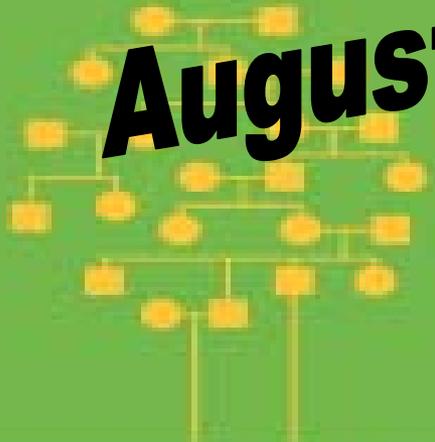
Surrogate markers

Effectiveness

---

Efficacy

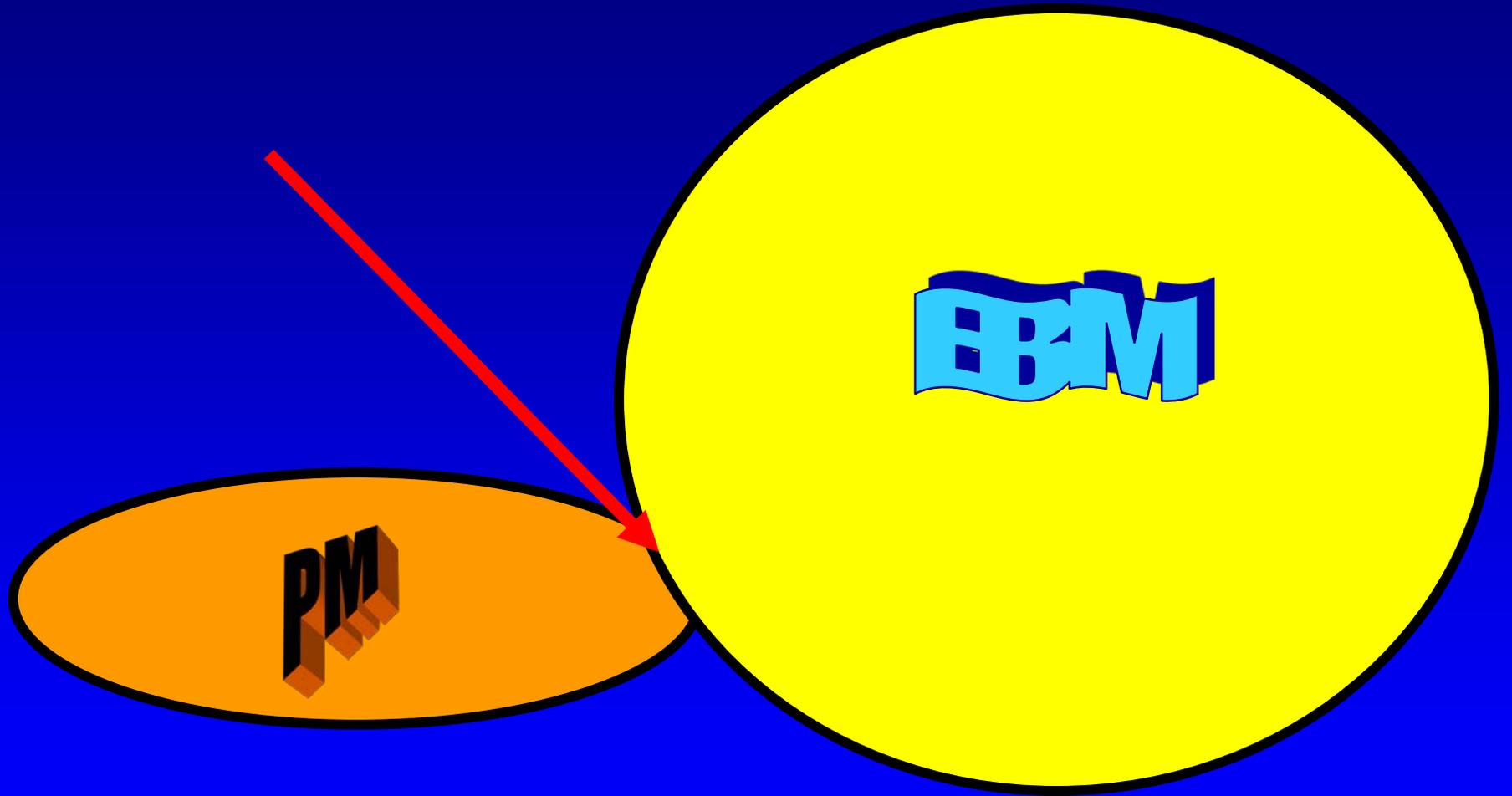
MINI STATE OF THE SCIENCE I CONFERENCE



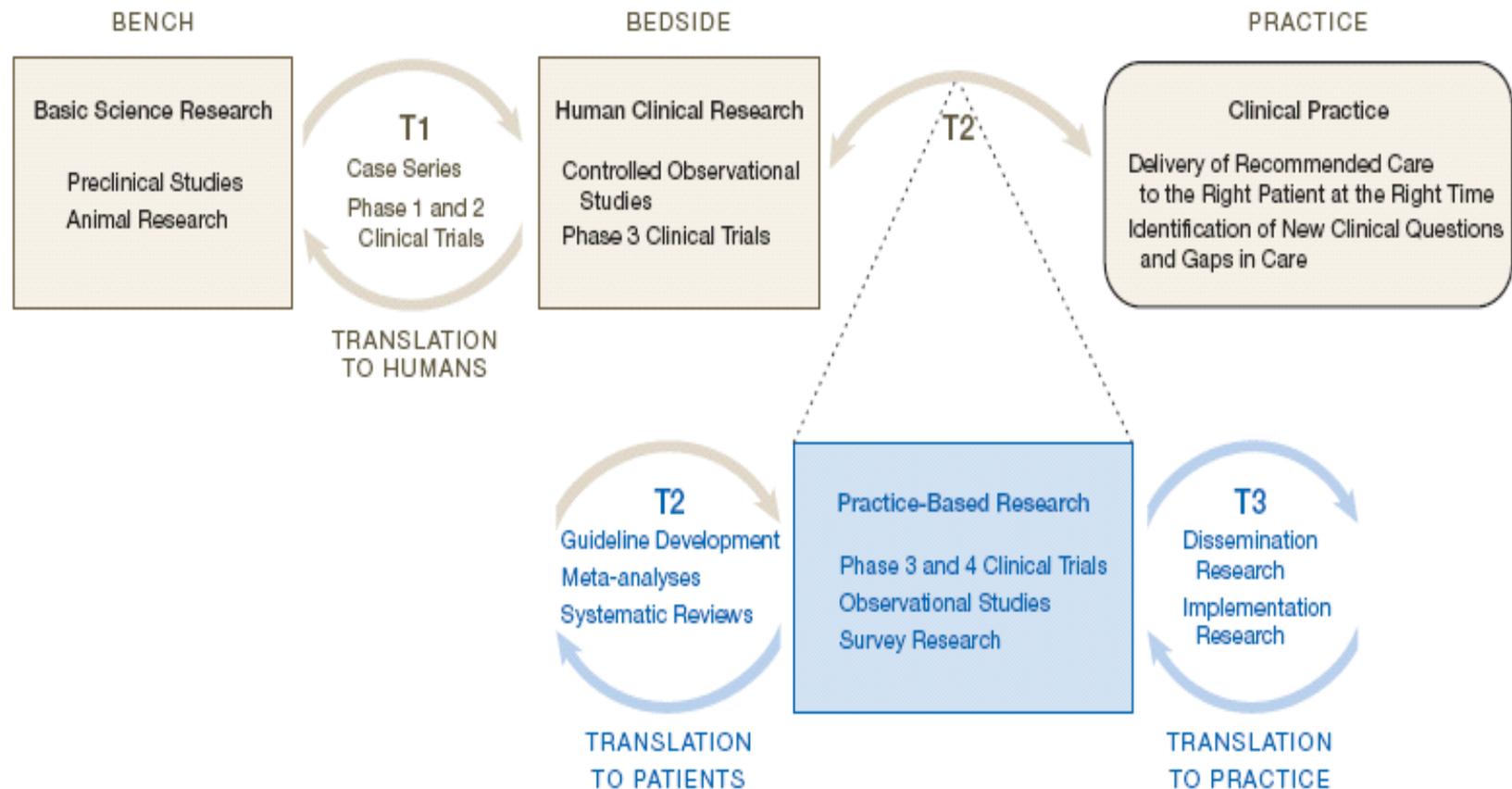
**August 24-26, 2009**

FAMILY HISTORY

and Improving Health



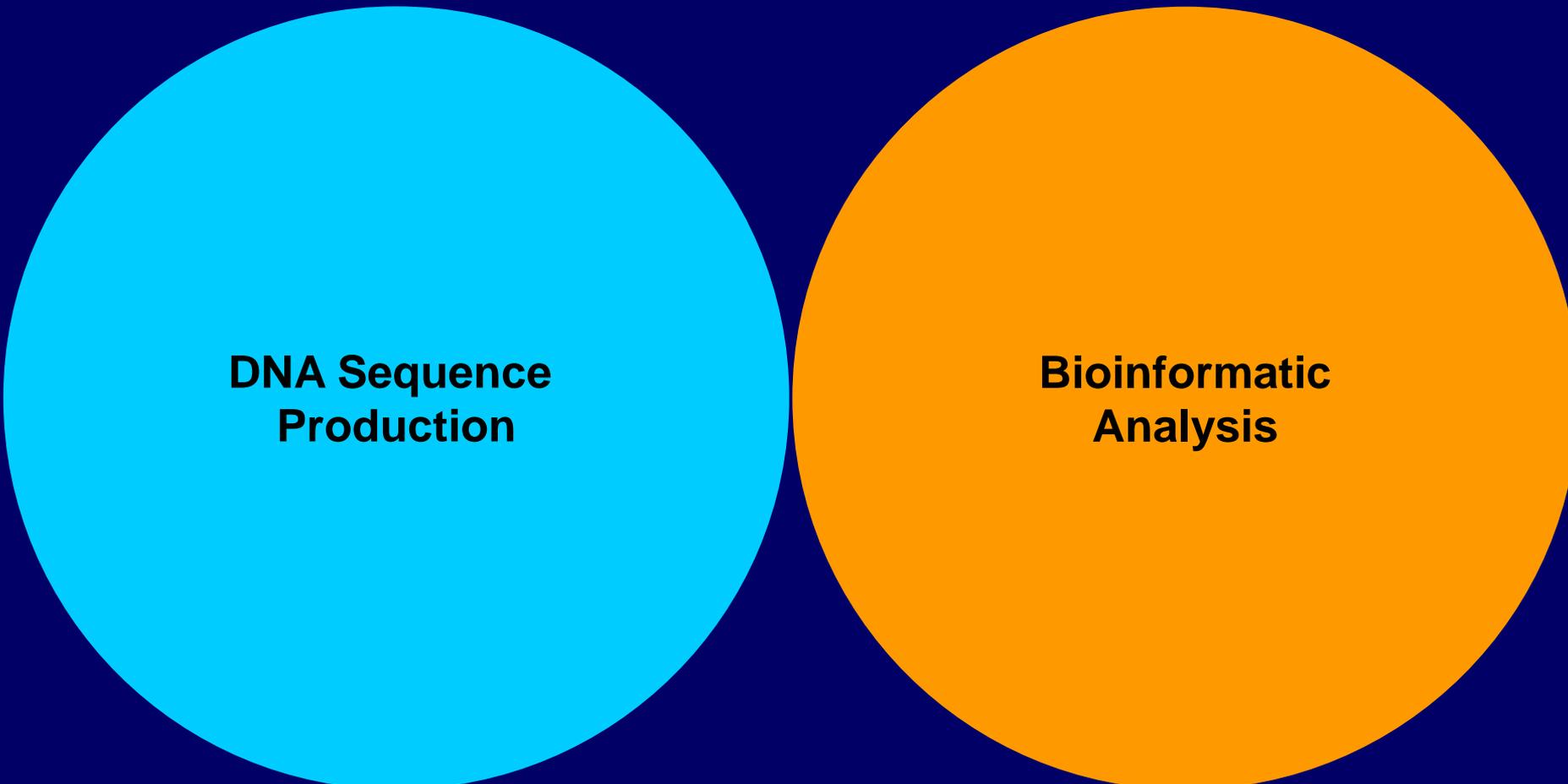
**Figure.** "Blue Highways" on the NIH Roadmap



*JM Westfall et al JAMA 2007;297:403.*

Getting to patient-oriented  
outcomes...

# Changing Infrastructure Requirements



**DNA Sequence  
Production**

**Bioinformatic  
Analysis**

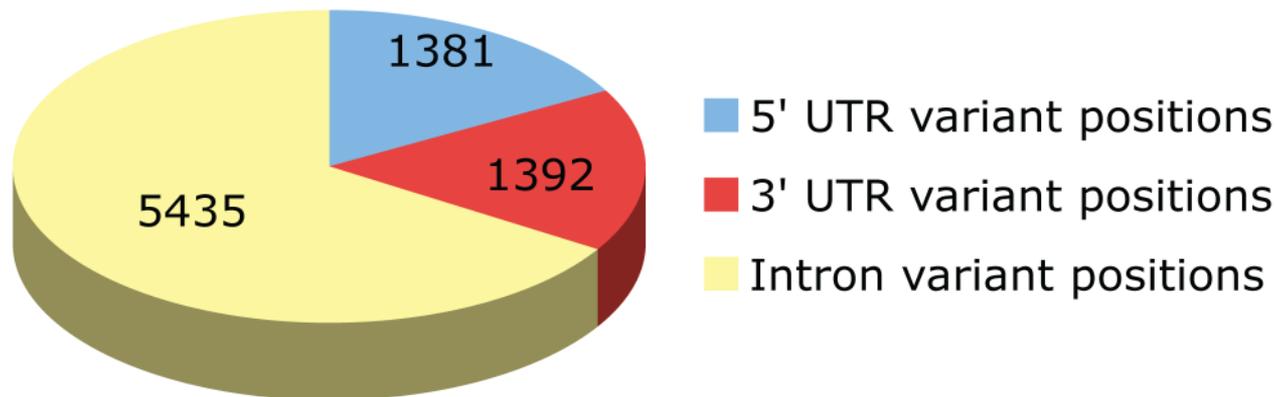


NHGRI Intramural Program  
Les Biesecker, PI

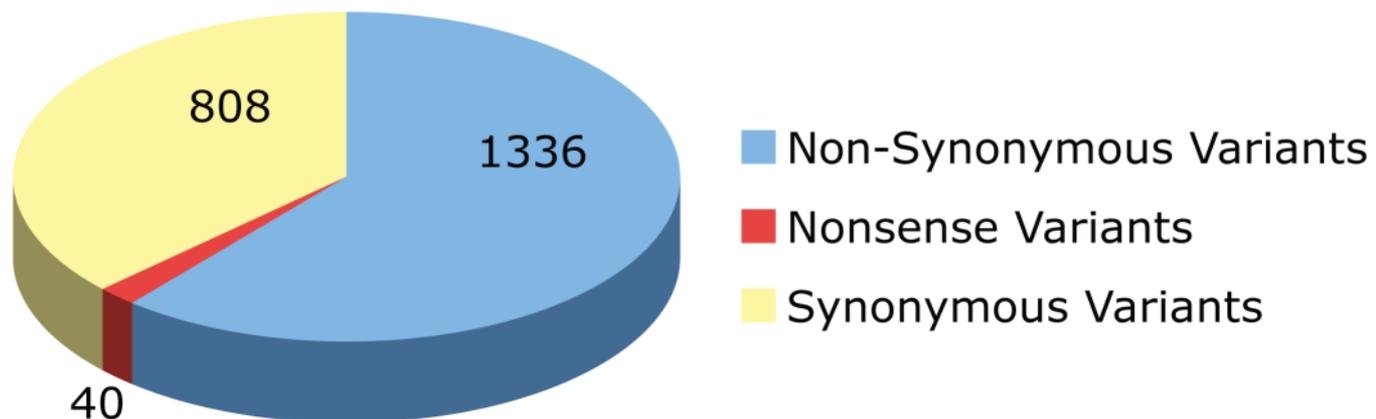
# Approach

- **Initial target phenotype: Atherosclerosis**
  - **Phenotype 1,000 subjects**
    - **Coronary calcium, lipidemia, & related indices**
  - **Sequence 200-400 candidate genes**
  - **Follow-up studies**
    - **Bioinformatic & bench**
  - **Interpret variants and validate *some***
  - **Return results**

## 8,208 Non-exon novel variants



## 2,185 Exonic novel variants



Data storage capacity?

“We currently have about 3,000 cores in our computational cluster and over 3 PB (3,000,000 GB) of storage online. When full of equipment in a few years, the data center will likely house tens of thousands of cores and on the order of 100 PB of storage.”

Oct., 2009

<http://news.wustl.edu/news/Pages/20393.aspx>



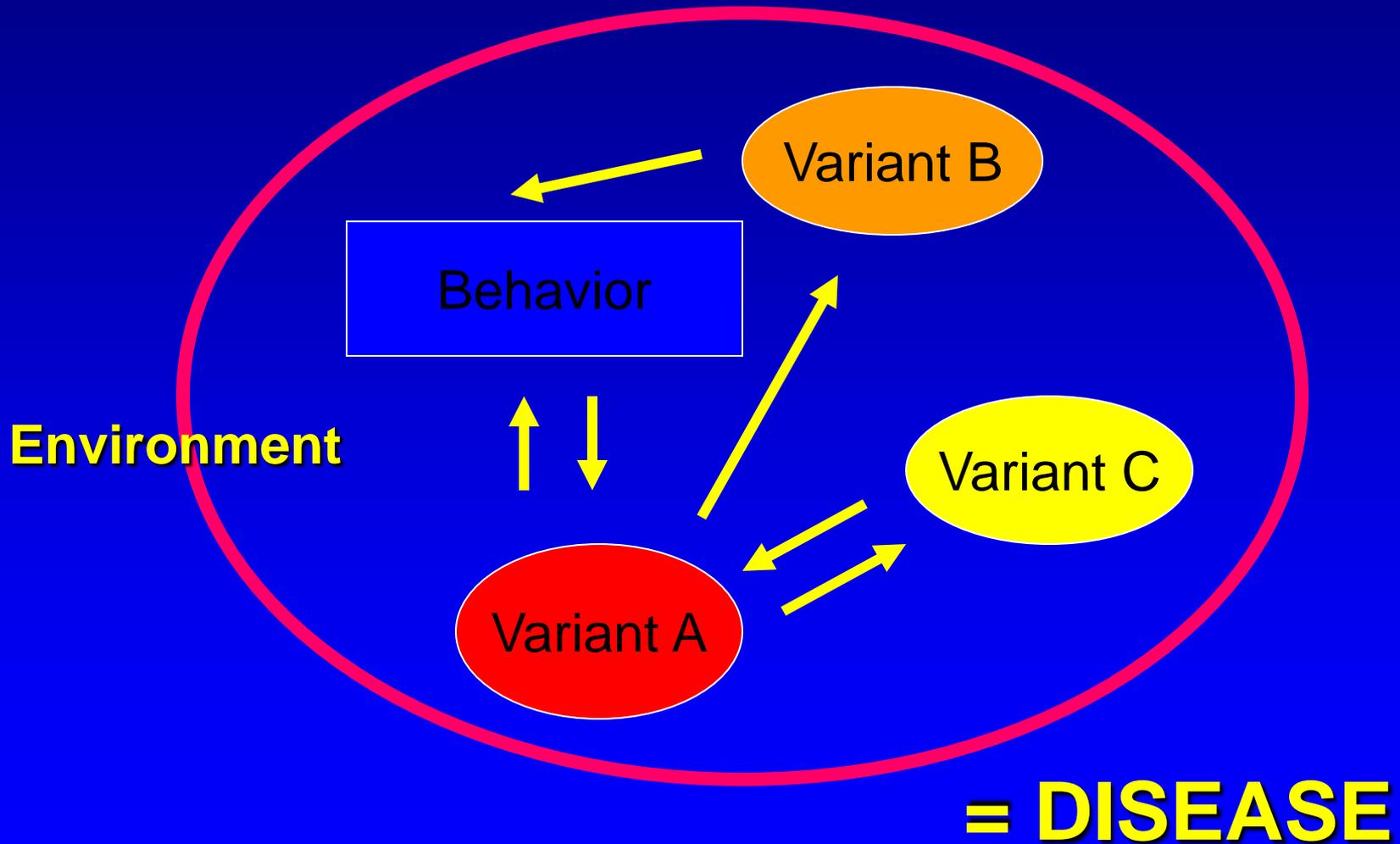
<http://www.flickr.com/photos/ddgenome/2166503922/in/set-72157603633991423/>

Sequencing accuracy?

A sequencing accuracy of 99.9999% will incur about 6,000 errors in a diploid human genome.



# Common complex disease:



What  
condition/exposure?



National Human  
Genome Research  
Institute



National  
Institutes of  
Health



U.S. Department  
of Health and  
Human Services

# **GENEVA, A Collaborative Network for Genome-Wide Association Studies**

**U.S. Department of Health and Human Services  
National Institutes of Health  
National Human Genome Research Institute**

**Teri A. Manolio, M.D., Ph.D.  
Director, Office of Population Genomics  
Senior Advisor to the Director, NHGRI,  
for Population Genomics**

**March 10, 2010**

# Phenotype Harmonization across GENEVA

GENE.ENVIRONMENT  
ASSOCIATION.STUDIES **GENEVA**

Search:

■ GENEVA Home

■ GENEVA Search

■ Study Overview

■ Calendar

■ Data Sets

■ Directory

■ Documents

■ IRB Documents

■ Meetings

■ Publications and Presentations

■ Statement on Incidental Findings

■ Subcommittees

## Phenotype Harmonization Committee Documents

■ [Members List](#)

■ [Summary listing by category of Ph II dbGaP variables](#)

■ [Template for working group cross-study analyses](#)

■ [Web-form Summary Phase II investigators](#)

■ [Individual responses to Phenotype Web Form](#)

■ [Smoking Variables Across Studies](#)

■ [Basic Phenotype Information by Study, October 2007](#)

## Call Summaries

■ [February 10, 2010](#)

■ [January 13, 2010](#)

# Phenotype Harmonization Summary Spreadsheet

	A	B	C	D	E	F	G	H	I	J	K	L	M	N
1		STUDY/SITE												
2	GEI SMOKING VARIABLES	t i o n	N H S	H P S	e a s e	U t a h	A s i a	k i n s	i g h t	P L C O	A T B C	A G L E	i r t h	U C S D
3	Current Smoker	X	V	V	V	X	X	X	V	V	X	V	X	V
4	Ever Smoker	V		X	V					V		V	X	V
5	100 cigs lifetime	V		X		X	X	X				X	X	
6	Daily smoking for month or more	V												
7	Cig years of smoking (ave. # day x years)	X			V									
8	Ave. # cigs smoked per day	X	X	X	X	X	X	X	X <sup>A</sup>	X	X	X	X	
9	Onset tobacco use	V										X		
10	Onset regular smoking	X			X	X	X	X		X	X		X	
11	Recency tobacco use	V		X	X	X	X	X		X <sup>B</sup>	X		X	
12	FTND	V										V		
13	Max cigs smoked in 24 hrs	V												
14	DSM-IV	V												
15	Pack Years									V	V	V		
16	Years smoking										X	X		
17	<sup>A</sup> during pregnancy only; <sup>B</sup> recency of regular smoking; <sup>C</sup> current smokers only													
18														
19	KEY: V=included on variable list; X=available from questionnaire(s)													

# The PhenX Project

- PI: Carol Hamilton, PhD (RTI International)
- **Goal:** Provide a resource of standard measures that can be incorporated into study protocols
- Selecting 15 high-priority measures for each of 21 research domains
- Measures made available to research community via the PhenX Toolkit

# Example Measure: Nicotine Dependence

Home Browse Search My Account Resources Help About

Review Measure

Top » Alcohol, Tobacco and Other Substances » Tobacco - Nicotine Dependence

◀ **MEASURE:** Tobacco - Nicotine Dependence #031000

**Definition:** Questions query respondent on whether s/he has symptoms of nicotine dependence.

**Purpose:** This measure can be used to assess the participant's dependence on nicotine.

**Essential Data:**  [Current Age, Tobacco - Smoking Status](#)

**Keywords:** tobacco, smoking, cigarette, nicotine dependence, nicotine, pack years, Fagerstrom Test for Nicotine Dependence, Fagerstrom Tolerance Questionnaire, Fagerstrom

 [Add this measure\\* to the cart](#)

 [Add this measure\\* + all essential data to the cart](#)

\* Adding this measure to the cart will add all protocols associated with this measure (listed below)

 **Protocols Associated with Measure**

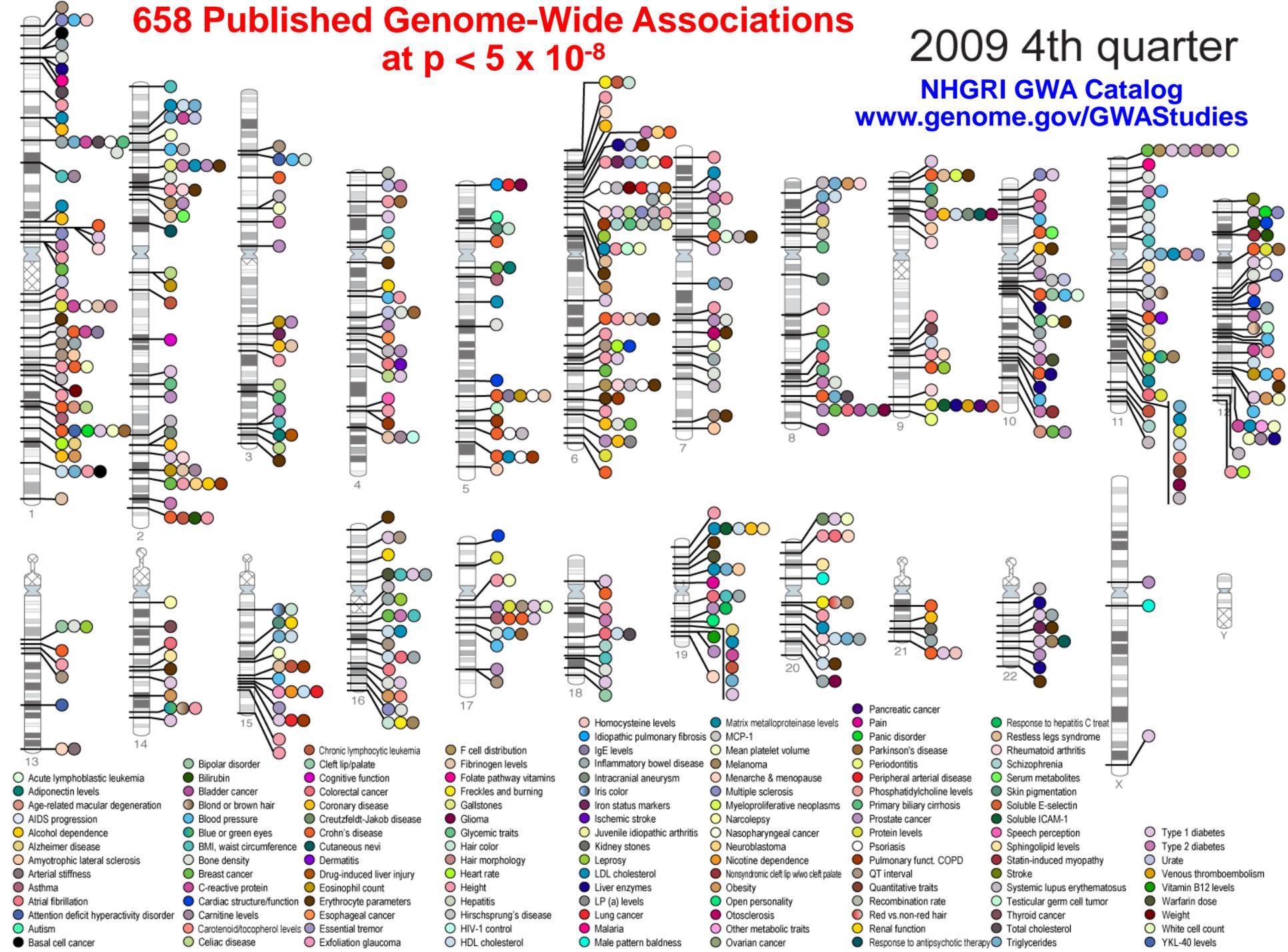
[Add to Cart](#) #031001 [Tobacco - Nicotine Dependence](#) »

What variant?

# 658 Published Genome-Wide Associations at $p < 5 \times 10^{-8}$

2009 4th quarter

NHGRI GWA Catalog  
[www.genome.gov/GWAStudies](http://www.genome.gov/GWAStudies)



- |  |                                |                                |                              |                                 |                                   |                              |                                 |
|--|--------------------------------|--------------------------------|------------------------------|---------------------------------|-----------------------------------|------------------------------|---------------------------------|
| ● Acute lymphoblastic leukemia             | ● Bipolar disorder             | ● Chronic lymphocytic leukemia | ● F cell distribution        | ● Homocysteine levels           | ● Matrix metalloproteinase levels | ● Pancreatic cancer          | ● Response to hepatitis C treat |
| ● Adiponectin levels                       | ● Bilirubin                    | ● Cleft lip/palate             | ● Fibrinogen levels          | ● Idiopathic pulmonary fibrosis | ● MCP-1                           | ● Pain                       | ● Restless legs syndrome        |
| ● Age-related macular degeneration         | ● Bladder cancer               | ● Cognitive function           | ● Inflammatory bowel disease | ● IgE levels                    | ● Mean platelet volume            | ● Panic disorder             | ● Rheumatoid arthritis          |
| ● AIDS progression                         | ● Blood pressure               | ● Colorectal cancer            | ● Intracranial aneurysm      | ● Iris color                    | ● Menarche & menopause            | ● Parkinson's disease        | ● Schizophrenia                 |
| ● Alcohol dependence                       | ● Blue or green eyes           | ● Coronary disease             | ● Iron status markers        | ● Juvenile idiopathic arthritis | ● Multiple sclerosis              | ● Periodontitis              | ● Serum metabolites             |
| ● Alzheimer disease                        | ● BMI, waist circumference     | ● Creutzfeldt-Jakob disease    | ● Kidney stones              | ● LDL cholesterol               | ● Myeloproliferative neoplasms    | ● Phosphatidylcholine levels | ● Skin pigmentation             |
| ● Amyotrophic lateral sclerosis            | ● Bone density                 | ● Crohn's disease              | ● Leprosy                    | ● Lip (a) levels                | ● Nasopharyngeal cancer           | ● Primary biliary cirrhosis  | ● Soluble E-selectin            |
| ● Arterial stiffness                       | ● Breast cancer                | ● Cutaneous nevi               | ● Menstrual cycle            | ● Lung cancer                   | ● Neuroblastoma                   | ● Prostate cancer            | ● Soluble ICAM-1                |
| ● Asthma                                   | ● C-reactive protein           | ● Dermatitis                   | ● Nicotine dependence        | ● Malaria                       | ● Narcoclepsy                     | ● Psoriasis                  | ● Speech perception             |
| ● Atrial fibrillation                      | ● Cardiac structure/function   | ● Drug-induced liver injury    | ● Obesity                    | ● Male pattern baldness         | ● Nasopharyngeal cancer           | ● QT interval                | ● Sphingolipid levels           |
| ● Attention deficit hyperactivity disorder | ● Carnitine levels             | ● Eosinophil count             | ● Open personality           | ● MCP-1                         | ● Neuroblastoma                   | ● Pulmonary funct. COPD      | ● Stroke                        |
| ● Autism                                   | ● Carotenoid/tocopherol levels | ● Erythrocyte parameters       | ● Other metabolic traits     | ● Nasopharyngeal cancer         | ● Neuroblastoma                   | ● Recombination rate         | ● Systemic lupus erythematosus  |
| ● Basal cell cancer                        | ● Celiac disease               | ● Essential tremor             | ● Other metabolic traits     | ● Nasopharyngeal cancer         | ● Neuroblastoma                   | ● Red vs. non-red hair       | ● Testicular germ cell tumor    |
|  |                                | ● Exfoliation glaucoma         | ● Other metabolic traits     | ● Nasopharyngeal cancer         | ● Neuroblastoma                   | ● Renal function             | ● Thyroid cancer                |
|  |                                |                                | ● Other metabolic traits     | ● Nasopharyngeal cancer         | ● Neuroblastoma                   | ● Total cholesterol          | ● Total cholesterol             |
|  |                                |                                | ● Other metabolic traits     | ● Nasopharyngeal cancer         | ● Neuroblastoma                   | ● Triglycerides              | ● Type 1 diabetes               |
|  |                                |                                | ● Other metabolic traits     | ● Nasopharyngeal cancer         | ● Neuroblastoma                   |                              | ● Type 2 diabetes               |
|  |                                |                                | ● Other metabolic traits     | ● Nasopharyngeal cancer         | ● Neuroblastoma                   |                              | ● Urate                         |
|  |                                |                                | ● Other metabolic traits     | ● Nasopharyngeal cancer         | ● Neuroblastoma                   |                              | ● Venous thromboembolism        |
|  |                                |                                | ● Other metabolic traits     | ● Nasopharyngeal cancer         | ● Neuroblastoma                   |                              | ● Vitamin B12 levels            |
|  |                                |                                | ● Other metabolic traits     | ● Nasopharyngeal cancer         | ● Neuroblastoma                   |                              | ● Warfarin dose                 |
|  |                                |                                | ● Other metabolic traits     | ● Nasopharyngeal cancer         | ● Neuroblastoma                   |                              | ● Weight                        |
|  |                                |                                | ● Other metabolic traits     | ● Nasopharyngeal cancer         | ● Neuroblastoma                   |                              | ● White cell count              |
|  |                                |                                | ● Other metabolic traits     | ● Nasopharyngeal cancer         | ● Neuroblastoma                   |                              | ● YKL-40 levels                 |

Nature June 9, 2010

LETTERS

---

# Functional impact of global rare copy number variation in autism spectrum disorders

A list of authors and their affiliations appears at the end of the paper.

The autism spectrum disorders (ASDs) are a group of conditions characterized by impairments in reciprocal social interaction and communication, and the presence of restricted and repetitive behaviours<sup>1</sup>. Individuals with an ASD vary greatly in cognitive development, which can range from above average to intellectual disability<sup>2</sup>. Although ASDs are known to be highly heritable (~90%)<sup>3</sup>, the underlying genetic determinants are still largely unknown. Here we analysed the genome-wide characteristics of rare (<1% frequency) copy number variation in ASD using dense genotyping arrays. When comparing 996 ASD individuals of European ancestry to 1,287 matched controls, cases were found to carry a

called by both algorithms in an individual (Fig. 1, Supplementary Tables 1–3 and Supplementary Fig. 3). This stringent data set of 5,478 rare CNVs in 996 cases and 1,287 controls of European ancestry (Supplementary Table 4) had the following characteristics: (1) CNV present at <1% frequency in the total sample (cases and controls); (2) CNV  $\geq 30$  kb in size (because >95% of these could be confirmed); and (3) all CNVs further verified using combined evidence from the PennCNV algorithm<sup>18</sup> and child–parent intensity fold changes, genotype proportions (to verify deletions) and visual inspection (for chromosome X).

We assessed the impact of rare CNV in cases compared to controls

Environment?

# Genes, Environment, and Health Initiative (GEI)

- Proposed in President's budget for FY07
- Aims to accelerate understanding of genetic and environmental contributions to health and disease
- Led by NIH-wide Coordinating Committee
- Two components:
  - Genetic analyses of case-control studies of common disease (\$26M per year for four years)
  - Development of innovative technologies to measure environmental exposures, diet, and physical activity (\$14M per year for four years)

# Gene–environment interactions in 7610 women with breast cancer: prospective evidence from the Million Women Study



Ruth C Travis, Gillian K Reeves, Jane Green, Diana Bull, Sarah J Tipper, Krys Baker, Valerie Beral, Richard Peto, John Bell, Diana Zelenika, Mark Lathrop, for the Million Women Study Collaborators\*

## Summary

**Background** Information is scarce about the combined effects on breast cancer incidence of low-penetrance genetic susceptibility polymorphisms and environmental factors (reproductive, behavioural, and anthropometric risk factors for breast cancer). To test for evidence of gene–environment interactions, we compared genotypic relative risks for breast cancer across the other risk factors in a large UK prospective study.

**Methods** We tested gene–environment interactions in 7610 women who developed breast cancer and 10 196 controls without the disease, studying the effects of 12 polymorphisms (*FGFR2*-rs2981582, *TNRC9*-rs3803662, 2q35-rs13387042, *MAP3K1*-rs889312, 8q24-rs13281615, 2p-rs4666451, 5p12-rs981782, *CASP8*-rs1045485, *LSP1*-rs3817198, 5q-rs30099, *TGFB1*-rs1982073, and *ATM*-rs1800054) in relation to prospectively collected information about ten established environmental risk factors (age at menarche, parity, age at first birth, breastfeeding, menopausal status, age at menopause, use of hormone replacement therapy, body mass index,

Published Online

June 2, 2010

DOI:10.1016/S0140-6736(10)60636-8

See Online/Comment

DOI:10.1016/S0140-6736(10)60876-8

\*Collaborators listed at end of paper

Cancer Epidemiology Unit

(R C Travis DPhil, G K Reeves PhD, J Green MD, D Bull,



Back to: [Common Fund Home](#) > [Programs](#)

## New Models for Large Prospective Studies

▶ [Overview](#)

▶ [Planning Group Members](#)

▶ [Meetings](#)

### OVERVIEW

Large prospective cohorts and biobanks are ideal models for defining disease burden in a population and for launching studies to examine the many genetic and environmental factors that contribute to disease, paving the way for personalized medicine. Advantages of large-scale approaches over smaller scale or retrospective study designs include greater generalizability of the research findings and efficiencies in time and resources because a single large, well defined cohort can be built to address multiple research questions within a single research framework.

The U.K. Biobank is a large-scale national resource initiated in 2004 to assess trends in disease burden and examine genetic and environmental risk factors for specific diseases. The approach used by the Biobank has enabled its leaders to achieve exceptional efficiencies in recruitment, assessment and record linkage. By spring of 2010, the UK Biobank will have reached its goal of 500,000 participants and begin closing down its field centers. To explore this model while it was still actively in place, the NIH convened a symposium to hear first hand from the leadership of the U.K. Biobank, and from leaders of other large national and international cohort studies and biobanks, about novel study designs, lessons learned, and opportunities that may inform NIH programs.

The NIH hosted the *New Models for Large Prospective Studies* symposium on January 22, 2010 in Bethesda, Maryland to:

- Hear from national and international leaders and funders of large cohort studies and biobanks, in particular the UK Biobank, about novel study designs; approaches to achieve efficiencies in recruitment, examination, and sample collection and handling; strategies to address consent and confidentiality; and lessons learned from pilot studies and full-scale implementations; and
- Identify areas of opportunity and collaboration, and identify possible next steps.

[View the meeting agenda and list of participants...](#)

It is difficult to envision how WGS data will be sorted out and used without a concerted effort to develop the infrastructure needed to follow and study large numbers of individuals longitudinally using electronic health records.

# The eMERGE Network

## electronic Medical Records & Genomics

*A consortium of biorepositories linked to electronic medical records data for conducting genomic studies*

### Main Menu

[Home](#)

[About eMERGE Network](#)

[GWAS Project](#)

[Network Publications](#)

[Calendar](#)

[Links](#)

[Contact Us](#)

### User Menu

[Log in](#)

## The eMERGE Network

The eMERGE Network is a national consortium formed to develop, disseminate, and apply approaches to research that combine DNA biorepositories with electronic medical record (EMR) systems for large-scale, high-throughput genetic research.

The mapping of the human genome has enabled new exploration of how genetic variations contribute to health and disease. To better realize this promise, researchers must now determine ways in which genetic make-up gives some individuals a greater chance of becoming sick with chronic conditions such as diabetes, Alzheimer's, or heart disease, in order to ultimately improve patient care.

There are a number of studies conducted routinely to uncover the association between disease and a person's genetic make-up, but they are typically costly and take a long time to complete. This consortium will use data from the EMR – clinical systems that represent actual health care events, an alternative methodology, which is highly cost and time-efficient, to propel this research. Electronic medical records are one of the most exciting potential resources for research data.

Each center participating in the consortium, organized by the National Human Genome Research Institute with additional funding from the National Institute of General Medical Sciences, has proposed studying the relationship between genome-wide genetic variation and a common human trait. (More detailed information about the

[www.gwas.net](http://www.gwas.net)



Will society act on the  
information in  
productive ways?

Embargoed for Release  
Monday, June 7, 2010  
3 p.m. EDT

Contact:  
[Raymond MacDougall](#)  
301-402-0911

## NIH Researchers Explore How Healthy, Young Adults View the Role Genetics Plays in Improving Health

Most healthy young adults place greater emphasis on health habits than on genetic risk factors when considering what causes common diseases, a research team from the National Human Genome Research Institute (NHGRI) and the Henry Ford Health System in Detroit has found. The study, based on a survey of 25- to-45-year-olds, was released June 8, 2010 in an early online edition of the *Annals of Behavioral Medicine*.

The research is part of the Multiplex Initiative, a large, population-based study of how healthy young people use genetic risk-susceptibility tests. Multiplex genetic testing, for which a single blood sample per individual is used to detect multiple genetic results, provides individuals with a slightly higher or lower comparative risk value for certain conditions compared to the general population.

"Genetic information is quickly becoming an important part of the equation for making individual health choices," said NHGRI Director Eric Green, M.D., Ph.D. "It is important that we understand how genetic test recipients react and use such information so that the public, clinicians and public health experts have realistic expectations of how genetic risk awareness may affect the health information landscape."

Participants in the study were offered multiplex genetic testing designed to yield information about 15 different genes that play roles in eight preventable conditions, including type 2 diabetes, coronary heart disease, high blood cholesterol, high blood pressure, osteoporosis, lung cancer, colorectal cancer and malignant melanoma.

In the not-too-distant future, with wider availability of genetic testing, genetic risk information will more often be an added element in the health risk equation.

### Related Links

-  [News & Events](#)
-  [Print this Page](#)
- [Subscribe](#)
-  [RSS](#)

### Bookmark & Share

-  [Delicious](#)
-  [Digg](#)
-  [Facebook](#)
-  [Add to Favorites](#)
-  [Google Bookmarks](#)
-  [MySpace](#)
-  [Twitter](#)
-  [Yahoo Bookmarks](#)
-  [E-mail this Page](#)

Search:  [Home](#)[Common Fund Programs](#)[Funding Opportunities](#)[Funded Research](#)[News & Events](#)[About the Common Fund](#)Back to: [Common Fund Home](#) > [Programs](#)

## Science of Behavior Change

[▶ Overview](#)[▶ Implementation Group Members](#)[▶ Funding Opportunities](#)[▶ Meetings](#)

### OVERVIEW

Human behavior accounts for almost 40% of the risk associated with preventable premature deaths in the United States. Health-injuring behaviors such as smoking, drinking, and drug abuse, as well as inactivity and poor diet are known to contribute to many common diseases and adverse health conditions. Unfortunately, there are few tried and true approaches to motivate people to adopt and maintain healthy behaviors over time. It is difficult for people to begin to change unhealthy behavior, even when they intend to do so, and even more difficult for them to maintain positive behavior changes in the long run. Effective and personalized approaches to achieve sustained behavior change are typically outside the routine practice of medical care. We often use terms like “willpower” and “self-control” to explain behavior change, although the underlying biological, social, and cultural contexts for these terms are not clear. It is clear, however, that understanding the basic underpinnings of motivation change across a broad range of health-related behaviors can lead to more effective and efficient approaches to behavioral intervention and ultimately improve the health of our nation.

The Common Fund is launching the Science of Behavior Change program to improve our understanding of human behavior change across a broad range of health-related behaviors. The program will support research that integrates basic and translational science and cuts across disciplines of cognitive and affective neuroscience, neuroeconomics, behavioral genetics, and behavioral economics. The program will establish the groundwork for a unified science of behavior change that capitalizes on both the emerging basic science and the progress already made in the design of behavioral interventions in specific disease areas. This will be accomplished by supporting basic research to improve our understanding of human motivation and maintenance of behavior change across multiple diseases and conditions, and using this knowledge to develop more effective and economical behavioral interventions.

The NIH hosted a [Science of Behavior Change Meeting](#) in June 2009 to garner input from experts about opportunities and needs in behavior change research.

The program is announcing a new funding opportunity: [RFA-RM-10-002 - Science of Behavior Change: Funding Mechanisms of Change in the Laboratory and in the Field \(R01\)](#).

Can the rubber meet  
the road?

# Are electronic health records ready for genomic medicine?

*Maren T. Scheuner, MD, MPH<sup>1</sup>, Han de Vries, MS<sup>1</sup>, Benjamin Kim, MD<sup>1,4</sup>, Robin C. Meili, MBA<sup>1</sup>, Sarah H. Olmstead, MS<sup>1</sup>, and Stephanie Teleki, PhD<sup>1</sup>*

---

**Purpose:** The goal of this project was to assess genetic/genomic content in electronic health records. **Methods:** Semistructured interviews were conducted with key informants. Questions addressed documentation, organization, display, decision support and security of family history and genetic test information, and challenges and opportunities relating to integrating genetic/genomics content in electronic health records. **Results:** There were 56 participants: 10 electronic health record specialists, 18 primary care clinicians, 16 medical geneticists, and 12 genetic counselors. Few clinicians felt their electronic record met their current genetic/genomic medicine needs. Barriers to integration were mostly related to problems with family history data collection, docu-

mation has the potential to not only radically change the way personal health care is delivered but also how public health is maintained and realized.

However, a recent review of the literature has found that clinicians are not prepared to integrate genetic information into routine clinical practice, including collection, documentation, and interpretation of family history for risk assessment and recommendation of risk-specific interventions, and knowing when to offer genetic tests.<sup>1</sup> Experts believe that electronic health records (EHRs) could enable adoption of genetic information into clinical practice, including the effective use of family history and genetic information to guide clinical practice.

Debate

Open Access

## A national clinical decision support infrastructure to enable the widespread and consistent practice of genomic and personalized medicine

Kensaku Kawamoto\*<sup>1,2</sup>, David F Lobach<sup>1</sup>, Huntington F Willard<sup>2</sup> and Geoffrey S Ginsburg<sup>2</sup>

Address: <sup>1</sup>Division of Clinical Informatics, Department of Community and Family Medicine, Box 104007, Duke University Medical Center, Durham, North Carolina 27710, USA and <sup>2</sup>Duke Institute for Genome Sciences & Policy, Duke University, Durham, North Carolina, USA

Email: Kensaku Kawamoto\* - [kawam001@mc.duke.edu](mailto:kawam001@mc.duke.edu); David F Lobach - [david.lobach@duke.edu](mailto:david.lobach@duke.edu); Huntington F Willard - [Hunt.Willard@duke.edu](mailto:Hunt.Willard@duke.edu); Geoffrey S Ginsburg - [geoffrey.ginsburg@duke.edu](mailto:geoffrey.ginsburg@duke.edu)

\* Corresponding author

Published: 23 March 2009

Received: 6 October 2008

*BMC Medical Informatics and Decision Making* 2009, **9**:17 doi:10.1186/1472-6947-9-17

Accepted: 23 March 2009

This article is available from: <http://www.biomedcentral.com/1472-6947/9/17>

# nature



## THE HUMAN GENOME AT TEN

Growing pains of the  
genomics age

**THE FAINT YOUNG SUN**  
A climate paradox revisited

**QUANTUM MECHANICS**  
Controlling objects you can see

**SLEEPING SICKNESS**  
Drug target for a neglected disease

**NATUREJOBS**  
Getting published



A Venn diagram on a blue background with three overlapping ovals. The largest oval is green and labeled 'Quality Health Care'. A smaller, light green oval overlaps its left side and is labeled 'Genomics'. A smaller orange oval overlaps the bottom of the green oval and is labeled 'Personalized Medicine'. The 'Genomics' and 'Personalized Medicine' ovals also overlap each other.

**Quality Health Care**

**Genomics**

**Personalized Medicine**

# THANKS

**Some slides courtesy of:**

**Les Biesecker, NHGRI**

**Teri Manolio, NHGRI**

**Laura Rodriguez, NHGRI**