



# An update on family history.

June 12, 2009

SACGHS

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April 16, 2009

## Genes Show Limited Value in Predicting Diseases

By [NICHOLAS WADE](#)

The era of personal genomic medicine may have to wait. The genetic analysis of common disease is turning out to be a lot more complex than expected.

Since the human genome was decoded in 2003, researchers have been developing a powerful method for comparing the genomes of patients and healthy people, with the hope of pinpointing the DNA changes responsible for common diseases.

This method, called a genomewide association study, has proved technically successful despite many skeptics' initial doubts. But it has been disappointing in that the kind of genetic variation it detects has turned out to explain surprisingly little of the genetic links to most diseases.

A set of commentaries in this week's issue of The [New England Journal of Medicine](#) appears to be the first public attempt by scientists to make sense of this puzzling result.

One issue of debate among researchers is whether, despite the prospect of diminishing returns, to continue with the genomewide studies, which cost many millions of dollars apiece, or switch to a new approach like decoding the entire genomes of individual patients.

**Family history** is still the cheapest, most accessible, most time-tested way to get a rough estimate of the genetic component of disease risk.

# Oregon 2007 General Knowledge Survey

Phone survey of ~ 2000 individuals >18yrs old part of CDC Behavioral Risk Factor Surveillance System (BRFSS)

- 99.2% agree that family history is important to their health
- 61.4% have collected family history information from relatives to develop a family health history
- 95.2% think that having a close relative with diabetes, heart disease or cancer raises their own risk

<http://egov.oregon.gov/DHS/ph/genetics>

# 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

## **FAMILY HISTORY**

Cancer syndromes:  
BRCA1

Pgx:  
Abacavir  
hypersensitivity

Pgx:  
Warfarin  
metabolism

Treatment selection:  
EGFR/breast cancer



# USPSTF and Family History: Breast Cancer

The USPSTF recommends that women whose **family history** is associated with an increased risk for deleterious mutations in *BRCA1* or *BRCA2* genes be referred for genetic counseling and evaluation for *BRCA* testing. **B recommendation**



*The* NEW ENGLAND JOURNAL *of* MEDICINE

Perspective  
JUNE 19, 2008

## Keeping Pace with the Times — The Genetic Information Nondiscrimination Act of 2008

Kathy L. Hudson, Ph.D., M.K. Holohan, J.D., and Francis S. Collins, M.D., Ph.D.

**L**aws and institutions must go hand in hand with the progress of the human mind. As that becomes more

sumers from discrimination by health insurers and employers on



## THE HIPAA PRIVACY RULE



# Frequently Asked Questions About Family Medical History Information

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U.S. Department of Health and Human Services • Office for Civil Rights

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**1. Does the HIPAA Privacy Rule limit an individual's ability to gather and share family medical history information?**

*No.* The HIPAA Privacy Rule may limit how a covered entity (for example, a health plan or most health care providers) uses or discloses individually identifiable health information, but does not prevent individuals, themselves, from gathering medical information about their family members or from deciding to share this information with family members or others, including their health care providers. Thus, individuals are free to provide their doctors with a complete family medical history or communicate with their doctors about conditions that run in the family.

[www.hhs.gov/ocr/privacy/familyhealthhistoryfaqs.pdf](http://www.hhs.gov/ocr/privacy/familyhealthhistoryfaqs.pdf)

# The Medicare Improvements for Patients and Providers Act (MIPPA)

July 15, 2008

**“MIPPA 2008 (Section 101) gives authority to DHHS Secretary to consider additional preventive services benefits (e.g., those with an “A” or “B” rating from US Preventive Services Task Force) through Medicare NCD process”**

**Barry M. Straube, MD SACGHS Meeting March 12, 2009**

# Development of Evidence: CDC Family Healthware™ Trial

Family Healthware™ is a web-based consumer tool that provides risk assessment and health messaging for 6 common complex conditions:

- Heart disease
- Stroke
- Diabetes
- Colorectal cancer
- Breast cancer
- Ovarian cancer

– Yoon PW, et al. *Prev Chronic Dis* 2009;6(1).

# CDC's Family Healthware™ Trial

3585 participants used Family Healthware™; 41 primary care practices, 187 clinicians in 13 states

– Oneil SM, et al. Am J Prev Med. 2009 Jun;36(6):506-14.

- High burden of familial risk
  - 82% had a strong / moderate familial risk for at least one of the 6 diseases assessed
- Electronic data collection improved data present in chart
  - 23% of those unable to have risk assessed using chart had moderate or strong risk for one of the 6 diseases
- Family Healthware™ influenced risk perception among underestimators (low → high)
  - CHD, DM, Stroke, Colon cancer
- Paradoxical effects, Control > Intervention
  - Adherence significantly improved in Controls vs. Intervention for Cholesterol screening and Clinical breast exam

# Development of Evidence

- CDC, NCI, NIDDK genomics translational research RFAs
  - Family history projects included
- NHGRI ARRA “Challenge Grants”
  - 10-HG-101
- NCI DCCPS 2009 “Priorities”
  - “There is a crucial need to incorporate family history and genomic information into population modeling models to investigate the population impact of using family history and genetic testing to stratify the population to more appropriately target prevention, early detection and treatment strategies.”

# Evidence Synthesis: AHRQ

Two reports produced by McMaster University

– *Collection and Use of Cancer Family History in Primary Care* 10/07

[www.ahrq.gov/clinic/tp/famhisttp.htm](http://www.ahrq.gov/clinic/tp/famhisttp.htm)

– *Clinical Utility of Cancer Family History Collection in Primary Care* 4/09

[www.ahrq.gov/Clinic/tp/famhist2tp.htm](http://www.ahrq.gov/Clinic/tp/famhist2tp.htm)

# *Collection and Use of Cancer Family History in Primary Care*

(AHRQ/McMaster University EPC 10/07)

<u>Disorder</u>	<u>Specificity</u>	<u>Sensitivity</u>
Breast Cancer	95-98%	85-90%
Colorectal Cancer	91-92%	57-90%
Ovarian Cancer	96-99%	67-83%
Prostate Cancer	93-99%	69-79%

**(accuracy across all relatives)**

# Clinical Utility of Cancer Family History Collection in Primary Care

**“Conclusions:** Our review indicates a very limited evidence base with which to address all four of the research questions:

1. The few evaluations of cancer risk prediction models do not suggest useful individual predictive accuracy.
2. The experimental evidence base for primary and secondary cancer prevention is very limited.
3. There is insufficient evidence to assess the effect of FHx-based risk assessment on preventive behaviors.
4. There is insufficient evidence to assess whether FHx-based personalized risk assessment directly causes adverse outcomes.”

# Evidence Synthesis: NIH



NIH EVIDENCE SYNTHESIS CONFERENCE

**August 24-26, 2009**

EVIDENCE SYNTHESIS

and Improving Health

The logo features a green background with a yellow horizontal band at the bottom. On the left, there is a stylized tree with yellow nodes and branches. The text 'EVIDENCE SYNTHESIS' is written vertically in white, with 'EVIDENCE' on the left and 'SYNTHESIS' on the right. The date 'August 24-26, 2009' is written in bold black text across the center. The words 'NIH EVIDENCE SYNTHESIS CONFERENCE' are at the top, and 'and Improving Health' is at the bottom.

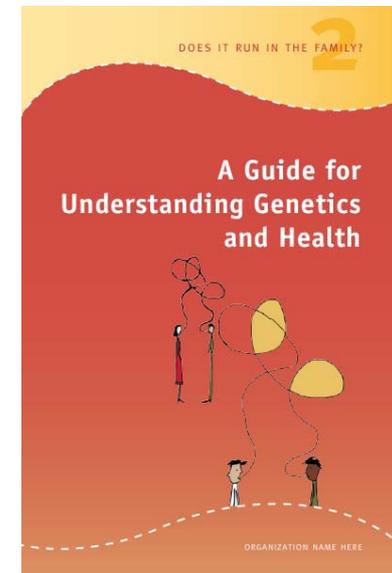
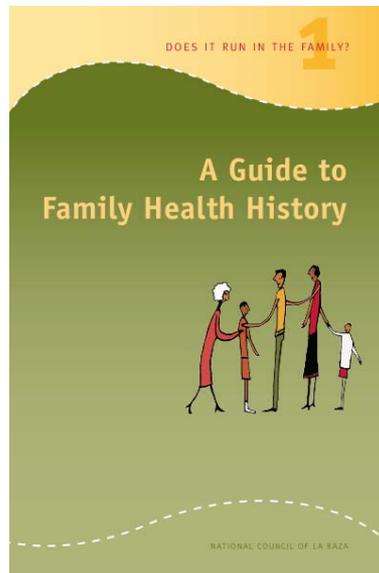
Register at:

[http://consensus.nih.gov/  
2009/familyhistory.htm](http://consensus.nih.gov/2009/familyhistory.htm)

The pragmatic approach....

# HRSA:

## Community Centered Family Health History



[www.geneticalliance.org/familyhealthhistory](http://www.geneticalliance.org/familyhealthhistory)

Supported through a cooperative agreement from the Genetic Services Branch of the Maternal and Child Health Bureau/HRSA/DHHS.

U33MC06836-

# Southcentral Foundation Family History Demonstration Project

- Engaged employees
- Educational Module on Importance of Family History
- Filmed video

**Family History and Your Health:  
Making the Connection**

Family health history is a story of your family's health.



**Importance of Family Health History**  
Knowing your family health history can help you identify whether diseases like diabetes, heart disease or other health conditions may be present in your family.



**Role of Family**  
If you don't know about your family health history, asking family members can help you with that journey.



**You**  
Knowledge about your family health history can help you take steps to better health.



**Your Provider**  
Talk to your provider about your family health history concerns.

**Join a Southcentral Foundation Project on Family Health History**

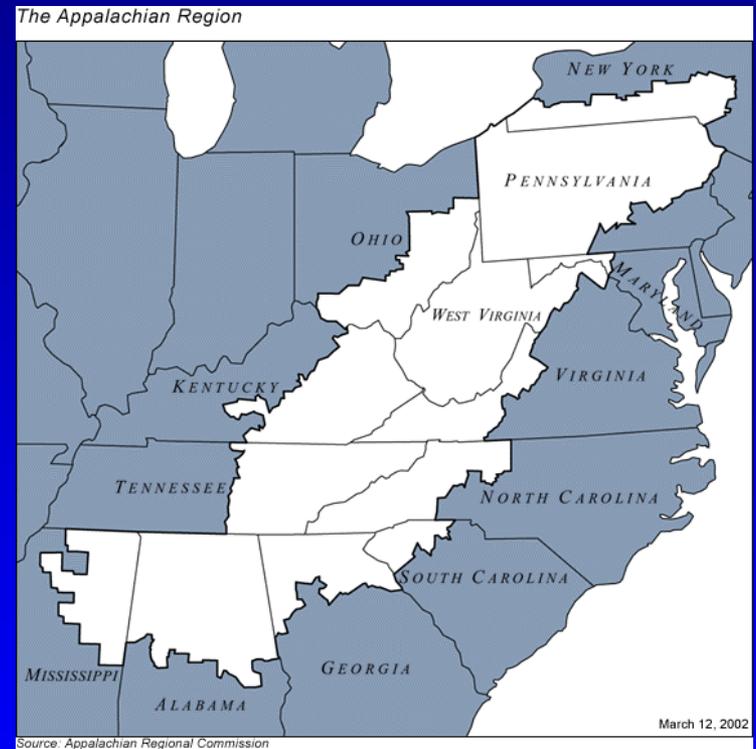
- All Southcentral Foundation employees are welcome
- After viewing an educational module simply complete a survey and return it to the Research Department
- You will receive a stainless steel travel mug and a \$5 coffee card
- It will take less than one hour to complete

Southcentral Foundation  
Research Department  
Neon Building  
4201 Tudor Centre Drive, Suite 108  
Anchorage, AK 99508  
(907) 729-8623

Southcentral Foundation 

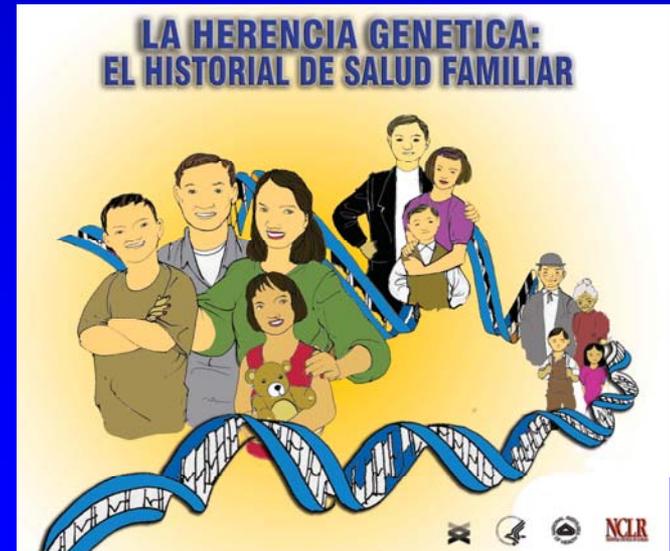
# Urban Appalachian Community

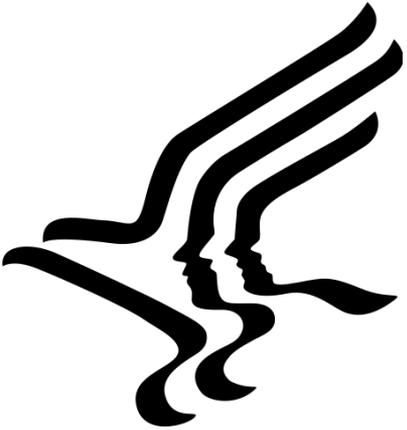
- Higher poverty rates related to:
  - High-school drop out rates
  - Unemployment
- Appalachian whites reported higher rates of:
  - Lung disease
  - Heart trouble
  - Diabetes
  - Hypertension
  - High Cholesterol
  - Stroke



# National Council of La Raza/ Institute for Hispanic Health:

## Developing a Community-Driven Model to Mobilize Latino Communities around Genetics Information





# My Family Health Portrait

A tool from the Surgeon General

## Family Health History

### *Surgeon General's Initiative*

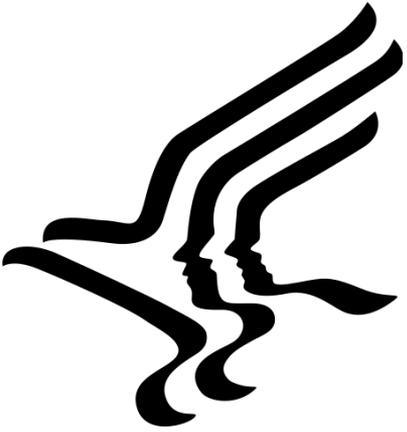
- In 2004, the Surgeon General introduced the first version of the web-based tool, “My Family Health Portrait.”
- This tool helped consumers by enabling them to complete histories at home.
- However – the original tool was NOT standards-based, interoperable, or EHR-ready.



# Sponsoring Federal Agencies

- Indian Health Service
- National Human Genome Research Institute
- National Cancer Institute
- Agency for Healthcare Research and Quality
- National Institute of Diabetes and Digestive and Kidney Disorders
- Office of Rare Diseases, National Institutes of Health
- Substance Abuse and Mental Health Services Administration
- National Office of Public Health Genomics,
- Centers for Disease Control and Prevention
- Office of the National Coordinator for Health Information Technology
- Office of Minority Health
- Office of the Surgeon General
- Office of the Assistant Secretary for Planning and Evaluation
- Federal Health Architecture (Veterans Health Administration and Department of Defense)
- Health Resources and Services Administration





# My Family Health Portrait

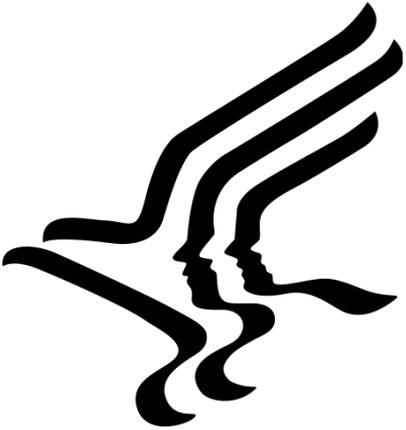
A tool from the Surgeon General

## Family Health History

### *Standards-based*

- XML-based
- HL7 family history model
- LOINC
- SNOMED-CT
- HL7 Vocabulary
- Minimum core data set
- Compatible with existing electronic genealogy tools





# My Family Health Portrait

A tool from the Surgeon General

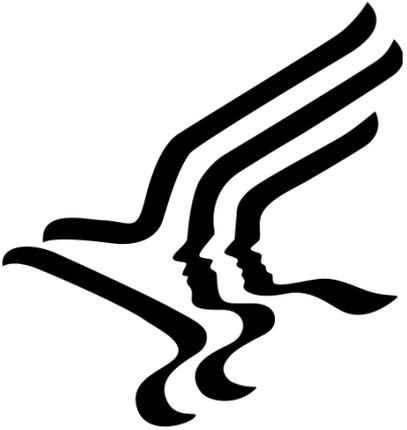
## Family Health History

### *Arriving at standards*

- Achieved in 2008 by a public/private task force convened by the AHIC Personalized Health Care workgroup
- Defined the minimum FHH data elements that every EHR and PHR should be able to capture
- HITSP approved interoperability Dec. 2008
  - See: [http://hitsp.org/ConstructSet\\_Details.aspx?&PrefixAlpha=1&PrefixNumeric=08](http://hitsp.org/ConstructSet_Details.aspx?&PrefixAlpha=1&PrefixNumeric=08)

*New Standards and Enhanced Utility for Family Health History Information in the Electronic Health Record: An Update from the American Health Information Community's Family Health History Multi-Stakeholder Workgroup*

W. Gregory Feero, Mary Beth Bigley, Kristin M. Brinner The Family Health History Multi-Stakeholder Workgroup of the American Health Information Community J Am Med Inform Assoc 2008; 15: 723-728.



# My Family Health Portrait

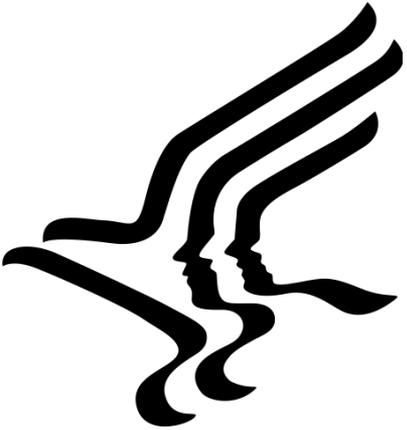
A tool from the Surgeon General

## Family Health History

### *New interoperable tool*

- In January 2009, the Surgeon General launched a new “2.0” family health history tool.
- The new tool is standards-based, interoperable and EHR-ready.
- Consumers can share histories electronically – with other family members and doctors.





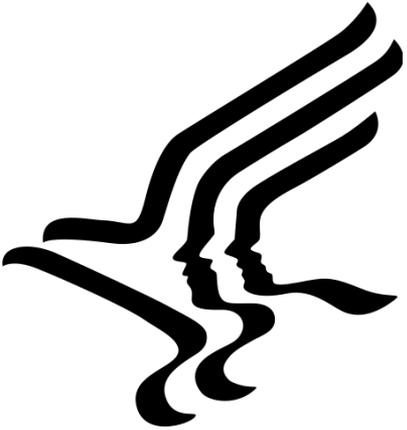
# My Family Health Portrait

A tool from the Surgeon General

## Family Health History

### *Openly-available source code*

- The Surgeon General's new tool is openly-available for other organizations to adopt.
- Source code for the tool is available without charge. No attribution to Surgeon General is needed.
- However, the adopted tool must preserve interoperability features.



# My Family Health Portrait

A tool from the Surgeon General

## Family Health History *Adopting Source Code*

- Source code is available at:
  - <https://gforge.nci.nih.gov/projects/fhh>
- Help desk assistance is available.
  - Email: [ncicb@pop.nci.nih.gov](mailto:ncicb@pop.nci.nih.gov)
  - Phone: (888) 478-4423/(301) 451-4384

# AHRQ:RTI DEcIDE

## **“Computer-based Clinical Decision Support Tools for Gene-based Tests Used in Breast Cancer:**

The **Patient BRCA Tool** will (1) assist in collection of family history of cancer to assess the risk of carrying the BRCA1 or BRCA2 gene mutations and (2) assist in making decisions for appropriate follow up. The tool will be completed by women in their homes prior to their office visit to a primary care provider.

The **Integrated BRCA Tool** will use family history information to guide primary care providers through the process of discussing the woman's family history, cancer risk, and the recommendation for or against genetic counseling and BRCA testing.”

# **HRSA:** New project: U33MC12786-01-00 Family Health History for Prenatal Care Providers

National Coalition for Health Professionals Education in Genetics in partnership with Genetic Alliance, Harvard Partners, and March of Dimes

## Goals of the project are:

- Adapt HP's existing family history tool to the prenatal setting
- Incorporate existing prenatal and women's health content, including newborn screening

## Goals continued:

- Address cultural sensitivity and ethical issues
- Provide decision-support algorithms that will highlight genetic red flags for the provider.
- Develop a continuing-education program for providers.
- “Just-in-time” information as defined by significant intervention points and teachable moments for provider and patient during the patient’s entire life course (e.g., first ob/gyn visit, prenatal/preconception, and mature adult).
- Develop related family history and educational materials for the public.

## Next stop for MFHP:

Developing open-source, interoperable interpretive capabilities for CRC risk.

CDC

NCI

NHGRI

OSG

?

# Conclusions

- Family history is a versatile and potentially powerful tool for improving health care.
- Family history will not be supplanted by genetic testing in the near future.
- Though the evidence base should be expanded, family history is a solid cornerstone for delivering clinical care.
- Ongoing Federal activities support expanding the evidence base and enhancing adoption.

# THANKS

**Slides courtesy of:**

**Greg Downing, OS**

**Alan Guttmacher, NHGRI**

**Sara Harding, NHGRI**

**Genetic Alliance**

**HRSA**