

MCH/CSHCN Directors  
May 2005 Webcast

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THE U.S. SURGEON  
GENERAL'S FAMILY  
HISTORY INITIATIVE



Alan E. Guttmacher, M.D.  
MCHCOM.COM Webcast  
May 12, 2005



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What We Will Talk About

- Past use of the family history
- Current use of the family history
- The U.S. Surgeon General's Family History Initiative
- Use of the family history in the future

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## Family History – Past Use

- Genealogy

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## Matthew 1:2-16

Abraham begat Isaac; and Isaac begat Jacob; and Jacob begat Judas and his brethren; and Judas begat Phares and Zara of Thamar; and Phares begat Esrom; and Esrom begat Aram; and Aram begat Aminadab; and Aminadab begat Naasson; and Naasson begat Salmon; and Salmon begat Booz of Rachab; and Booz begat Obed of Ruth; and Obed begat Jesse; and Jesse begat David the king; and David the king begat Solomon of her that had been the wife of Urias; and Solomon begat Roboam; and Roboam begat Abia; and Abia begat Asa; and Asa begat Josaphat; and Josaphat begat Joram; and Joram begat Ozias; and Ozias begat Joatham; and Joatham begat Achaz; and Achaz begat Ezekias; and Ezekias begat Manasses; and Manasses begat Amon; and Amon begat Josias; and Josias begat Jechonias and his brethren, about the time they were carried away to Babylon. And after they were brought to Babylon, Jechonias begat Salathiel; and Salathiel begat Zorobabel; and Zorobabel begat Abiud; and Abiud begat Eliakim; and Eliakim begat Azor; and Azor begat Sadoc; and Sadoc begat Achim; and Achim begat Eliud; and Eliud begat Eleazar; and Eleazar begat Matthan; and Matthan begat Jacob; and Jacob begat Joseph the husband of Mary, of whom was born Jesus, who is called Christ

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## THE TWO AGES OF THE FAMILY HEALTH HISTORY ?

- 1) Today: Used primarily as surrogate for individual's genetic makeup
- 2) Future: Used in conjunction with genetic testing to predict disease risk, prognosis, and response to therapy

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In the “Age of Genomics,”  
Why Family History?

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In the “Age of Genomics,”  
Why Family History?

- Most diseases are due to interactions of multiple genes and environmental factors.

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**≥ 9 of the 10 Leading Causes of Mortality Have Genetic and Environmental Components**

- 1. Heart disease (28.9% of U.S. deaths in '01)
- 2. Cancer (22.9%)
- 3. Cerebrovascular diseases (6.8%)
- 4. Chronic lower respiratory dis. (5.1%)
- ? 5. Injury (4.0%)
- 6. Diabetes (2.9%)
- 7. Pneumonia/Influenza (2.6%)
- 8. Alzheimer disease (2.2%)
- 9. Kidney disease (1.6%)
- 10. Septicemia (1.3%)

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**GENES + ENVIRONMENT =  
HEALTH AND DISEASE**

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**Family History**

- Family history is a record of shared health and disease
- But, remember:  
    Health and disease = genes + environment
- Therefore,  
    Family history is a record of shared genes + shared environment  
    - Q.E.D.

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### Family History Changes Population Screening Guidelines

- Visual Impairment
- Hearing Impairment
- Thyroid Disease
- Thromboembolism
- Hypertension
- Diabetes
- Coronary Artery Disease
- Dyslipidemia
- Breast Cancer
- Colon Cancer
- Prostate Cancer
- Liver Cancer
- Hip Dysplasia
- Iron Def Anemia
- Osteoporosis
- Cardiomyopathy

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### Family History Changes Management of Common Disease

- Coronary Heart Disease
- Hypertension
- Heart Failure
- Emphysema & COPD
- Syncope
- Pancreatitis
- Diabetes
- Thromboembolism
- Thyroid Cancer
- Breast Cancer
- Colon Cancer
- Urticaria
- Developmental Delay
- Pancreatitis

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### But Do People Care About Family History?

- In the Parade/Research!America 2004 survey of 1,000 individuals:
  - 65% have found health information on the Internet to be very or somewhat helpful

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### But Do People Care About Family History?

- In the Parade/Research!America 2004 survey of 1,000 individuals:
  - 65% have found health information on the Internet to be very or somewhat helpful
  - But, 96% thought, in thinking of their own health, that knowledge of family health history was very or somewhat important

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### But Do People Care About Family History?

- In a CDC-based survey of over 4,000 individuals:
  - 97% thought that knowledge of family health history was important

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### But Do People Care About Family History?

- In a CDC-based survey of over 4,000 individuals:
  - 97% thought that knowledge of family health history was important
  - But, only 30% had actually ever collected health information from relatives to develop a family health history.

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### In the “Age of Genomics,” Why Family History?

- Although we will gain important new genomic tools, family history will remain highly relevant for years.
- Family history helps predict risk for such varied health concerns as heart disease, colorectal cancer, breast cancer, ovarian cancer, osteoporosis, atopy or asthma, type 2 diabetes, suicide, etc.
- Yet, many people are unaware of relatives’ medical histories, and many health professionals underutilize this information in advising patients on how to maintain good health.

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### If Family History Is So Useful, Why Don’t Health Professionals Use It More Consistently and Effectively?

- 1) Underestimation by clinicians of the utility of the family history
  - Requires better teaching and more pervasive role modeling of effective use of the family history
- 2) Not enough time to obtain, organize, and analyze family history information
  - Requires creative approaches to the family history that demand less practitioner time

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### So, What Can We Do?



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### Goals of U.S. Surgeon General's Family History Initiative

- Increase the American public's awareness of the importance of family history in health;
- Give the American public tools to gather, understand, evaluate, and use family history to improve their health;
- Increase the awareness of health professionals about the importance of family history;
- Give health professionals tools to gather, evaluate, and use family history information; and to communicate with their patients about family history;
- Increase genomics and health literacy;
- Prepare both the American public and their health professionals for the coming era in which genomics will be an integral part of regular health care.

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### So, What Can We Do?

[www.hhs.gov/familyhistory](http://www.hhs.gov/familyhistory)

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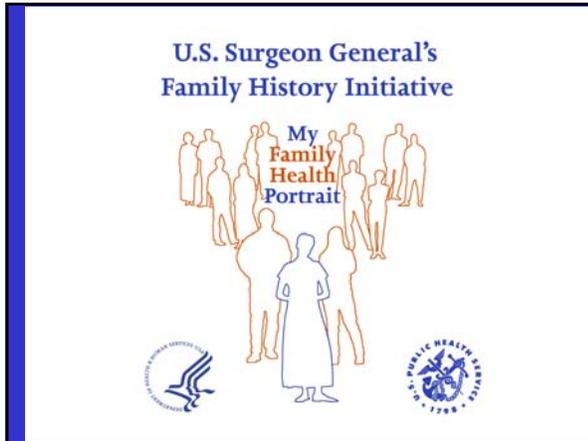
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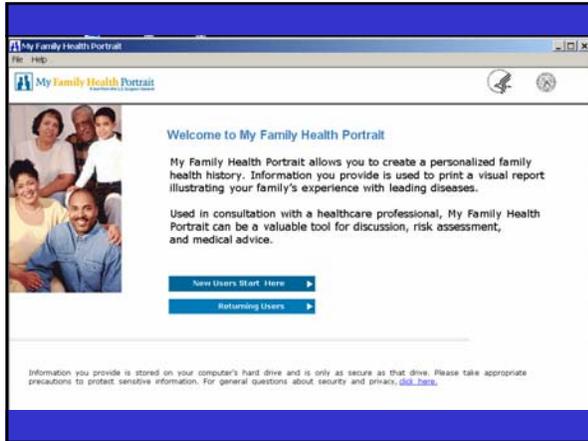
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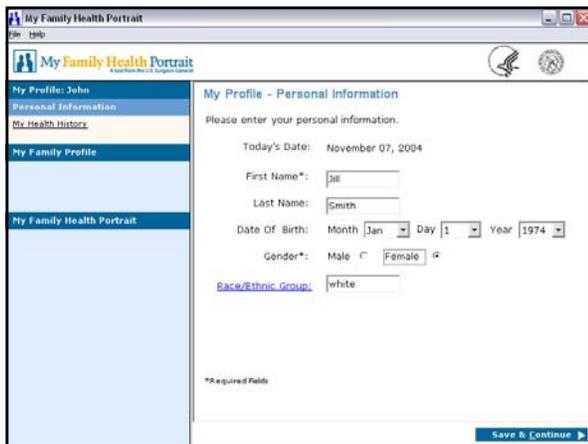
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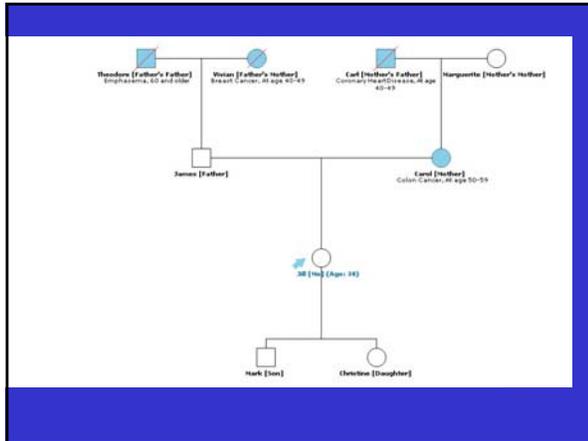


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**My Family Health Portrait - Text Report**  
Jill Smith - November 07, 2004

My Family Health Portrait should only be used in consultation with a healthcare professional. It can be a valuable tool for discussion, risk assessment, and medical advice.

**My Profile - Personal Information**

Name: Jill Smith  
Date of Birth: January 01, 1974  
Gender: F  
Race/Ethnic Group: white

**My Profile - My Health History (Disease, Age at first diagnosis)**

Coronary Heart Disease: No  
Stroke: No  
Diabetes: No  
Colon Cancer: No  
Breast Cancer: No

	Coronary Heart Disease	Stroke	Diabetes	Colon Cancer	Breast Cancer	Ovarian Cancer	Other
Jill [me]	No	No	No	No	No		
Christine [Daughter]	No	No	No	No	No	No	
Mark [Son]	No	No	No	No	No	No	
Carol [Mother]	No	No	No	At age 58-59	No	No	
Margaret [Mother's Mother]	No	No	No	No	No	No	
Earl [Mother's Father]*	At age 48-49	No	No	No	No		
James [Father]	No	No	No	No	No		
Theodore [Father's Father]*	No	No	No	No	At age 40-49	No	Emphysema [14 and other]
Helen [Father's Mother]*	No	No	No	No	No		

\*Deceased

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**Next Steps**

- Improve “My Family Health Portrait”
- Continued media attention and outreach
- New projects and uses by any and all
- Make Thanksgiving the Annual Family History Day

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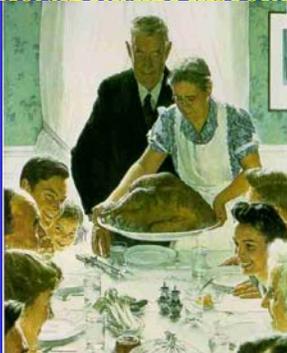
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**THANKSGIVING DAY IS THE ANNUAL  
NATIONAL FAMILY HISTORY DAY**



Now that we've finished the family history, let's eat!

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SOUNDING BOARD

The Family History — More Important Than Ever

Alan E. Guttmacher, M.D., Francis S. Collins, M.D., Ph.D., and Richard H. Carmona, M.D., M.P.H.

For many observers, the term "genomic medicine" conjures up space-age images of microarray chips, bioinformatics, and designer drugs. Today, with medicine poised at the dawn of the genomic era, it is seductive to believe that such high-tech options have already become the most important genomic tools in health care. However, as so often happens in medicine, new developments do not eclipse the tried-and-true method; instead, they give it new meaning and power.

Most diseases are the result of the interactions of multiple genes and environmental factors. Although these interactions are complex, almost every patient today has access to a free, well-proven, personalized genomic tool that captures many of these interactions and can serve as the cornerstone for individualized disease prevention. This valuable tool is the family history.<sup>1,2</sup>

Although advances arising from the Human Ge-

disease. Knowledge that both parents are carriers for sickle cell disease can lead to early diagnosis in an asymptomatic but affected newborn, prompt introduction of prophylactic antibiotic therapy, and careful surveillance for painful crises, and thus improving the likelihood of decreasing the baby's disease burden. Knowledge that a woman has a brother and a maternal uncle with the fragile X syndrome affects both prenatal counseling and the evaluation of her child who has a developmental delay. The approach to a patient with shortness of breath changes dramatically if an alert physician obtains a history of a first-degree relative with frequent epistaxis and telangiectasia, suggesting that the patient may have hereditary hemorrhagic telangiectasia. As a case reported in this issue of the *Journal* illustrates, knowledge of a parental balanced chromosomal inversion can abbreviate an otherwise lengthy workup for failure to thrive.<sup>1,4</sup>

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## THE TWO AGES OF THE FAMILY HEALTH HISTORY ?

- 1) Today: Used primarily as surrogate for individual's genetic makeup
- 2) Future: Used in conjunction with genetic testing to predict disease risk, prognosis, and response to therapy

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### Today

- We use family history as a surrogate for genes - in the absence of genetic testing of the individual (until recently, that was almost always the case; now usually the case), we use family history as the best available surrogate for actual genetic makeup.

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### Tomorrow

- We will use genetic testing to assay genes directly.



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## Future

- We will use genetic testing to assay genes directly.
- So, we will no longer need to use family history as a surrogate for genetic status.
- However, for many years, we will not know exactly how the specific genetic status interacts with environment to create health and disease.
- So, we will use family history to help guide analysis and use of genetic tests.
  - e.g., interpretation of *BRCA1* mutation

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## What Might We Do to Give Us Better Tools for This Future?

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## An American Gene x Environment Study?



**insight commentary**

### The case for a US prospective cohort study of genes and environment

Francis S. Collins

National Human Genome Research Institute, National Institutes of Health, Building 31, Room 409A, MSC 2152, 31 Center Drive, Bethesda, Maryland 20892-2152, USA (e-mail: fsc@nih.gov)

Information from the Human Genome Project will be vital for defining the genetic and environmental factors that contribute to health and disease. Well-designed case-control studies of people with and without a particular disease are essential for this, but rigorous and unbiased conclusions about the causes of diseases and their population-wide impact will require a representative population to be monitored over time (a prospective cohort study). The time is right for the United States to consider such a project.

Identification of the genetic and environmental factors that contribute to health, disease and response to treatment is essential for the reduction of disease. This, of course, is the primary goal of biomedical research. Several auspicious recent developments suggest that progress in this area could be quite rapid. The sequencers of the human genome<sup>1</sup> and increasing understandings about the genome's function have provided a robust foundation for the investigation of human health and disease. Likewise, results from the exploration of human genetic variation through the International HapMap Project<sup>2</sup> will soon furnish researchers with a powerful tool for identifying variants that contribute to common disease. This information will be especially useful when combined with well-established methods for measuring environmental exposures. These techniques promise to extend the range of epidemiological investigation<sup>3</sup>. There is growing recognition that a change in the environment, in combination with genetic disposition, has produced most recent epidemics of chronic disease, and may hold the key for reversing the course of some diseases<sup>4</sup>. For example, consider the interaction of gene-environment-protective genetic predispositions with a modern environment in which there is a ready availability of excess calories. This has probably contributed to the current obesity epidemic in the United States. Development of robust analytical methods for assessing disease-risk relationships and interactions is beginning to allow researchers to disentangle such complex effects from a population as a whole<sup>5</sup>.

Collins FS. Nature 2004. 429:475-7.

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### Eventually

- We will need to perfect (and make cheap and accessible) genetic testing.
- We will need to develop effective and efficient techniques for gathering appropriate information about all three environment: physical, built, dietary, behavioral, social, cultural, etc.
- We will need to do all sorts of real world research to determine how best to collect, analyze, communicate, and employ all this information.

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### Eventually

- Perhaps the family history can return to genealogy, from whence it came...

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### Matthew 1:2-16

Abraham begat Isaac; and Isaac begat Jacob; and Jacob begat Judas and his brethren; and Judas begat Phares and Zara of Thamar; and Phares begat Esrom; and Esrom begat Aram; and Aram begat Aminadab; and Aminadab begat Naasson; and Naasson begat Salmon; and Salmon begat Booz of Rachab; and Booz begat Obed of Ruth; and Obed begat Jesse; and Jesse begat David the king; and David the king begat Solomon of her that had been the wife of Urias; and Solomon begat Roboam; and Roboam begat Abia; and Abia begat Asa; and Asa begat Josaphat; and Josaphat begat Joram; and Joram begat Ozias; and Ozias begat Joatham; and Joatham begat Achaz; and Achaz begat Ezekias; and Ezekias begat Manasses; and Manasses begat Amon; and Amon begat Josias; and Josias begat Jechonias and his brethren, about the time they were carried away to Babylon. And after they were brought to Babylon, Jechonias begat Salathiel; and Salathiel begat Zorobabel; and Zorobabel begat Abiud; and Abiud begat Eliakim; and Eliakim begat Azor; and Azor begat Sadoc; and Sadoc begat Achim; and Achim begat Eliud; and Eliud begat Eleazar; and Eleazar begat Matthan; and Matthan begat Jacob; and Jacob begat Joseph the husband of Mary, of whom was born Jesus, who is called Christ.

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Family History Can Be a Lifesaver...  
But It Does Have Limits...  
Limits We Can Extend

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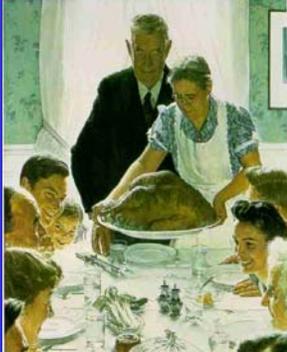
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