

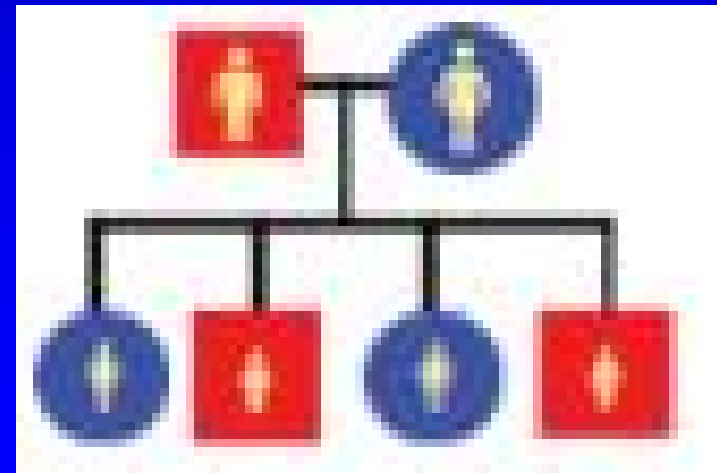
# THE GENETIC FAMILY HISTORY OF THE FUTURE ONLINE, INTERACTIVE, RISK ASSESSMENT

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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.

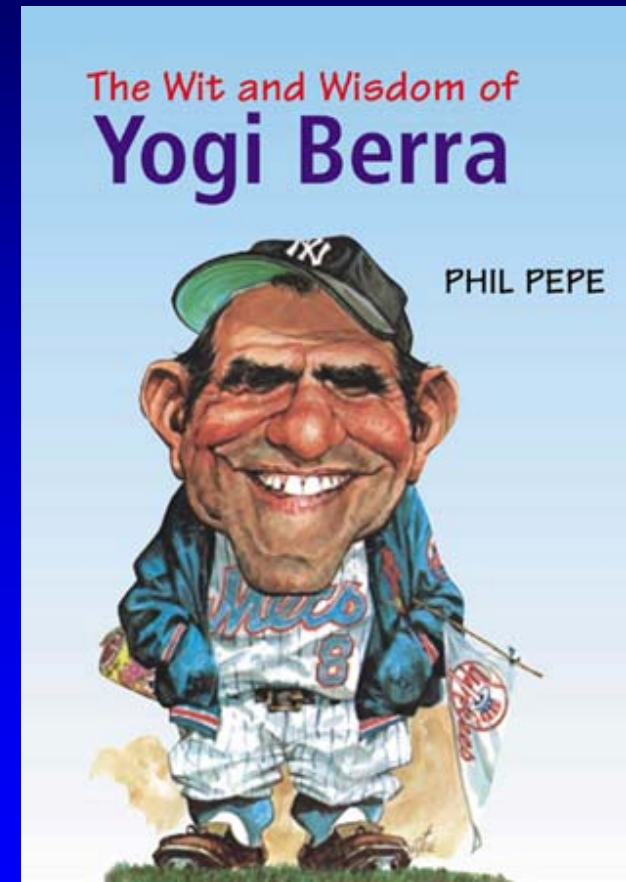
For many observers, the term “genomic medicine” conjures up space-age images of microarray chips, bioinformatics, and designer drugs. Today, with medicine poised at the dawn of the genomic era, it is seductive to believe that such high-tech options have already become the most important genomic tools 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.

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, 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 ap-

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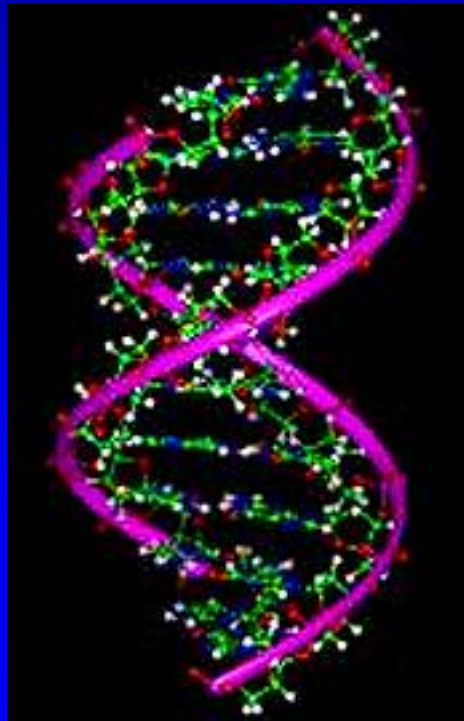
# The Family History - Philosophy

- “It’s tough to make predictions, especially about the future....the future ain’t what it use to be”.  
– Yogi Berra



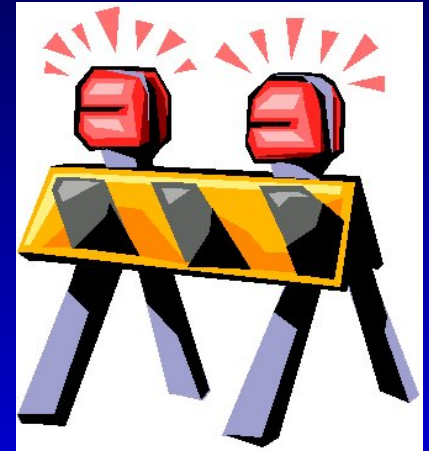
# The Problem

- Clinical Genetics: “The health care of a few”
- Genomic Medicine: “The health care of all”



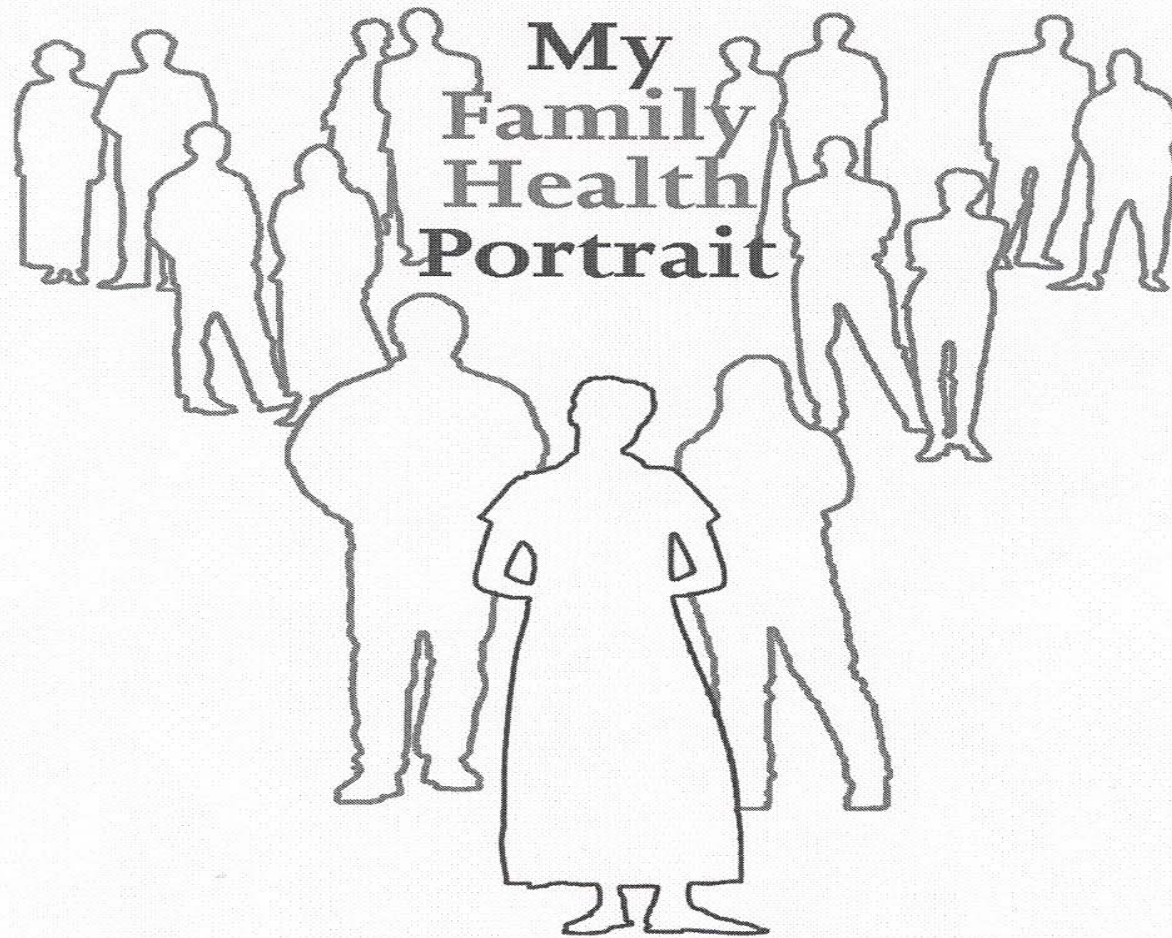
# Family History - Barriers

- Inadequate time of primary care physician
- Underestimation of its value by clinician
- Insufficient knowledge of basic genetics
- Patient may withhold critical family history
- Patient may not know critical family history
- Insufficient knowledge to do risk assessment



# The Solution

- A more efficient clinical genetics infrastructure
- Increase Primary Care genetic services
  - Genetic Counselor within Primary Care Department
  - The “Genetic Inbox”
- Incorporate the internet into genetic services:
  - Patient triage
  - Collection of medical history information
  - Pedigree construction and family history
  - Patient and provider education



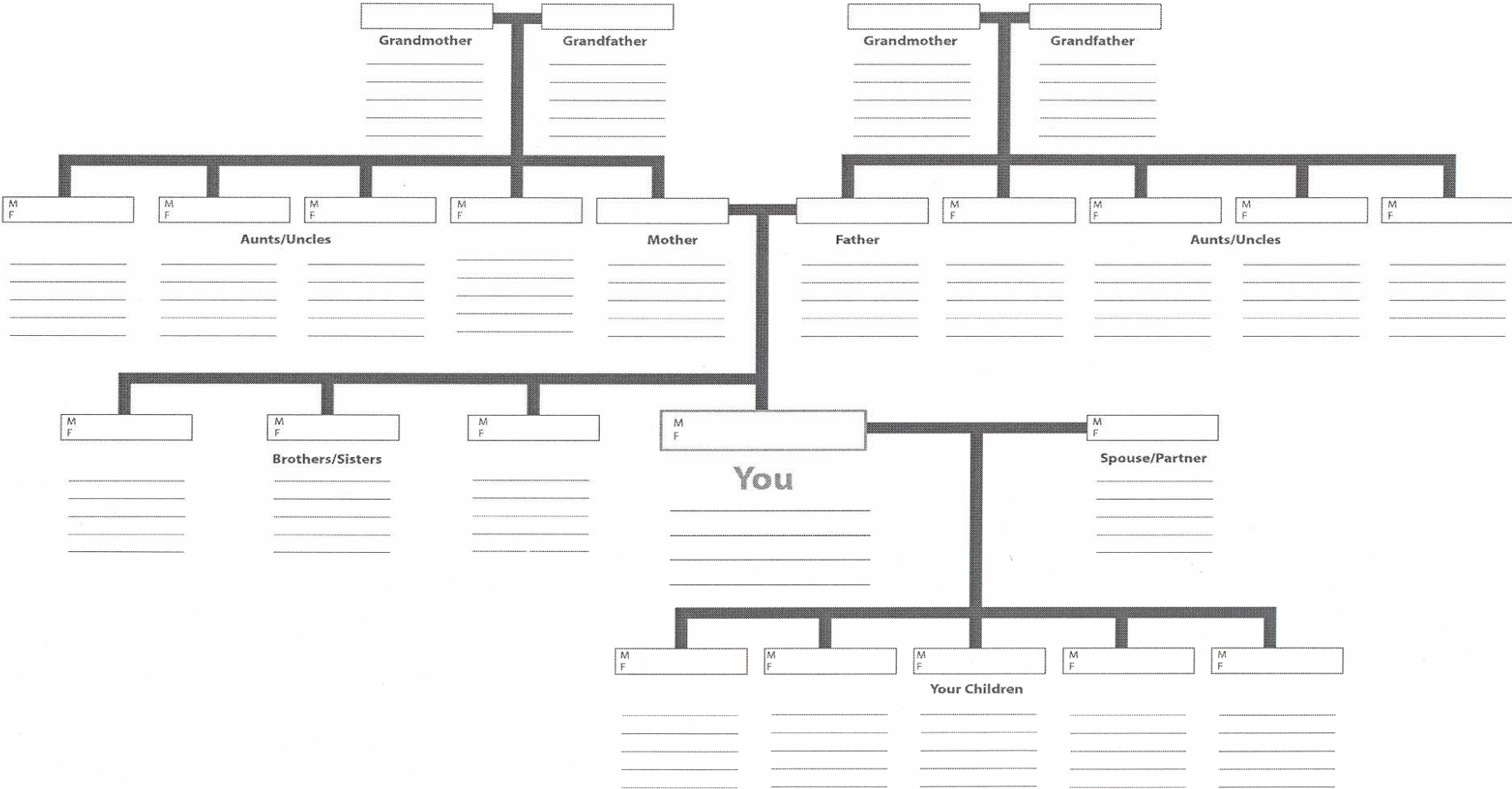
**THE SURGEON GENERAL'S  
FAMILY HISTORY INITIATIVE**

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

# MY FAMILY HEALTH PORTRAIT

Name: \_\_\_\_\_

Date: \_\_\_\_\_



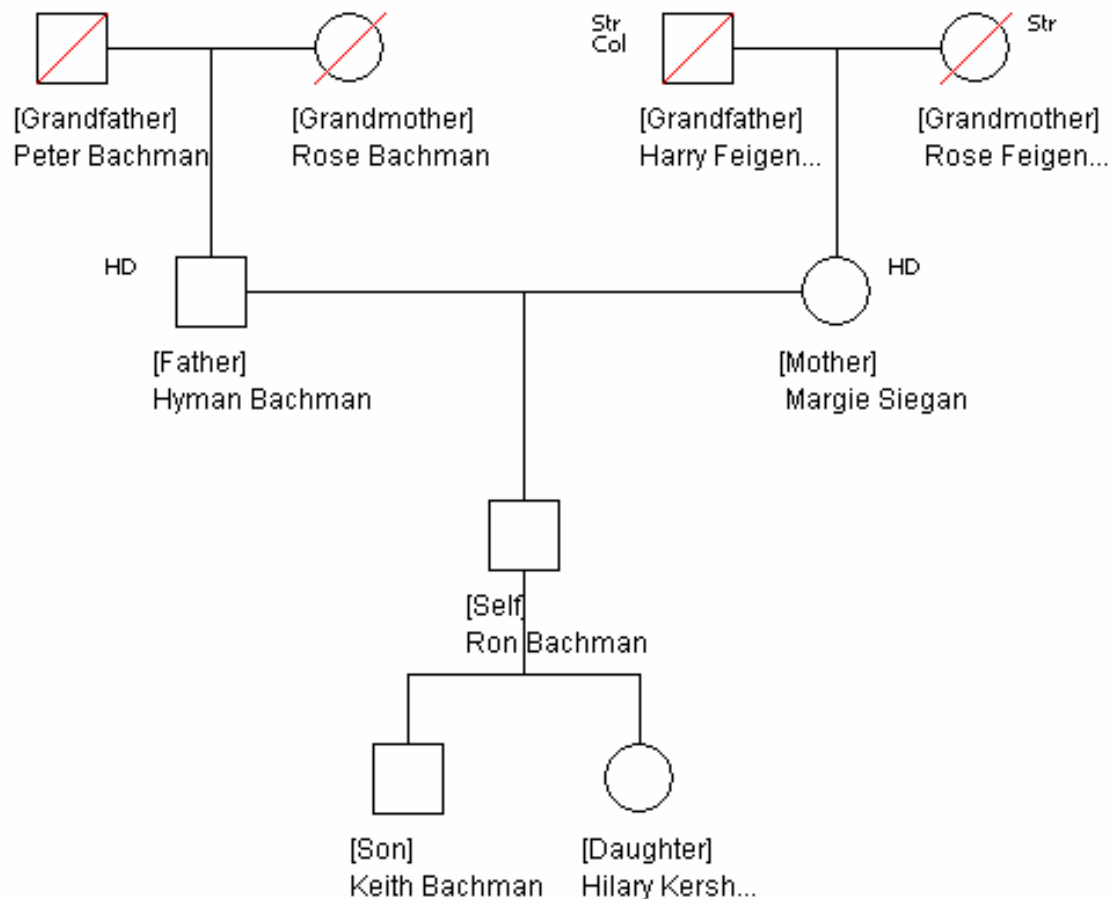


# My Family Health Portrait – The Surgeon General’s Family History Initiative

## My Family Health Portrait - Drawing Report

Ron Bachman - November 30, 2005

□ - Male Family Member	■ ● - Family Members with a History of Disease	
○ - Female Family Member	◻ ◯ - Deceased Family Member	
HD - Heart Disease	Str - Stroke	Dia - Diabetes
Col - Colon Cancer	BrC - Breast Cancer	OvC - Ovarian Cancer



# THE FUTURE

- The “virtual” visit
- The “online” family history
  - interactive software
  - “dynamic”
- The computerized pedigree
- The electronic medical record
- Risk assessment and stratification

# THE “VIRTUAL” VISIT

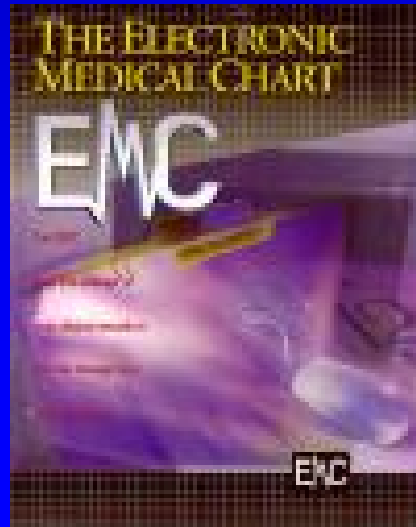
- Online family history questionnaire
- Construction of a 3 generation pedigree
- Risk stratification
- Available to primary care physician at visit

# The Virtual Visit – Step 2

- Risk assessment
- Online health education materials
  - e.g. weight management information
- Importation of established clinical guidelines
  - Amsterdam criteria for colorectal cancer
  - adult or pediatric cholesterol management guideline
- Can be downloaded to “PDA”

# Electronic Medical Record

- The electronic genetic family history
- Risk stratification
- Results to primary care physician
- High genetic risk cases to geneticist's "inbox"
- Reference materials for patient



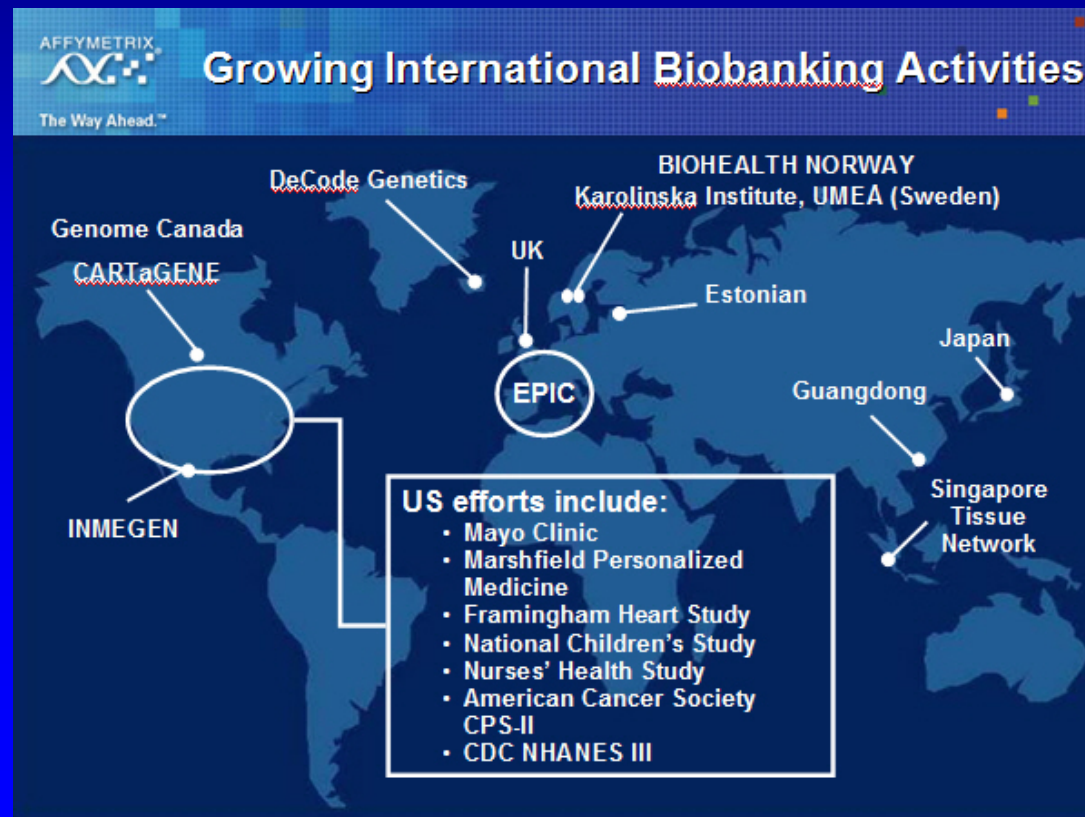
# The Future Future

- Integration of “SNP” map into family history
  - “SNP” = Single Nucleotide Polymorphism
- Integration of the “HapMap” into family history
  - “HapMap” = a catalog of human genetic variations
- Treatments: stem cells, gene therapy



# The Future Future Future Accelerating Personalized Medicine

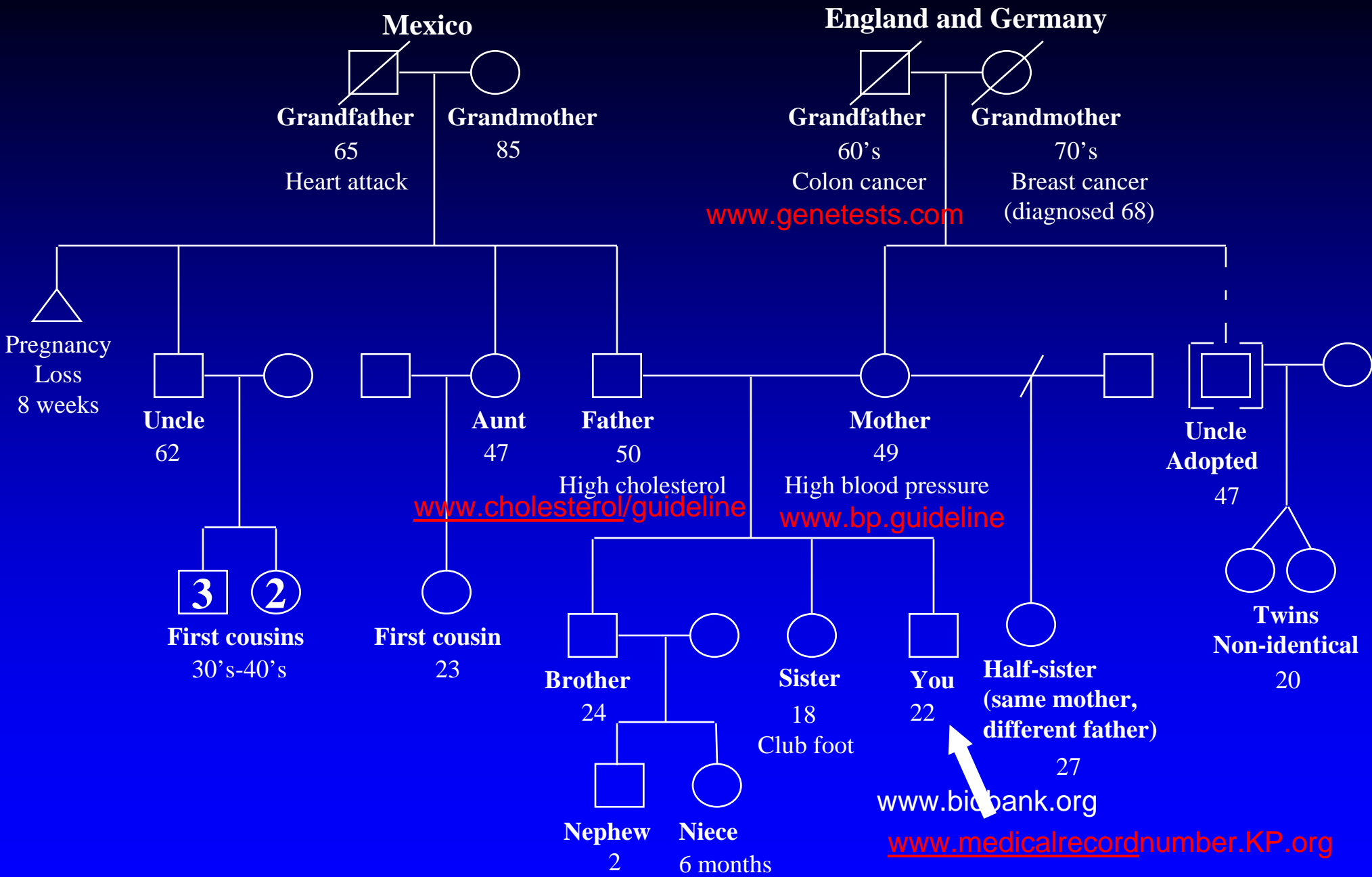
## ■ Integration of BioBanking data



# The “Dynamic” Family History

- Periodic family updates
- Linkage of medical reports of family members\*
- Linkage to laboratory studies of patient/family \*
- Linkage to imaging studies of patient/family \*
- Linkage to health education resources
- Linkage to established clinical guidelines
- (\* with appropriate permission/informed consent)

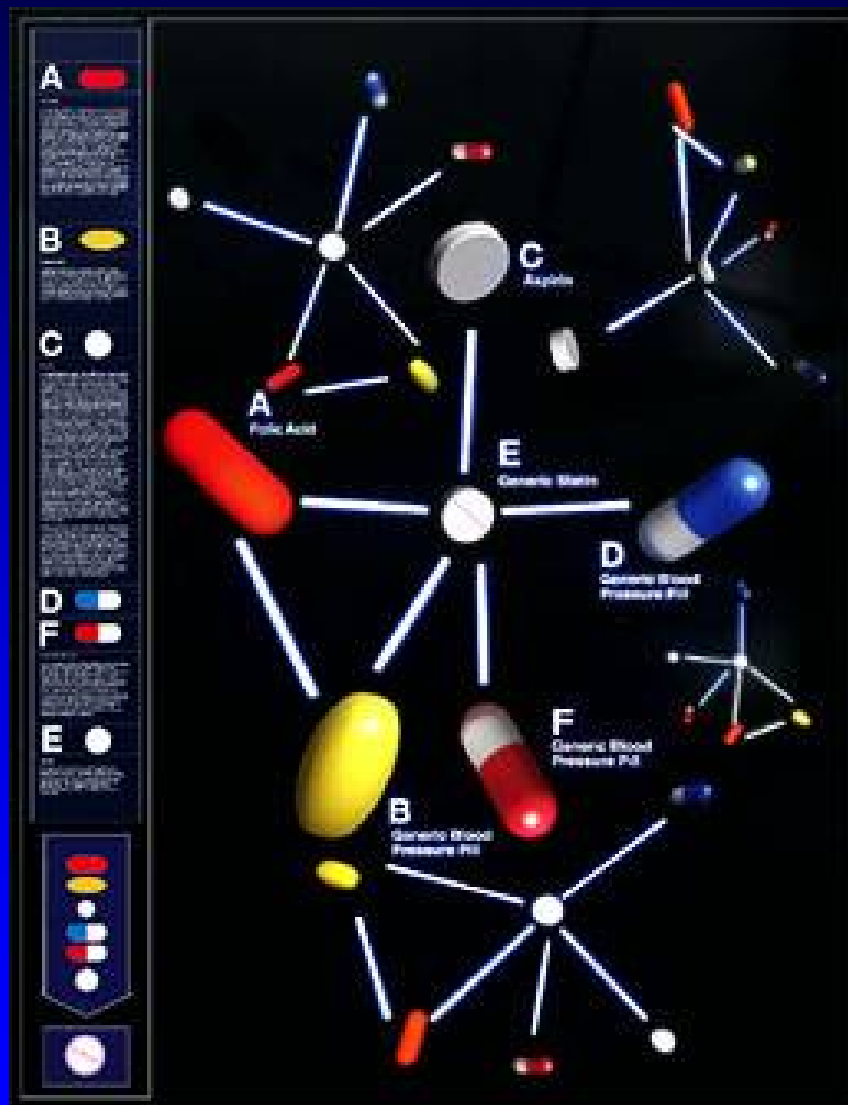




## The Dynamic Family History and Pedigree

# Treating the Family History: The “Polypill”

- A = Folic Acid 0.8 mg
  - B = Blood Pressure Pill (thiazide)
  - C = Aspirin 75 mg
  - D = Blood Pressure Pill (beta blocker)
  - E = Statin Drug (simvastatin 40 mg)
  - F = Blood Pressure Pill (ACE inhibitor)
- (all generic)



# The Polypill Prescription

- Folic Acid
  - lowers homocysteine, prevent birth defects
- Blood pressure reduction
  - prevent heart disease and stroke
- Aspirin
  - prevent heart attack, stroke
- Statin
  - prevent atherosclerotic heart disease

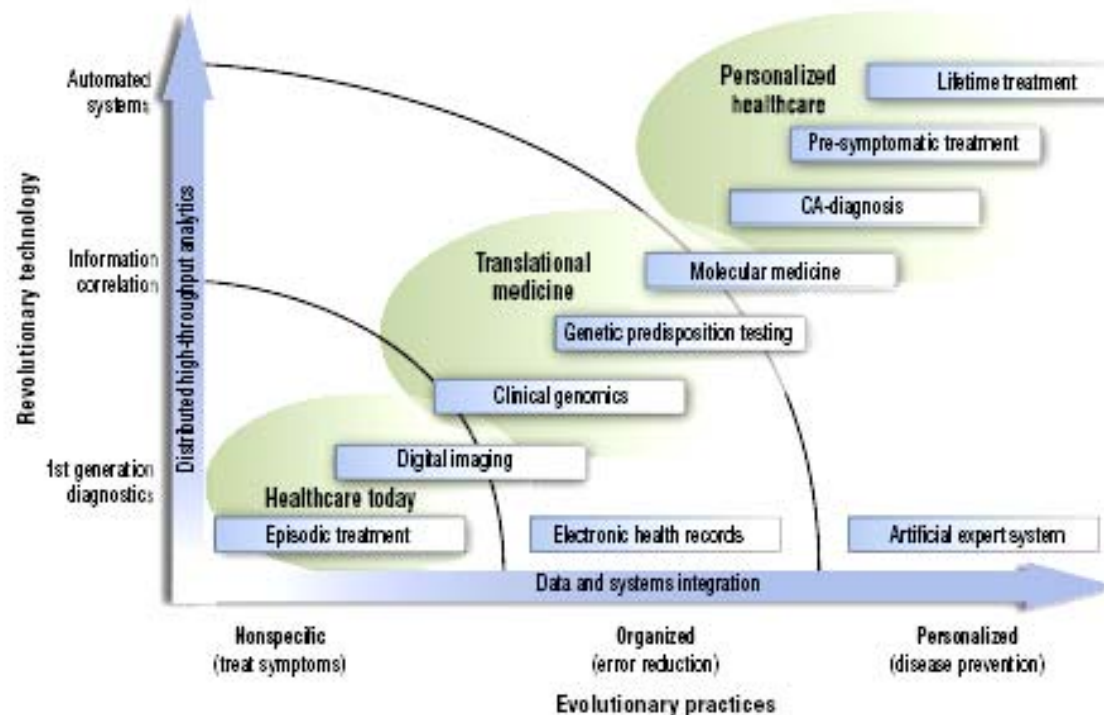
# The Polypill Predictions

- Reduces ischemic heart disease: 88%
- Reduces stroke: 80%
- These predictions apply if >55 years old or at higher risk for these disorders
  - personal or family history
- Adverse reactions to “Polypill”: 8-15%

(Wald, N J et al, A Strategy to reduce cardiovascular disease by more than 80%, BMJ, June 28, 2003)

# Market Trends and Drivers

Figure 1: Market trends and drivers: revolutionary technologies and evolutionary practices.



Source: IBM Life Sciences Solutions

# CONCLUSION

information and guidance that is based on the latest evidence-based medicine. As Rich et al. have written, "One can envision a future where portable electronic family history data integrated through electronic medical records to PDAs [personal digital assistants] and evidence-based practice guidelines could overcome current barriers to the thorough collection, accurate interpretation, and wise application of the family history in primary care practice."<sup>1</sup>

Rich EC, et al. J Gen Intern Med, 2004

# Family History Websites

- [www.hhs.gov/familyhistory/](http://www.hhs.gov/familyhistory/)
  - Surgeon General's online family history
- [www.nchpeg.org](http://www.nchpeg.org)
  - Family history newsletter
- <http://www.nsgc.org/consumer/familytree/>
  - National Society of Genetic Counselors