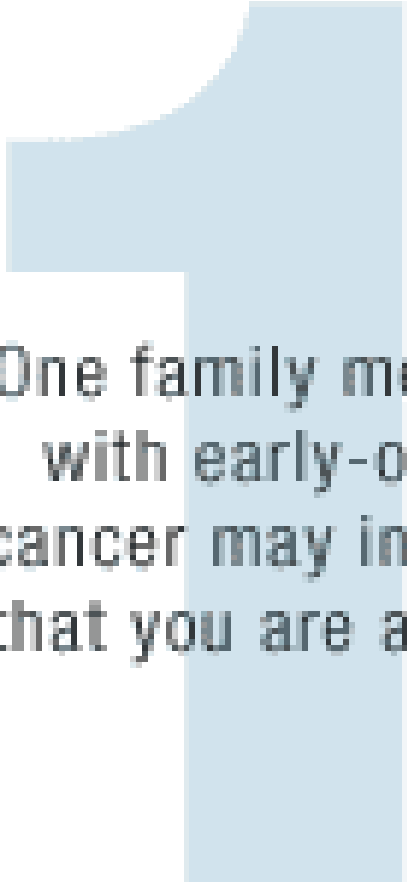


**ONE in  
300.**

The number of people  
with mutations that  
predispose them to  
breast, colon,  
endometrial (uterine) or  
ovarian cancer.



One family member  
with early-onset  
cancer may indicate  
that you are at risk.

# *Genetic Testing*

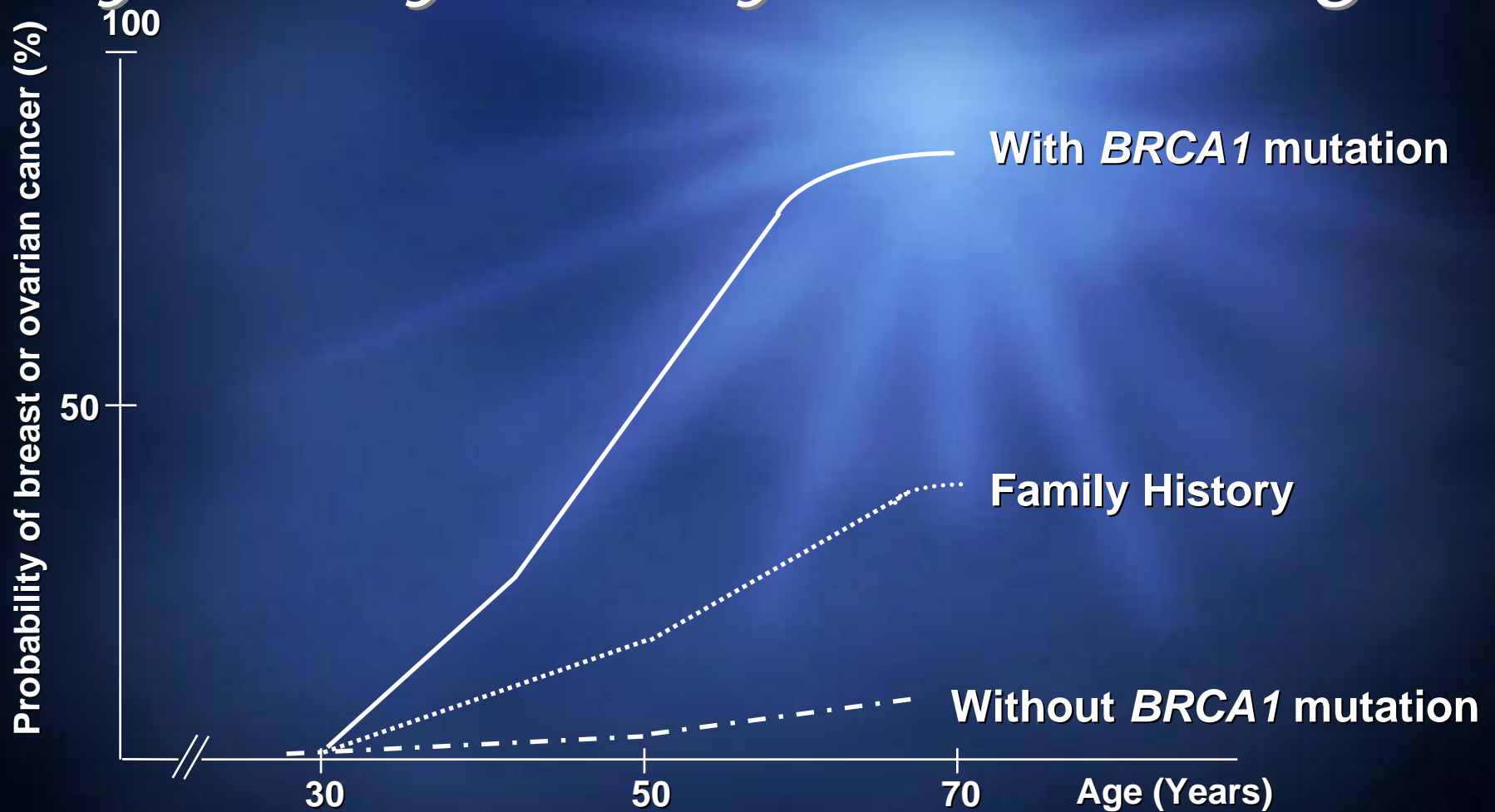
## ⊕ Benefits

- ⊕ Allows for individualized medical management
- ⊕ Accurate risk assessment
- ⊕ Covered by insurance
- ⊕ Alleviates uncertainty and anxiety

## ⊕ Limitations

- ⊕ Positives and true negatives are most informative results
- ⊕ Genetic testing for *BRCA1* and *BRCA2* does not identify all causes of HBOC

# Why Family History is Not Enough



Reprinted with permission from Ponder B: Genetic Testing for Cancer Risk. *Science* 1997; 278:1050-4.  
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# PRIOR TO GENETIC TESTING



NORMAL NORMAL



NORMAL MUTATED



50%

**Autosomal Dominant  
Not Sex Linked!**



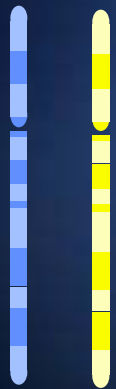
50%

# GENETIC TESTING



NORMAL NORMAL

NORMAL MUTATE  
D



NORMAL NORMAL

11%



NORMAL MUTATED

50-87%

# *Insurance Coverage of Genetic Testing*

- ⊕ Most insurers provide coverage for genetic testing
  - ⊕ On average, a patient will pay \$300 or less out-of-pocket
  
- ⊕ Established guidelines
  - ⊕ Medicare
  - ⊕ Most major carriers

# *Genetic Discrimination Myth versus Reality*

- ⊕ Federal and state laws prohibit the use of genetic information as a 'pre-existing condition'
  - ⊕ Federal HIPAA legislation
  - ⊕ The majority of states have additional laws
- ⊕ No well-documented cases of genetic discrimination
- ⊕ Over 50,000 clinical tests performed to date

# *“Genetic Discrimination” in Health Insurance is Illegal*

- ⊕ Health Insurance Portability and Accountability Act (HIPAA)
  - ⊕ Prohibits group health insurance plans from discriminating on the basis of genetic information.
- ⊕ Most states have enacted additional protections
- ⊕ Family members do not have to disclose whether a relative has undergone genetic testing

# *"The perceptions of discrimination far exceed the reality."*

*Genetic Testing 1:91-98, 1997*

- ✦ Follow-up study of women tested for hereditary breast and ovarian cancer risk found that "none has had medical insurance cancelled or insurance rates raised."  
✦ *Am Soc Clin Oncol Program/Proceedings 1999;18:611a*
- ✦ "Like so-called urban legends that are built on rumor rather than fact, the perception of insurance company bias against patients who undergo predictive genetic testing seems to be largely unsubstantiated."  
✦ *JAMA December 15 1999;282:2197-2198*
- ✦ "We find that there are almost no well-documented cases of health insurers either asking for or using presymptomatic genetic test results in their underwriting decisions."  
✦ *Am J Hum Genet 2000;66:293-307*

# Genetic Anti-Discrimination Bill Clears Congress

- On May 1, 2008, the House passed the Genetic Information Nondiscrimination Act (GINA) by a vote of 414 to 1.
- The act will protect Americans against discrimination based on their genetic information when it comes to health insurance and employment.
- The Senate unanimously approved the legislation a week prior to the House
- It's anticipated that President Bush will sign the bill soon.
- The long-awaited measure, which has been debated in Congress for 13 years, will pave the way for people to take full advantage of the promise of personalized medicine without fear of discrimination.

## *Red Flags for Hereditary Breast and Ovarian Cancer*

- ⊕ Breast cancer before age 50
- ⊕ Ovarian cancer at any age
- ⊕ Two or more cancers in the family involving breast and/or ovary
- ⊕ Male breast cancer
- ⊕ Bilateral Breast Cancer
- ⊕ Breast and ovarian cancer in the same person
- ⊕ Ashkenazi jewish ancestry

## *"Red Flags" for HNPCC*

- Early onset colorectal cancer (<50y)
- Early onset endometrial cancer (<50y)
- Two or more HNPCC cancers in an individual or family\*

\*HNPCC cancers: colorectal, endometrial, gastric, **ovarian**, ureter/renal pelvis, biliary tract, small bowel, pancreas, brain, sebaceous adenoma

# *Autosomal Dominant Inheritance* *not sex-linked!*

Father with  
mutation



OR



Mother  
With mutation



Each child has a **50%** chance of inheriting the mutation

# *Gene Therapy*

to alter a person's genetic material  
to fight or prevent disease

# *Cancer Gene Therapy*

- ⊕ replace missing or altered genes with healthy genes
- ⊕ to improve a patient's immune response to cancer.
- ⊕ To insert genes into cancer cells to make them more sensitive to chemotherapy, radiation therapy,
- ⊕ To makes normal cells more resistant to the side effects of high doses of anticancer drugs
- ⊕ to introduce "suicide genes" into cancer cells
- ⊕ to prevent cancer cells from developing new blood vessels (angiogenesis)

*Francis S. Collins, M.D., Ph.D.*  
*Director of the Human Genome Project*

"The Language of God:  
A Scientist Presents Evidence for Belief"

I see DNA .....as God's language!

DNA provides the strongest possible proof  
of our relatedness to all other living things

*Francis S. Collins, M.D., Ph.D.*  
*Director of the Human Genome Project*

I have found there is a wonderful harmony in the complementary truths of science and faith. **The God of the Bible is also the God of the genome.** God can be found in the cathedral or in the laboratory. By investigating God's majestic and awesome creation, **science** can actually be **a means of worship.**

*II Timothy 1:6 says:  
"for God did not give us a  
spirit of fear, but rather a  
spirit of power, and of love  
and a sound mind."*