The First Line of Defense:

Application of Breast Cancer Genomics Standards in Primary Care

Friday, Feb. 21, 2014 | The Lodge and Spa at Callaway Gardens

GEORGIA BREAST CANCER
GENOMIC HEALTH CONSORTIUM
EDUCATION SURVEILLANCE AND POLICY

Thank You!


DPH
Georgia Department of Public Health


CDC
CENTERS FOR DISEASE CONTROL AND PREVENTION


Georgia State University


MOREHOUSE
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RESEARCH & EDUCATION

MORE Continuing Education Credit Opportunities

Earn more credits with our MARCH 2014 WEBINAR on USPSTF Standards and Guidelines featuring Mark H. Ebell, MD, MS.*

More information is available at www.GeorgiaCORE.org.

*Dr. Ebell is an associate professor of epidemiology and biostatistics at The University of Georgia with a background in family medicine. His expertise and research interests include primary care research, point-of-care decision support, health information technology for the primary care setting, evidence-based medicine, and systematic reviews of screening and diagnostic tests. Dr. Ebell joined the U.S. Preventive Services Task Force in January 2012.

MORE Continuing Education Credit Opportunities

Are you interested in earning free CME credits?

Physicians can earn up to 2 AMA PRA Category 1 Credits for completing this course.

This case-based program is based on 3 patient scenarios, through which participants can navigate through various scenarios and outcomes of patients at risk for inherited cancer predisposition. It was developed for primary care providers including physicians, physician assistants and nurse practitioners, although the content is likely to be appropriate for other health professionals as well.

Visit the link below to access this free CME opportunity:

http://www.nchpeg.org/hboc/

Thank you to our attendees and presenters!

GEORGIA BREAST CANCER
GENOMIC HEALTH CONSORTIUM
EDUCATION SURVEILLANCE AND POLICY

Planning Committee

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Nancy M. Paris, MS, FACHE
Rachel Webster
Roland Matthews, MD
Victoria Green, MD, MHSA, MBA, JD

Our CME/CNE Providers

Continuing Medical Education

Jointly sponsored by the Morehouse School of Medicine and the Georgia Breast Cancer Genomic Health Consortium.

Continuing Nurses Education

This continuing nursing education activity was approved by the Georgia Nurses Association, an accredited approver by the American Nurses Credentialing Center's Commission on Accreditation.

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Karen Minyard, PhD
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MOREHOUSE SCHOOL OF MEDICINE

Roland Matthews, MD
Ijeoma Azonobi, MD

Family Health History

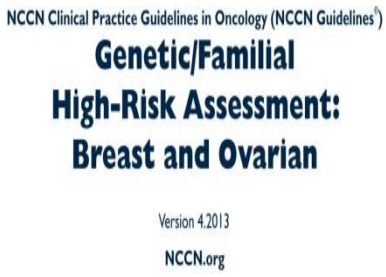
Genetics

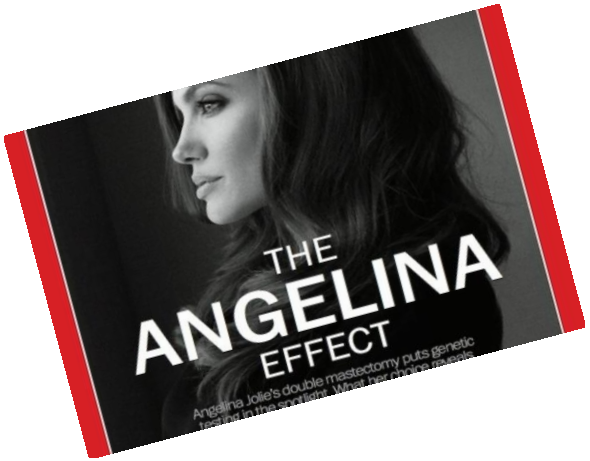
Lifestyle

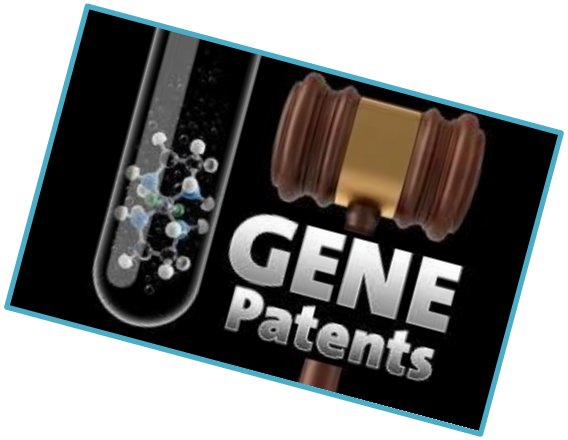
Environment

www.hhs.gov/familyhistory

Hereditary Risk Assessment Drivers of Change







The Emerging Role of Primary Care Providers in Cancer Genetics

Roland Matthews, MD
Professor and Chairman
Morehouse School of Medicine, Department of OB/GYN
Georgia Cancer Coalition Distinguished Cancer Scholar
Director, Georgia Cancer Center for Excellence at Grady Hospital

Faculty Disclosure

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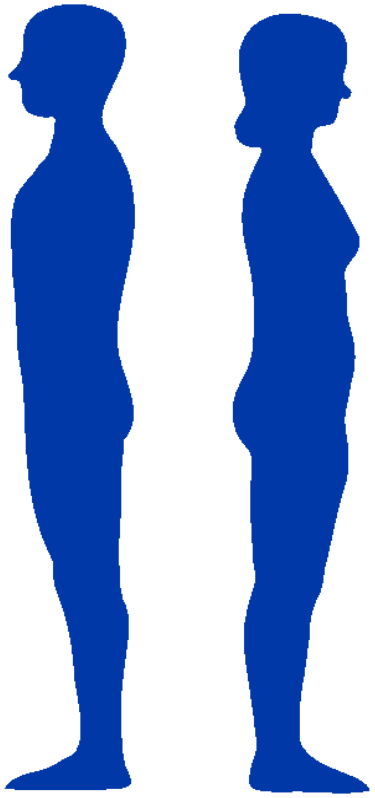
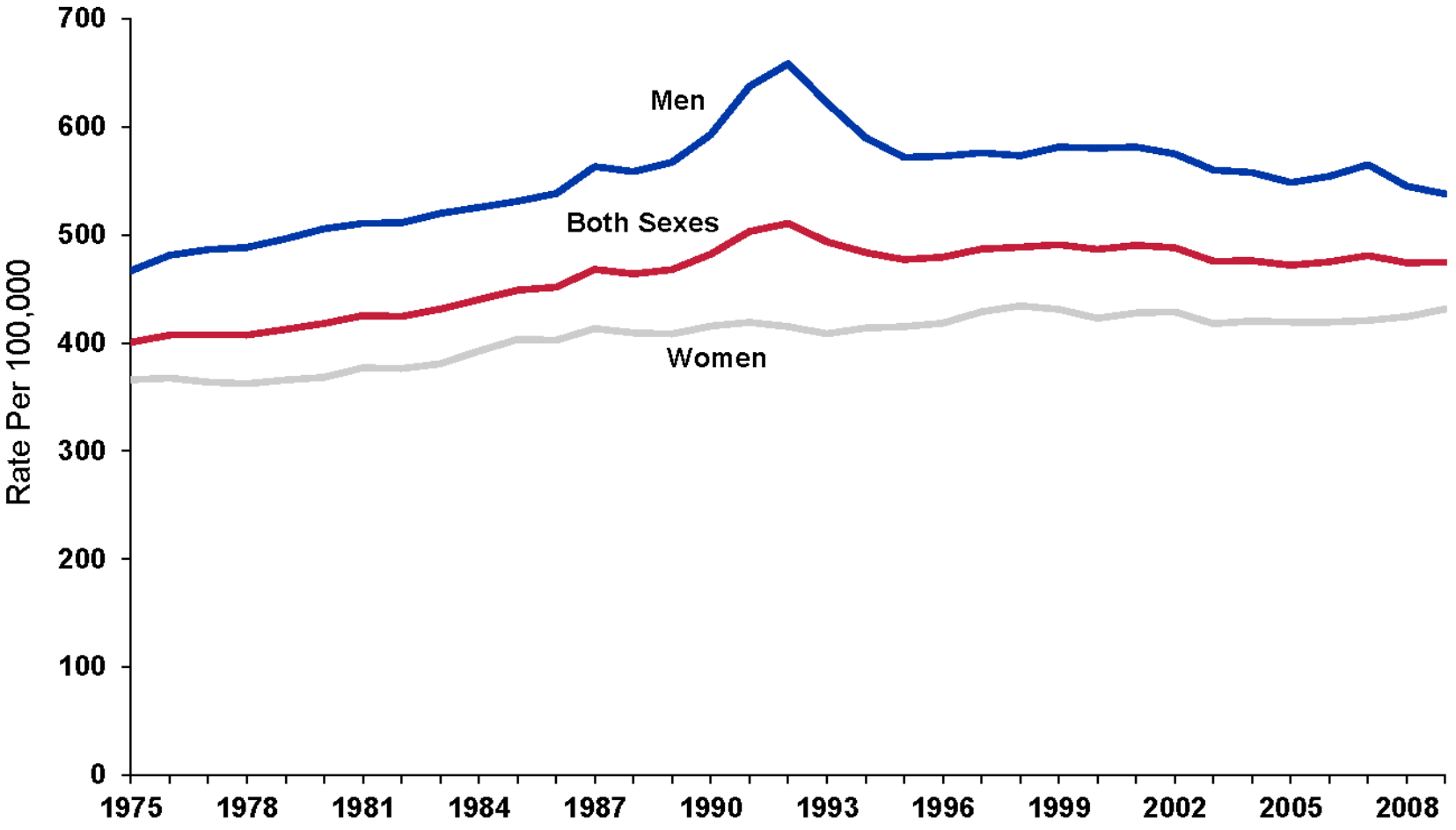
Estimated New Cancer Cases* in the US in 2013



Men		Women	
Prostate	28%	29%	Breast
Lung & bronchus	14%	14%	Lung & bronchus
Colon & rectum	9%	9%	Colon & rectum
Urinary bladder	6%	6%	Uterine corpus
Melanoma of skin	5%	6%	Thyroid
Kidney & renal pelvis	5%	4%	Non-Hodgkin lymphoma
Non-Hodgkin lymphoma	4%	4%	Melanoma of skin
Oral cavity	3%	3%	Kidney & renal pelvis
Leukemia	3%	3%	Pancreas
Pancreas	3%	3%	Ovary
All Other Sites	20%	19%	All Other Sites

*Excludes basal cell and squamous cell skin cancers and in situ carcinoma except urinary bladder.

Cancer Incidence Rates* by Sex, US, 1975-2009



*Age-adjusted to the 2000 US standard population and adjusted for delays in reporting.
Source: Surveillance, Epidemiology, and End Results Program, Delay-adjusted Incidence database: SEER Incidence Delay-adjusted Rates, 9 Registries, 1975-2009, National Cancer Institute, 2012.

The Lifetime Probability of Developing Cancer for Men, 2007-2009*

Site	Risk
All sites†	1 in 2
Prostate	1 in 6
Lung and bronchus	1 in 13
Colon and rectum	1 in 19
Urinary bladder‡	1 in 26
Melanoma §	1 in 35
Non-Hodgkin lymphoma	1 in 43
Kidney	1 in 49
Leukemia	1 in 63
Oral Cavity	1 in 66
Stomach	1 in 92

* For those free of cancer at beginning of age interval.

† All sites exclude basal and squamous cell skin cancers and in situ cancers except urinary bladder.

‡ Includes invasive and in situ cancer cases

§ Statistic for white men.

Source: DevCan: Probability of Developing or Dying of Cancer Software, Version 6.6.1 Statistical Research and Applications Branch, National Cancer Institute, 2012.

The Lifetime Probability of Developing Cancer for Women, 2007-2009*

Site	Risk
All sites†	1 in 3
Breast	1 in 8
Lung & bronchus	1 in 16
Colon & rectum	1 in 21
Uterine corpus	1 in 38
Non-Hodgkin lymphoma	1 in 52
Urinary bladder‡	1 in 87
Melanoma §	1 in 54
Ovary	1 in 72
Pancreas	1 in 69
Uterine cervix	1 in 147

* For those free of cancer at beginning of age interval.
† All sites exclude basal and squamous cell skin cancers and in situ cancers except urinary bladder.
‡ Includes invasive and in situ cancer cases
§ Statistic for white women.
Source: DevCan: Probability of Developing or Dying of Cancer Software, Version 6.6.1 Statistical Research and Applications Branch, National Cancer Institute, 2012.

Genetic Testing for Hereditary Cancer Syndromes

- Genetic mutations play a role in the development of all cancers.
- Inherited mutations play a major role in the development of about 5 to 10 percent of all cancers.
- The genetic mutations associated with more than 50 hereditary cancer syndromes have been identified.
- A genetic counselor, doctor, or other health care professional trained in genetics can help an individual or family understand genetic test results.

http://www.cancer.gov/

Cancer genetics in clinical practice

- Although less than 5% of cancers are linked to highly predisposing gene mutations, because of the high incidence of cancers one individual among 200 is affected by cancer genetics.
- Cancer genetics is one of the emblematic fields of personalized genomic medicine and is prone to take part in the renewed practices of genetic medicine.
- The management of high-risks individuals, still needs to be translated to clinical practice.

European Journal of Human Genetics (2013)
21, 120; doi:10.1038/ejhg.2011.69

Roles Of The Primary Care Provider

- Incorporating genetics into primary care involves a shift of paradigm in medical thinking.
- It involves thinking about the genetic makeup of each individual to develop differential diagnoses for disease or for preventive counseling.
- Primary care providers will need to be reeducated and updated on new knowledge in genetics.

"Primary Care Pediatrics: 2004 and Beyond"; Pediatrics, Vol. 113 No. 6 June 2004, pp. 1802-1809

The potential roles of primary care providers in genetic medicine can include:

- **Providing a medical home:** Individuals with complex genetic service needs can have a large care team that works together to meet the patient's needs.
- **Recognizing the special psychosocial issues**: For a family in which one or more members are affected with a genetic disorder or susceptibility primary care providers need to recognize and address relevant psychosocial issues.
- **Possessing knowledge of how to access the full range of genetics services from which patients might benefit**
- **Appropriately referring patients**: Primary care providers are in the position to know which patients with additional genetics services needs require referral and are able to refer appropriately.
- **Facilitating the use of genetics services.**

The potential roles of primary care providers in genetic medicine can include:

- **Identifying individuals who may benefit from genetic services**: These individuals include those with a genetic disorder, as well as those at increased risk for having or transmitting a genetic disorder.
- **Recognizing historical and physical features of common genetic conditions**
- **Monitoring the health of individuals with a genetic disorder**: Primary care providers in collaboration with appropriate subspecialists work to monitor the health of patients with a genetic disorder or those with an increased risk for having a genetic disorder.
- **Providing basic genetics information to patients and families**: By providing information regarding genetics to patients and families, primary care providers are able to help their understanding and informed decision making.

Cancer genetics in primary care

- Primary care physicians are in a unique position to apply advances in cancer genetics to the improved care of their patients.
- Attributes of family and personal history are the most significant indicators of an increased risk of cancer in the individual patient.
- Genetic testing can be used to further assess risk and guide strategies for cancer screening, prevention, and treatment..

J Nutr. 2003 Nov;133(11 Suppl 1):3767S-3772S

The primary care physician role in cancer genetics: a qualitative study of patient experience

- Patients expected PCPs to play a role in referral for genetic testing
- They hoped that PCPs would have sufficient knowledge to appreciate familial risk and supportive attitudes towards genetic testing
- Patients had more difficulty in identifying a PCP role following receipt of genetic test results
- Cancer patients in particular emphasized this as a role for cancer specialists
- Still, some patients anticipated an ongoing PCP role comprising risk-appropriate surveillance or reassurance, especially as specialist care diminished.

Family Practice (2010) 27 (5): 563-569.

The primary care physician role in cancer genetics: a qualitative study of patient experience

- The potential PCP role in cancer genetics is quite broad
- Patients expect PCPs to play a role in risk identification and genetics referral
- In addition, some patients anticipated an ongoing role for their PCPs after receiving genetic test results
- Sustained efforts will be needed to support PCPs in this expansive

Family Practice (2010) 27 (5): 563-569.

What's the future?

Without question, genetic testing will play a larger role in cancer risk assessment, detection, and treatment in the future.

Thank you

HBOC Background, Family History Red Flags and Clinical Guidelines

Victoria L. Green, MD, JD, MBA
Associate Professor
Emory University
Atlanta, GA

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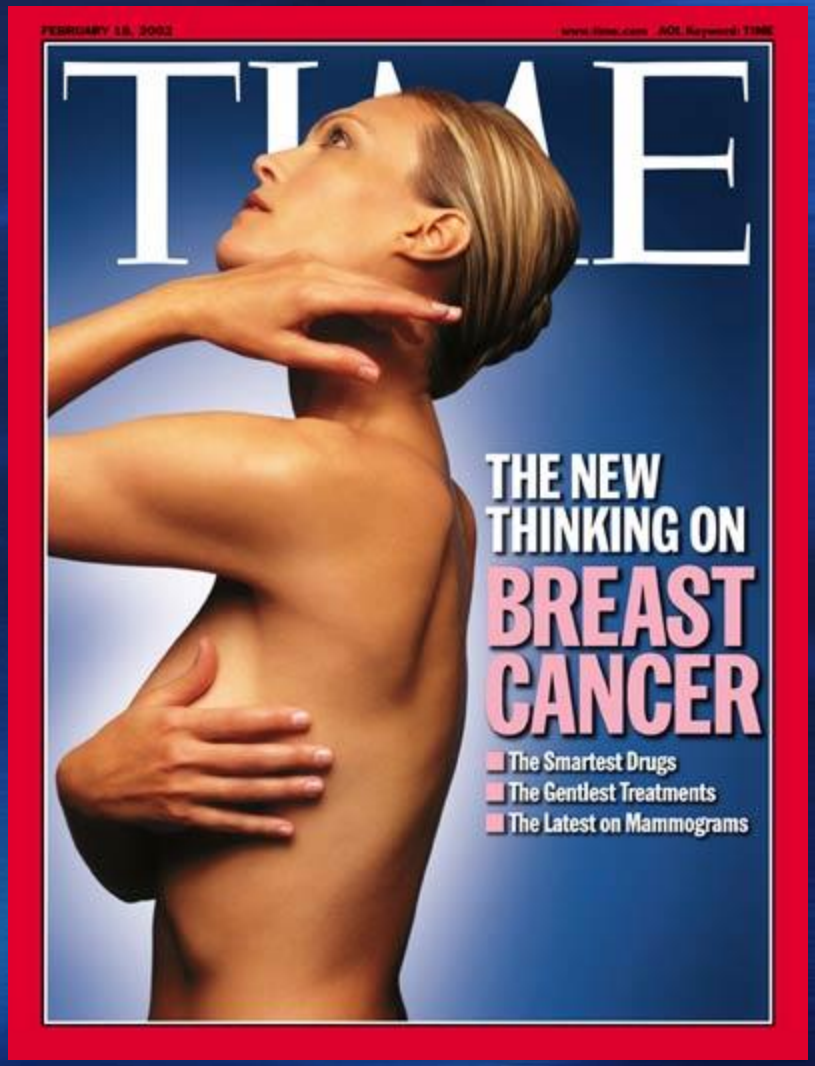
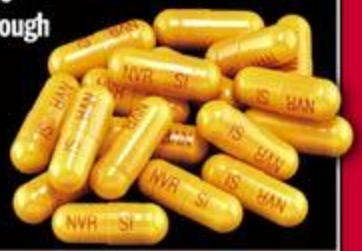

FEBRUARY 18, 2002
TIME
THE NEW THINKING ON
BREAST CANCER
The Smartest Drugs
The Gentlest Treatments
The Latest on Mammograms

MAY 28, 2001 www.time.com AOL Keyword: TIME

TIME

THERE IS NEW AMMUNITION IN THE WAR AGAINST CANCER. THESE ARE THE BULLETS.

Revolutionary new pills like GLEEVEC combat cancer by targeting only the diseased cells. Is this the breakthrough we've been waiting for?



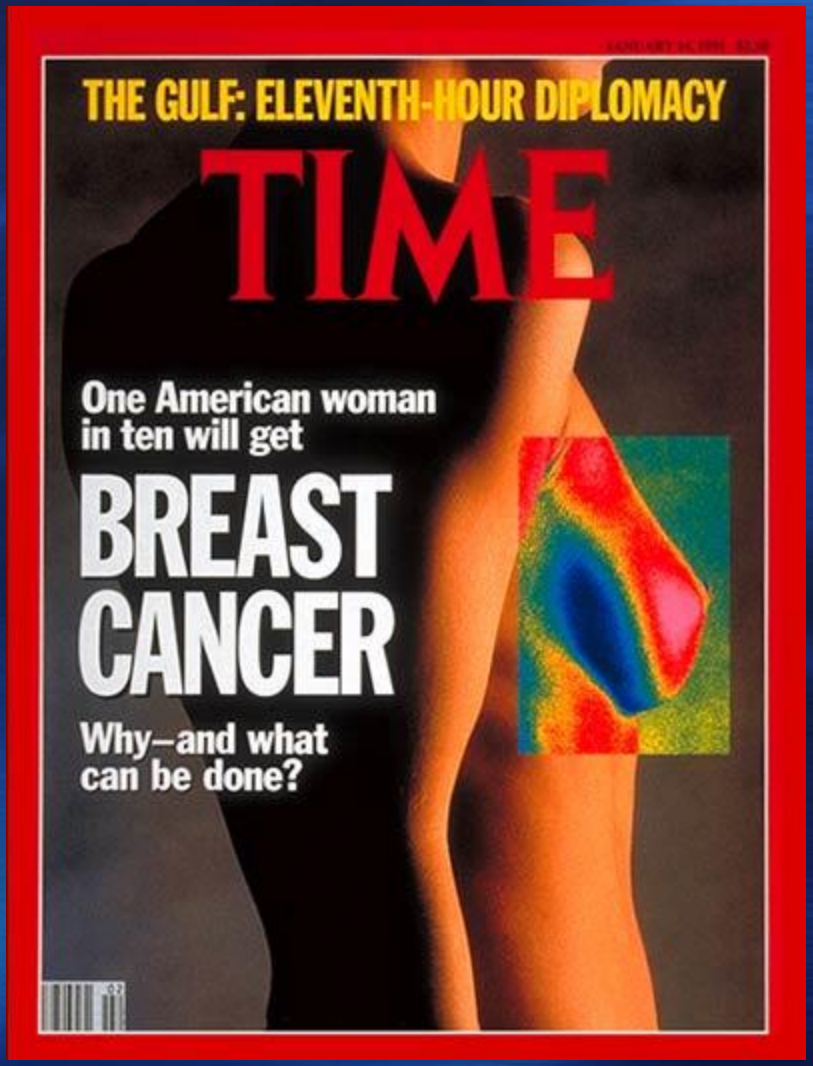
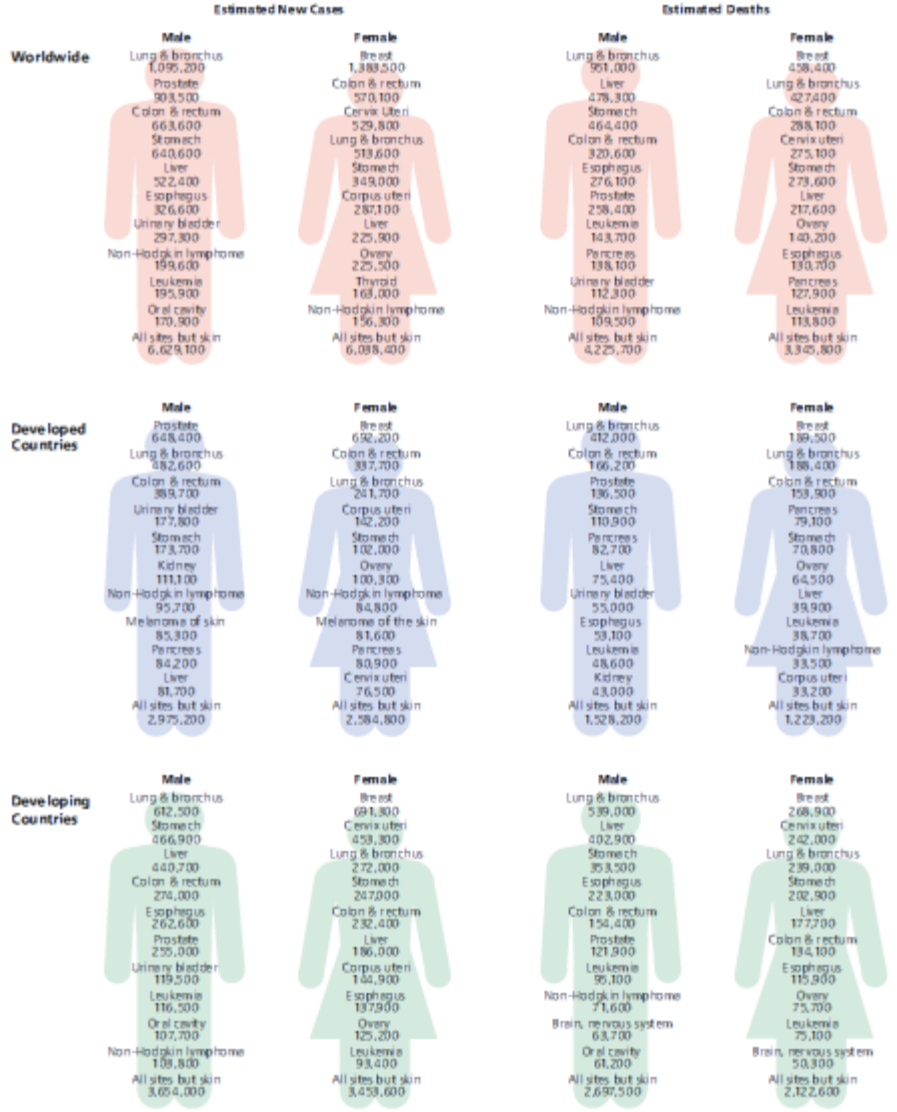

THE GULF: ELEVENTH-HOUR DIPLOMACY
TIME
One American woman in ten will get
BREAST CANCER
Why—and what can be done?

Figure 1. Estimated New Cancer Cases and Deaths Worldwide for Leading Cancer Sites by Level of Economic Development, 2008



Source: Globocan 2008

Leading New Cancer Cases and Deaths – 2012 Estimates

Estimated New Cases*

Male	Female
Prostate 241,740 (29%)	Breast 226,870 (29%)
Lung & bronchus 116,470 (14%)	Lung & bronchus 109,690 (14%)
Colon & rectum 73,420 (9%)	Colon & rectum 70,040 (9%)
Urinary bladder 55,600 (7%)	Uterine corpus 47,130 (6%)
Melanoma of the skin 44,250 (5%)	Thyroid 43,210 (5%)
Kidney & renal pelvis 40,250 (5%)	Melanoma of the skin 32,000 (4%)
Non-Hodgkin lymphoma 38,160 (4%)	Non-Hodgkin lymphoma 31,970 (4%)
Oral cavity & pharynx 28,540 (3%)	Kidney & renal pelvis 24,520 (3%)
Leukemia 26,830 (3%)	Ovary 22,280 (3%)
Pancreas 22,090 (3%)	Pancreas 21,830 (3%)
All sites 848,170 (100%)	All sites 790,740 (100%)

Estimated Deaths

Male	Female
Lung & bronchus 87,750 (29%)	Lung & bronchus 72,590 (26%)
Prostate 28,170 (9%)	Breast 39,510 (14%)
Colon & rectum 26,470 (9%)	Colon & rectum 25,220 (9%)
Pancreas 18,850 (6%)	Pancreas 18,540 (7%)
Liver & intrahepatic bile duct 13,980 (5%)	Ovary 15,500 (6%)
Leukemia 13,500 (4%)	Leukemia 10,040 (4%)
Esophagus 12,040 (4%)	Non-Hodgkin lymphoma 8,620 (3%)
Urinary bladder 10,510 (3%)	Uterine corpus 8,010 (3%)
Non-Hodgkin lymphoma 10,320 (3%)	Liver & intrahepatic bile duct 6,570 (2%)
Kidney & renal pelvis 8,650 (3%)	Brain & other nervous system 5,980 (2%)
All sites 301,820 (100%)	All sites 275,370 (100%)

*Excludes basal and squamous cell skin cancers and in situ carcinoma except urinary bladder.

1120 Georgia

15%

4017 California

2460 Texas

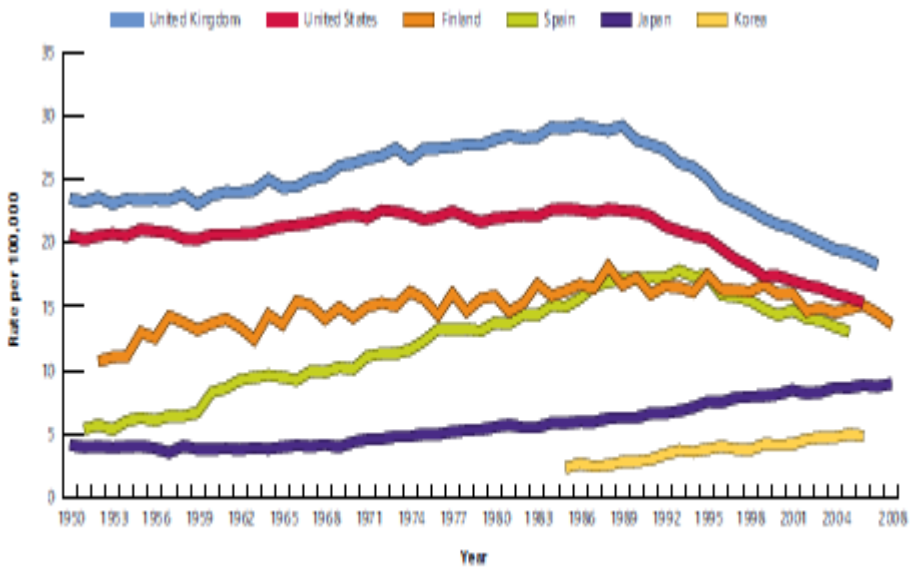
2760 New York

Press Room

Cancer Deaths Drop for Second Consecutive Year

Atlanta 2007/01/17 -An American Cancer Society report shows there was a drop of 3,014 cancer deaths in the United States from 2003 to 2004, the most recent year for which mortality data are available fom the National Center for Health Statistics. This drop was significantly larger than the 369 fewer deaths reported for the previous time period (2002 to 2003), which itself marked the first decline in actual number of cancer deaths in the more than 70 years since nationwide data began to be compiled.

Figure 5. Trends in Age-standardized Female Breast Cancer Death Rates in Select Countries



Source: WHO Mortality Database

Cancer Incidence and Death Rates* by Site, Race, and Ethnicity†, US, 2004-2008

Incidence	White	African American	Asian American or Pacific Islander	American Indian or Alaska Native‡	Hispanic/ Latino
All sites					
Male	545.0	626.2	332.4	427.8	423.4
Female	420.8	394.2	284.0	362.1	333.5
Breast (female)	122.3	116.1	84.9	89.2	92.3
Colon & rectum					
Male	54.6	66.9	42.4	51.5	48.6
Female	40.3	49.7	32.7	41.5	34.2
Kidney & renal pelvis					
Male	20.8	22.6	9.9	27.4	19.4
Female	10.9	11.7	4.9	16.8	11.2
Liver & intrahepatic bile duct					
Male	8.6	14.1	21.7	15.8	17.0
Female	2.9	4.0	8.2	7.6	6.4
Lung & bronchus					
Male	83.7	102.7	49.8	71.0	46.8
Female	57.2	51.4	28.1	51.7	27.0
Prostate	142.8	230.8	79.7	101.2	126.7
Stomach					
Male	8.5	16.4	16.8	13.9	13.8
Female	4.0	8.2	9.4	6.8	8.4
Uterine cervix	7.7	10.6	7.4	9.8	12.2

Mortality	White	African American	Asian American and Pacific Islander	American Indian and Alaska Native‡	Hispanic/ Latino
All sites					
Male	222.0	295.3	134.7	190.0	149.1
Female	152.8	177.7	94.1	138.4	101.5
Breast (female)	22.8	32.0	12.2	17.2	15.1
Colon & rectum					
Male	20.1	30.5	13.3	19.8	15.5
Female	14.0	20.4	9.9	14.0	10.3
Kidney & renal pelvis					
Male	6.0	6.0	2.6	8.9	5.2
Female	2.7	2.6	1.2	4.1	2.3
Liver & intrahepatic bile duct					
Male	7.2	11.5	14.7	11.9	11.6
Female	3.0	3.9	6.3	6.7	5.2
Lung & bronchus					
Male	66.9	85.4	36.7	50.5	31.9
Female	41.2	38.8	18.5	33.9	14.3
Prostate	22.4	54.9	10.5	20.7	18.5
Stomach					
Male	4.5	10.7	9.2	8.5	7.7
Female	2.3	5.0	5.4	3.9	4.5
Uterine cervix	2.2	4.3	2.1	3.4	3.1

*Per 100,000, age adjusted to the 2000 US standard population.

†Race and ethnicity categories are not mutually exclusive; persons of Hispanic origin may be of any race.

‡Data based on Contract Health Service Delivery Areas, comprising about 55% of the US American Indian/Alaska Native population; for more information, please see: Espey DK, Wu XC, Swan J, et al. Annual report to the nation on the status of cancer, 1975-2004, featuring cancer in American Indians and Alaska Natives.

Source: Incidence: NAACCR, 2011. Data are collected by cancer registries participating in the National Cancer Institute's SEER program and the Centers for Disease Control and Prevention's National Program of Cancer Registries. Mortality: National Center for Health Statistics 2011.

American Cancer Society Surveillance Research, 2012

Cancer Survival*(%) by Race,1996-2003

Site	White	African American	Absolute Difference
All Sites	67	57	10
Breast (female)	90	78	12
Colon	66	55	11
Esophagus	18	11	7
Leukemia	51	40	11
Non-Hodgkin lymphoma	65	56	9
Oral cavity	62	41	21
Prostate	99	95	4
Rectum	66	58	8
Urinary bladder	81	65	16
Uterine cervix	74	66	8
Uterine corpus	86	61	25

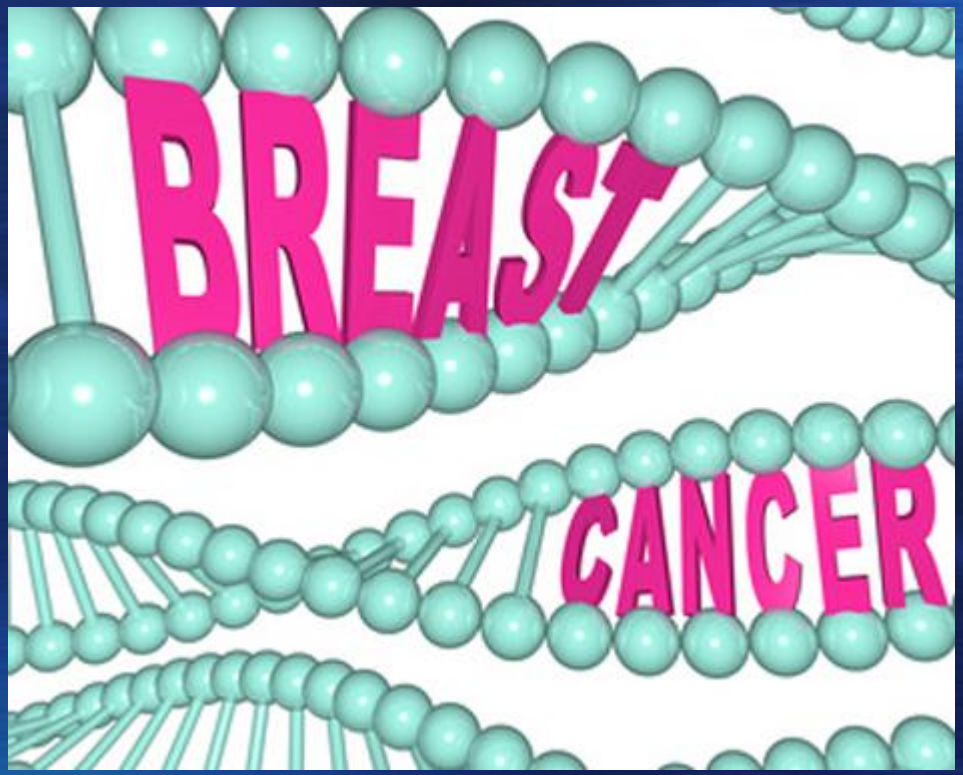
***5-year relative survival rates based on cancer patients diagnosed from 1996 to 2003 and followed through 2004. Source: Surveillance, Epidemiology, and End Results Program, 1975-2004, Division of Cancer Control and Population Sciences, National Cancer Institute, 2007.**





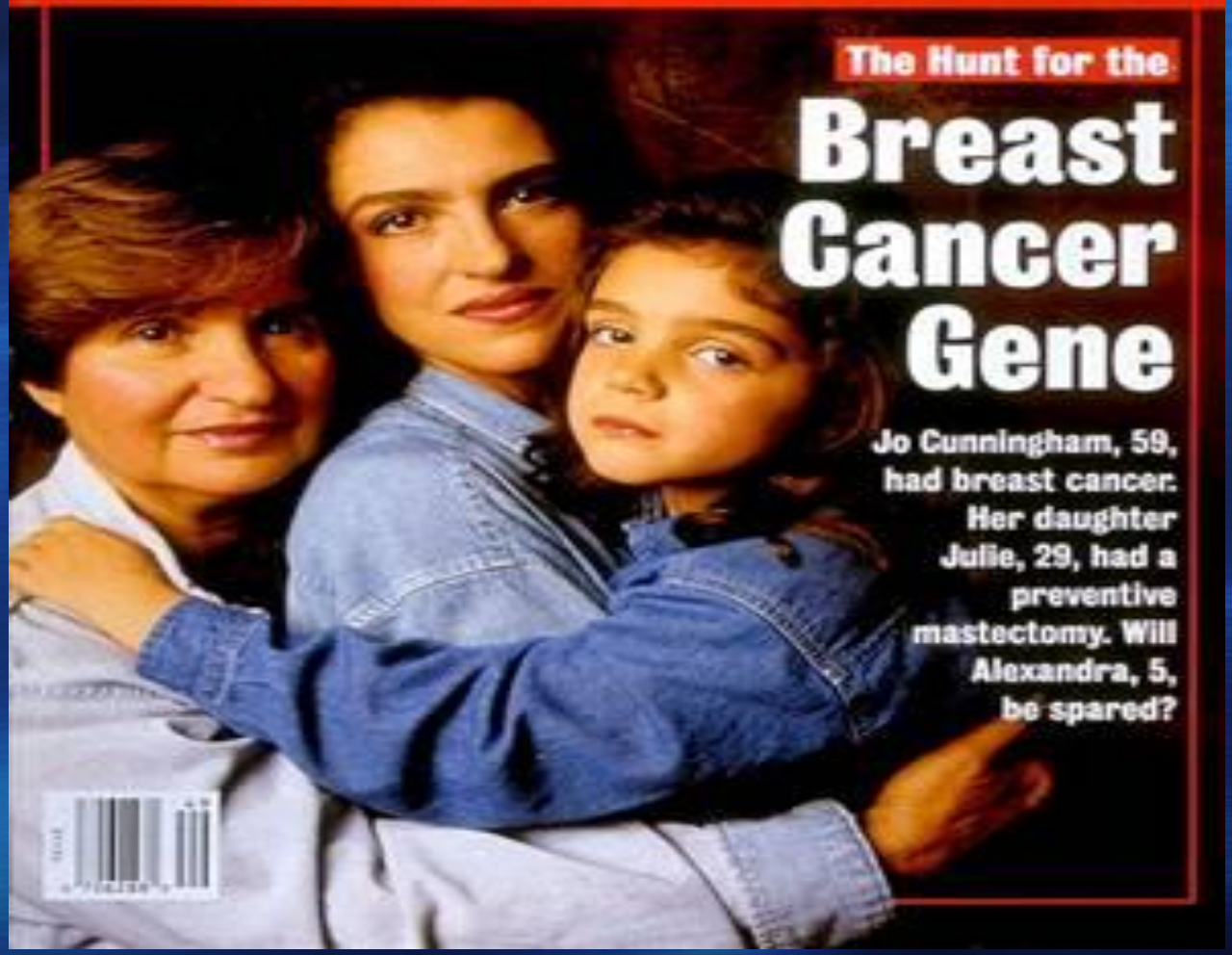

what's my risk
for cancer?

GEN Genetic Engineering & Biotechnology News



RETIREMENT BLUES: HOW SAFE IS YOUR PENSION?

Newsweek



The Hunt for the

Breast Cancer Gene

Jo Cunningham, 59, had breast cancer. Her daughter Julie, 29, had a preventive mastectomy. Will Alexandra, 5, be spared?

CNN Entertainment

Angelina Jolie undergoes double mastectomy

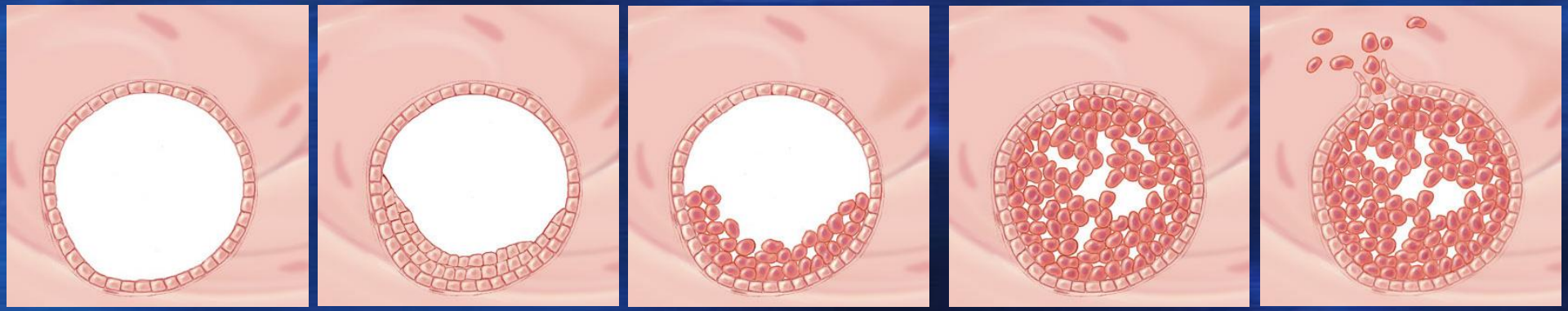
May 16, 2013

Objectives

- Discuss identification of women at high risk for breast cancer
- Review important aspects of the family history
- Become familiar with national guidelines for genetic referral and/or testing
- Review current status of risk assessment

A New Breast Cancer Paradigm

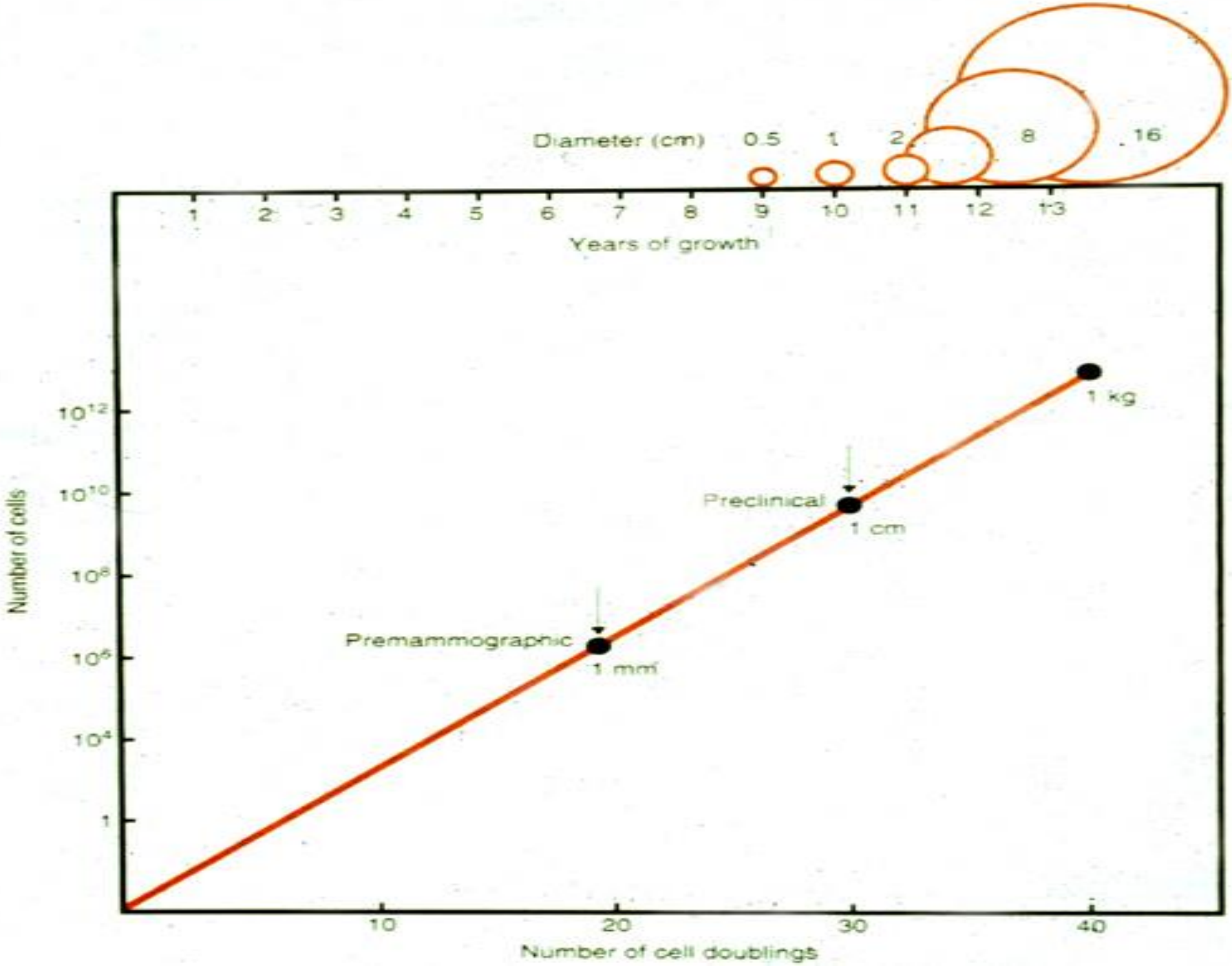
The goal: Identify women at highest risk so they can be targeted for a proactive risk management strategy.



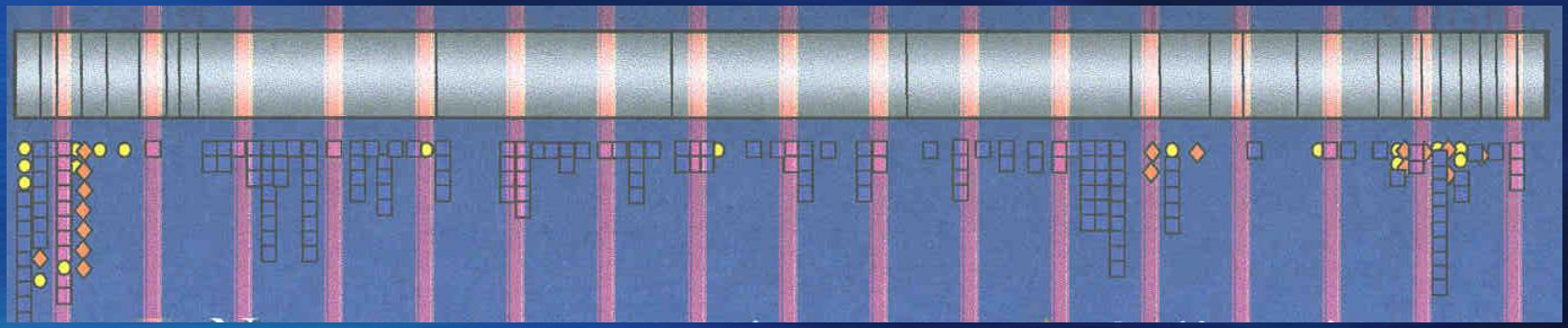
Normal Duct | **Intraductal Hyperplasia** | **Atypical Ductal Hyperplasia** | **Ductal Carcinoma *In Situ*** | **Invasive Ductal Carcinoma**

Predict and Prevent

Detect and Treat

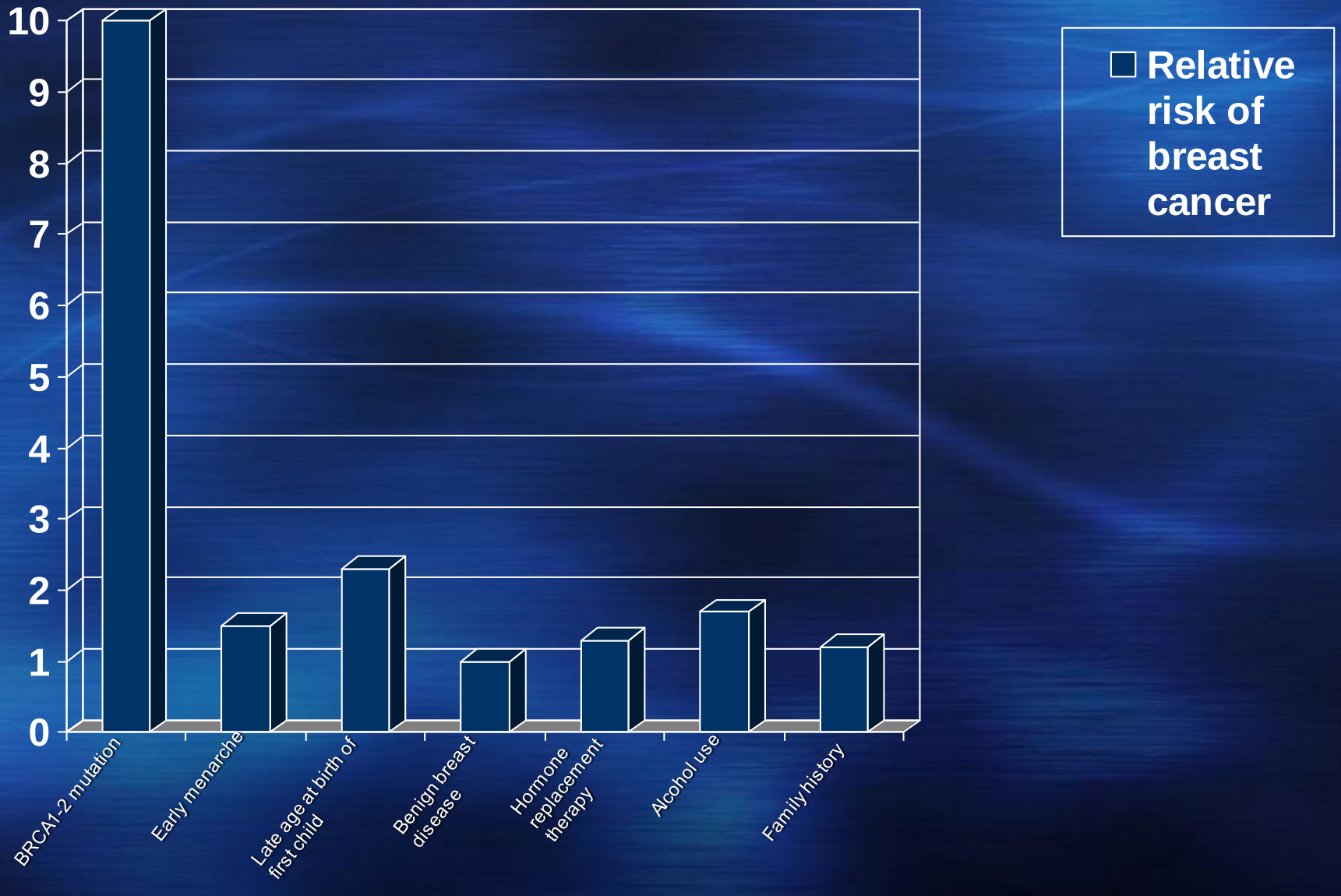

Diameter (cm)
0.5
1
2
8
16
1
2
3
4
5
6
7
8
9
10
11
12
13
Years of growth
10^{12}
10^{10}
10^{8}
10^{6}
10^{4}
1
Number of cells
1 kg
Preclinical
1 cm
Premammographic
1 mm
10
20
30
40
Number of cell doublings

Breast Cancer Susceptibility Genes



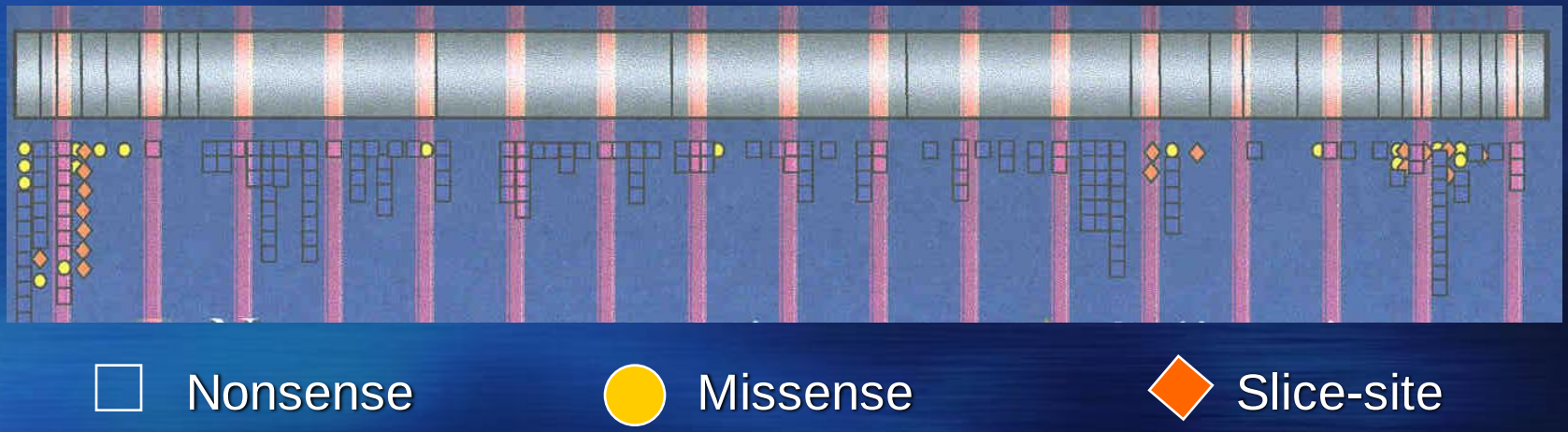
Breast Cancer Information Core

BRCA1-2 Mutations Increase the Risk of Cancer More than Other Factors



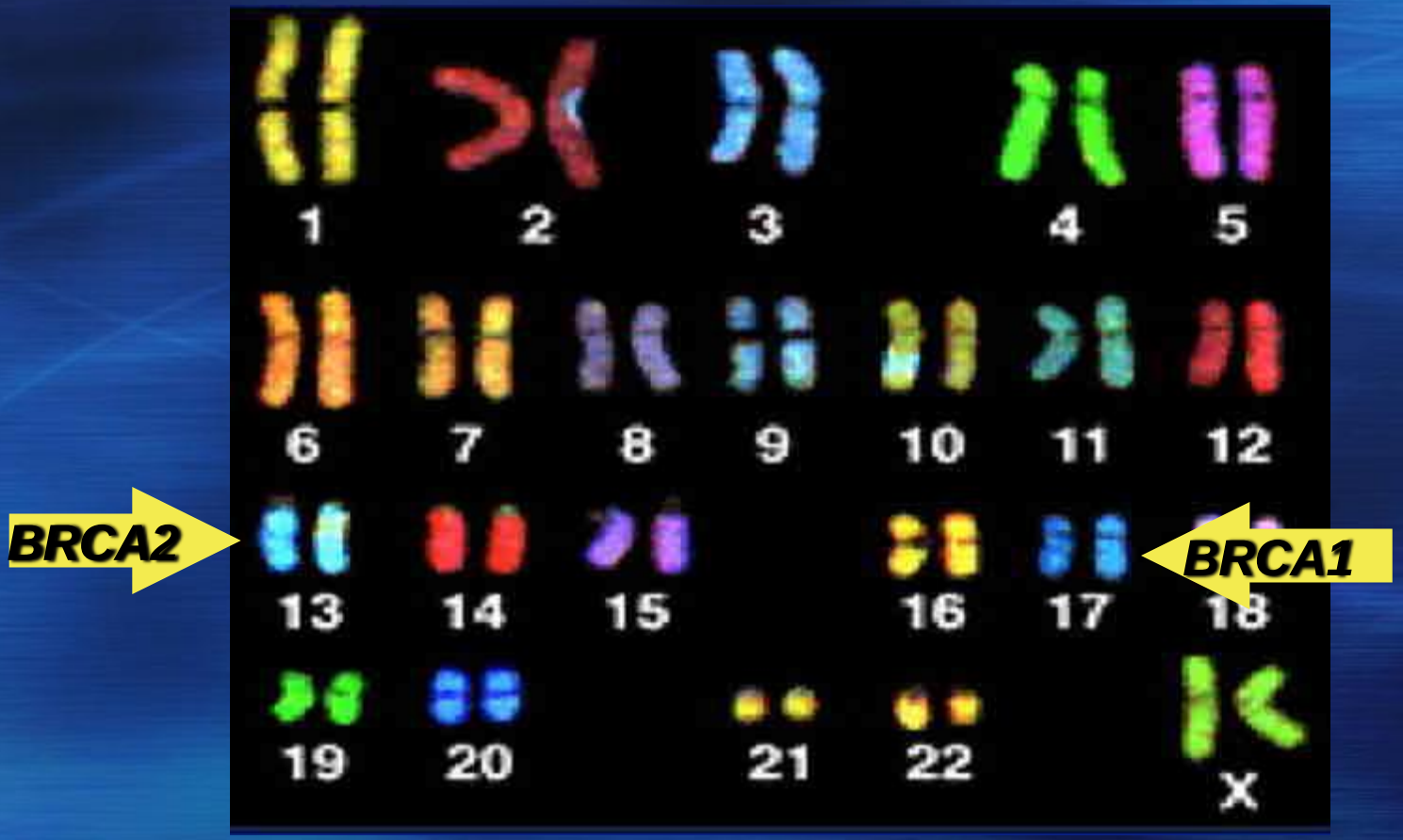
BRCA 1 and 2

- Tumor suppressor gene on chromosome 17,13
- Autosomal dominant transmission
- Protein has role in genomic stability
- ~500 different mutations reported



Breast Cancer Information Core

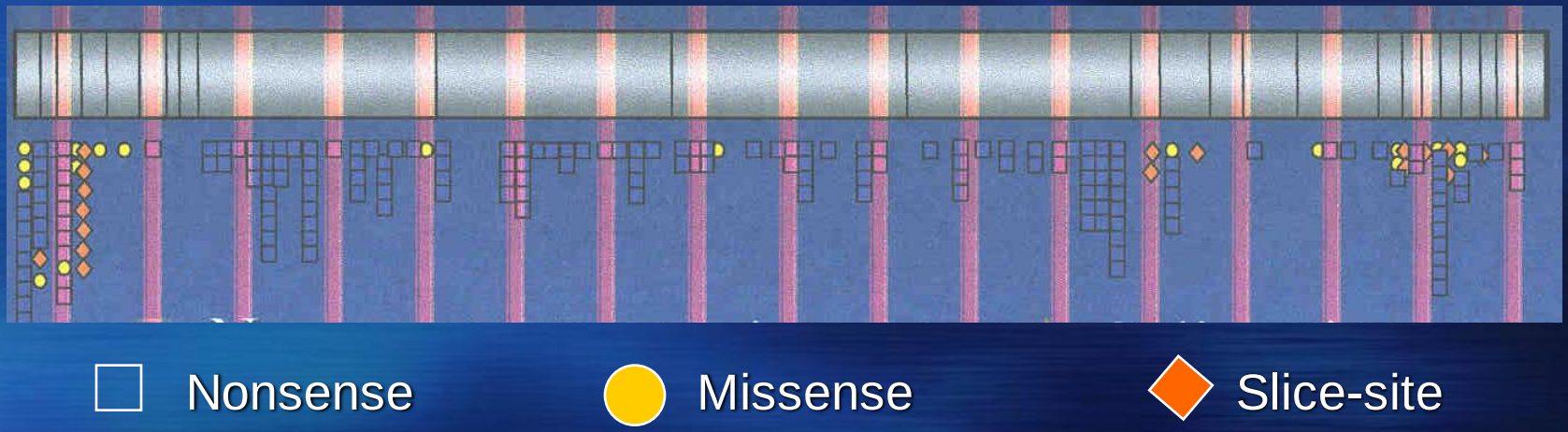
Cells have Two Copies of BRCA1 and BRCA2



Adapted from *Tools for Understanding Genetics*
National Human Genome Research Institute
Office of Science Education and Outreach
www.nhgri.nih.gov/DIR/VIP

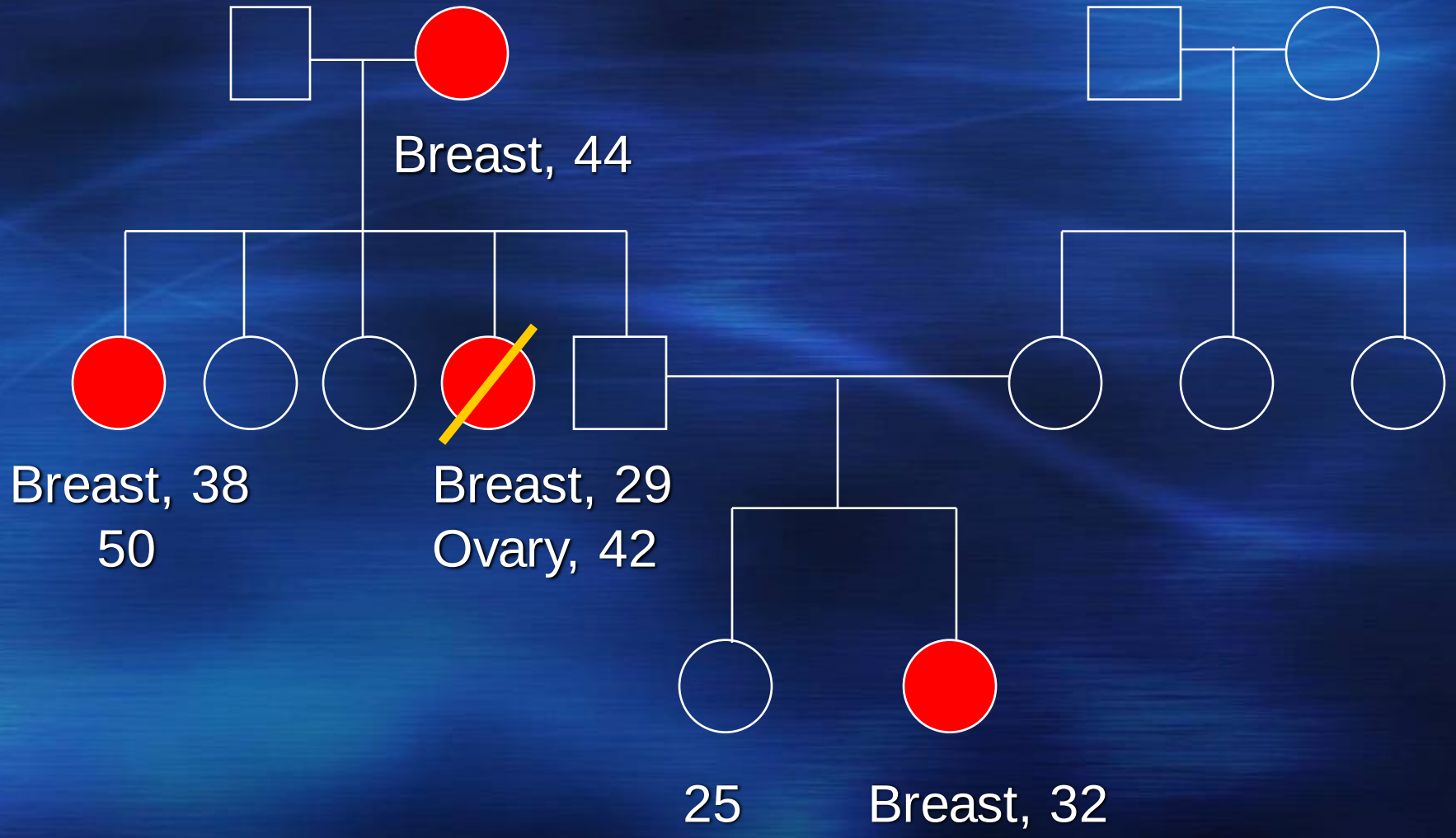
BRCA 1 and 2

- Tumor suppressor gene on chromosome 17,13
- Autosomal dominant transmission
- Protein has role in genomic stability
- ~500 different mutations reported



Breast Cancer Information Core

Example of BRCA1/2 Pedigree



BRCA 1 and 2

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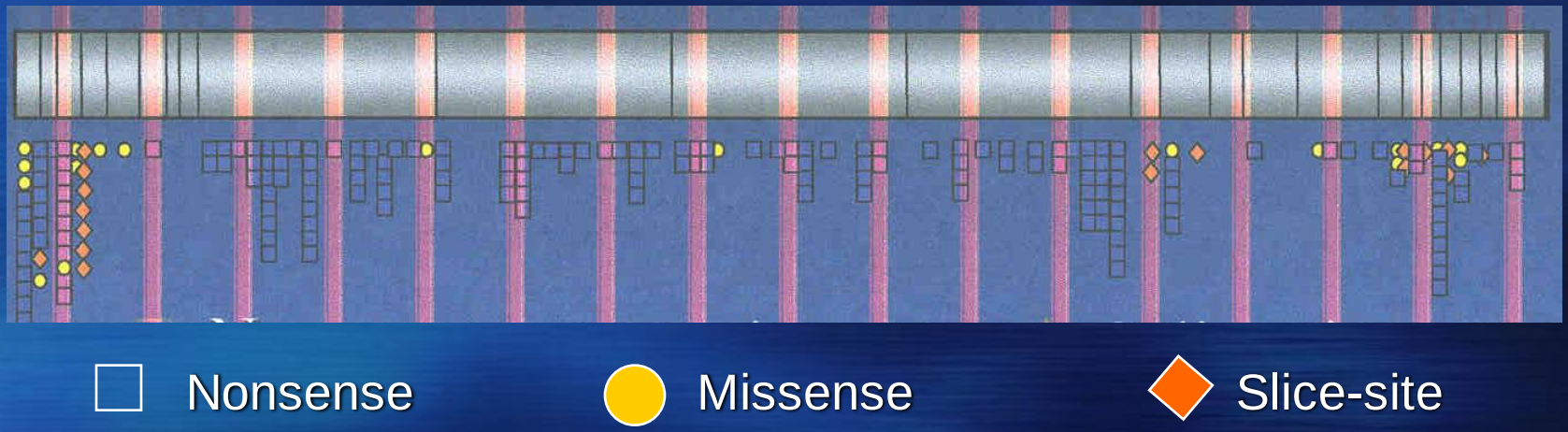
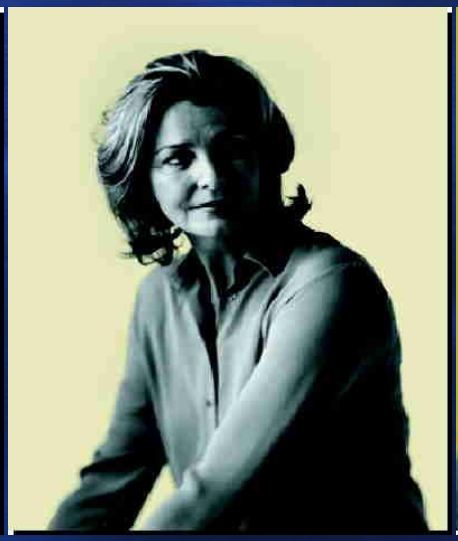
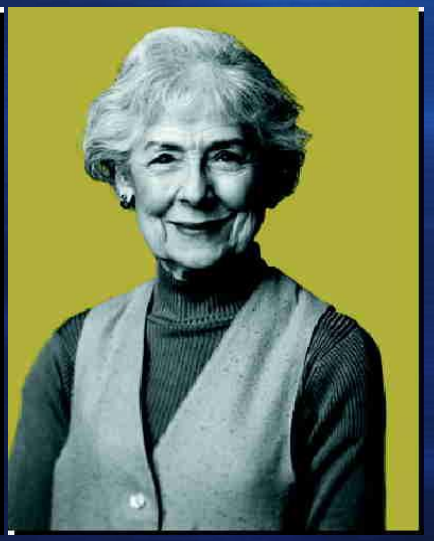


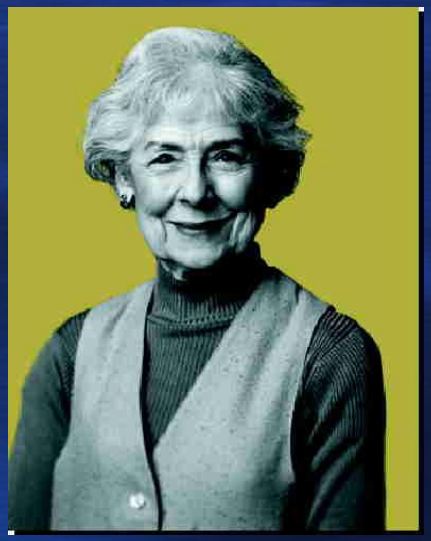
Breast Cancer Information Core

BRCA1-2 Mutations Increase the Risk of Early-Onset Breast Cancer

	By age 40	By age 50	By age 70
			
Population Risk	0.5%	2%	7%
Hereditary Risk	10%-20%	33%-50%	56%-87%

BRCA1-2 Mutations Increase the Risk of Ovarian Cancer

By age 70



Population Risk	1%
Hereditary Risk	**~44% (*BRCA1*)**
	27% (*BRCA2*)

The women's health provider may be the only healthcare practitioner that a woman sees during her reproductive years. This allows us to:

- Identify those at increased risk
- Refer for genetic counseling
- Follow closely for cancer surveillance

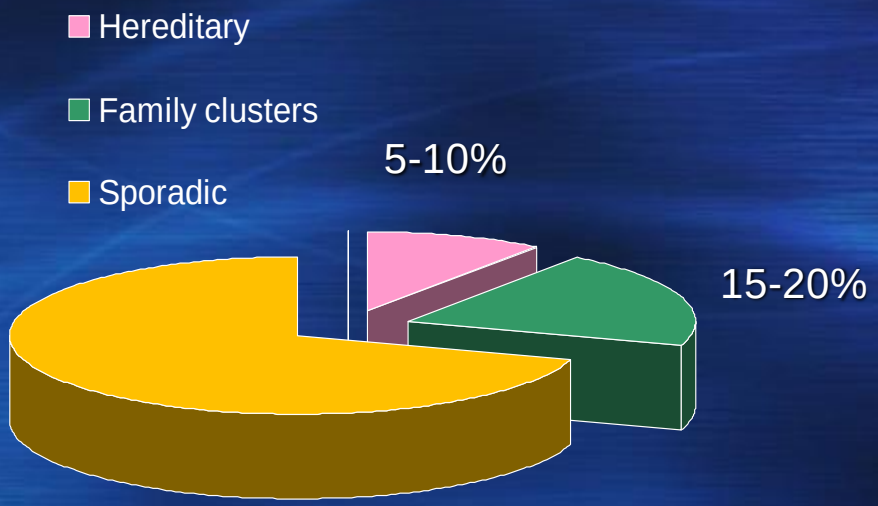
Truth or Myth

A majority of breast cancers are caused by the BRCA gene

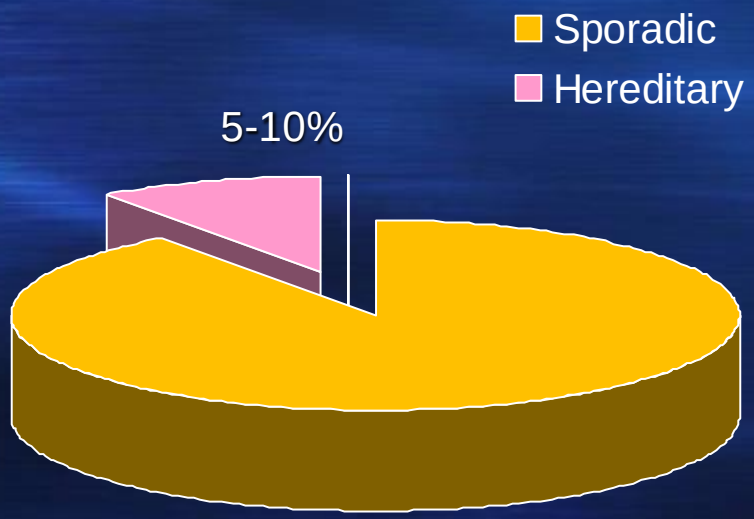
Truth or Myth

A majority of breast cancers are caused by the BRCA gene

How much breast and ovarian cancer is hereditary?



Breast Cancer



Ovarian Cancer

Frequency of BRCA mutations

- For those unaffected women:
  - 1/300- 1/800 in the general population
  - 1/40 in Jewish women with 3 specific mutations
  - ? Those with family history

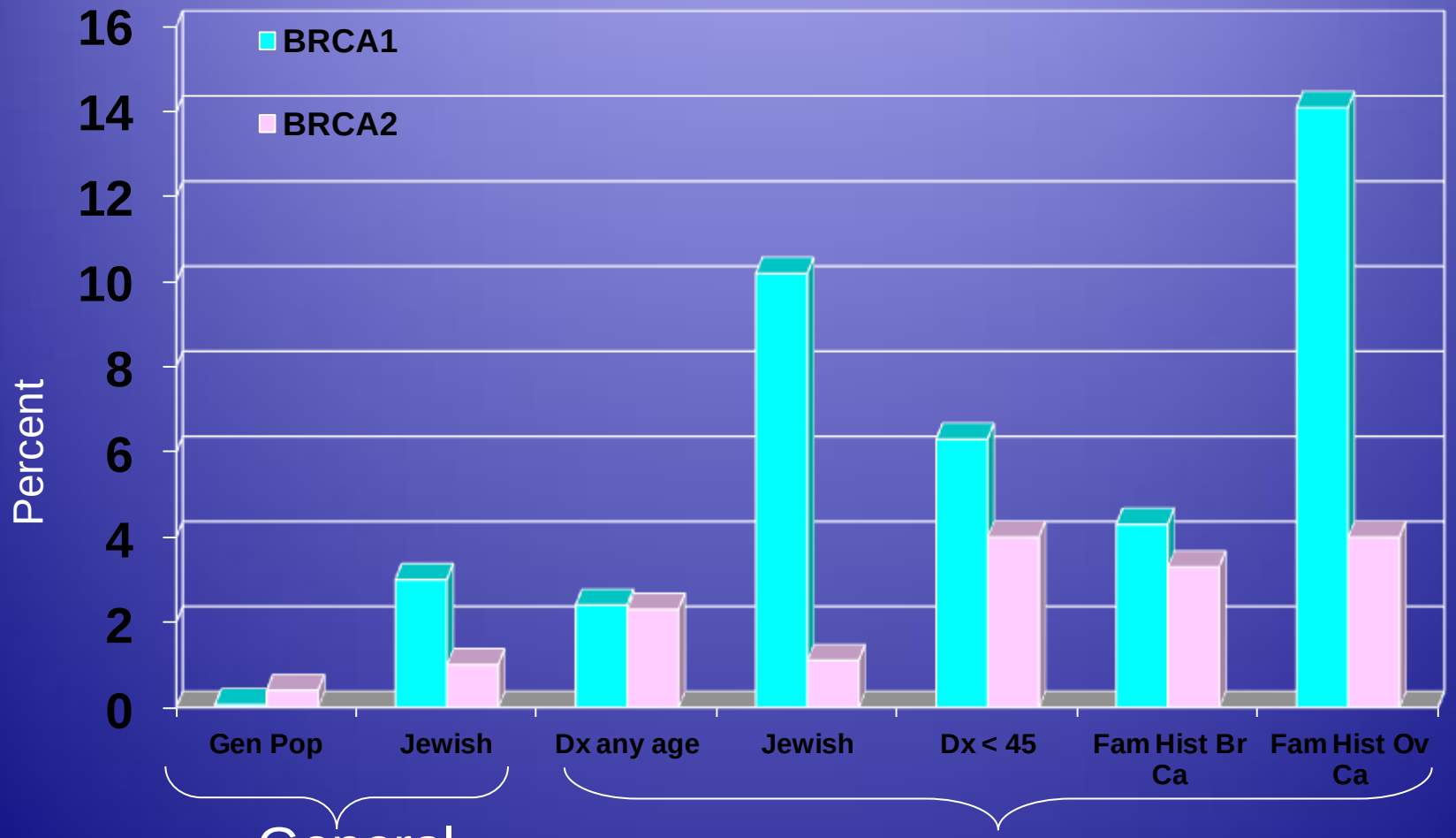
Saslow et al. CA Cancer J Clin2007:57: 75-89

AP via Wisconsin State Journal // Joseph M. Jackson III

Red Flags

- Strong family/personal history of breast/ovarian cancer especially premenopausal

Prevalence of *BRCA1/2* Mutations



Slide by Dr. Cecilia Bellcross

Predicted Probability of a *BRCA1* Mutation

Average age at DX of Br Ca	Probability (%) Br Ca only family	Probability (%) Br Ca/Ov Ca family
< 35	17.4	55.0
35 – 39	11.7	43.5
40 – 44	7.7	32.7
45 – 49	5.0	23.4
50 – 54	3.2	16.2
55 – 59	2.1	10.8
> 59	1.3	7.1

(Avg. of 3.5 cancers/family)

Couch FJ et al. NEJM. 1997; 336:1409

Frequency of BRCA mutations

- For those unaffected women:
  - 1/300- 1/800 in the general population
  - 1/40 in Jewish women with 3 specific mutations
  - ? Those with family history
- For women with breast cancer, risk of mutation varies by age of onset of cancer
  - The younger the age at diagnosis, the greater the chance of a mutation
- For women with ovarian cancer, 15-21% carry a BRCA mutation

Saslow et al. CA Cancer J Clin2007:57: 75-89

Conditions Associated with Hereditary Breast Cancer Risk

Conditions	Gene	Hereditary Br Ca Risk
Hereditary Breast Cancer 1	*BRCA1*	~45%
Hereditary Breast Cancer 2	*BRCA2*	~35%
CHEK2	*CHEK2*	~5%
Li-Fraumeni	*p53*	<1%
Cowden	*PTEN*	low
Ataxia-Telangectasia	*ATM*	low
Peutz-Jeghers	*STK11*	low
Hereditary Diffuse Gastric Cancer	*CHD1*	low
Undiscovered Genes		20%+

Red Flags

- Strong family/personal history of breast/ovarian cancer especially premenopausal
- Breast and ovarian cancer in the same person
- Diagnosis of bilateral breast cancer
- Male breast cancer
- Known BRCA1/2 mutation in family
- Ashkenazi ancestry

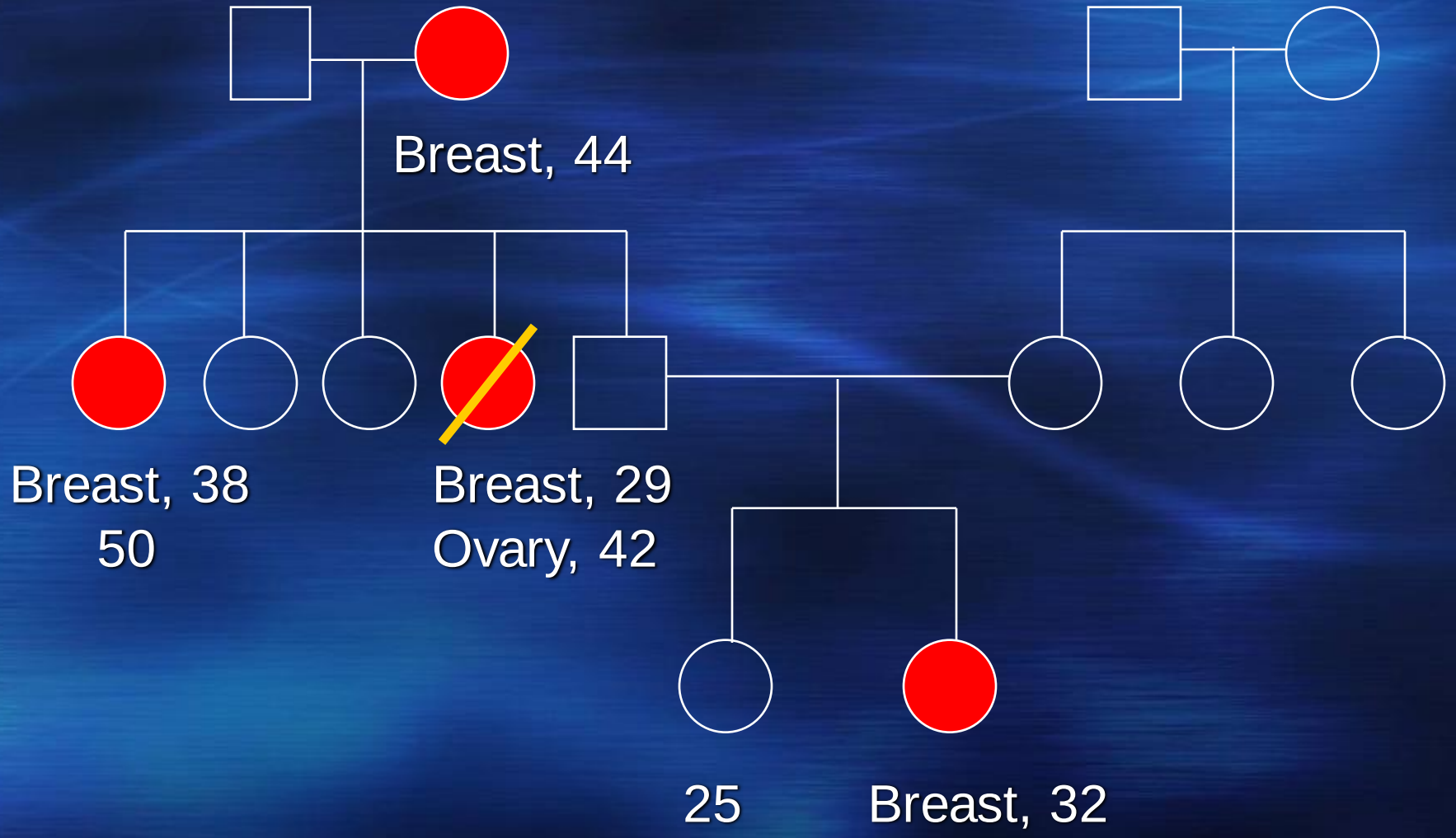
Truth or Myth

"The BRCA gene can only be passed from the mother's side of the family"

Truth or Myth

“The BRCA gene can only be passed from the mother’s side of the family”

Example of BRCA1/2 Pedigree



Truth or Myth

If you are tested and do not have the BRCA gene, you do not have to get a mammogram or an exam anymore because you are not at risk for breast cancer.

Truth or Myth

If you are tested and do not have the BRCA gene, you do not have to get a mammogram or an exam anymore because you are not at risk for breast cancer.

Implications for Healthcare Providers (HCP)

- All licensed HCP will have a role in the delivery of genetic services
- HCP will require genetic knowledge
- Assessment of family history will become increasingly important
- Important to be aware of specialized cancer care and risk counseling services

- OB/GYN Mgt July 2005

acog today

Published by the American College of Obstetricians and Gynecologists

Volume 47 Issue 9 October 2003

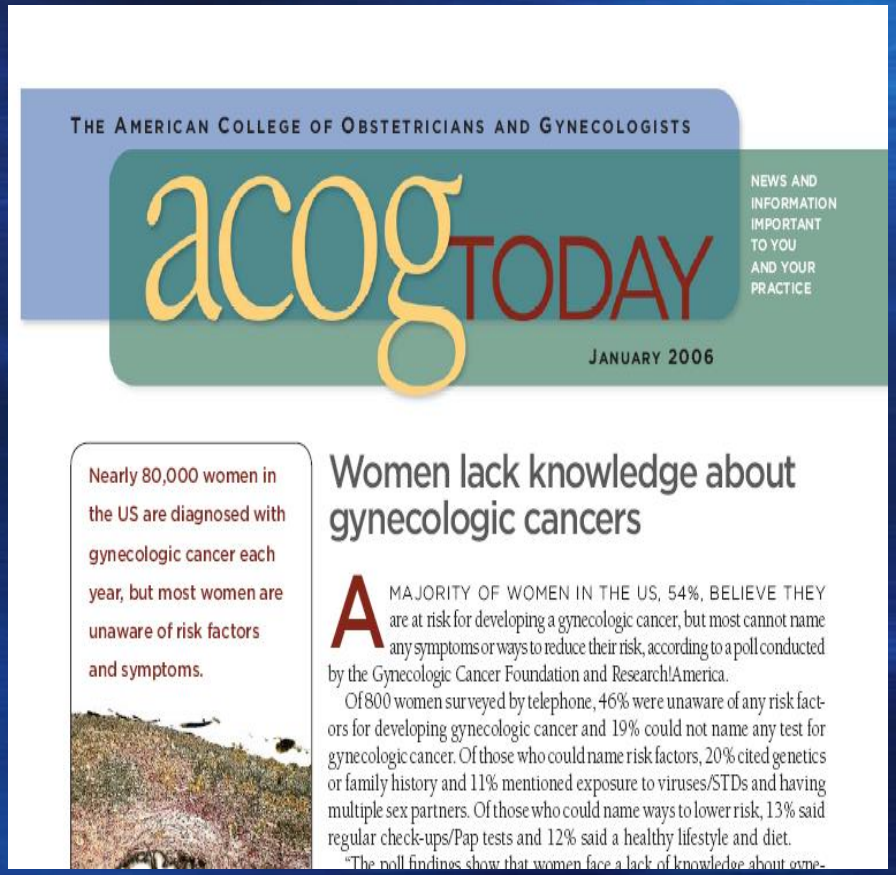
Breast cancer prevention and treatment: what's new, what's promising

How ob-gyns can help patients

- **Assessing and reducing risk:** "There's plenty that ob-gyns can do now to help their patients avoid and survive breast cancer," says Dr. Zujewski. "Evaluate risk factors and family history regularly. We need to move the focus to prevention rather than early detection. Ob-gyns should be familiar with the risk-assessment tool called the Gail Model. It helps, in a very simple way, to evaluate risk."

Women Lack Knowledge

- 46% unaware of any risk factors
  - 20% genetics/family history
  - 11% exposure to viruses/STDs/multiple partners
- 19% could not name any tests/lower risk
  - 13% regular check-ups
  - 12% healthy lifestyle/diet



THE AMERICAN COLLEGE OF OBSTETRICIANS AND GYNECOLOGISTS

acog TODAY

NEWS AND INFORMATION IMPORTANT TO YOU AND YOUR PRACTICE

JANUARY 2006

Nearly 80,000 women in the US are diagnosed with gynecologic cancer each year, but most women are unaware of risk factors and symptoms.

Women lack knowledge about gynecologic cancers

A MAJORITY OF WOMEN IN THE US, 54%, BELIEVE THEY are at risk for developing a gynecologic cancer, but most cannot name any symptoms or ways to reduce their risk, according to a poll conducted by the Gynecologic Cancer Foundation and Research!America.

Of 800 women surveyed by telephone, 46% were unaware of any risk factors for developing gynecologic cancer and 19% could not name any test for gynecologic cancer. Of those who could name risk factors, 20% cited genetics or family history and 11% mentioned exposure to viruses/STDs and having multiple sex partners. Of those who could name ways to lower risk, 13% said regular check-ups/Pap tests and 12% said a healthy lifestyle and diet.

"The poll findings show that women face a lack of knowledge about gyne-

ACOG PRACTICE BULLETIN

Number 103, April 2009, Reaffirmed 2013

CLINICAL MANAGEMENT GUIDELINES FOR OBSTETRICIAN–GYNECOLOGISTS

Hereditary Breast and Ovarian Cancer Syndrome

Hereditary breast and ovarian cancer syndrome is an inherited cancer-susceptibility syndrome. The hallmarks of this syndrome are multiple family members with breast cancer or ovarian cancer or both, the presence of both breast cancer and ovarian cancer in a single individual, and early age of breast cancer onset. Clinical genetic testing for gene mutations allows physicians to more precisely identify women who are at substantial risk of breast cancer and ovarian cancer. For these individuals, screening and prevention strategies can be instituted to reduce their risks. Obstetricians and gynecologists play an important role in the identification and management of women with hereditary breast and ovarian cancer syndrome.

BRCA1 and *BRCA2*

Germline mutations in *BRCA1* and *BRCA2* account for the vast majority of families with hereditary breast and ovarian cancer syndrome. Approximately 10% of cases of ovarian cancer and 3–5% of cases of breast cancer are due to germline mutations in *BRCA1* and *BRCA2* (1–3). *BRCA1* is found on chromosome 17, and *BRCA2* is on chromosome 13. More than 1,200 different mutations have been reported for *BRCA1*, and more than 1,300 different mutations have been reported for *BRCA2*. *BRCA1* and *BRCA2* are tumor suppressor genes that encode proteins that function in the DNA repair process (4, 5). Although individuals with hereditary breast and ovarian cancer syndrome inherit one defective allele in *BRCA1* or *BRCA2* from their father or mother, they have a second, functional allele. If the second allele becomes nonfunctional, cancer can develop through the accumulation of additional mutations. This is called the

Risk Assessment Recommended

- Personal history ovarian cancer **and** 1° or 2° relative with
  - Ovarian cancer **and/or**
  - Premenopausal breast cancer
- Ashkenazi Jewish ancestry
  - Personal history ovarian cancer **or**
  - Personal history breast cancer ≤ 40
- Personal history breast **and** ovarian cancer

Risk Assessment Recommended

- Personal history breast cancer ≤ 50 **and** 1° or 2° relative with
  - Ovarian cancer **or**
  - Male breast cancer
- 1° or 2° relative with known BRCA mutation

Risk Assessment May Be Helpful

- Personal history breast cancer ≤ 40
- Personal history breast cancer ≤ 50 with Ashkenazi Jewish ancestry
- Personal history breast cancer ≤ 50 **and** 1° or 2° relative with breast cancer ≤ 50
- Personal history bilateral breast cancer
- Personal history breast cancer any age **and ≥ 2** 1° or 2° relatives with breast cancer any age

Risk Assessment May Be Helpful

- Personal history ovarian cancer of high-grade, serous histology
- Unaffected women with 1° or 2° relative meeting above criteria

- 35 yo – personal history breast cancer and brother with history of breast cancer
- 27 G0 - mom history of postmenopausal breast cancer
- 37 yo - cousin premenopausal breast cancer
- 40 yo - mom aged 76 history triple negative breast cancer
- 16 yo maunt serous epithelial ovarian cancer
- 45 yo G4P4 – sister premenopausal BCA, mother ovarian CA. desires TAH 2/2 fibroids and irreg cycles

A LIFE IS NOT IMPORTANT EXCEPT IN THE IMPACT IT HAS ON OTHER LIVES

Jackie Robinson

Knowledge *is* Power

Knowledge *is* choice

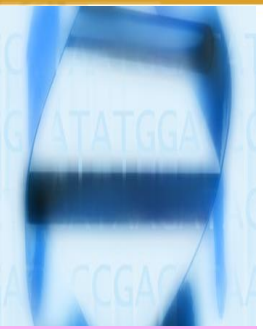
Knowledge *is* Hope

Breast Cancer Survivor

Conclusion

- Epidemiology of breast cancer
- Hereditary breast and ovarian cancer syndrome (*BRCA1* and *BRCA2* gene mutations) is associated with increased risk for breast and ovarian cancer
- Importance of a detailed family history
- Guidelines used to identify patients requiring referral/testing

Thank you for your attention



Hereditary Breast and Ovarian Cancer Screening & Genetic Counseling

Cecelia Bellcross, PhD, MS, CGC
Emory University School of Medicine
Department of Human Genetics

The First Line of Defense
February 21, 2014

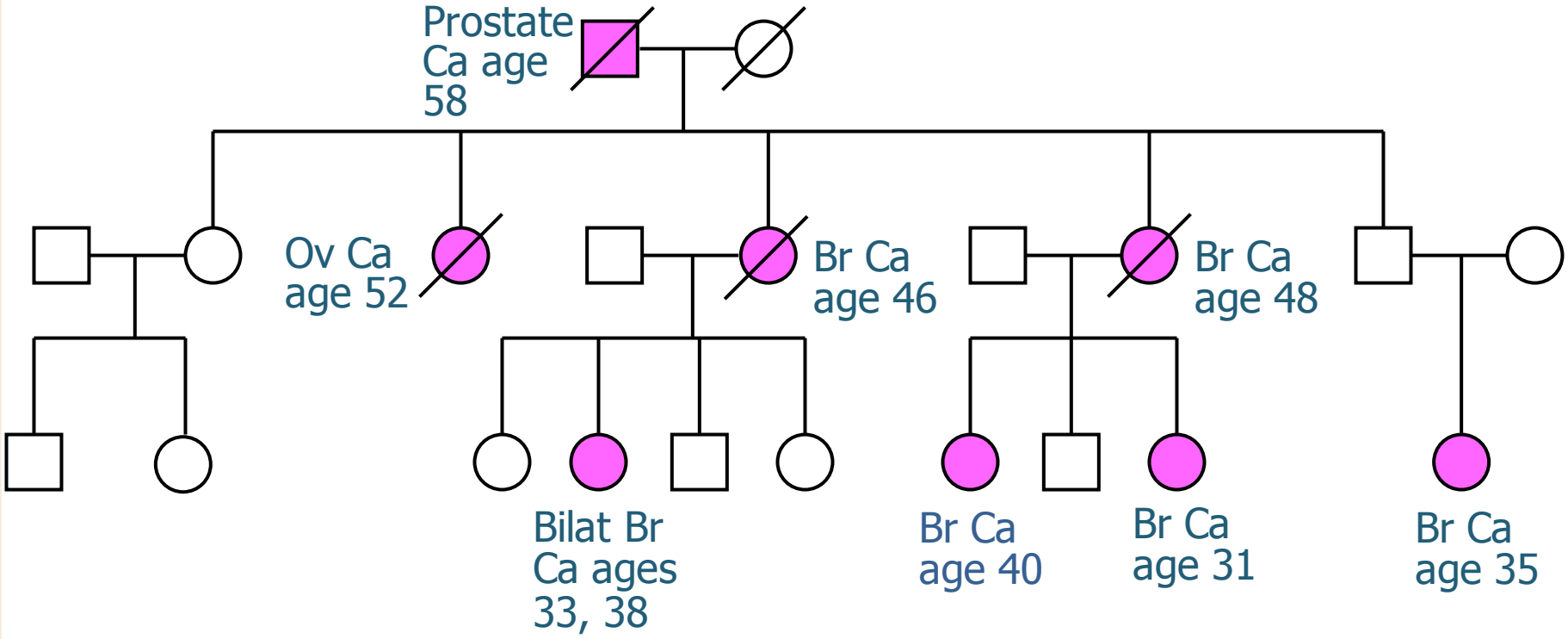
Faculty Disclosure

In compliance with ACCME Guidelines, I hereby declare:

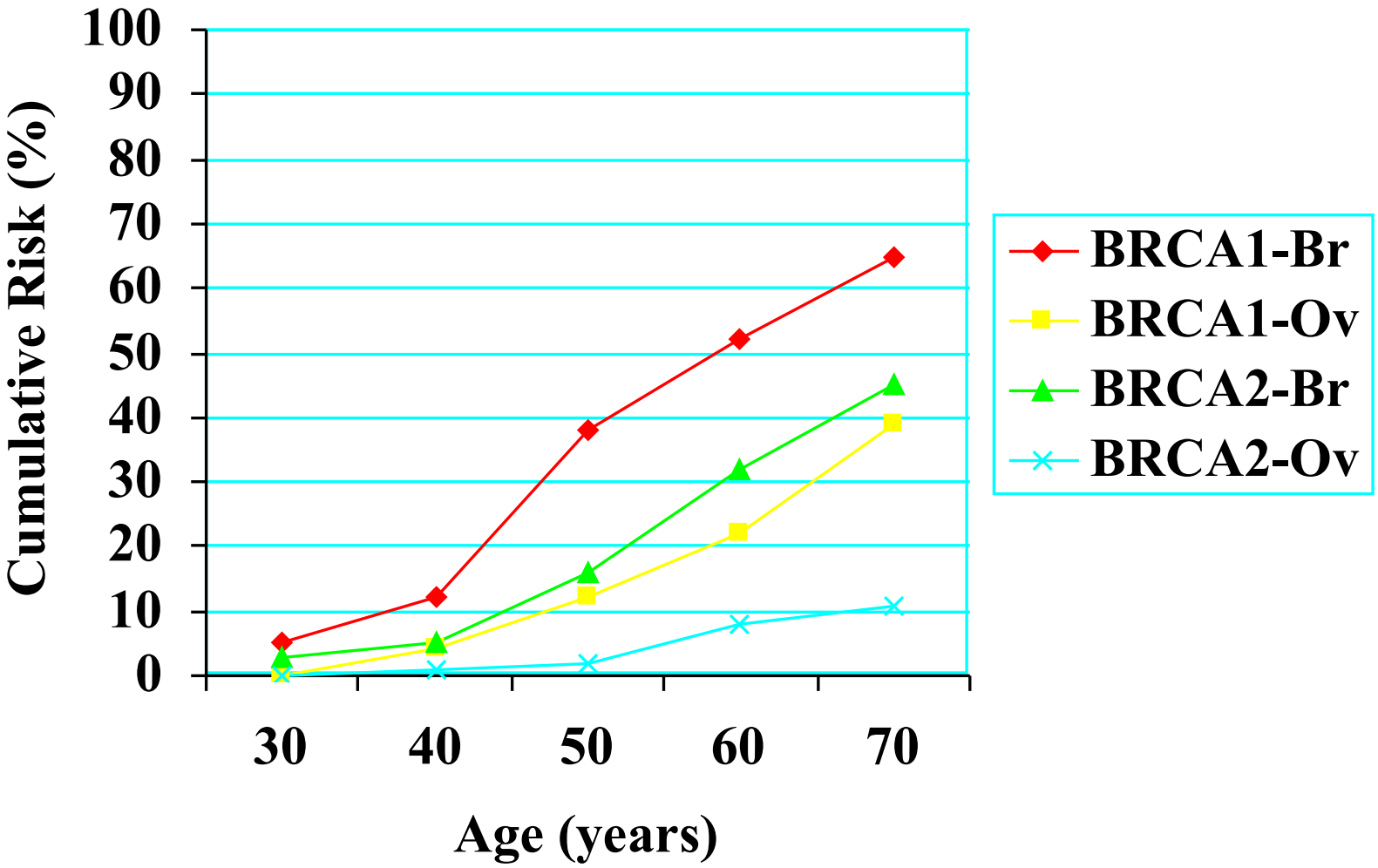
- I do not have financial or other relationships with the manufacturer(s) of any commercial services(s) discussed in this educational activity.

Cecelia A. Bellcross, PhD, MS, CGC
Board Certified Genetic Counselor
Assistant Professor
Director – Emory Genetic Counseling Training Program

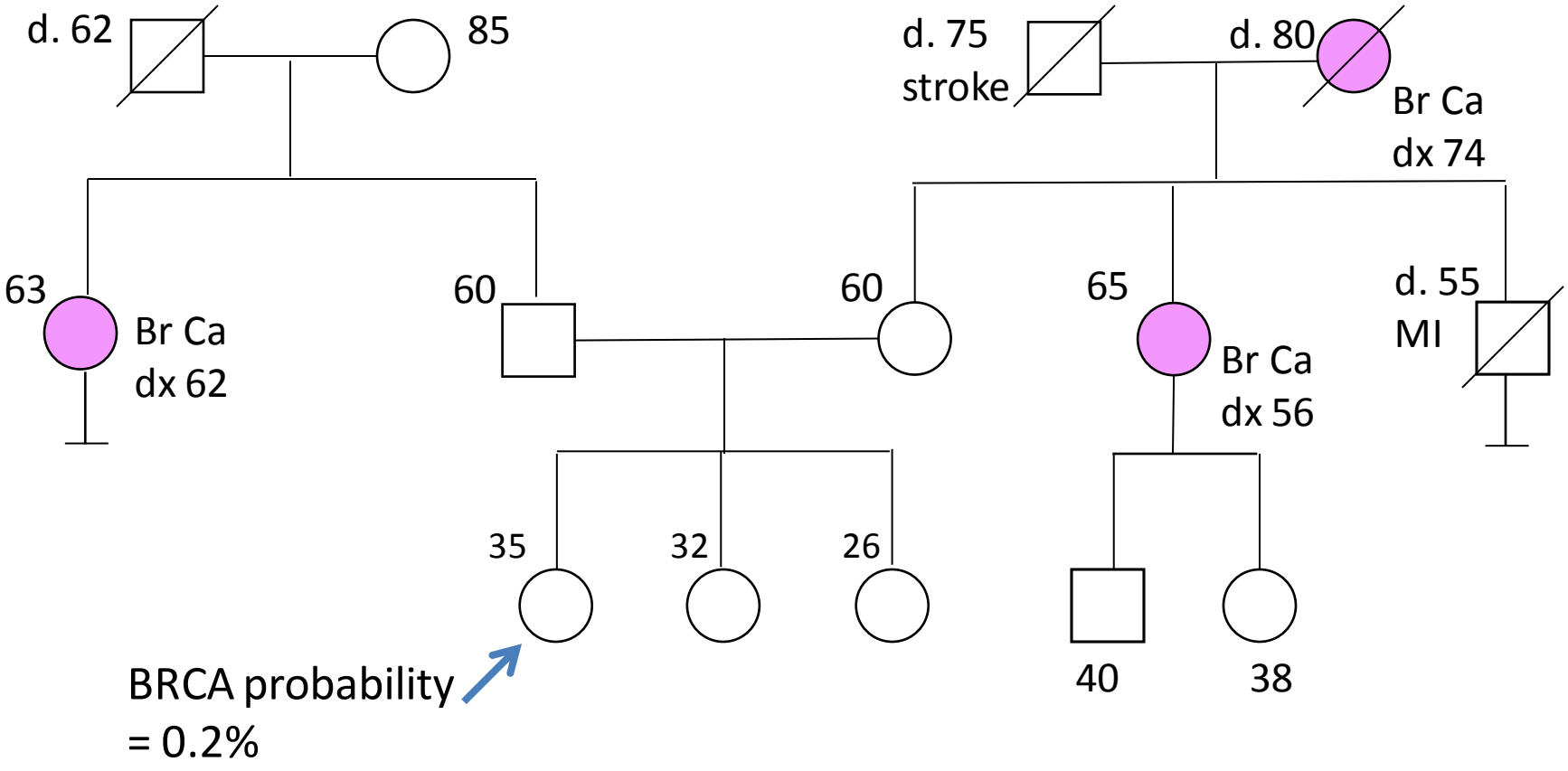
The "no-brainer" BRCA Family



BRCA1/2 Risks Over Time

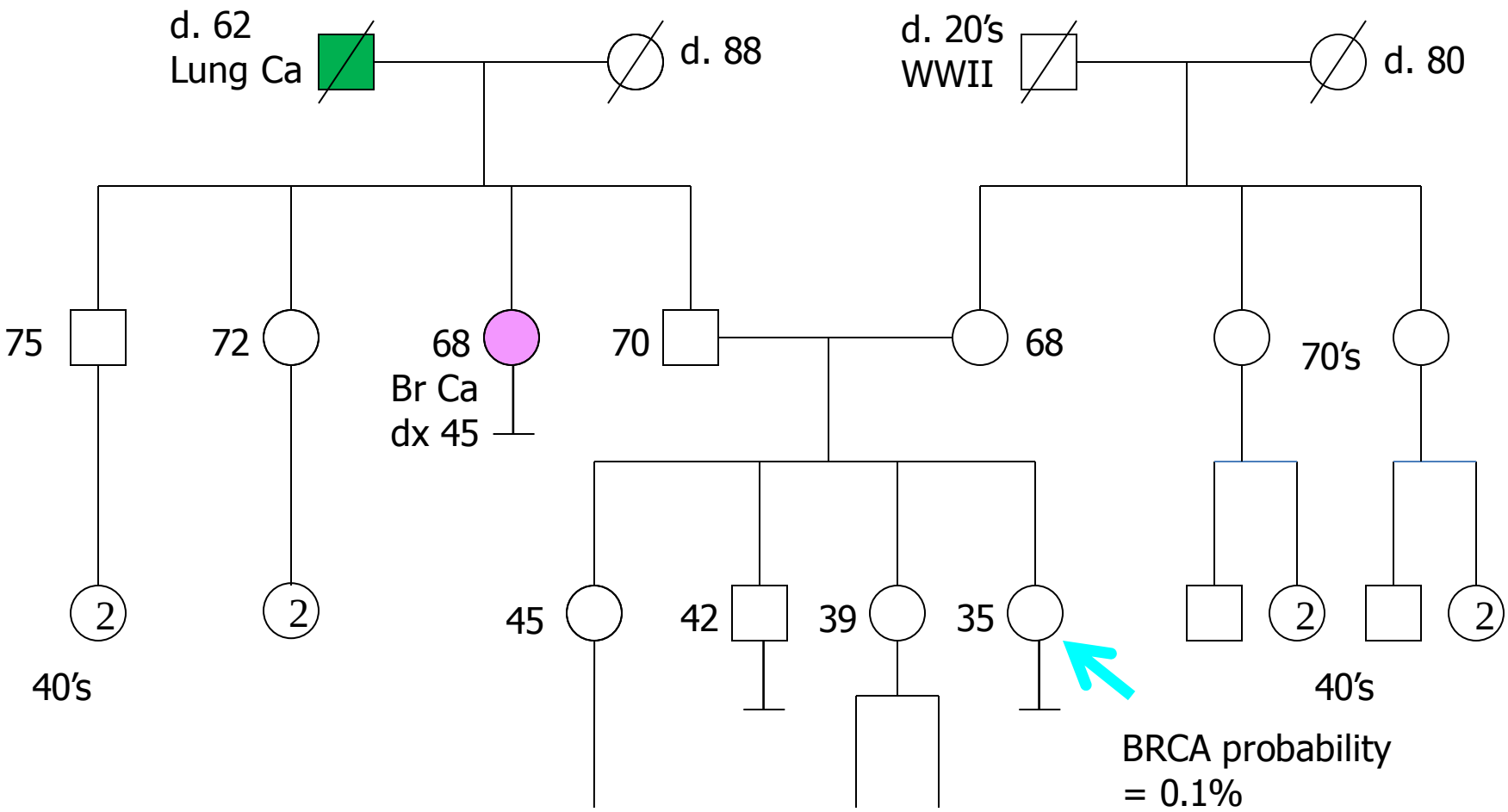


What about this family?



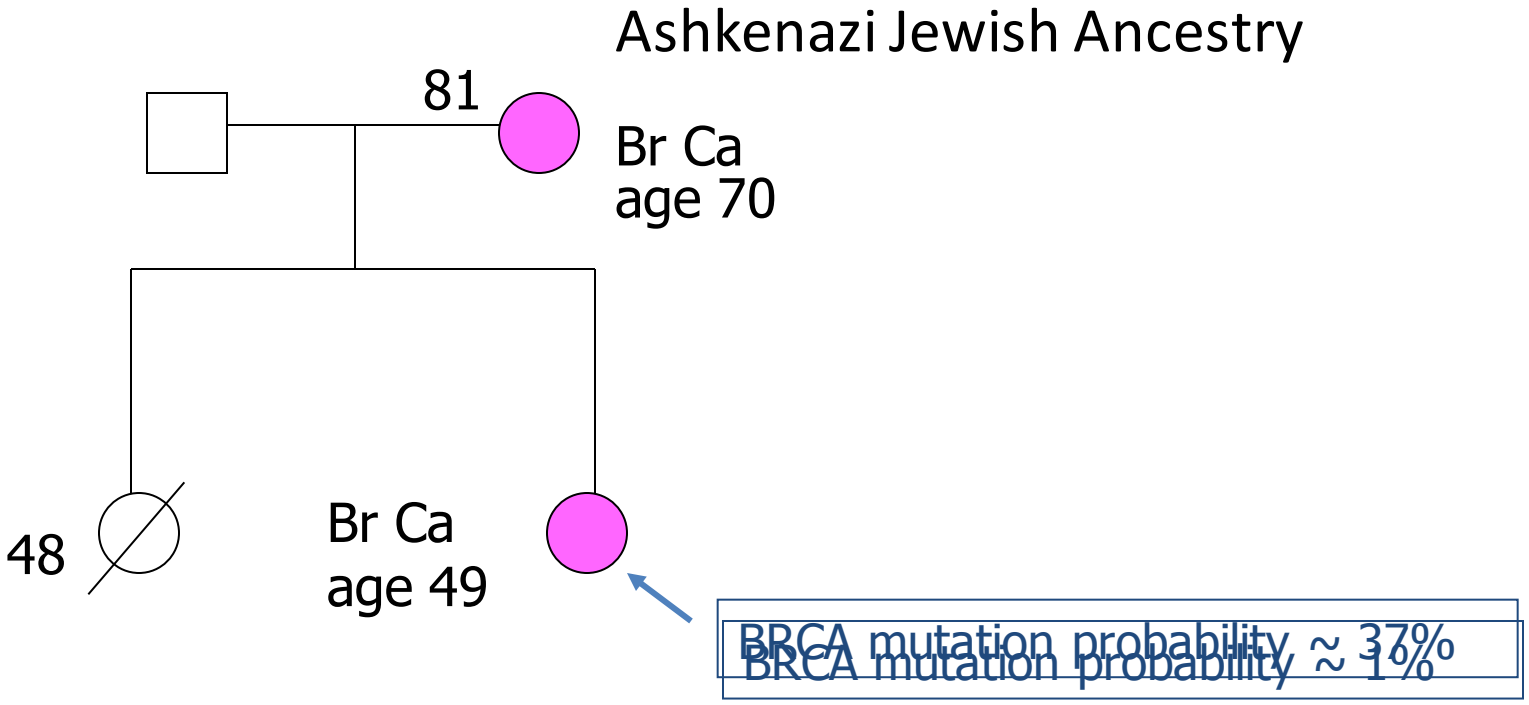
USPSTF 2005: … "a combination of 3 or more first- or second-degree relatives with breast cancer regardless of age at diagnosis…."

What about this family?



NCCN Criteria for further genetic risk evaluation:
Unaffected individual with 1st or 2nd-degree relative with br ca ≤ age 45

What about this family?



United States Preventive Services Task Force (USPSTF) 2005 Recommendation

- "….women whose family history is associated with an increased risk for deleterious mutations in the BRCA1 or BRCA2 genes [should] be referred for genetic counseling and evaluation for BRCA testing."

Research Gaps:

- Need to develop and validate tools feasible for use in primary care to screen for individuals appropriate for cancer genetics referral

REFERRAL SCREENING TOOL

History of **BREAST** *or* **OVARIAN** cancer in the family?

NO ☐ (stop)

YES ☐ (complete checklist)

TABLE

	Breast cancer at or before age 50	**Ovarian cancer at any age**
Yourself		
Mother		
Sister		
Daughter		
Mother's side		
Grandmother		
Aunt		
Father's side		
Grandmother		
Aunt		
Two (2) or more cases of breast cancer *(after age 50)* on the <u>same</u> side of the family		
Male breast cancer at *any age* in any relative		
Jewish Ancestry		

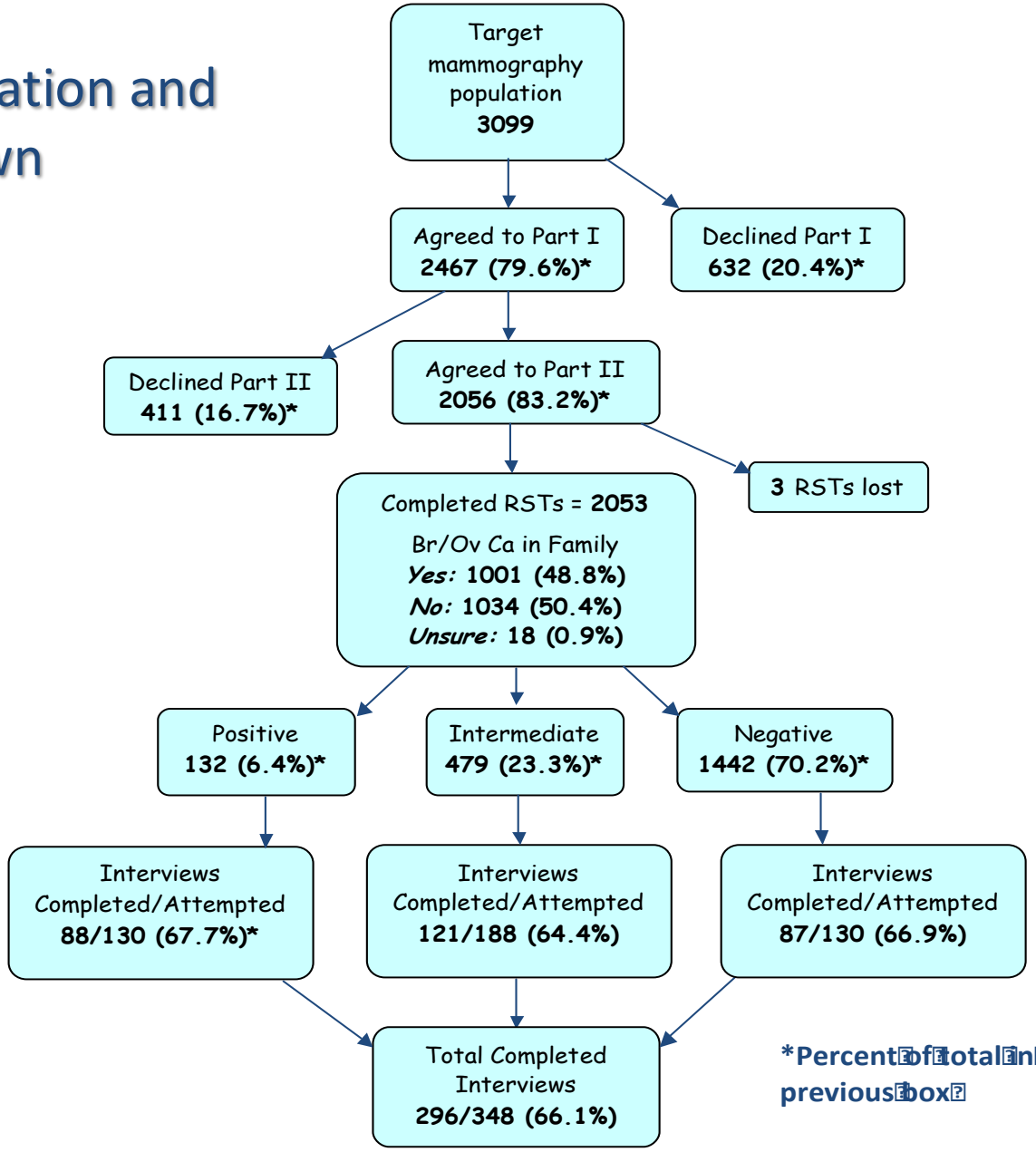
ASSESSMENT: **(Positive Screen =** Two [2] or more checks (✓) in above table.)

POSITIVE SCREEN _____ NEGATIVE SCREEN _____

Research Methods

- RST administered to unselected individuals undergoing a screening mammogram at one of three clinics
- Phone interview by GC to collect four-generation cancer pedigree on random subjects
- Pedigree analysis by 4 validated models: BOADICEA (BD), BRCAPRO (BR), Myriad II (MII) and Family History Assessment Tool (FH)
  - RST positive vs. model risk ≥ 10%

Study Participation and Risk Breakdown



Reliability - Agreement of RST Scores

Repeat administration of RST on random 6% of subjects

Comparison	Concordance	Kappa Statistic
Tech1 vs. Tech2 ***n*** **= 156**	0.96	0.75 (Good/Excellent)

Validation Measures

Measure	BD/BR/ MII/FH* vs. RST	BD/BR/ MII* vs. RST	BD/BR* vs. RST	Model Accuracy Measures†
Sensitivity %	81	87	88	78-94%
Specificity %	92	82	78	51-82%
ROC AUC	0.87	0.85	0.83	0.68-0.82

*High-risk by at least one of the models = actual positive state

†Range of *upper end* values reported for BD/BR/MII/FH models where positive state = identified deleterious *BRCA1/2* mutation

Percentiles of Model Values by RST Score

MODEL RST Score*	Median Value	Percentile			
		5th	25th	75th	95th
BOADICEA†					
RST Negative	0.5	0.0	0.2	0.7	3.8
RST Positive	4.3	0.5	1.8	11.3	41.8
BRCAPRO†					
RST Negative	0.5	0.0	0.0	0.5	2.4
RST Positive	3.4	0.5	1.1	10.2	48.4
Myriad II†					
RST Negative	2.8	0.0	2.2	4.5	6.8
RST Positive	8.7	2.9	5.3	12.2	15.8
FHAT‡					
RST Negative	4.0	0.0	0.8	6.0	11.0
RST Positive	13.0	5.0	9.8	16.0	21.0

*Negative n=210, Positive n=86

† Value corresponds to percent likelihood for a *BRCA1/2* mutation

‡ Value unique to model-reflects strength of family history

B-RST vs. RST

- Electronic application distinguishes between maternal and paternal lineages
- Nieces/nephews included in 2nd degree relatives assessed
- Bilateral breast cancer added
- Breast/ovarian cancer in the same person added (alone flags positive)
- Breast cancers over 50 incorporated

B-RST (Breast Cancer Genetics Refer...

Introduction

Breast Cancer Genetics Referral Screening Tool (B-RST)

This website is for individuals and healthcare providers who are wondering if they, or their patients, might be at risk for hereditary breast/ovarian cancer.

Introduction:

B-RST is a screening tool designed to quickly identify who should be referred for a cancer risk assessment/genetic consultation, to formally evaluate their family history and discuss the benefits and limitations of *BRCA1/2* genetic testing.

Health Care Providers **Consumers**

Disclaimer: Tool is provided for informational purposes only. It is not intended to provide medical advice or serve as a substitute for professional advice, medical diagnosis or treatment.

B-RST Validation

Measure	BD	BR	MII	FH	Overall*
Sensitivity %	100	100	96.9	90.0	89.4
Specificity %	75.1	74.0	76.1	89.8	91.5
AUC (95% CI)	0.88 (0.83-0.92)	0.87 (0.83-0.91)	0.87 (0.81-0.92)	0.90 (0.85 -0.94)	0.90 (0.85-0.95)

*High-risk by at least one of the models = actual positive state

Sensitivity increased for all model comparisons (+ 8.2% to +11.1%), while specificity decreased slightly (-0.4% to -2.3%)

Screening Site Login

www.breastcancergenescreen.org

Breast Cancer Genetics
Referral Screening Tool
(B-RST)

B-RST is a screening tool that asks questions about family history to assess if you (or your patient) may be at risk for Hereditary Breast and Ovarian Cancer.

This tool is designed to quickly identify who should be referred for cancer genetic counseling to formally evaluate their family history and discuss the benefits and limitations of genetic testing for Hereditary Breast and Ovarian Cancer.

HEALTH CARE PROVIDERS | GENERAL PUBLIC

GEORGIA BREAST CANCER GENOMIC CONSORTIUM
EDUCATION SURVEILLANCE AND POLICY

2013 USPSTF Recommendation

The USPSTF recommends that primary care providers screen women who have family members with breast, ovarian, tubal, or peritoneal cancer with one of several screening tools designed to identify a family history that may be associated with an increased risk for potentially harmful mutations in breast cancer susceptibility genes (BRCA1 or BRCA2). Women with positive screening results should receive genetic counseling and, *if indicated after counseling*, BRCA testing. (**B recommendation**)

Family History Screening

- PCPs should ask about specific types of cancer, cancer sites, which family members were affected, relatives with multiple primary cancers, age of diagnosis, sex of affected individuals

- For women who have at least 1 family member with breast, ovarian or other types of BRCA-related cancers, PCPs may use 1 of several brief familial risk stratification tools to determine the need for *in-depth genetic counseling*.

Familial Risk Stratification Tools

- Since 2005, …. tools have been developed and validated for use in PCP practice to guide referral for BRCA counseling
- Ontario Family History Assessment Tool, Manchester Scoring System, Referral Screening Tool (RST), Pedigree Assessment Tool
- "All of these tools seem to be clinically useful predictors of which women should be referred for genetic counseling because of increased risk of potentially harmful BRCA mutations…."
- "The Referral Screening Tool and the FHS7 are the simplest and quickest to administer."
- PCPs should not use general breast cancer risk assessment models (e.g. Gail)

Importance Evidence Updates

- Genetic Counseling
  - Increases the accuracy of risk perception
  - Decreases the intention for genetic testing among unlikely carriers
  - Decreases cancer-related worry, anxiety and depression

Cancer Genetic Counseling

- Collect and review detailed personal and family cancer histories
- Assess likelihood for a hereditary cancer syndrome and cancer risk probabilities
- Determine appropriateness of genetic testing, who best to test, and which test to order
- Educate patient about cancer genetics, personal risks, pros/cons/limitations of genetic testing, possible test results:
  - positive, true negative, uninformative negative, variant of uncertain significance

Cancer Genetic Counseling- cont.

- Address patient's risk perceptions and psychosocial concerns
- Review options of screening, management, risk reduction
- Coordinate testing and insurance coverage
- Post-test disclosure session
  - implications of test results for patient/family
  - management plan based on result
  - additional testing needed/available
  - psychosocial adjustment to results/implications

Cancer Program Standards 2012: Ensuring Patient-Centered Care

- Standard 2.3: Risk Assessment and Genetic Counseling
  - All programs are required to provide cancer risk assessment as well as genetic counseling and testing, on-site or by referral, by a qualified genetics professional [who has]
  - … extensive experience and educational background in genetics, cancer genetics, counseling, and hereditary cancer syndromes to provide risk assessment and empathetic genetic counseling to patients with cancer and their families

Cancer Program Standards 2012: Ensuring Patient-Centered Care

- Cancer Genetic Professionals:
  - ABMG/ABGC certified genetic counselors
  - ABMG certified medical geneticist
  - Advanced practice nurses with genetics certification/credentialing
  - Board certified physician with experience in cancer genetics (provides on a regular basis)
- *Educational seminars offered by commercial laboratories on how to perform genetic testing are not considered adequate training for cancer risk assessment and genetic counseling….*


THE
ANGELINA
EFFECT
Angelina Jolie's double mastectomy puts genetic testing in the spotlight. What her choice reveals about calculating risk, cost and peace of mind
BY JEFFREY KLUGER & ALICE PARK

The USPSTF recommends against routine genetic counseling or BRCA testing for women whose family history is not associated with an increased risk for potentially harmful mutations in the BRCA1 or BRCA2 genes. (**D recommendation**)

Finding a Cancer Genetics Professional

- National Society of Genetic Counselors: NSGC Find a Counselor
- National Cancer Institute's Cancer Information Service: 1-800-4-CANCER http://www.cancer.gov/search/geneticsservices/
- Tele-counseling Services
  - http://www.informeddna.com/
  - http://www.geneticcounselingservices.com/

Other High Penetrance Breast Cancer Susceptibility Genes

Condition	Gene (s)	Breast cancer risks	Other associated cancers
Li-Fraumeni Syndrome	*TP53*	56% by age 45, >90% lifetime	Soft tissue sarcomas, leukemias, brain tumors, osteosarcomas, adrenal
Cowden Syndrome	*PTEN*	30-50% lifetime	Thyroid, endometrial
Peutz-Jeghers Syndrome	*STK11*	8% by age 40, 32% by age 60	Colorectal, gastric, pancreatic
Hereditary Diffuse Gastric Cancer (HDGC)	*CDH1*	(lobular) 39% lifetime	Diffuse gastric cancer

Breast Cancer PLUS

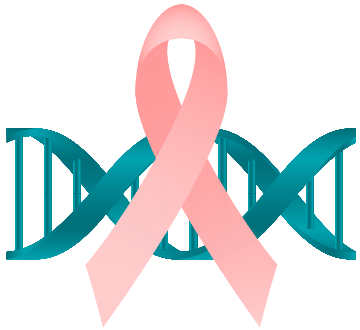
Ambry Genetics™

- Full sequencing plus deletion/duplication analysis of:
  - BRCA1
  - BRCA2
  - CDH1 (Hereditary Diffuse Gastric Cancer)
  - PTEN (Cowden syndrome)
  - STK11 (Peutz-Jegher syndrome)
  - TP53 (Li-Fraumeni syndrome)

And the complexity increases….

TEST CODE	TEST(S)	TAT*	PRICE**	NOTES
8836	BRCAplus (BRCA1, BRCA2, CDH1, PTEN, TP53, STK11) Gene Sequence and Deletion/Duplication Analyses	21 Days*	$3,300	BRCA1/2 gene sequence and deletion/duplication analyses and BRCAplus do not reflex to BreastNext, OvaNext or CancerNext.
8838	BRCA1/2 Gene Sequence and Deletion/Duplication Analyses (Concurrent)	14-21 Days*	$2,200	
8862	BRCA1/2 Analysis with reflex to BRCAplus if negative	14 - 21 Days*	$3,350	
5892	BRCA Ashkenazi Jewish 3-Site Mutation panel	7 - 10 Days*	$500	
5894	BRCA Ashkenazi Jewish 3-Site Mutation panel with reflex to BRCA1/2 Analysis if negitive	14 - 21 Days*	$2,250	
5890	BRCA1/2 Deletion/Duplication (ie. large rearrangement) Analysis	14 Days*	$500	
5864	BRCA1 Specific Site Analysis	7-14 Days*	$400	Patients with a previous history of BRCA1/2 testing can choose to blind BRCA 1/2 sequencing results on any of the NGS panels, by including a copy of the previous testing report with their sample submission.
5884	BRCA2 Specific Site Analysis	7-14 Days*	$400	
8820	BreastNext (16 genes)	6-10 Wks	$3,900	
8830	OvaNext (21 genes)	12-16 Wks	$3,900	
8824	CancerNext (24 genes)	12-16 Wks	$4,900	
8042	PancNext (13 genes)	12 Wks	$3,900	


GTACT
CAAGT
CTA
TAC
CCGA
TAGG
ATCC
TGG
ATCC



Breast Cancer Genetics
Referral Screening Tool

breastcancergenescreen.org

Thank You

cbellcr@emory.edu

Bellcross CA, Lemke AA, Pape LS, Tess AL, Meisner LF. Evaluation of a breast/ovarian cancer genetics screening tool in a mammography population. *Genet Med 2009;* 11(11):783

Bellcross CA. Further development and evaluation of a breast/ovarian cancer genetics referral screening tool. *Genet Med* 2010;12(4):240, 2010.

Embracing the Family Tree

Case Examples:

Applying the Evidence

Alice Kerber, MN, APRN, ACNS-BC, AOCN, APNG

Oncology Clinical Nurse Specialist

Georgia Center for Oncology Research and Education

Faculty Disclosure

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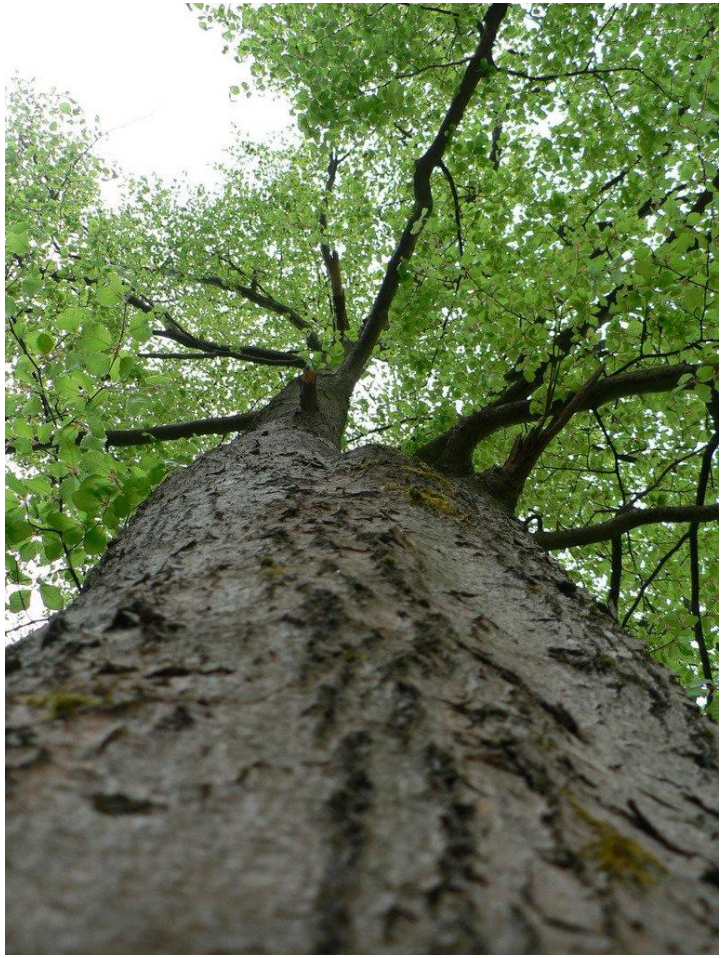
I am on the Speakers' bureau for Pfizer , Inc.

Alice Kerber, MN, APRN, ACNS-BC, AOCN, APNG

Hereditary Syndrome Patterns

- Hereditary = known genetic predisposition for cancer
- Assumed or putative hereditary = hallmarks and features of syndrome
- Familial = not clear pattern, may be nature/nurture
- Sporadic = acquired mutation or environmental exposure

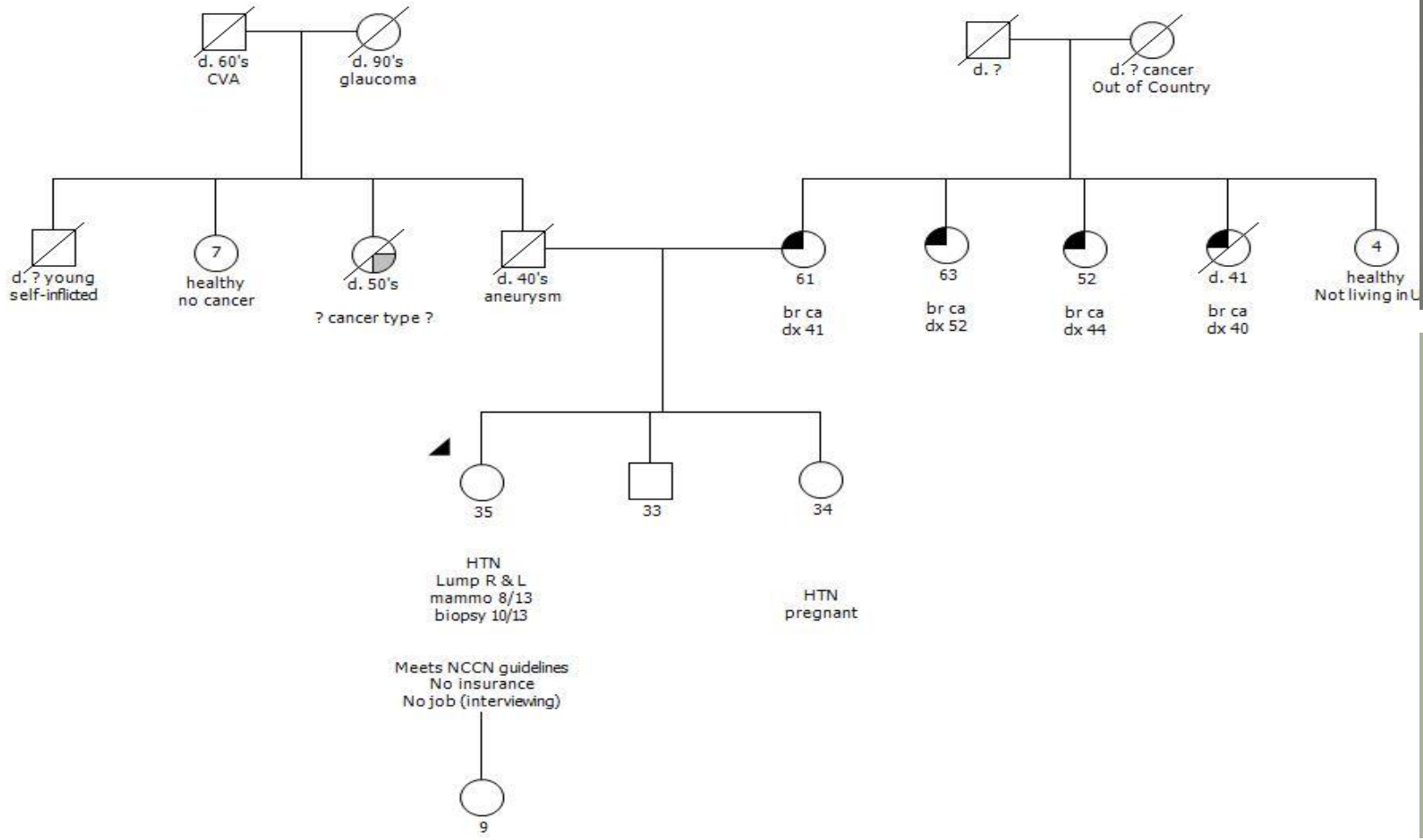


Case Study

- **35 year old Ashkenazi/African American/Hispanic female, unaffected**
- **<u>Family History</u>: Mother and 3 maternal aunts had breast cancer diagnosed before age 45**
  - **Breast findings, mammogram/ultrasound inconclusive**
  - **B-RST positive**
- **<u>Social</u>: no insurance, no job,**
- **One daughter**
- ***Recommendations?***

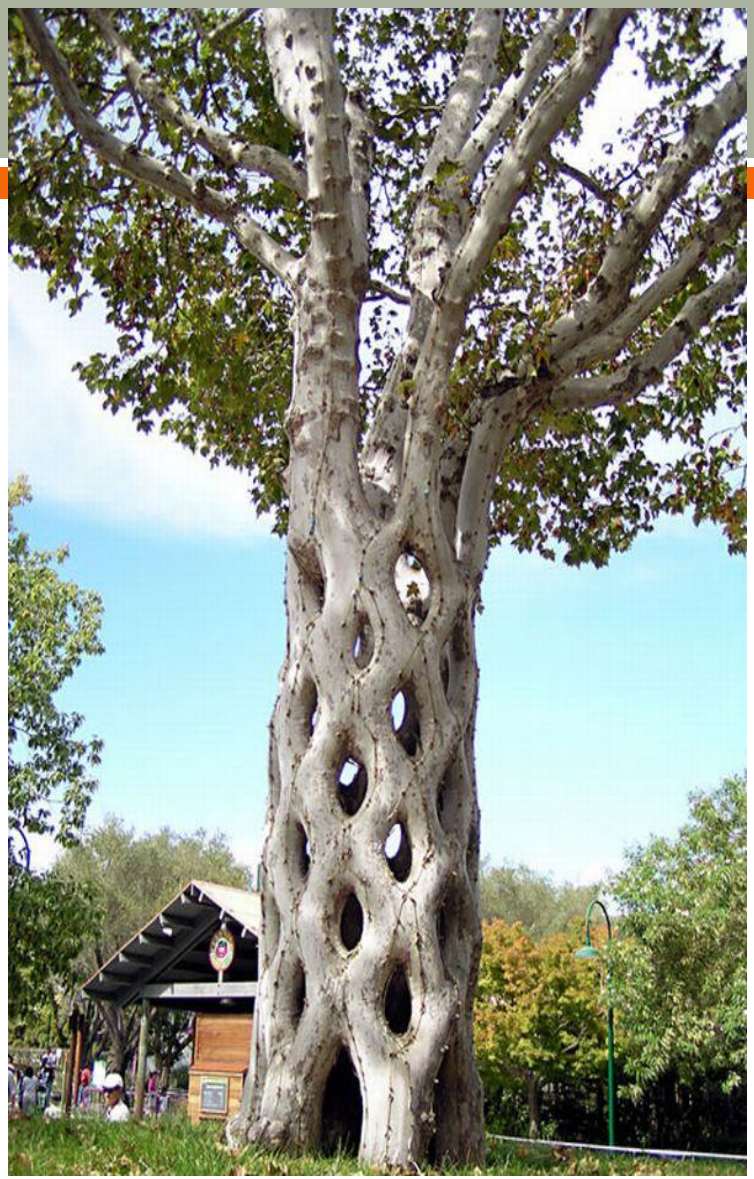
African American/ Native American/ German/ AJ/ Hispanic

2/20/2014



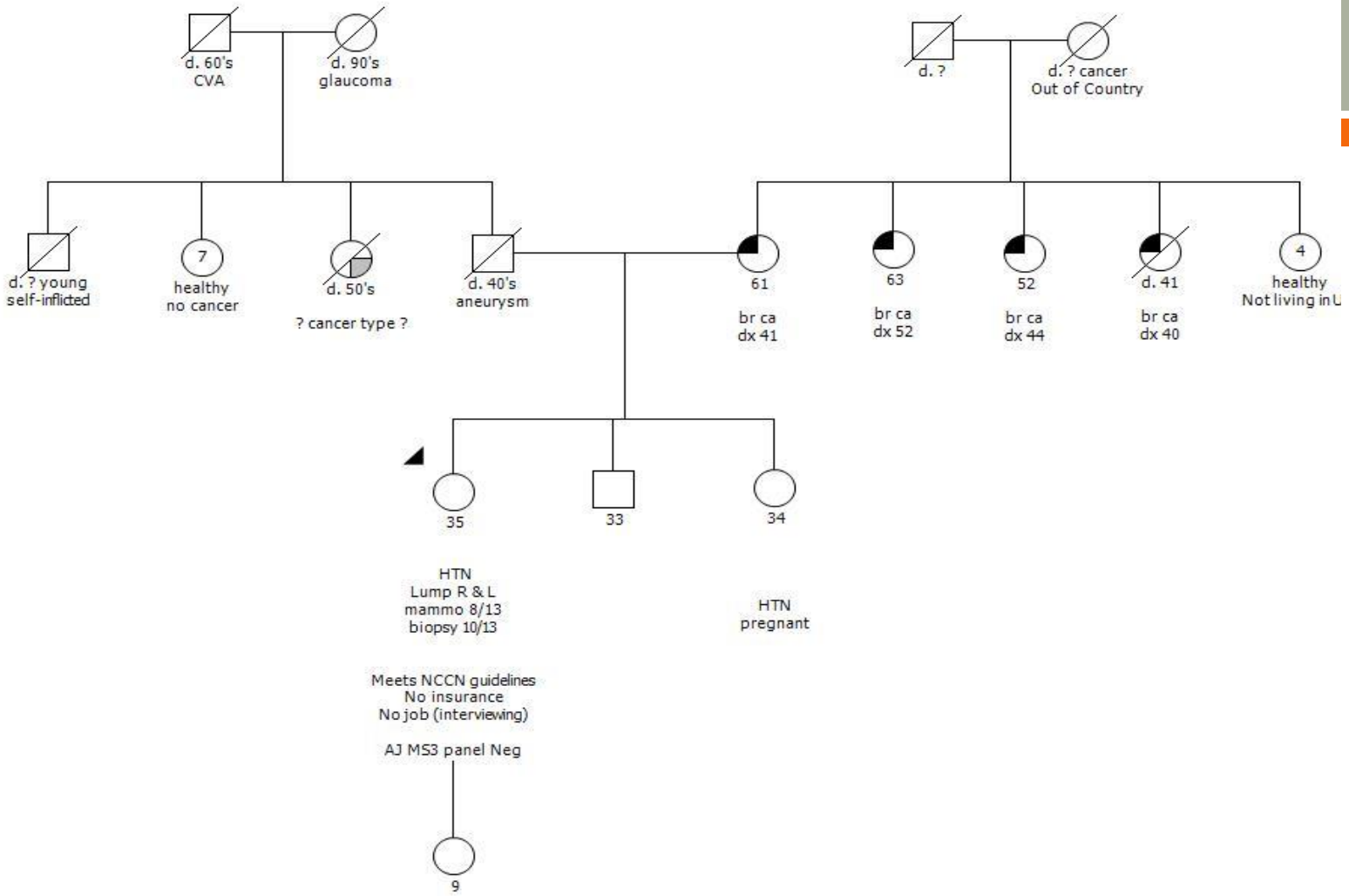
Questions to Ask

- **Three generations**
  - maternal and paternal
- **Major health conditions**
- **Age** of diagnosis and demise
- **Ethnic background**
- **Pregnancy problems**
- **Family lifestyle**

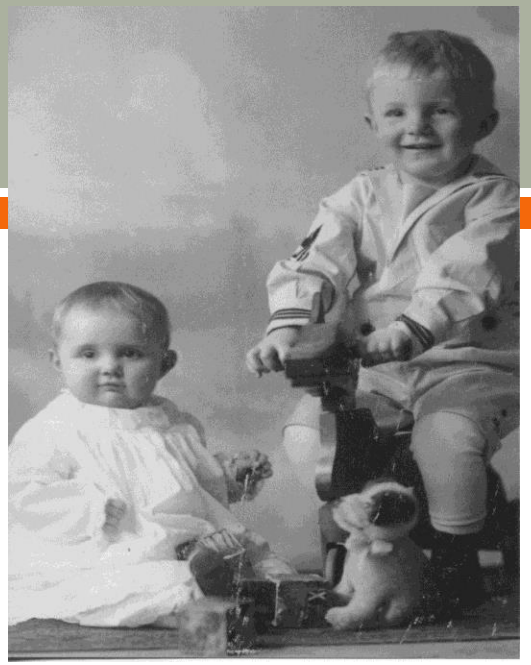


African American/ Native American/ German/ AJ/ Hispanic

2/20/2014



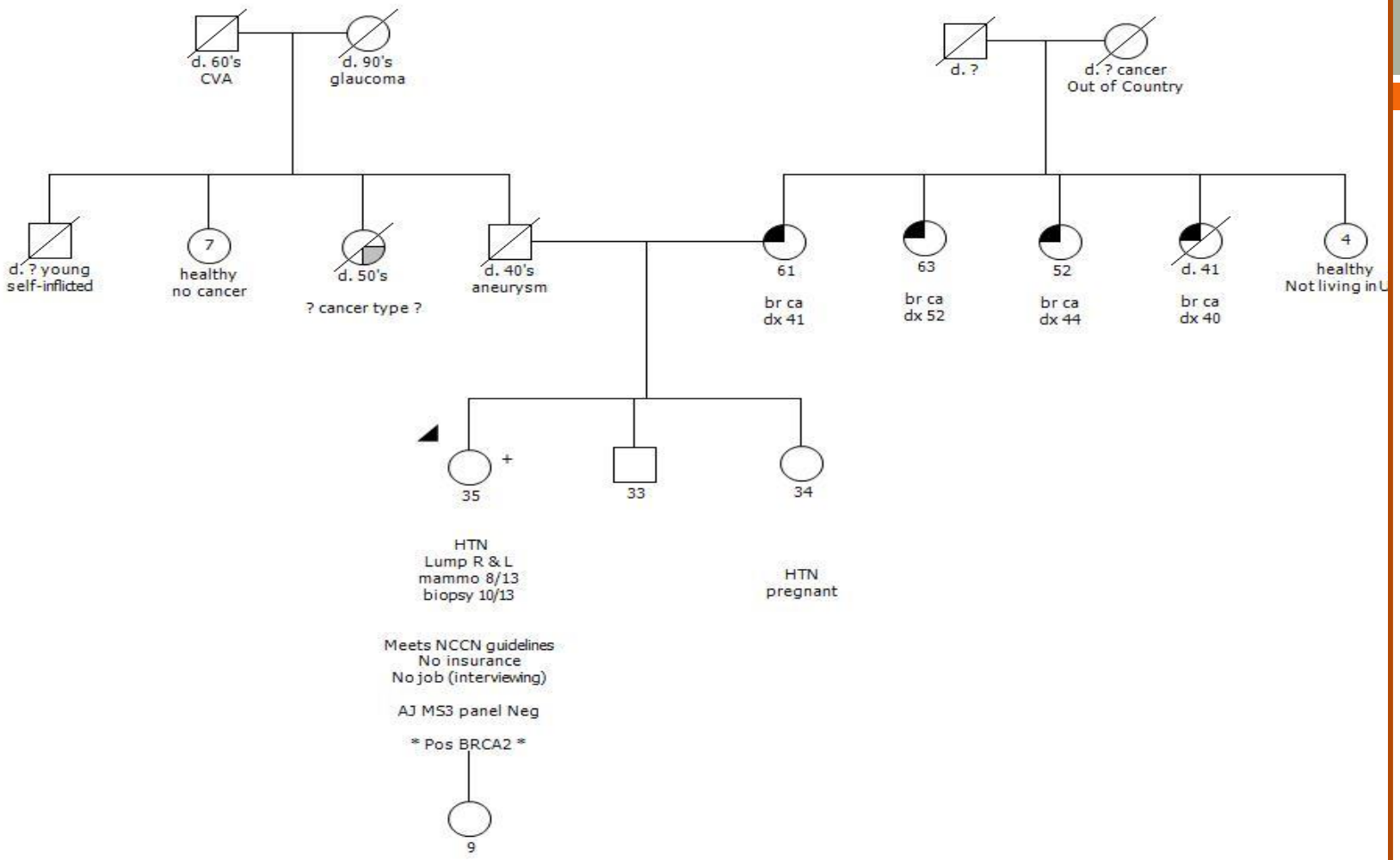
Relatives.....
Who to Include?



- **First degree = 50% of genes**
  - Parents, siblings, children
- **Second degree = 25% of genes**
  - Aunts, uncles, grandparents, nieces, nephews
- **Third degree = 12.5% of genes**
  - Cousins, great-aunts, great-uncles, great-grandparents

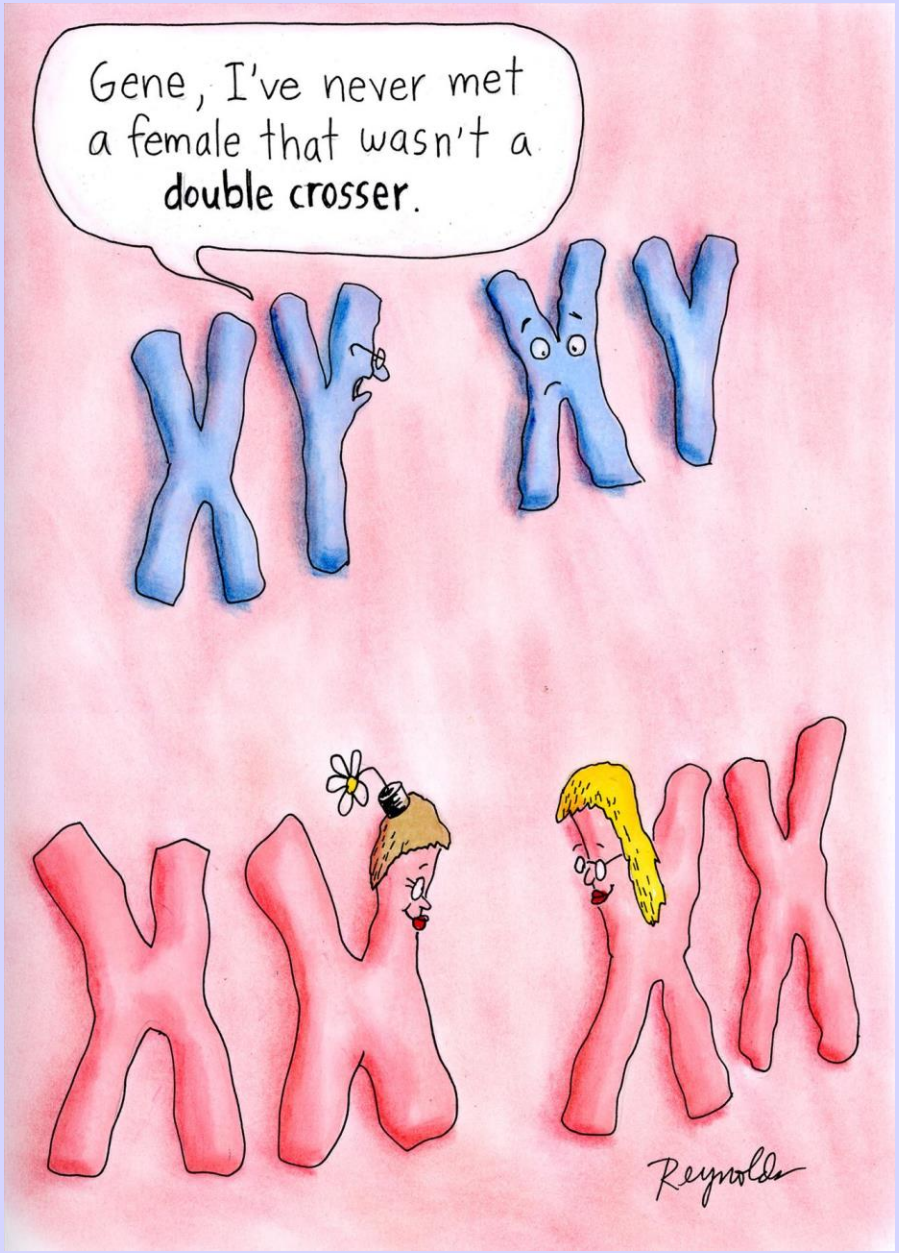
African American/ Native American/ German/ AJ/ Hispanic

2/20/2014



Limitations

- Privacy issues
- Small family
- Accidental deaths
- Absence of targeted organ
- Male dominance in female hereditary syndrome
- Family proximity
- Adoption


Gene, I've never met a female that wasn't a double crosser.
XY
XY
XX
XX
Reynolds

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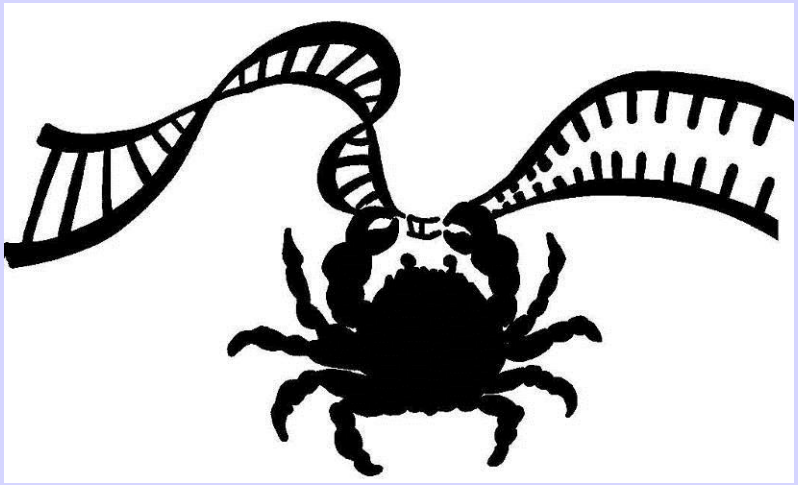
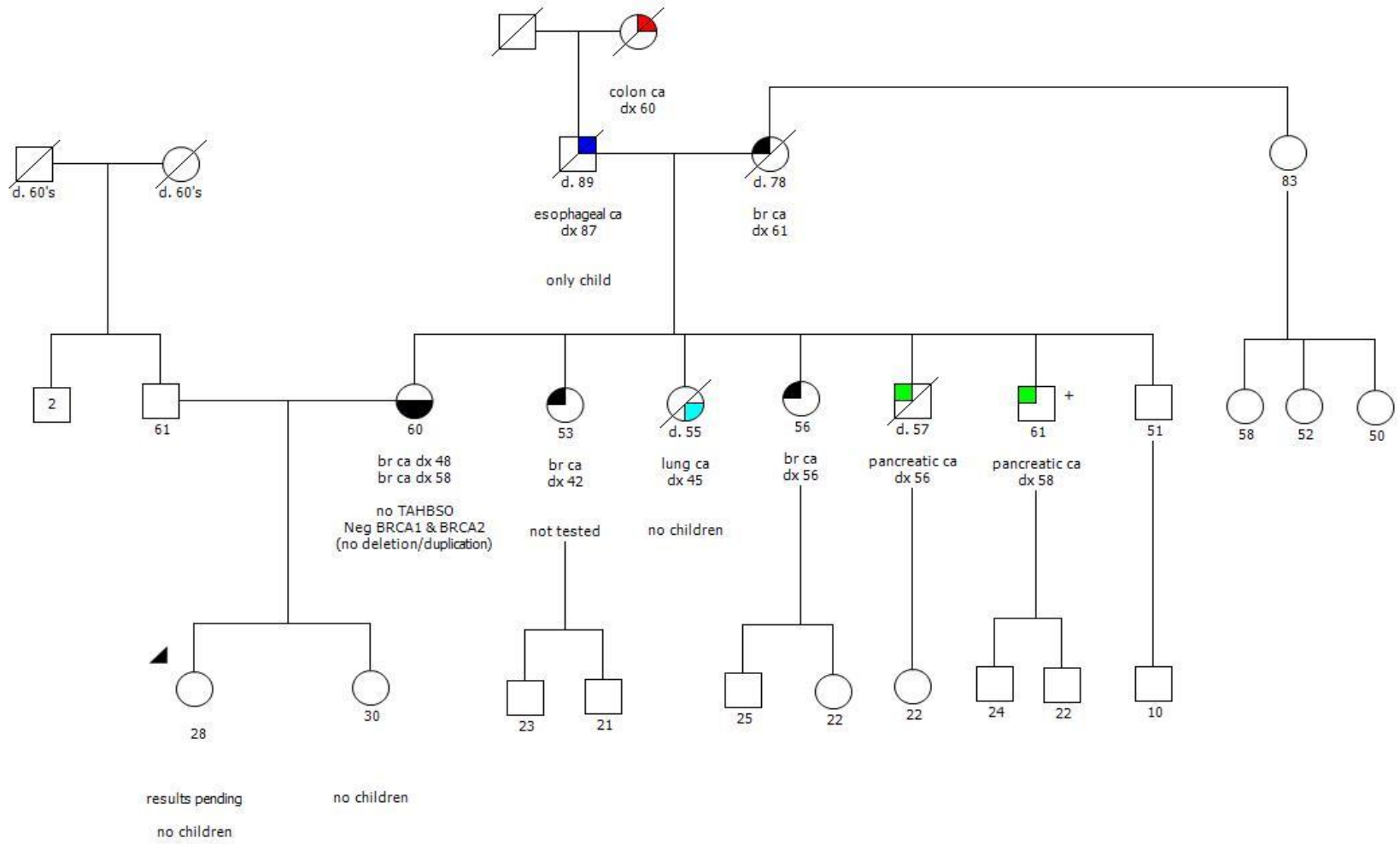
Molly Klein, MS CGC

Cancer Genetic Testing



Molly H. Klein, MS, CGC
1800 Howell Mill Road, suite 625
Atlanta, GA 30318
(404) 425-7949

2/11/2014


colon ca
dx 60
d. 60's
d. 60's
d. 89
esophageal ca
dx 87
only child
d. 78
br ca
dx 61
83
2
61
60
br ca dx 48
br ca dx 58
no TAHBSO
Neg BRCA1 & BRCA2
(no deletion/duplication)
53
br ca
dx 42
not tested
d. 55
lung ca
dx 45
no children
56
br ca
dx 56
d. 57
pancreatic ca
dx 56
61
+
pancreatic ca
dx 58
51
58
52
50
28
results pending
no children
30
no children
23
21
25
22
22
24
22
10

Panel Testing for Other Genes

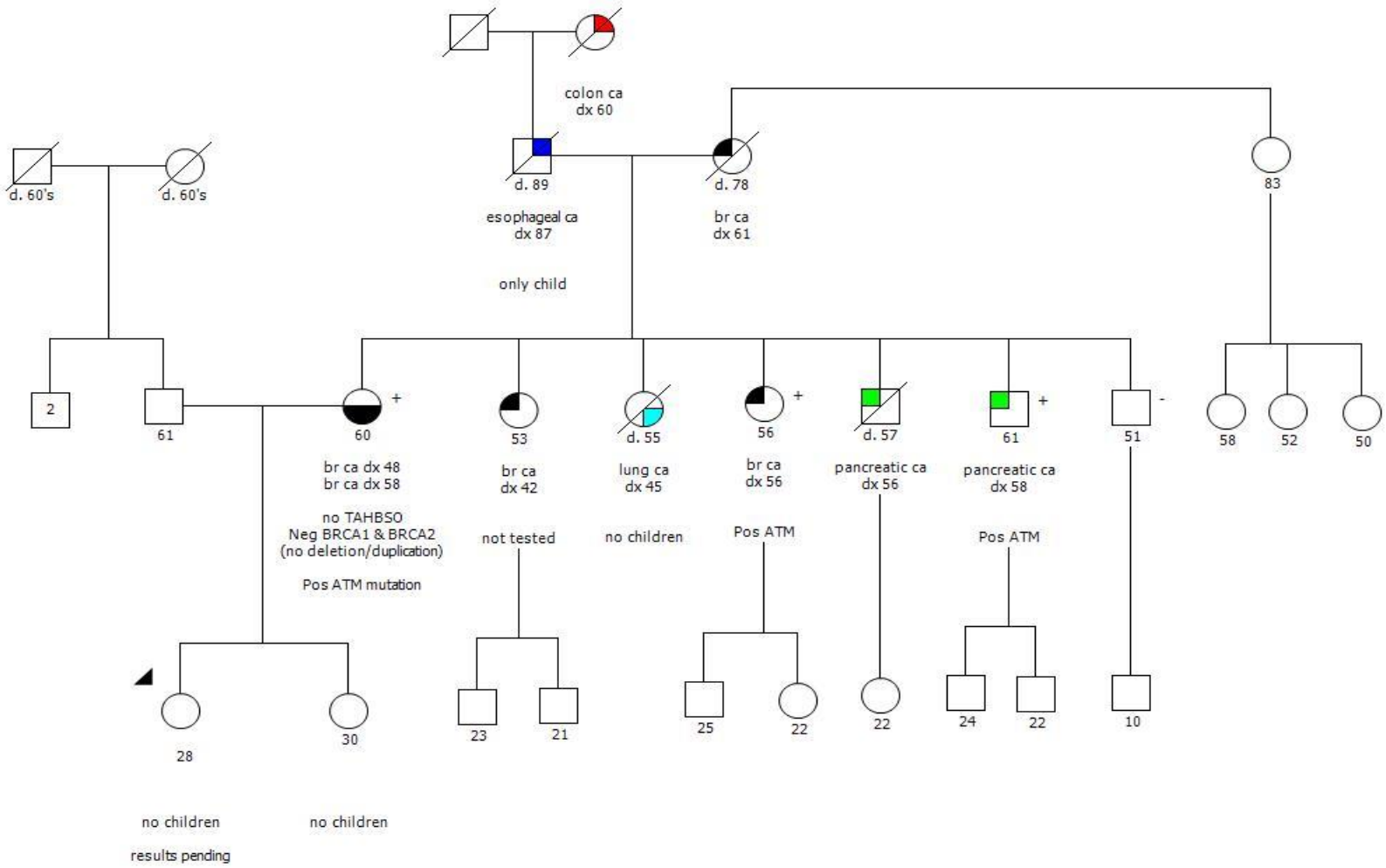
Benefits:

- More cancer genes tested at once (6-35 genes)
- Most current technology available
- Less expensive than ruling out single genes one at a time

Limitation

- Long turn-around times 10-16 weeks
- Higher incidence of an uncertain test result
- Testing positive for a gene that has been newly associated with cancer – no treatment guidelines

2/11/2014


colon ca
dx 60
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d. 60's
d. 89
esophageal ca
dx 87
only child
d. 78
br ca
dx 61
83
2
61
60
+
br ca dx 48
br ca dx 58
no TAHBSO
Neg BRCA1 & BRCA2
(no deletion/duplication)
Pos ATM mutation
53
br ca
dx 42
not tested
d. 55
lung ca
dx 45
no children
56
+
br ca
dx 56
Pos ATM
d. 57
pancreatic ca
dx 56
61
+
pancreatic ca
dx 58
Pos ATM
51
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58
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no children
results pending
no children
23
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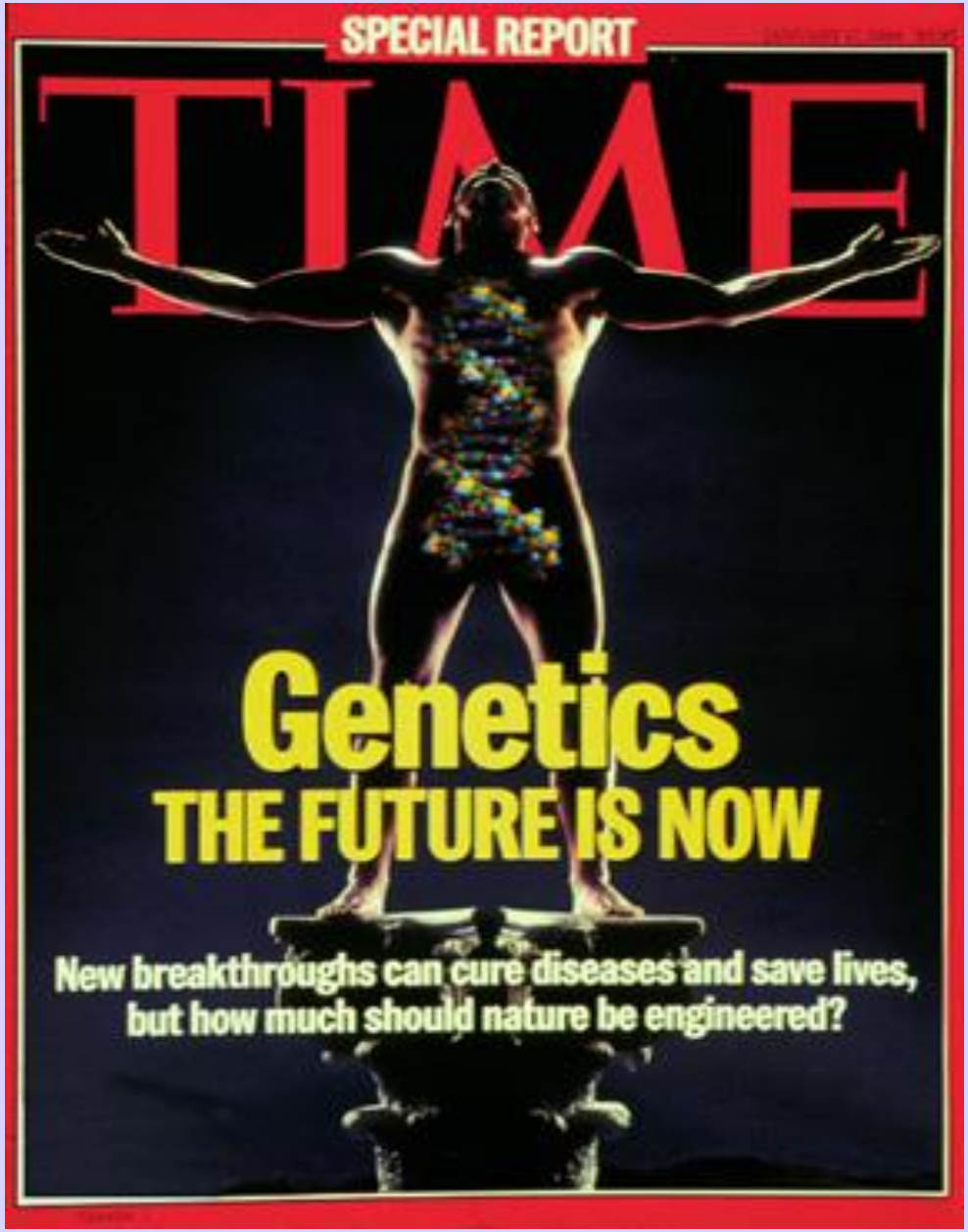
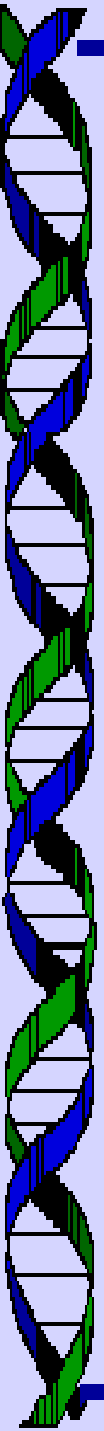

SPECIAL REPORT
TIME
Genetics
THE FUTURE IS NOW
New breakthroughs can cure diseases and save lives,
but how much should nature be engineered?



Spectrum of Care Options for Women at High Risk for Breast and Ovarian Cancer

Sheryl G.A. Gabram, MD, MBA, FACS
Professor of Surgery, Emory University
Director, High Risk Assessment Program
Winship Cancer Institute of Emory University
Surgeon-in-Chief, Grady Memorial Hospital

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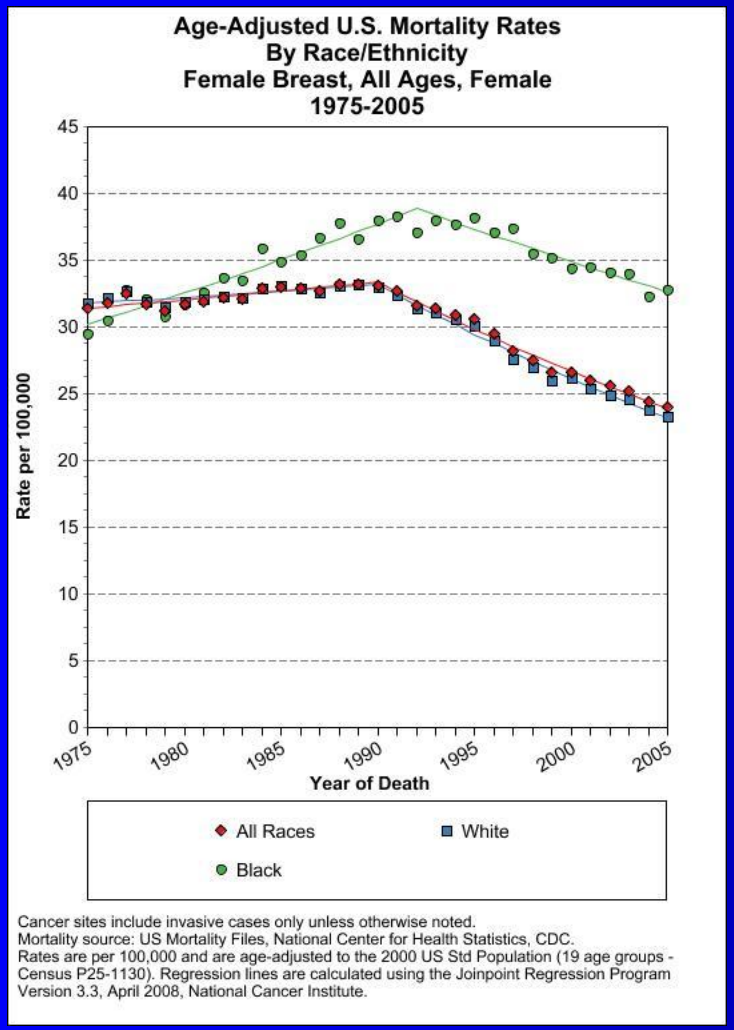
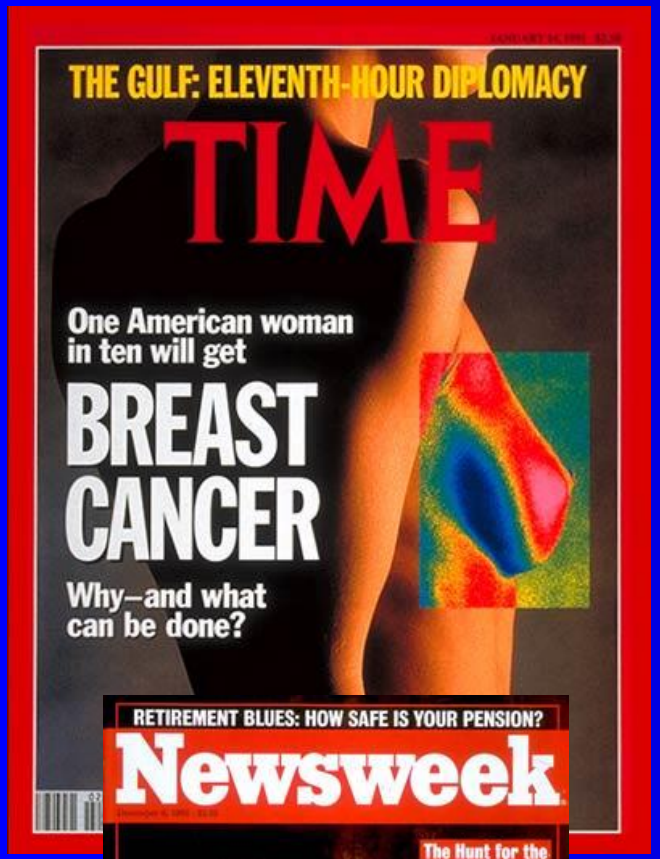
Sheryl G. A. Gabram MD
Professor of Surgery, Emory University

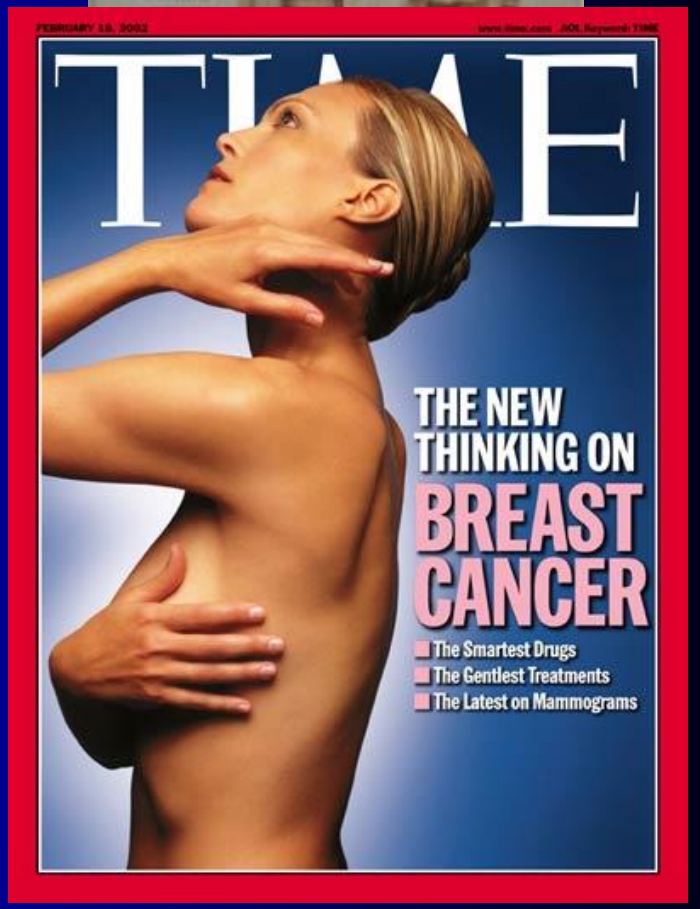
Impacting further on mortality…



Public

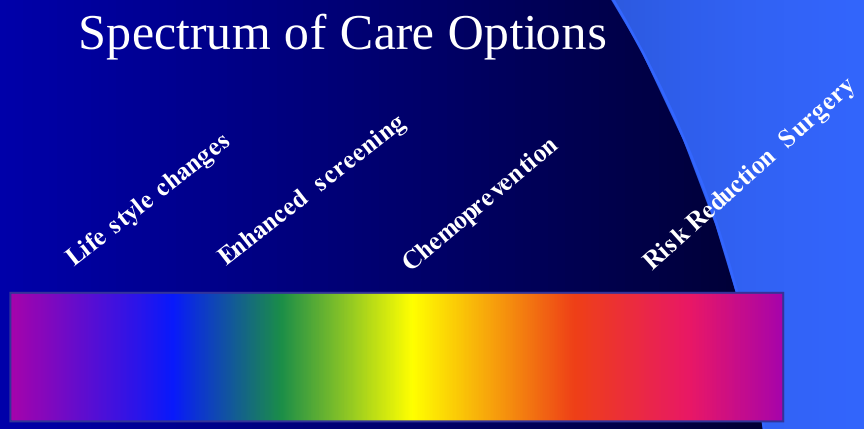
Awareness





Objective

- Discuss recommendations and treatment options for women at high risk for breast and/or ovarian cancer that include:
  - Lifestyle Changes
  - Enhanced Screening
  - Chemoprevention
  - Risk Reduction Surgery



Winship (Emory) High Risk Clinic

- *Clinical:* *One stop Shopping* started 2007
  - Over 200 patients to date

Team: genetic counselor, medical/surgical oncologists, breast imagers/pathologists, medical geneticist

- *Research:* Access to clinical trials
- Education: Genetic Counseling Training Program started Sept 2012

Bridging the Grady GAP

Genetic

Access

Program

AA ♀ 78% less likely genetic testing

Clinic Coordination, Genetic Counseling, Genetic Testing for high-risk patients and cover BRCA 1/2 test costs for uninsured

Armstrong K et al JAMA 2005;293:1729

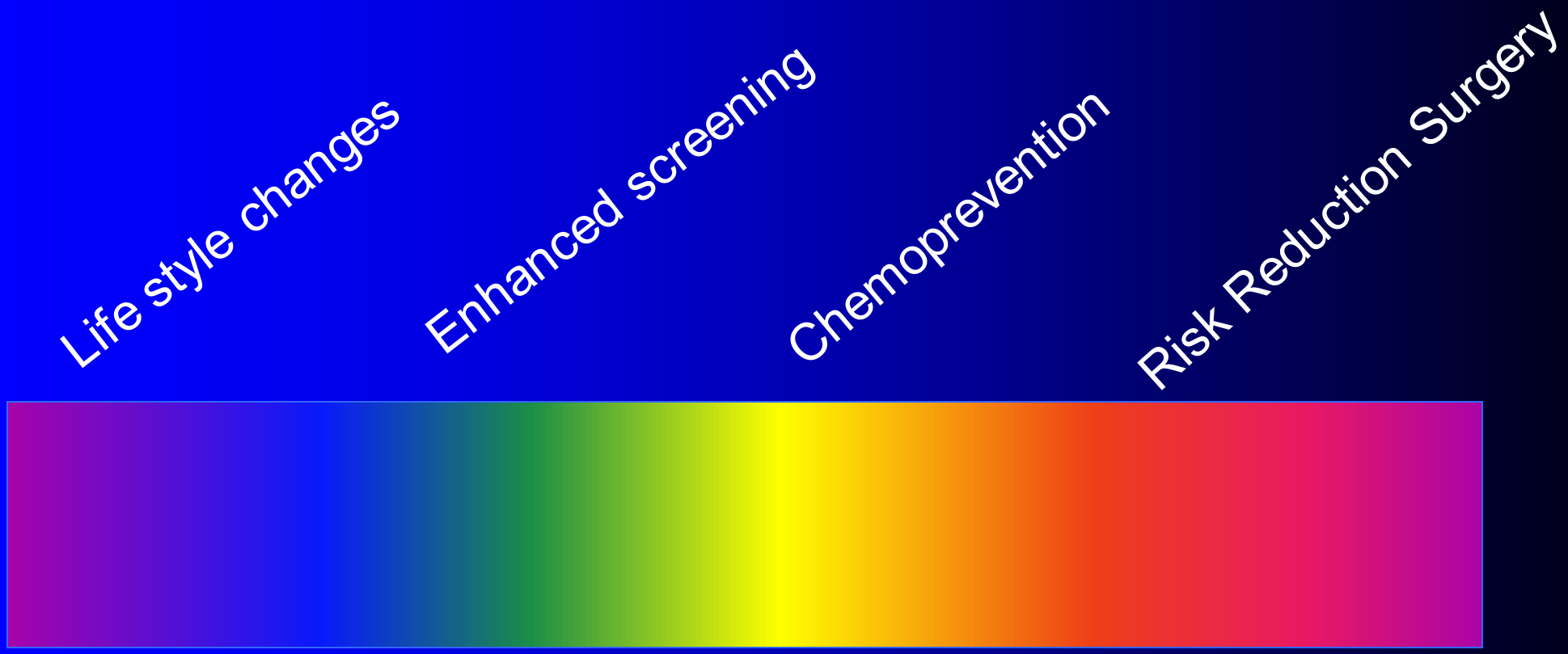
*Patients at High Risk Breast CA**

- Pedigree suggestive of genetic predisposition or lifetime risk >20%
- History of LCIS
- Prior thoracic radiation therapy (< age 30)
- 5 year Breast Cancer risk ≥ 1.70%

  http://www.cancer.gov/bcrisktool/

National Comprehensive Cancer Network
http://www.nccn.org/default.aspx

Spectrum of Care Options



Role of referral for genetic counseling/testing

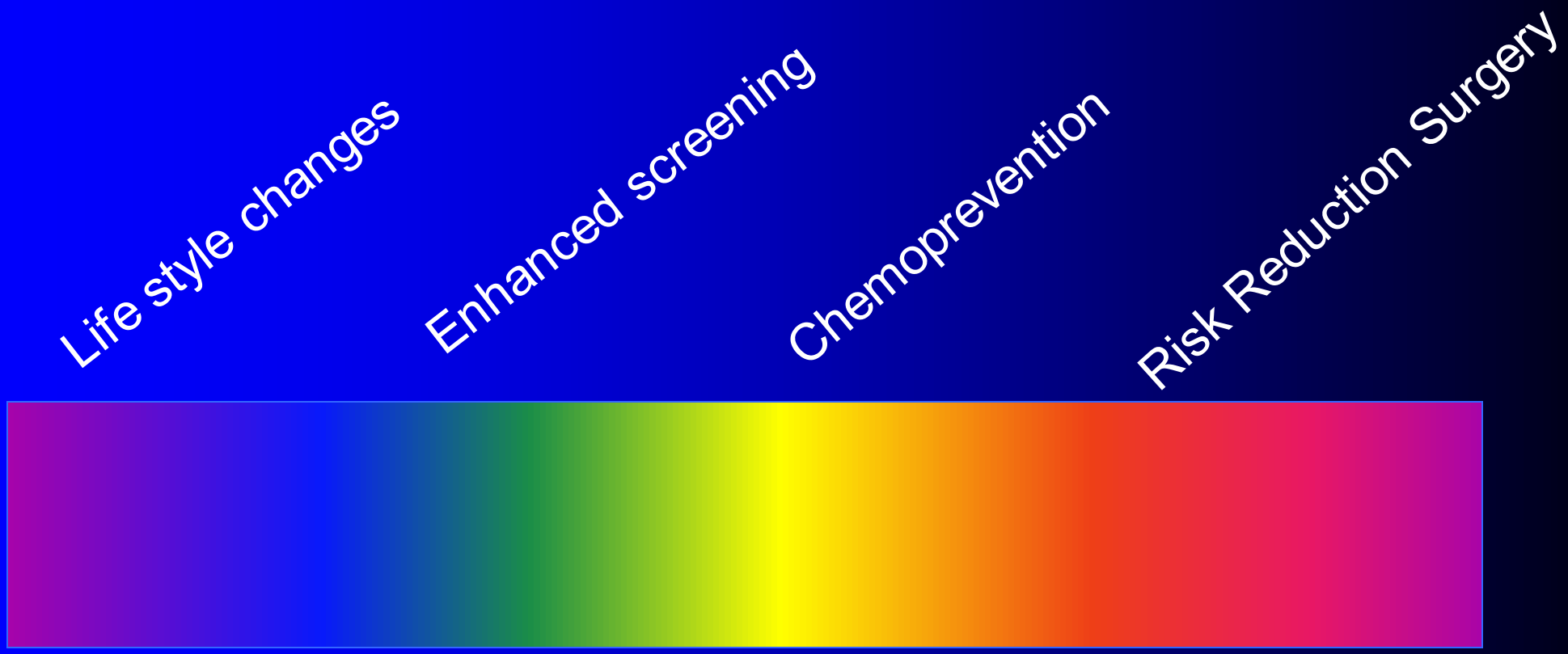
Gabram SGA et al: Breast J 2009; Suppl 1: S39-45.

Lifestyle modifications

- Strong Evidence
  - Alcohol, combination HT, weight gain, ionizing radiation
  - Physical activity (for decreasing risk)
- Unclear Evidence
  - Diet and Vitamin intake (fat intake)
  - Tobacco smoke
  - Chemicals in environment
  - Night work
- No scientific evidence
  - Antiperspirants, bras, induced abortion, breast implants

http://www.cancer.org/cancer/breastcancer/detailedguide/breast-cancer-risk-factors

Spectrum of Care Options



Role of referral for genetic counseling/testing

Gabram SGA et al: Breast J 2009; Suppl 1: S39-45.

HBOC Syndrome
Enhanced Screening Women

- Breast Awareness starting age 18
- CBE every 6-12 months starting age 25
- Annual Mammo/MRI starting age 25 or FH
- Transvaginal ultrasound/CA-125 age 30 or FH
- Consider investigational imaging and screening studies when available, in context of clinical trials

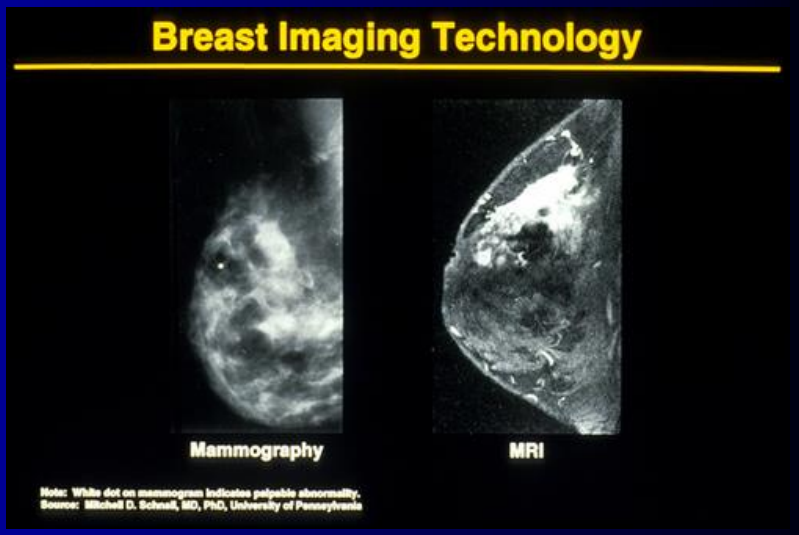
National Comprehensive Cancer Network Guidelines 4.2013

HBOC Syndrome Enhanced Screening Men

- BSE starting age 35
- CBE every 6-12 months starting age 35
- Consider baseline mmg age 40; annual if gynecomastia or parenchymal density
- Consider prostate CA screening age 40
- No guidelines: pancreatic/melanoma (may individualize)

National Comprehensive Cancer Network Guidelines 4.2013

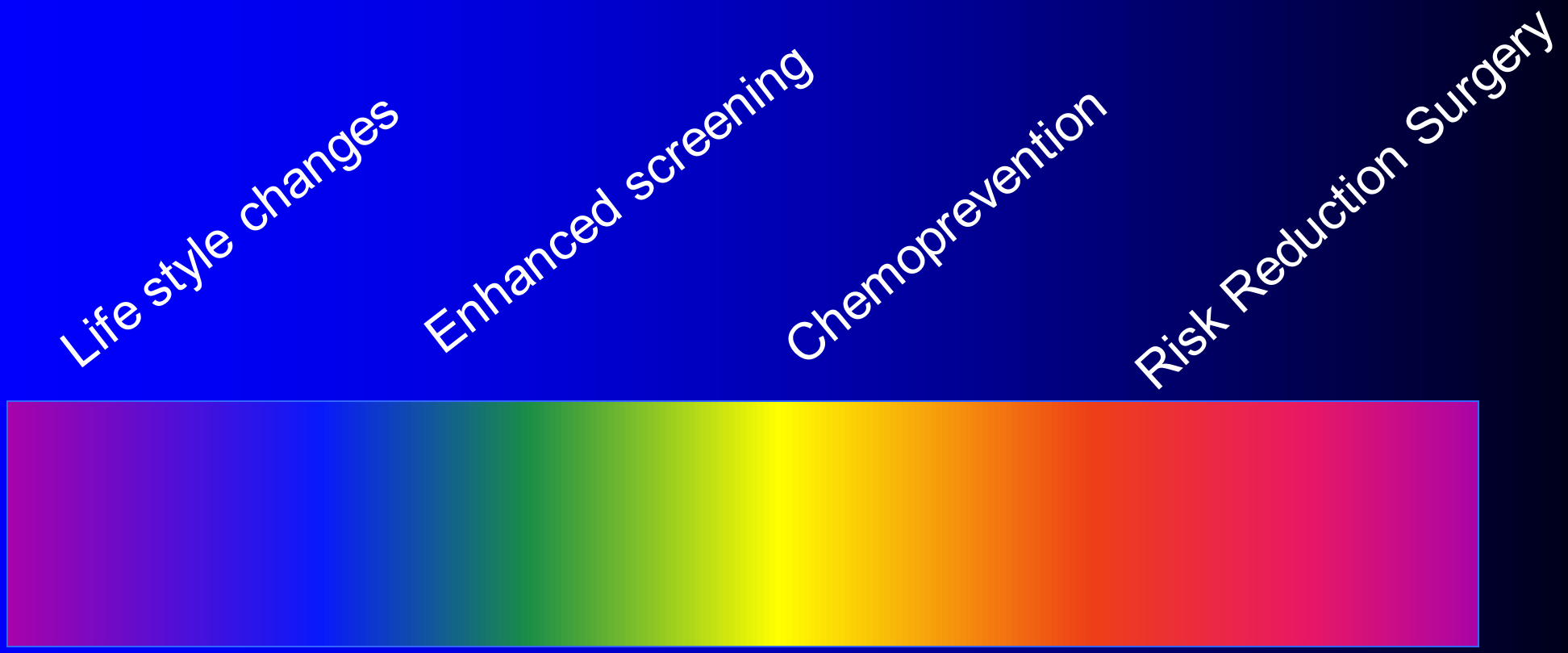
American Cancer Society Guidelines for Breast MRI



- Recommend (Evidence)
  - *BRCA1* or *BRCA2* mutation
  - 1st degree relative BRCA carrier, untested
  - Lifetime risk 20-25%
- Recommend (Expert Consensus)
  - Radiation to chest between ages 10 and 30
  - Li-Fraumeni and 1st degree
  - Cowden's and 1st degree

Saslow D, et al: CA Cancer J Clin 57:75-89, 2007

Spectrum of Care Options



Role of referral for genetic counseling/testing

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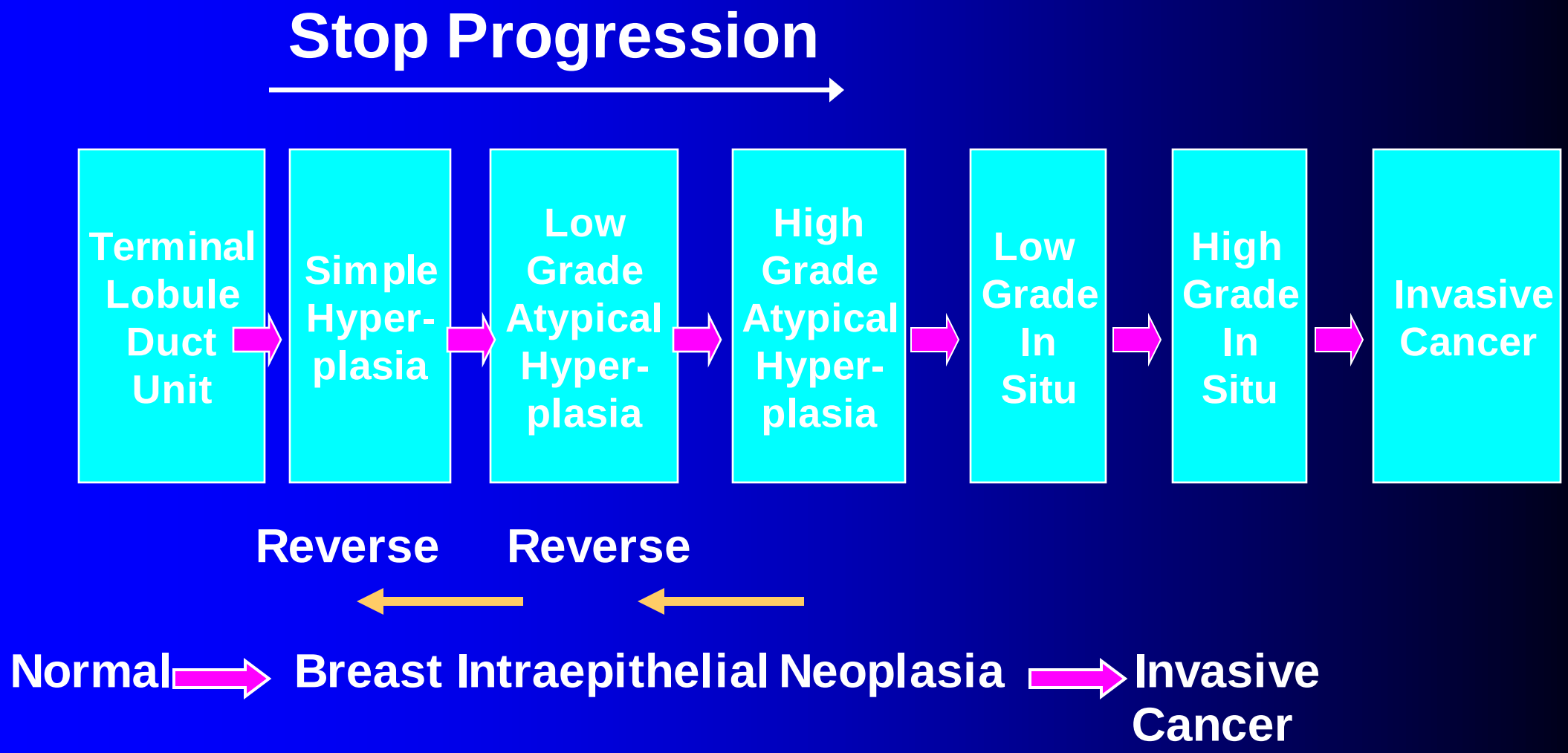
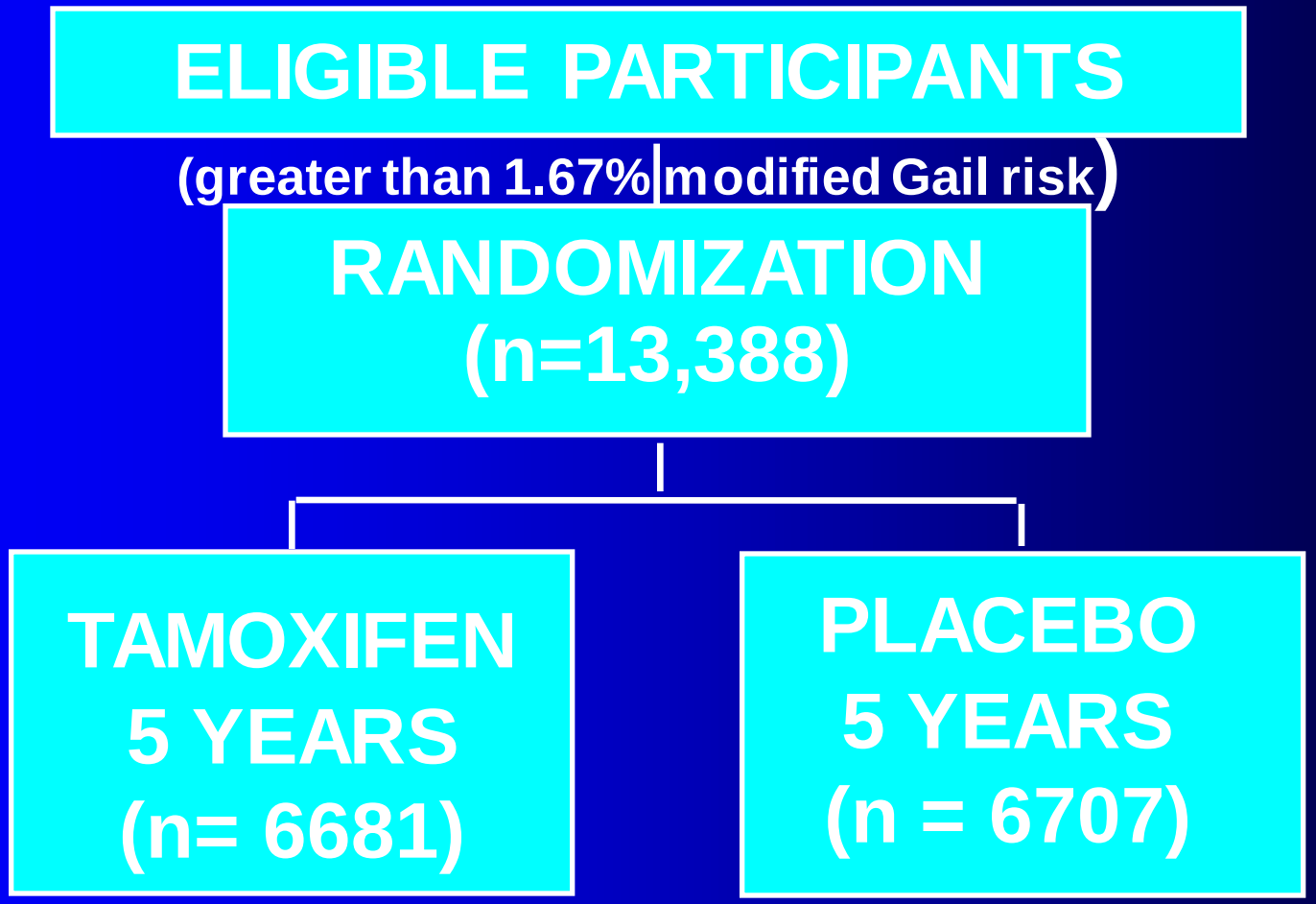
BREAST CANCER CHEMOPREVENTION

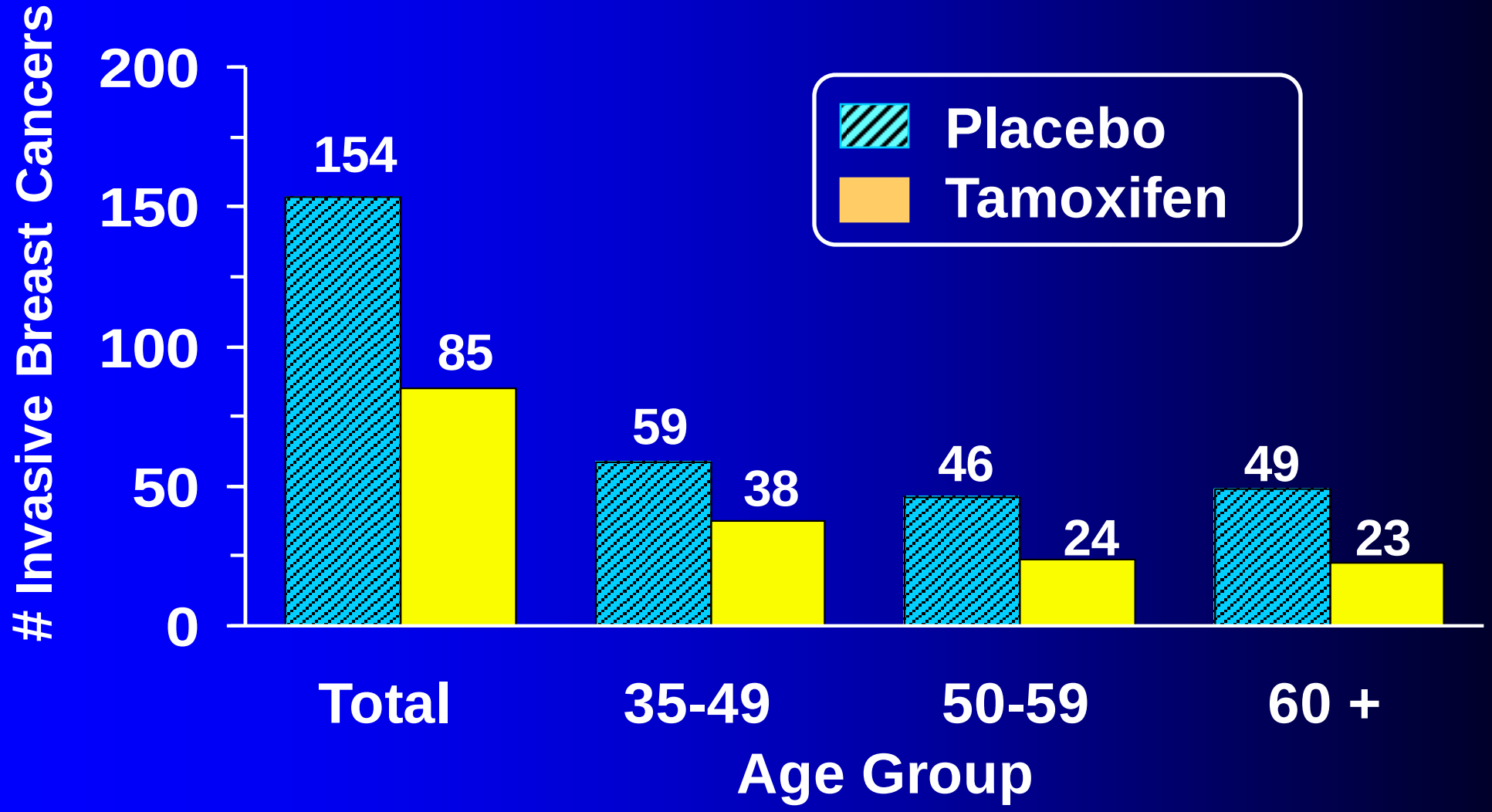


BCPT SCHEMA

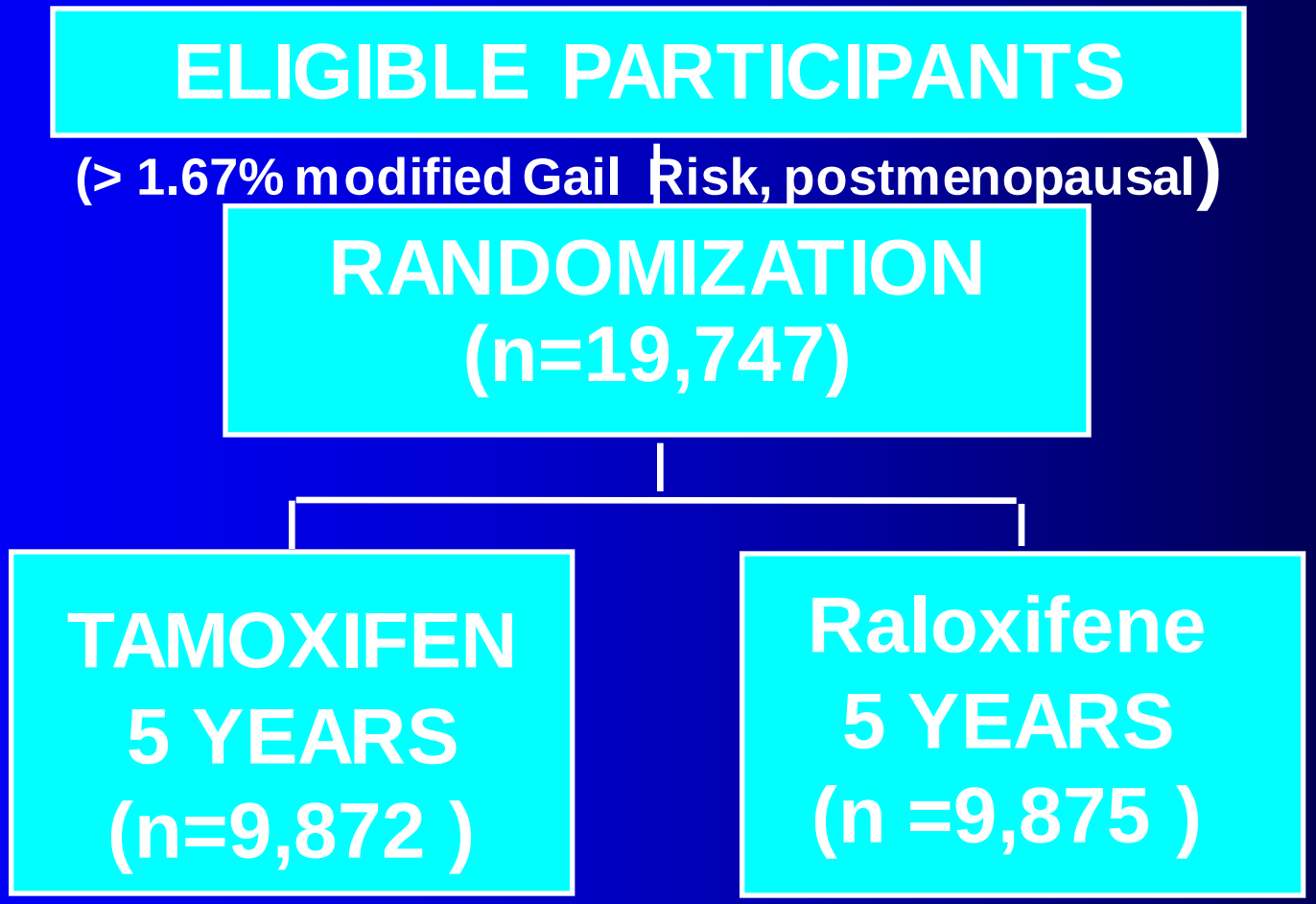


Fisher B et al: J Natl Cancer Inst 90:1371, 1998

Tamoxifen Reduced Invasive Breast Cancer in All Ages BCPT



STAR SCHEMA



Vogel V et al: JAMA 295(23): 2727-2741, 2006

Update of the NSABP Project Study of Tamoxifen and Raloxifene (STAR) P-2 Trial: Preventing Breast Cancer

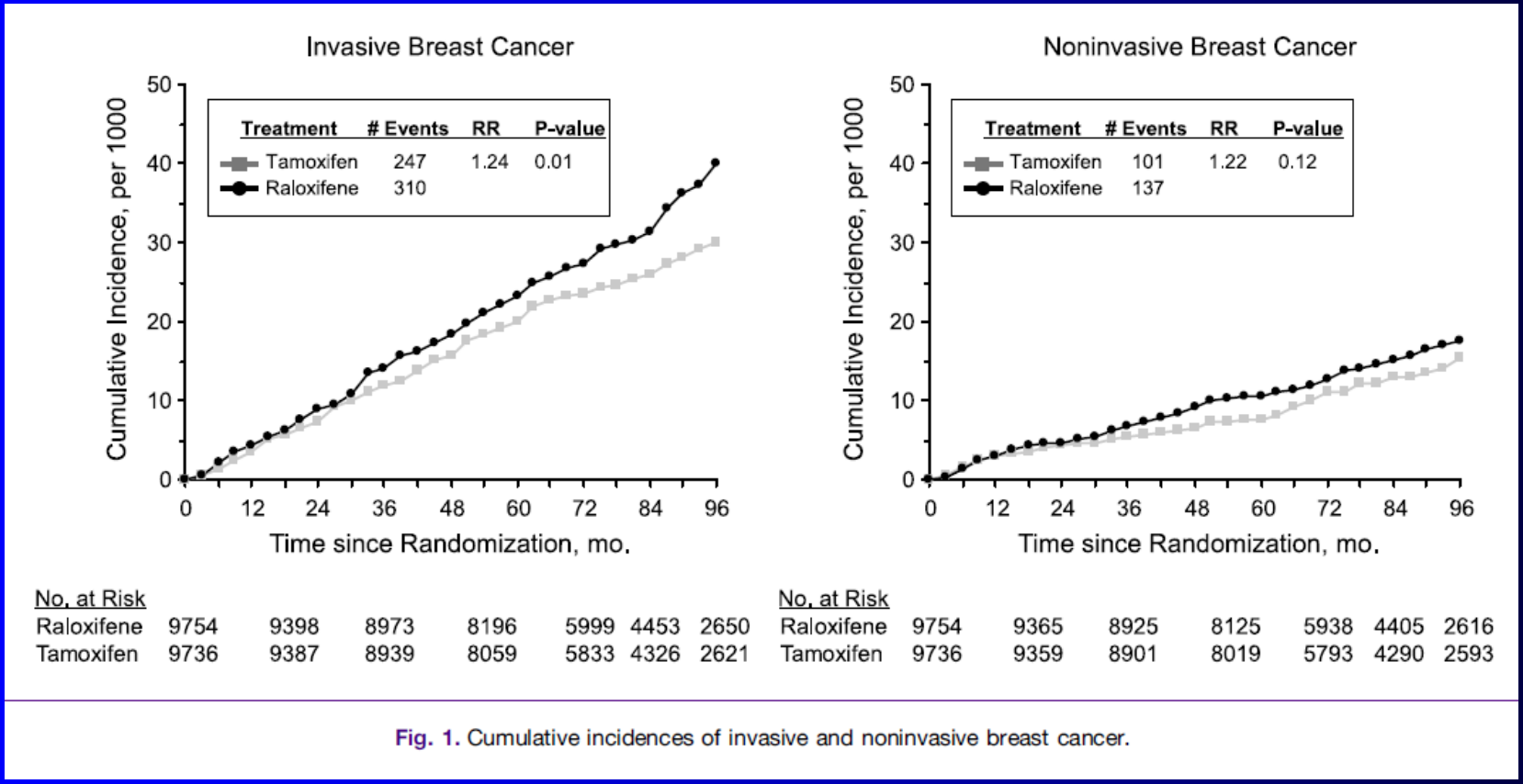


Fig. 1. Cumulative incidences of invasive and noninvasive breast cancer.

Vogel V et al: Cancer Prev Res; 3(6) June 2010

Use of tamoxifen and raloxifene for breast cancer chemoprevention in 2010

- 2000: 120,737 ♀ tamoxifen
  2010: 20,598 ♀ tamoxifen and 96,890 ♀ raloxifene
- Why so low?
  - PCPs reluctant because of side effects and lack of sufficient information about risk reduction
  - Patients reluctant: perceive more risks versus benefits, especially if informed
- Conclusion: consider medical, psychosocial and personal factors for each patient

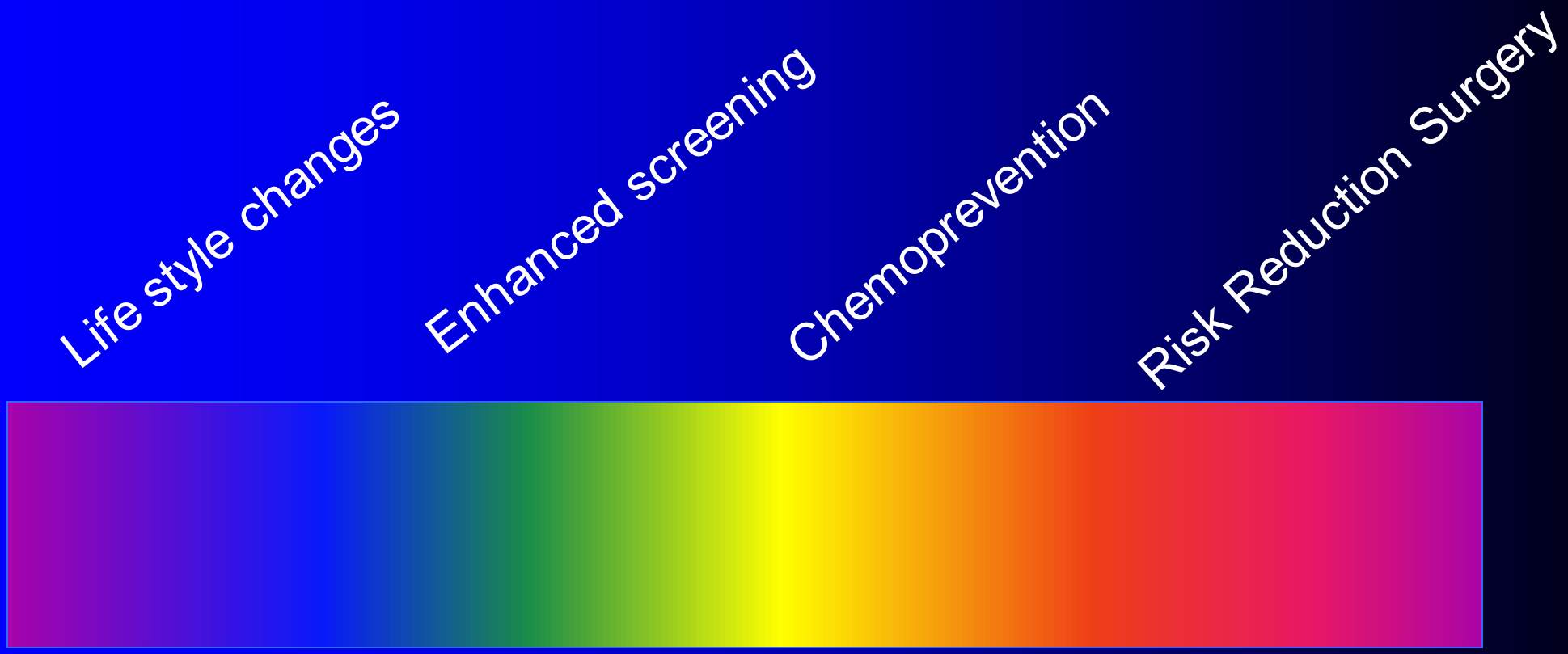
Waters EA et al: Br Can Res Treat 2012;134:875-880.

Exemestane for Breast-Cancer Prevention in Postmenopausal Women

- RCT: post menopausal (>35) one risk factor:
  - 60 yrs or >, Gail 5-year risk> 1.66%, prior ADH, ALH, LCIS or DCIS with mastectomy
- 4,560 ♀ (62.5 yrs, Gail risk 2.3%)
  - 35 month f/u, 65% relative reduction in Inv BC
  - Adverse events 88% (exem) versus 85% (plac)
  - No difference: skeletal fxn, CV events, other cancers or treatment related deaths

Goss PE et al: NEJM 2011; 364(25:) 2381-2391.

Spectrum of Care Options



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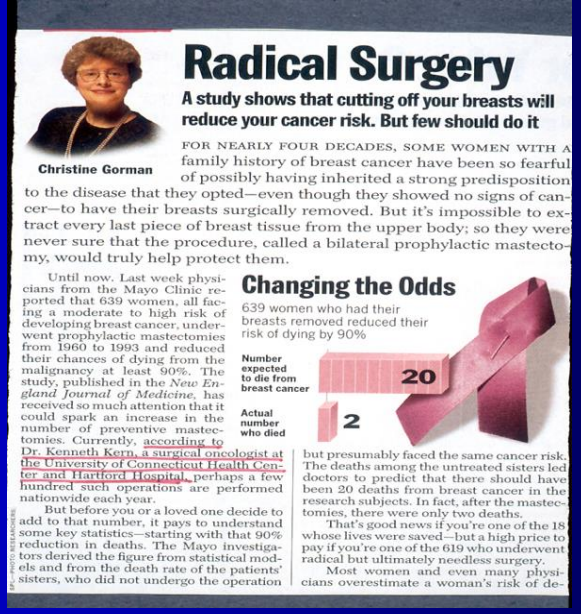
Risk Reduction Surgery



Radical Surgery

A study shows that cutting off your breasts will reduce your cancer risk. But few should do it

Christine Gorman

FOR NEARLY FOUR DECADES, SOME WOMEN WITH A family history of breast cancer have been so fearful of possibly having inherited a strong predisposition to the disease that they opted—even though they showed no signs of cancer—to have their breasts surgically removed. But it's impossible to extract every last piece of breast tissue from the upper body; so they were never sure that the procedure, called a bilateral prophylactic mastectomy, would truly help protect them.

Until now. Last week physicians from the Mayo Clinic reported that 639 women, all facing a moderate to high risk of developing breast cancer, underwent prophylactic mastectomies from 1960 to 1993 and reduced their chances of dying from the malignancy at least 90%. The study, published in the *New England Journal of Medicine*, has received so much attention that it could spark an increase in the number of preventive mastectomies. Currently, according to Dr. Kenneth Kern, a surgical oncologist at the University of Connecticut Health Center and Hartford Hospital, perhaps a few hundred such operations are performed nationwide each year.

But before you or a loved one decide to add to that number, it pays to understand some key statistics—starting with that 90% reduction in deaths. The Mayo investigators derived the figure from statistical models and from the death rate of the patients' sisters, who did not undergo the operation

Changing the Odds

639 women who had their breasts removed reduced their risk of dying by 90%

but presumably faced the same cancer risk. The deaths among the untreated sisters led doctors to predict that there should have been 20 deaths from breast cancer in the research subjects. In fact, after the mastectomies, there were only two deaths.

That's good news if you're one of the 18 whose lives were saved—but a high price to pay if you're one of the 619 who underwent radical but ultimately needless surgery.

Most women and even many physicians overestimate a woman's risk of de-

Should not be performed on healthy women before offering genetic counseling/testing

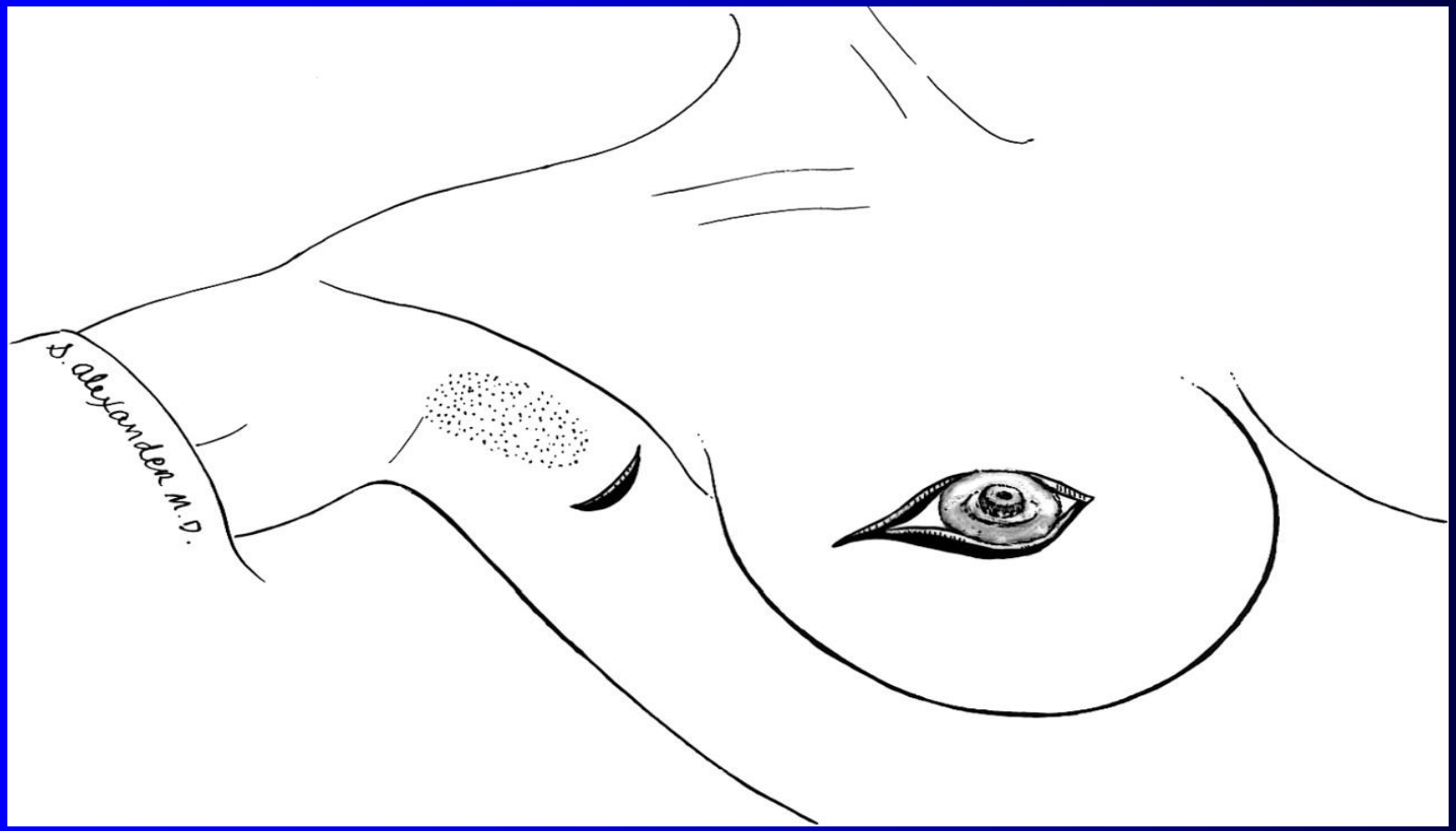
Women tend to overestimate their risk

Never an emergent or urgent procedure

Body image and effect on sexuality

Wood WC: Oncology 18: 28-32, 2004.

Skin Sparing Mastectomy

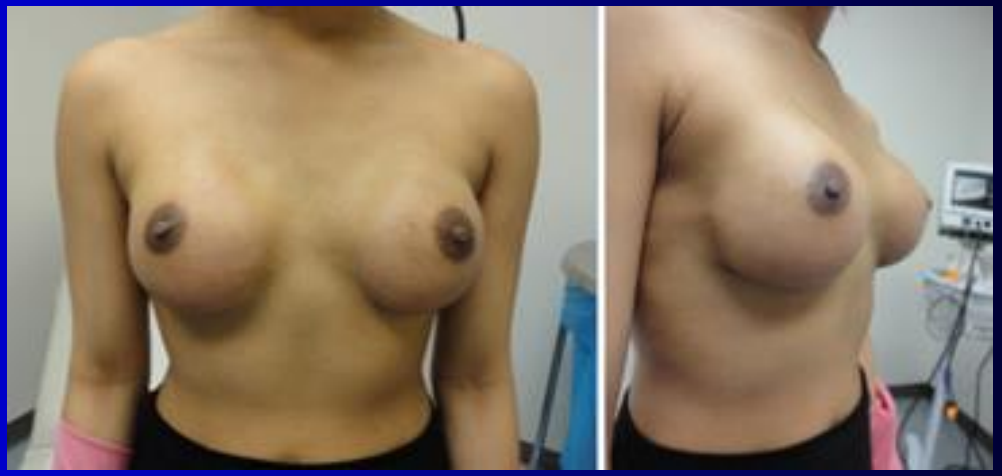


Stradling B, Ahn M, Angelats J, Gabram SGA: Less is More: Skin-Sparing mastectomy with sentinel lymph node dissection. Arch Surg 2001;136:1069-1075.

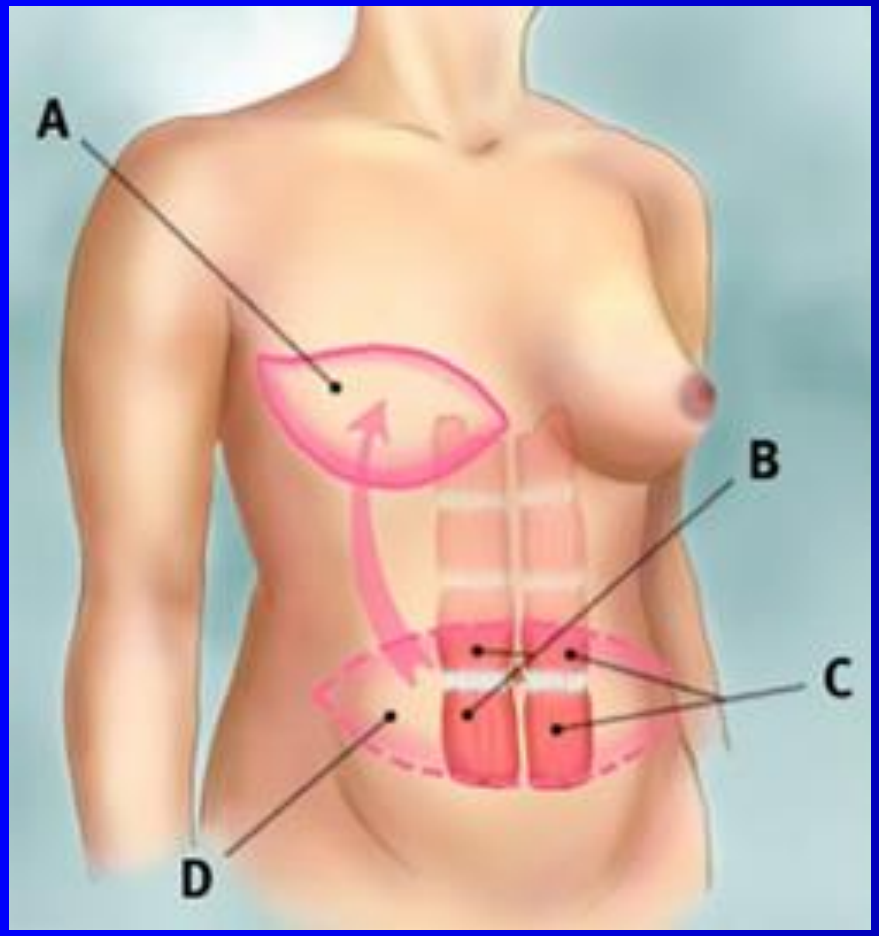
ARCHIVES OF SURGERY

Bilateral Implant/Expanders

- Skin Sparing
- Nipple sparing

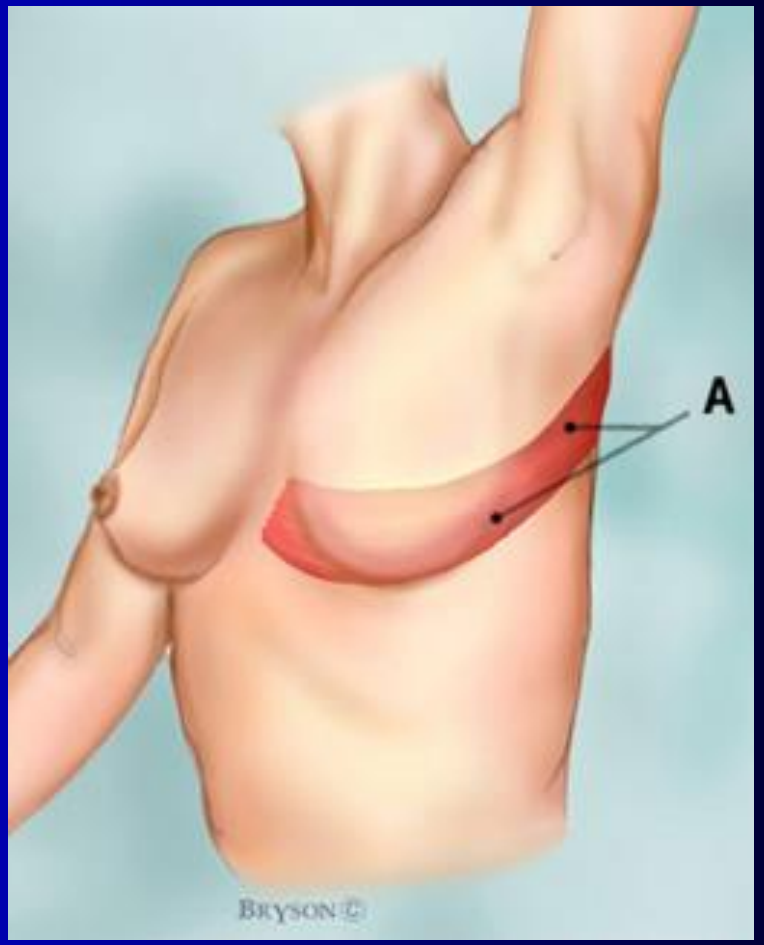


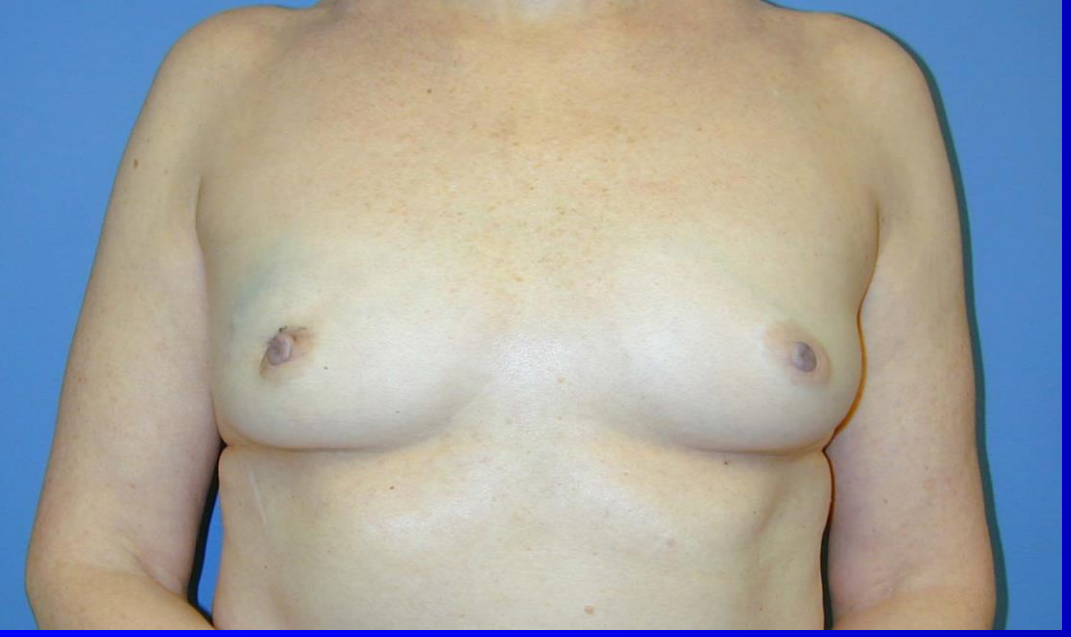
Coopey SB et al: Ann Surg Oncol 2013;20:3218-3222

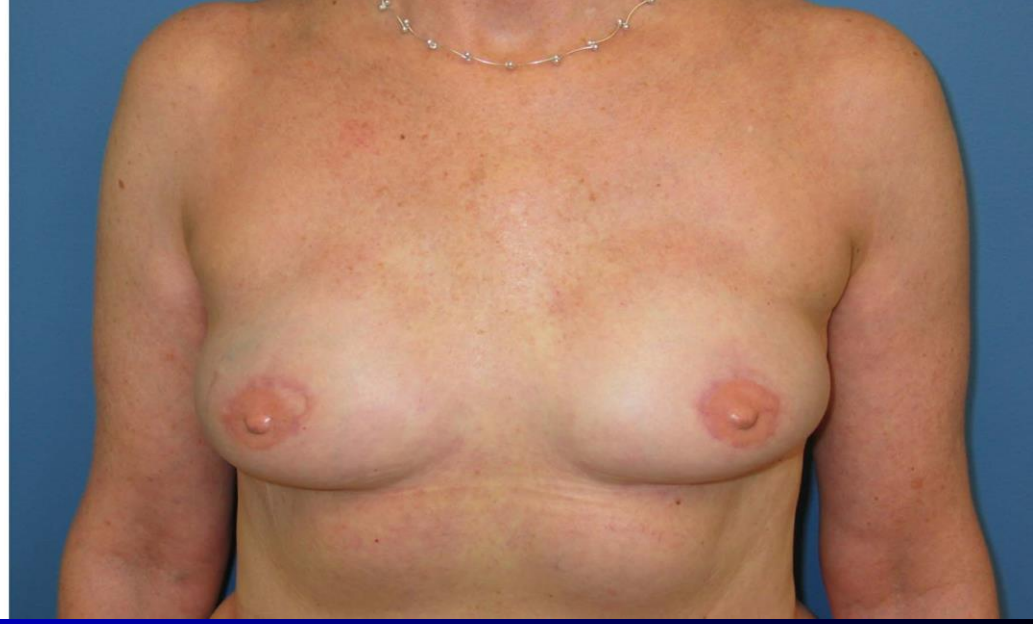


TRAM Flap

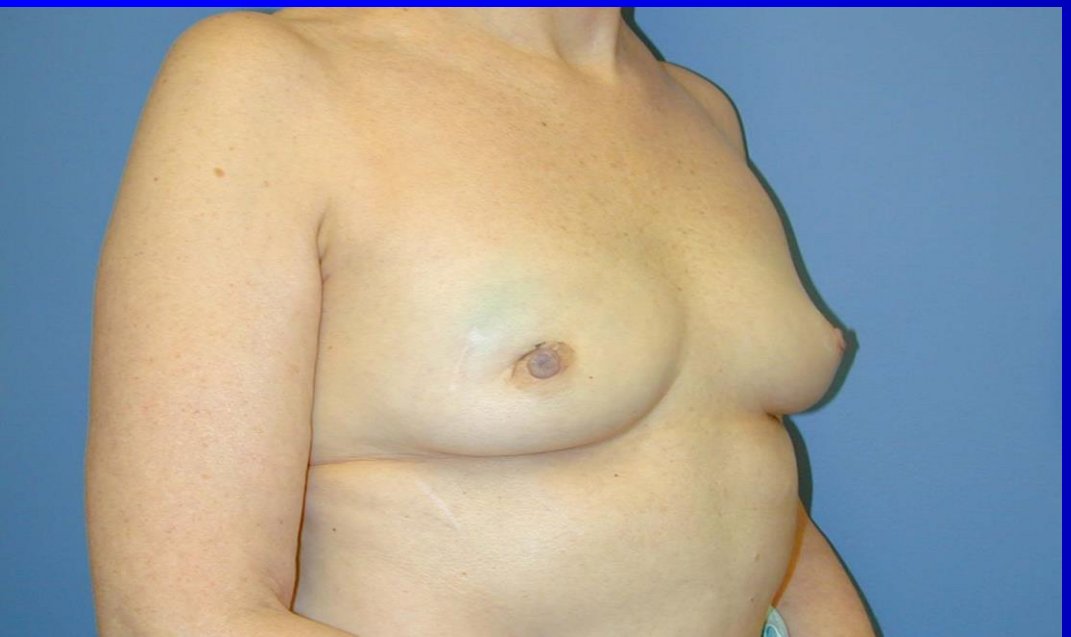
Latissimus Dorsi Flap

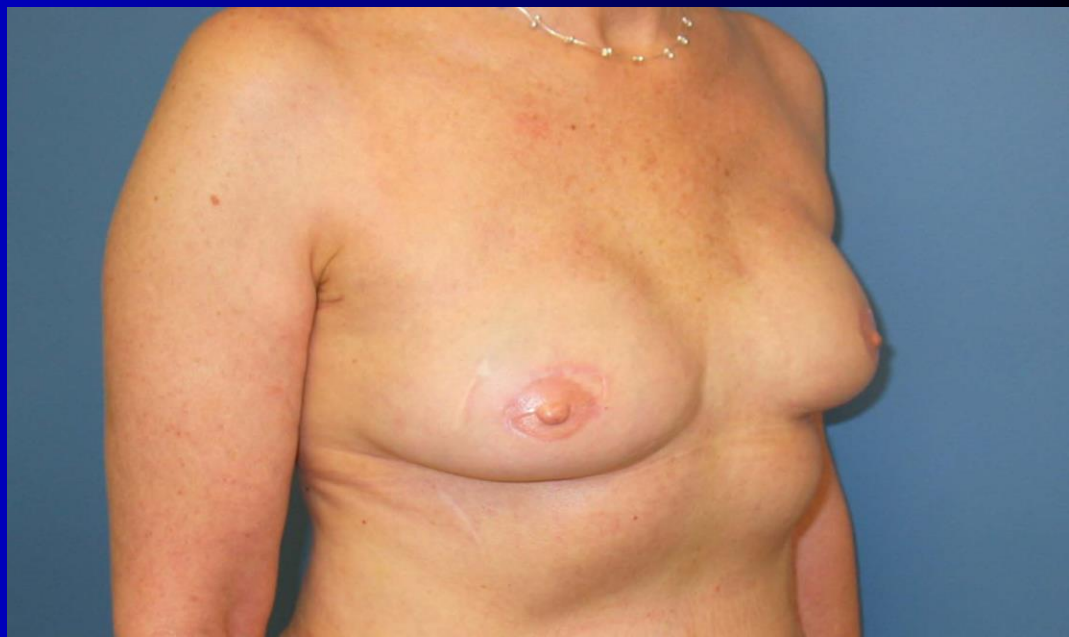






Bilateral TRAM





Association of Risk-Reducing Surgery in *BRCA1* or*BRCA2* Mutation Carriers with Cancer Risk and Mortality

- Prospective cohort study 2,482 from 1974-2008 (22 centers N. America and Europe)
- Outcome measures: Breast and ovarian CA risk, cancer-specific mortality, overall mortality
- Women followed to end of 2009: median f/u
  Surg: 3.65 yrs (.52-27.4 yrs)
  Surveillance: 4.29 yrs (.5-27.9 yrs)

Domchek S: JAMA 304: 967, Sept 2010.

Results and Conclusions

- For RRM, 0% developed BC vs 7% surveillance (3 years follow up)
- For RRSO, 1.1% developed Ovar CA vs 3% surveillance (6 years follow-up)
- RRSO: associated with reduction in all cause mortality by 60%, BC mortality by 56%, and Ovarian CA mortality by 79%
- Only 10% and 38% ♀ chose RRM and RRSO

With this data, ♀ can make more informed choices

Domchek S: JAMA 304: 967, Sept 2010

We have come a LONG Way…
Thank You and
Questions?

Insurance Protections and Coverage

The First Line of Defense: Application of Breast Cancer Genomics Standards in Primary Care

Friday, February 21, 2014

Presenter: Lisa K. Liang, Esq.
Health Law Unit (Breast Cancer Legal Project)
Atlanta Legal Aid Society, Inc.

Faculty Disclosure

In compliance with ACCME Guidelines, I hereby declare:

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Presenter: Lisa K. Liang, Esq.
Health Law Unit (Breast Cancer Legal Project)
Atlanta Legal Aid Society, Inc.

GEORGIA BREAST CANCER
GENOMIC HEALTH CONSORTIUM
EDUCATION SURVEILLANCE AND POLICY

Atlanta Legal Aid Society, Inc.

- **Private**, non-profit law firm
  - Funding for our work comes from various sources
  - Not a governmental entity
- **Free** civil (non-criminal) legal services to eligible low- income residents living in metro-Atlanta (Fulton, Clayton, Cobb, Gwinnett, and DeKalb)
- **Eligibility:** household size and income, assets, US citizens & legal residence, case priorities
- **Services:** direct legal representation, brief service, self-help, legal advice, and referral
- **Programs:** General Law, Family Law, Grandparent/ Relative Caregiver Project, Disability Integration Project, Senior Citizens Law Project, Home Defense, Health Law Partnership (HeLP), and Long-Term Care Ombudsman Program

Health Law Unit

We provide free civil legal services to low-income cancer patients and survivors so that they can focus on treatment and wellness, not on the legal issues they confront.

Types of Legal Issues We Address:

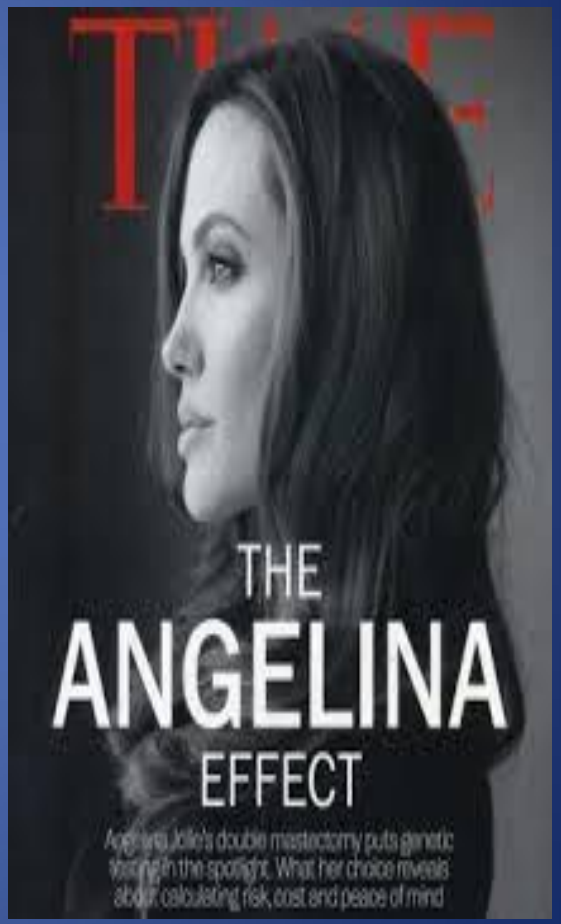
- Obtaining and maintaining public benefits (TANF, Food Stamps, Housing, Unemployment)
- Access to health care
  - Medicare/ Medicaid coverage and access issues
  - Appealing insurance coverage denials (health, long-term/short-term disability)
- Social Security Disability/SSI (initial application advocacy, appeals, overpayments)
- Debt Relief (bankruptcy and consumer advocacy)
- Landlord/ Tenant (conditions issues & evictions)
- Wills, Advance Directives for Health Care, POAs, Nominations of Guardians for Minor Children
- Family Law (domestic violence, divorce, separation, child custody, child support)
- Employment Issues (FMLA, Reasonable Accommodations under ADA, COBRA)
- Predatory Mortgage Lending, Foreclosure Issues, and Consumer Fraud

Breast Cancer Legal Project
Atlanta Legal Aid Society • Health Law Unit

CANCER LEGAL INITIATIVE
A Division of Atlanta Legal Aid Society

Today's Topics

- How to get screened/tested?
  - Impact of Affordable Care Act (ACA) on access to BRCA gene screening and testing
- What happens after screening/testing?
  - Genetic Information Nondiscrimination Act (GINA)
- What if my patient doesn't have insurance?



Affordable Care Act Provisions: Access to Testing

- Must cover 22 preventative health care without charging co-pay or co-insurance (even if deductible hasn't been met)
  - BRCA genetic test counseling and Breast Cancer Chemoprevention counseling for women at higher risk
  - Mammography (every 1-2yrs for women 40+)
  - Well-woman visits to get recommended services for women under 65
  - *Provision is being challenged because it includes contraceptive health care (only enjoined as to contraception)*
- In/Out-of-Network: If a plan doesn't have an in-network provider to provide the service, plan must cover service provided by an out-of-network provider and cannot impose cost-sharing

Affordable Care Act: BRCA Counseling and Testing

- If one has a high risk for breast cancer, plan must cover genetic counseling and evaluation AND the BRCA test itself (w/o cost-sharing).
- High risk means those who have/are:
  - Two+ first-degree relatives (parent, children or siblings) who had breast cancer with at least one being diagnosed before the age of 50,
  - Three + first- or second-degree relatives (grandparent, grandchild, uncle, aunt, nephew, niece, half-sibling) with breast cancer,
  - A combination of breast and ovarian cancer among first- and second-degree relatives,
  - A male family member who had breast cancer OR
  - Ashkenazi Jewish women with one first-degree relative or two second-degree relatives with either breast or ovarian cancer.
- If BRCA gene mutation present, ACA requires coverage of counseling to help decide course of treatment

Affordable Care Act: Post-Testing and Counseling

- Prohibits insurance companies from refusing to cover people, or charging more, based on any preexisting condition
- Elimination of annual and lifetime caps on insurance coverage
- Caps out-of-pocket healthcare expenditures (sliding scale)
- Coverage for those enrolled in clinical trials for the prevention, detection and treatment of cancer and other life-threatening illnesses
- *Note: Women's Health and Cancer Rights Act: mastectomy benefits must cover reconstruction of the breast removed and surgery/reconstruction of other breast to look symmetrical/balanced post-mastectomy, any external breast prostheses and treatment of any physical complications*

Affordable Care Act: Exclusions

- Genetic testing limited to those at high-risk for BRCA gene mutation but NOT for
  - those with a low-risk for BRCA gene mutation,
  - those with Lynch Syndrome or other hereditary cancer syndromes,
  - diseases other than breast cancer,
  - women who have already been diagnosed with cancer or
  - men.

BRCA Testing under Medicare

- **Similar to ACA, BRCA genetic counseling and testing available for those with:**
  - personal history of breast cancer + one or more of the following:
  - Diagnosed age ≤45 y, with or without family history
  - Diagnosed age ≤50 y or two breast primaries, with ≥1 close blood relative(s) with breast cancer ≤50 y or ≥1 close blood relative(s) with ovarian cancer/fallopian tube/primary peritoneal cancer
  - Two breast primaries when first breast cancer diagnosis occurred prior to age 50
  - Diagnosed at any age, with ≥2 close blood relatives with breast and/or epithelial ovarian/fallopian tube/primary peritoneal cancer, at any age
  - Close male blood relative with breast cancer
  - Personal history of epithelial ovarian/fallopian tube/primary peritoneal cancer
  - If of certain ethnicity associated with higher mutation frequency, (eg, founder populations of Ashkenazi Jewish, Icelandic, Swedish, Hungarian or other) no additional family history required
  - a close relative with a known BRCA1 or BRCA2 gene mutation
  - Personal history of epithelial ovarian/fallopian tube/primary peritoneal cancer.
  - Personal history of male breast cancer.
- **Some states offer BRACAnalysis for genetic counseling through Medicaid (not GA); Georgia Medicaid covers chemoprevention counseling**

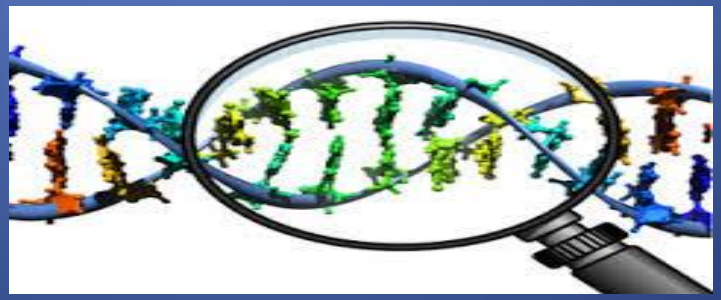
My patient knows she has the BRCA gene mutation – now what?

What are the legal ramifications of having the BRCA genetic information?

- Provides federal protection from genetic discrimination in health insurance (Title I) and employment (Title II)
- Genetic information
  - ≠ Symptomatic, diagnosed, manifested condition
  - Includes genetic information of individual and family members (up to fourth-degree)
- Only applies to health insurance (not life, disability, etc.); not applicable to US Military/VA benefits (but similar legislation in place for those benefits)
- Signed into law 2008, effective by end of 2009 (applies to genetic information obtained prior to law but no retroactive remedy)

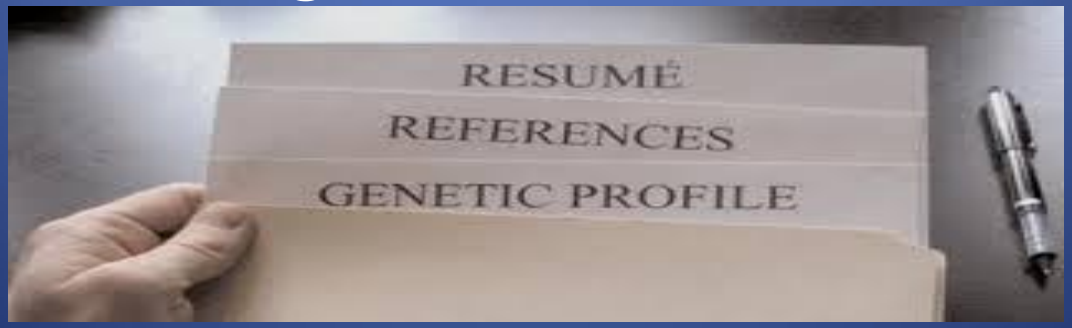
Title I of GINA

- Title I: Health insurance companies cannot use or require genetic information to make decisions about a person's insurance eligibility, coverage or cost
  - Exception: health insurer can ask for genetic information to make a decision about whether it will pay for a requested test, treatment, or procedure, in order to determine medical need for the service



Title II of GINA

- Employers CANNOT use genetic information:
  - To make decisions about hiring, promotion, etc.
  - To request, require, purchase employee's genetic information
  - During any part of employment – on applications, post-offer or fitness-for-duty examinations
- If inadvertently or otherwise receives genetic information, must keep information confidential
- Applies to employers of 15+ employees, employment agencies, labor organizations, etc.



Remedies for Title I Violations

- State: Georgia Office of Insurance and Safety Fire Commissioner (www.oci.ga.gov) – Consumer Services Division
- U.S. Department of Labor has jurisdiction over employer health benefit plans (Sec'y of Labor can fine employer-sponsored health plan)
- IRS can assess tax penalties on employer-sponsored health plans
- U.S. Department of Health and Human Services has jurisdiction over individual policies, employer-provided plans & Medigap plans

Remedies for Title II Violations

- GINA enforced by Equal Employment Opportunities Commission (EEOC) enforces Title II of GINA
- File charge of discrimination with EEOC within 180 days of alleged violation
- EEOC will investigate and either initiate suit or issue Right to Sue letter
- Since 2009, 700+ charges received by EEOC

Other Genetics Laws

- HIPAA: applies to employer-based and commercially issued health insurance only
  - Prohibits use of any health status-related factor as a basis for limiting or denying eligibility for coverage or for charging individual more for coverage

Uninsured Patients: Access to Genetic Counseling and Testing

- Women's Health Medicaid Program does exist for women with breast and cervical cancers
- Problematic for Georgians for:
  - Those who earn under 138%FPL are not Medicaid eligible nor are they eligible for subsidies for private insurance. ACA intended for them to have Medicaid coverage but Georgia is not expanding Medicaid. This population segment is uninsured and has limited access to BRCA counseling and testing.
- Other sources for BRCA counseling and testing include Public Health Departments. Seek out Patient Navigators!

GEORGIA core CENTER *for* ONCOLOGY RESEARCH & EDUCATION

For Patients:
Accessing Our Services

Telephone Intake

- Confidential Voice Mail Line
- (404) 614-3969
- 9:00am – 12:00pm Mon., Tues., Wed., Thur.

Walk-in

- 151 Spring Street, NW, Atlanta, GA
- 9:00am – 12:00pm Mon., Tues., Wed., Thur.

Outreach

- Grady GCCE (2nd Wednesday and 4th Monday 9:30 am - 11:30am)DeKalb Medical, Decatur Campus (3rd Monday, 12pm – 2pm)

QUESTIONS?

Contact Information for Providers:

Lisa K. Liang, Attorney at Law

Atlanta Legal Aid Society, Inc.

151 Spring Street NW

Atlanta, GA 30303

(404) 614-3971 (direct)

lkliang@atlantalegalaid.org

Always comfy in jeans, but is there comfort in genes?

Sarah Iwanski

February 21, 2014

Faculty Disclosure

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Sarah Iwanski

Presentation

- 29 year old female
- 2 months post-partum and breastfeeding
- Essentially negative chest CT with incidental finding of bilateral lymphadenopathy
- Lymph nodes non-tender and mobile on exam

Family History

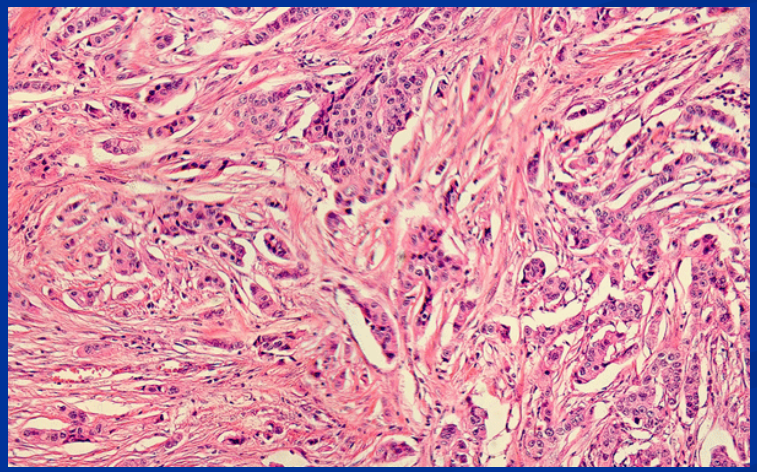
- Ovarian Cancer: maternal grandmother, 76 years
- Breast Cancer: maternal cousin, 32 years
- Prostate Cancer: paternal uncle, 55 years

Road to Diagnosis

- Pulmonologist: physician dictates "most likely non-pathologic, ultrasound in a few months if needed"
- OB/GYN: lymph node ultrasound ordered, negative results per secretary over the phone
  - BIRADS 2
- Oncologist: ordered lymph node biopsy per patient request

Pathology

- Stage IIIA invasive ductal carcinoma
- T1b N2 M0
  - 0.7 cm breast tumor
  - 4 lymph nodes macroscopically positive
  - No evidence of metastasis by PET scan



Making testing decision

- COST!!!
- Current treatment plan
- Future treatment
- Children's medical care

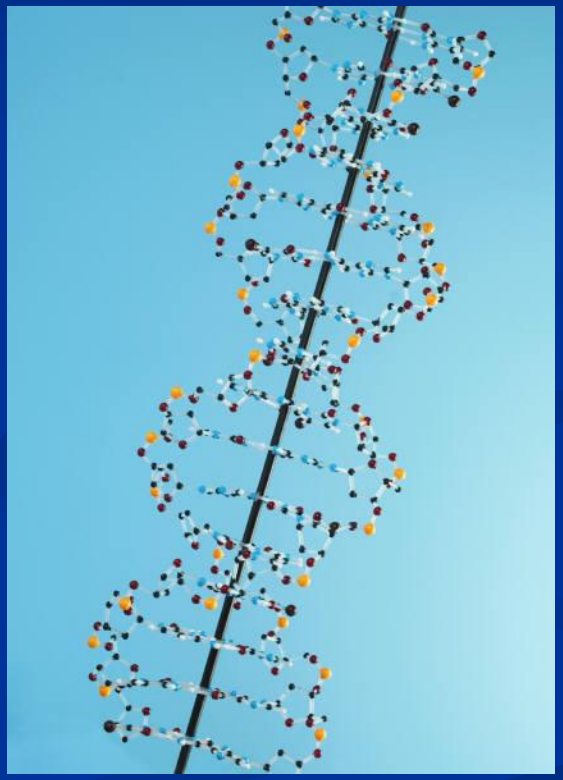
Genetic counseling and testing

- Informed DNA provided a phone interview and online preparation of risk factors, medical, and family histories.
- Blood specimen was collected, processed and shipped by a local hospital
- The results were given by phone and a copy emailed to the patient.

Results

- BRCA 1/2: No mutation detected by comprehensive gene sequencing and rearrangement (BART) analysis.

- Myriad Integrated BRACAnalysis



Other genetic causes

- Li Fraumeni syndrome
- Cowden syndrome
- Hereditary hemorrhagic telangiectasia

Affordable Care Act

- Genetic counseling
- BRCA testing covered if appropriate
- Covered for high-risk patients, as determined by clinical expertise.
- New regulations do not specify coverage for men, survivors, or testing for rarer genetic causes of breast cancer.

- OUR MISSION: to provide navigation to women to help eliminate barriers to quality healthcare, to provide emotional support, and to coordinate patients across a disconnected health system.

www.survivingsisters.org

Questions

GEORGIA BREAST CANCER
GENOMIC HEALTH CONSORTIUM
EDUCATION SURVEILLANCE AND POLICY

The First Line of Defense: Application of Breast Cancer Genomics Standards in Primary Care

February 21, 2014

Monique Martin, MPH, CHES
Health Education and Communication Specialist
Georgia Center for Oncology Research and Education (Georgia CORE)

FACULTY DISCLOSURE

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Monique Martin, MPH, CHES

Health Education and Communication Specialist

Georgia Center for Oncology Research and Education (Georgia CORE)

OUTLINE

- ❑ Consortium's Background
- ❑ Why we are here
- ❑ Policy Accomplishment
- ❑ Screening Women in Public Health
- ❑ Challenges & Successes
- ❑ Post-test

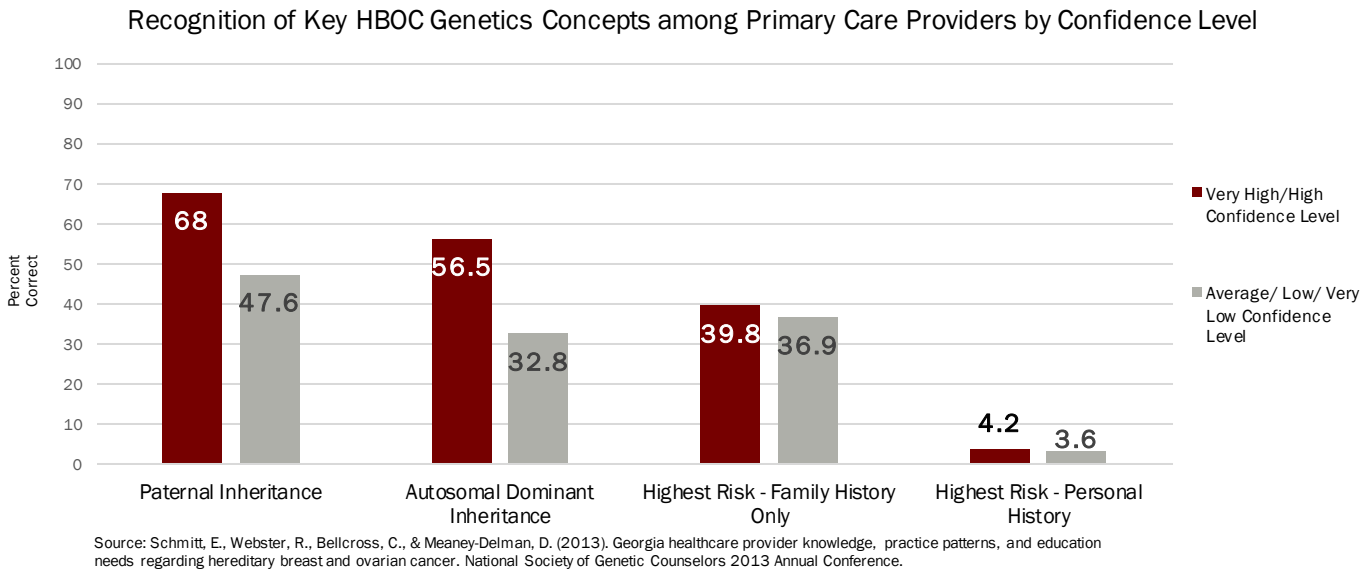
WHY ARE YOU HERE???

Callaway Gardens®

You're here because…

Needs Assessments:

- Young Breast Cancer Survivors
- Primary Care Residencies
- Cancer Genetic Counselors
- **Primary Care Providers**

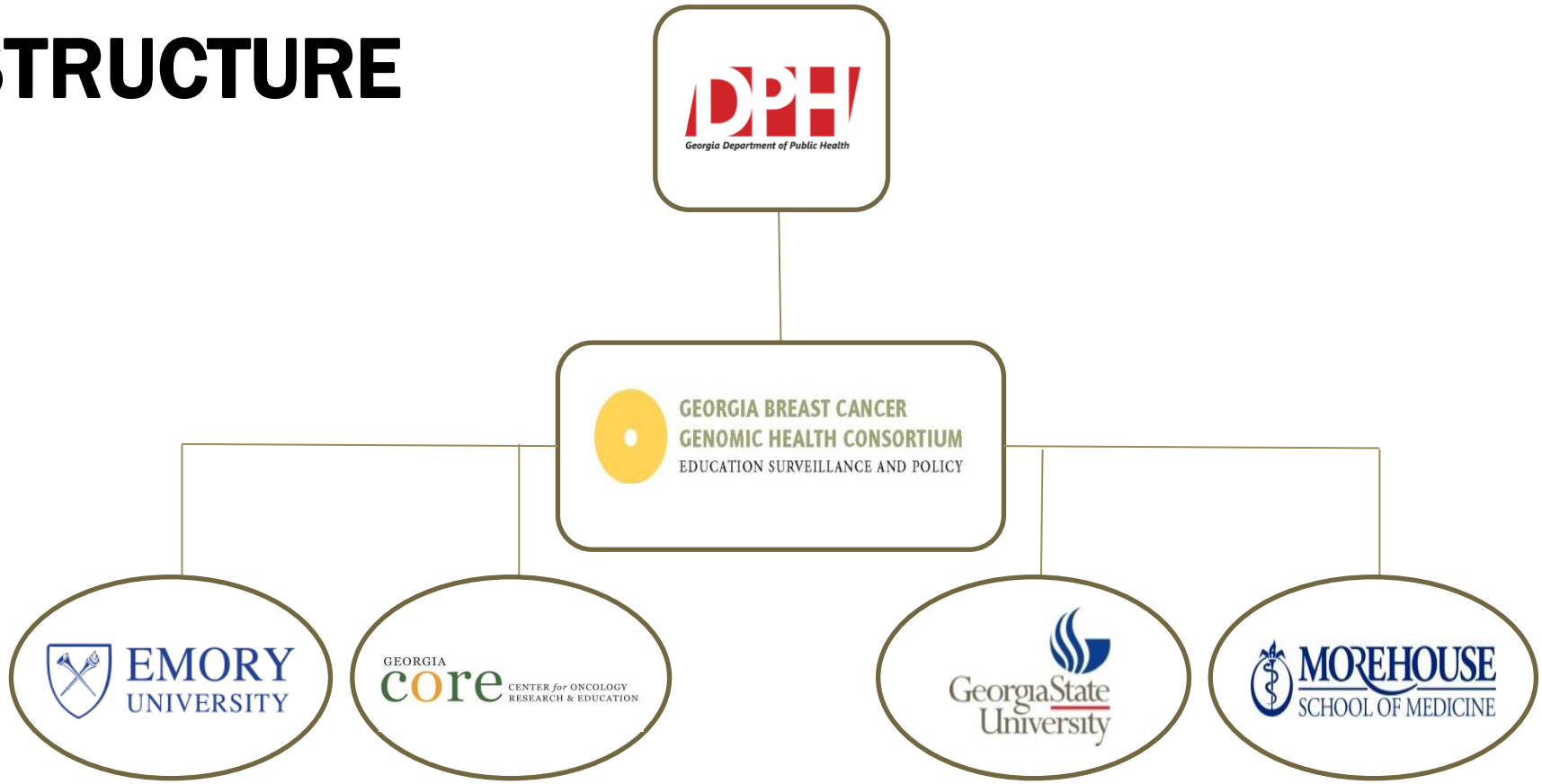


Source: Schmitt, E., Webster, R., Bellcross, C., & Meaney-Delman, D. (2013). Georgia healthcare provider knowledge, practice patterns, and education needs regarding hereditary breast and ovarian cancer. National Society of Genetic Counselors 2013 Annual Conference.

- 275 licensed Primary Care Providers were surveyed.
- Found a significant HBOC knowledge deficit among Georgia PCPs, particularly with respect to identification of high-risk individuals.

GEORGIA BREAST CANCER GENOMIC HEALTH CONSORTIUM
EDUCATION SURVEILLANCE AND POLICY

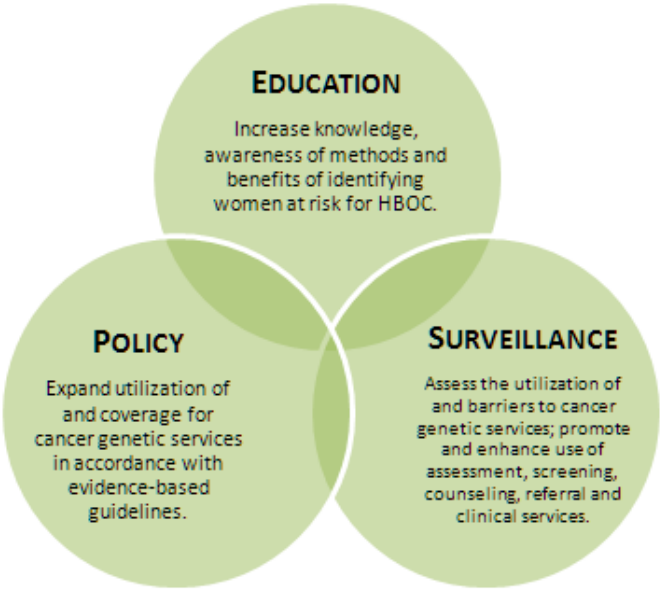
CONSORTIUM'S STRUCTURE



A Public Private Partnership

PURPOSE

To promote use of evidence-based guidelines for breast and ovarian cancer genetic risk assessment, counseling and testing, and improve the identification of young women at genetic risk for these cancers.



Policy

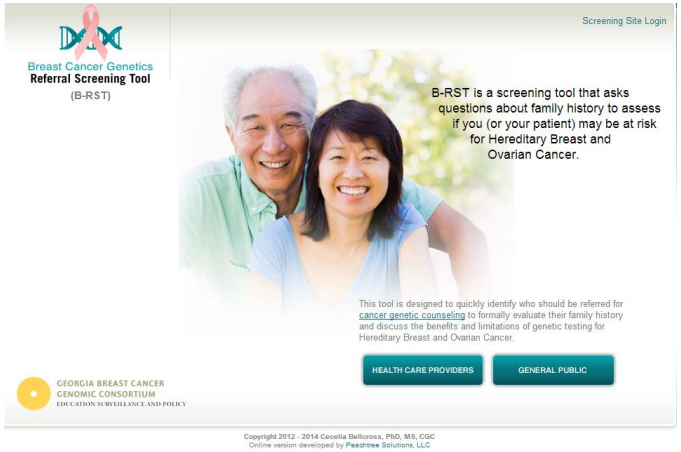
Approval of $46,000 **Genetic Testing Fund** with proceeds from GA Breast Cancer License Tag

Every $22 Dollars from the purchase or renewal of a GA Breast Cancer License Tag is deposited in the Indigent Care Fund for breast health programs for the underserved

Creates a new stream of funding for the underinsured for genetic testing for hereditary breast and ovarian cancer in Georgia.

Made Available January 2014

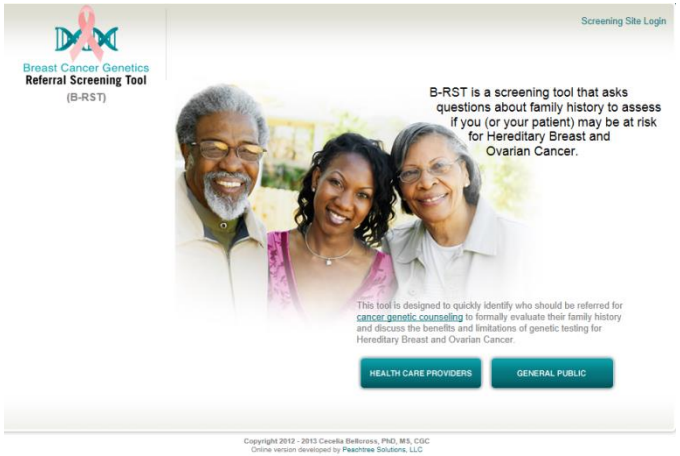
BREASTCANCERGENESCREEN.ORG



- Website developed using B-RST Algorithm
- 6 Questions
- 3 Results:
  - Negative
  - Moderate
  - Positive
- Targeted provider and public information

GEORGIA BREAST CANCER GENOMIC HEALTH CONSORTIUM
EDUCATION SURVEILLANCE AND POLICY

BREASTCANCERGENESCREEN.ORG

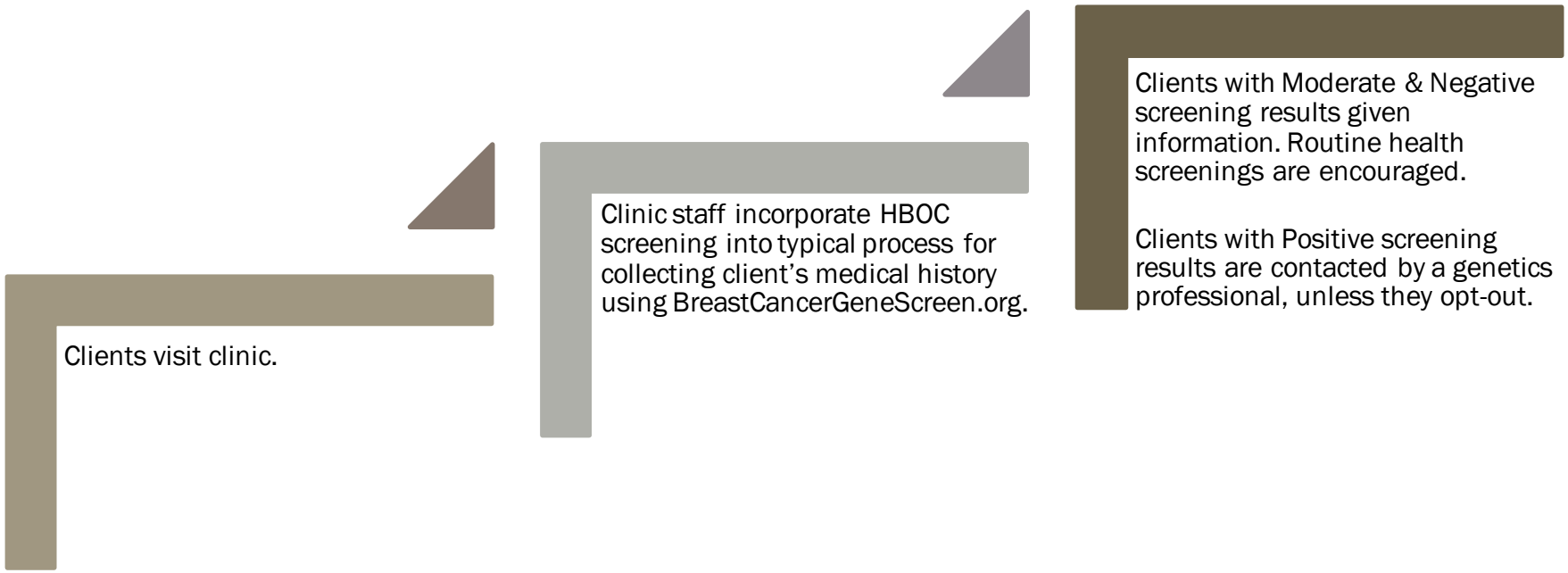


- USPSTF's recommendation cites Breast Cancer Genetics Referral Screening Tool (B-RST*) and www.BreastCancerGeneScreen.org
  - "Simplest and quickest to administer."

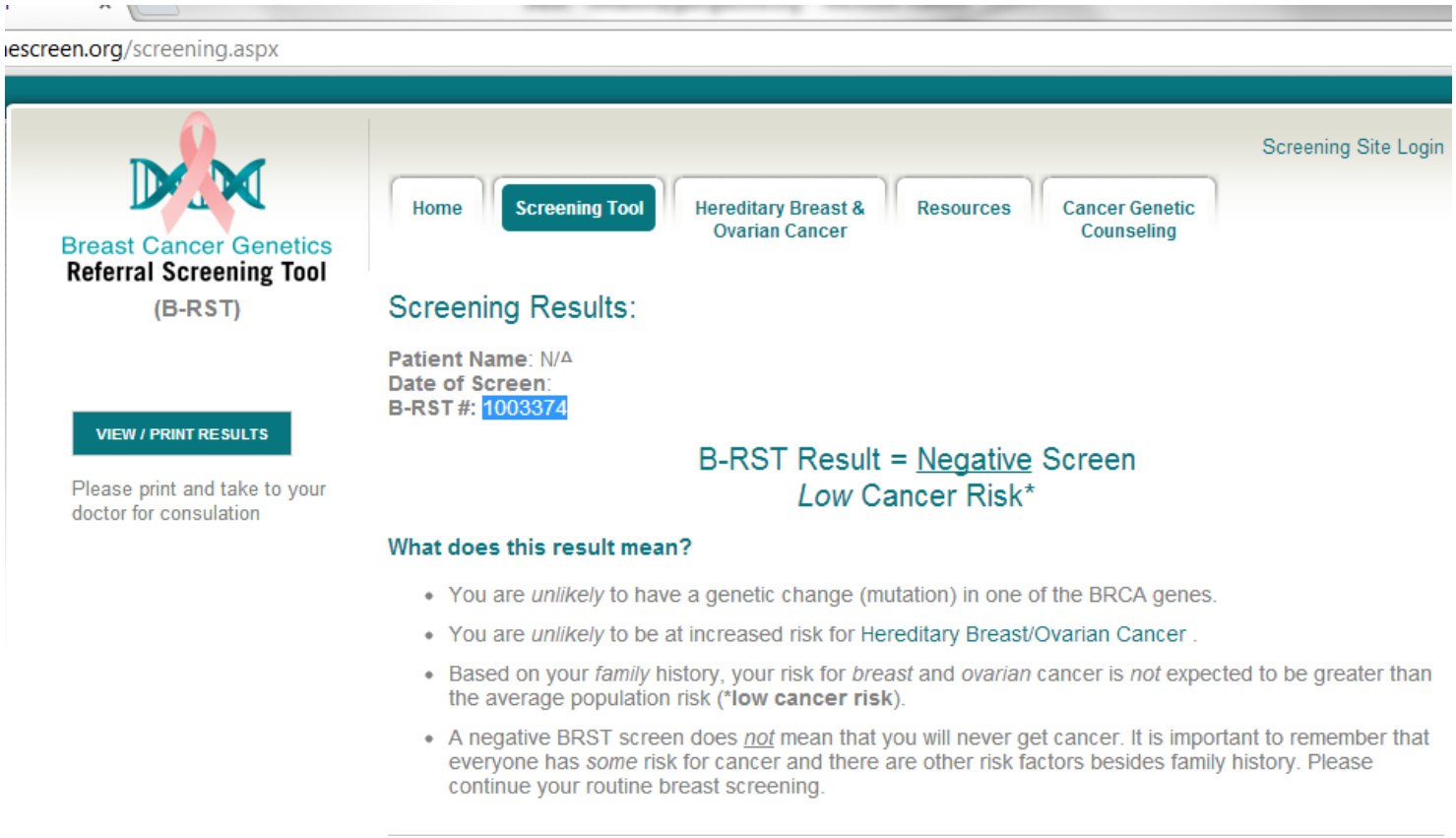
*Developed and validated by Cecelia Bellcross, PhD, MS, CGC, Emory University School of Medicine, Department of Human Genetics, Member of Winship Cancer Institute

SCREENING STEPS

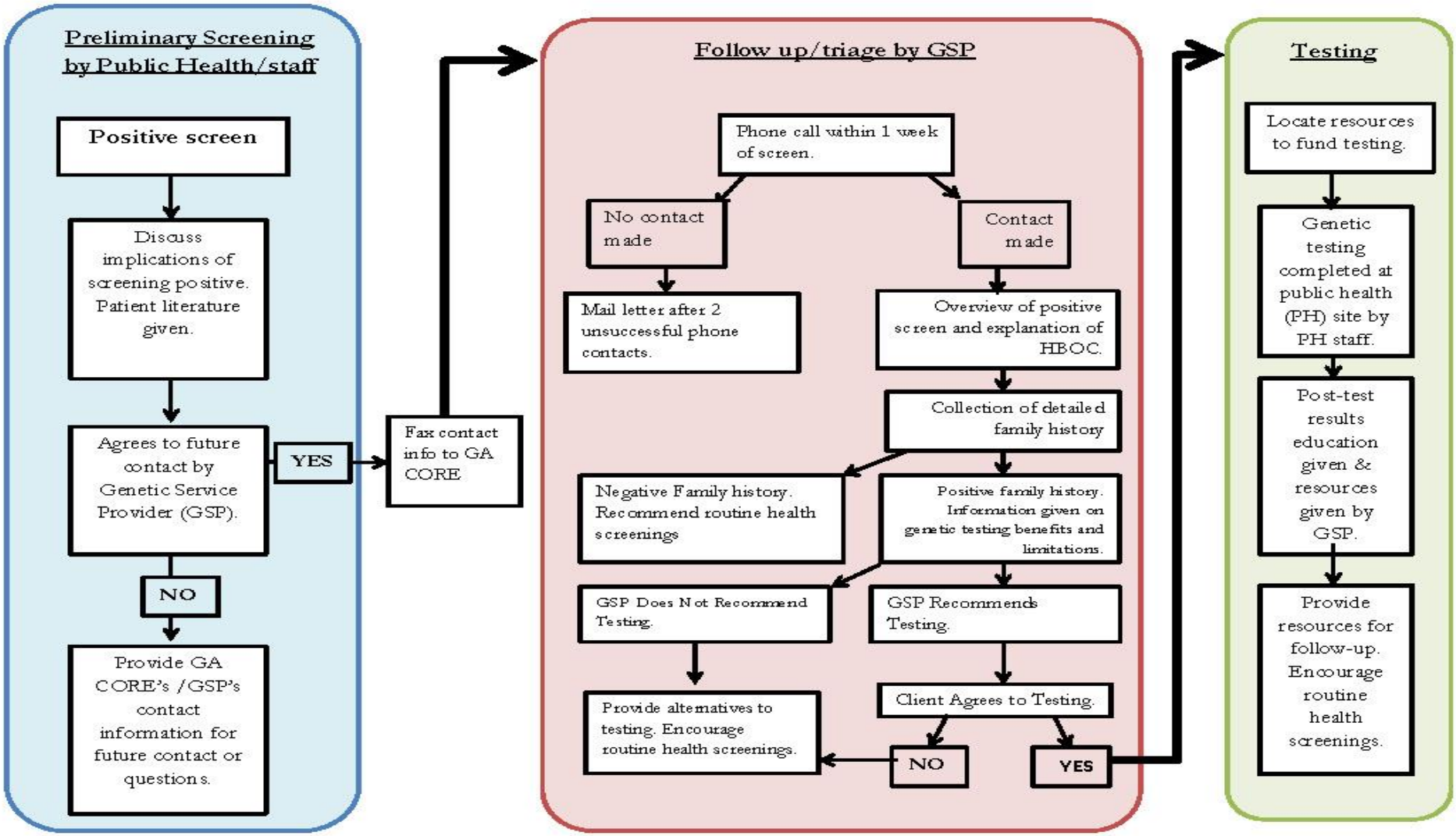
USE of B-RST in GA PUBLIC HEALTH CLINICS



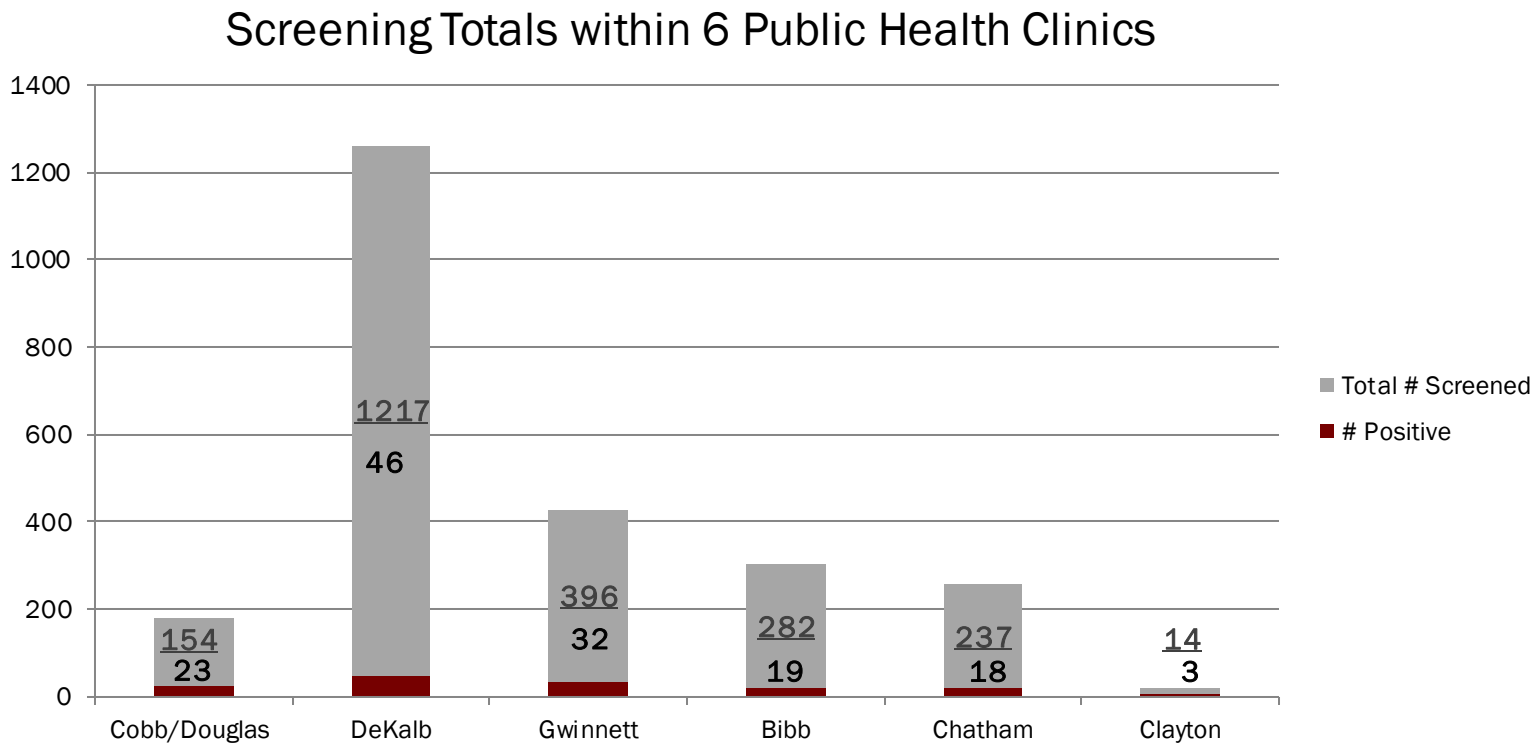
EDUCATION & RESULTS



POSITIVE PATIENT FOLLOW UP



SURVEILLANCE

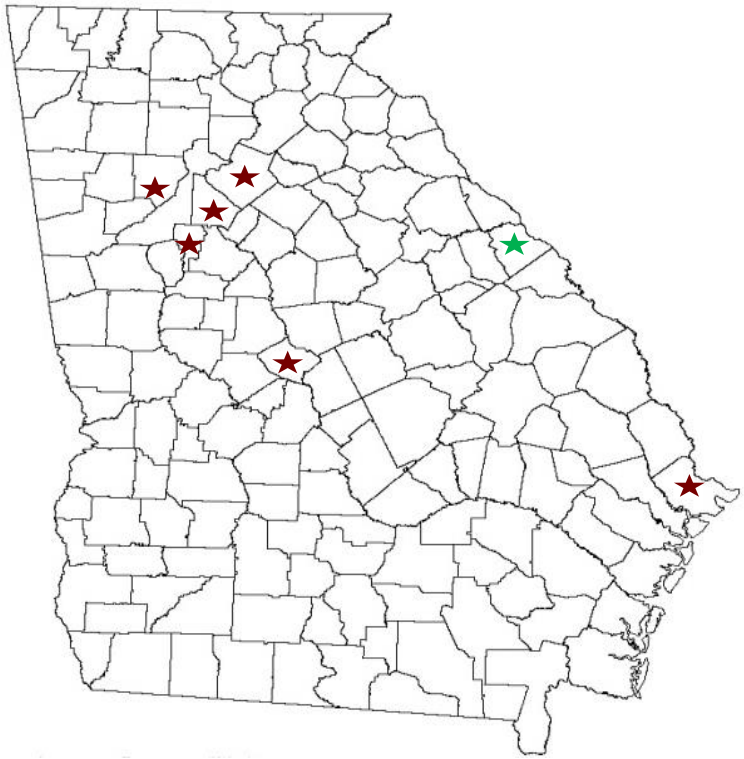


- Data presented collected from 2012 – 2013
- Clinics have varying:
  - Implementation dates
  - Structures
  - Number of providers

GEORGIA BREAST CANCER GENOMIC HEALTH CONSORTIUM
EDUCATION SURVEILLANCE AND POLICY

SURVEILLANCE

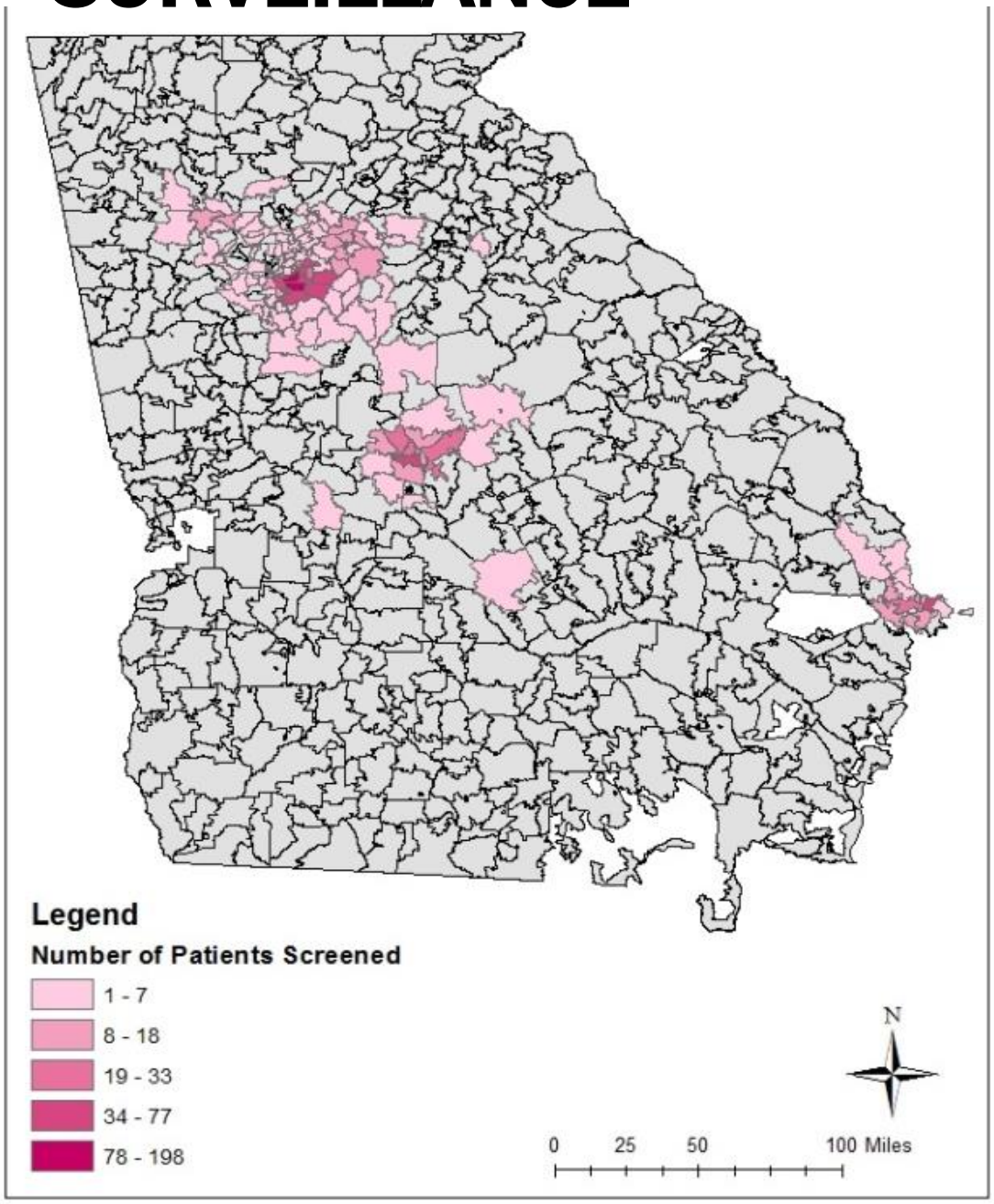
Location of counties with clinics using B-RST screening*



* From Top to Bottom: Gwinnett, Cobb/Douglas, DeKalb, Clayton, Bibb, Columbia, Chatham

GEORGIA BREAST CANCER
GENOMIC HEALTH CONSORTIUM
EDUCATION SURVEILLANCE AND POLICY

SURVEILLANCE



Patients screened using the B-RST in 6 public health clinics by Zip Code

Source: Map by E. Dauria, 2013

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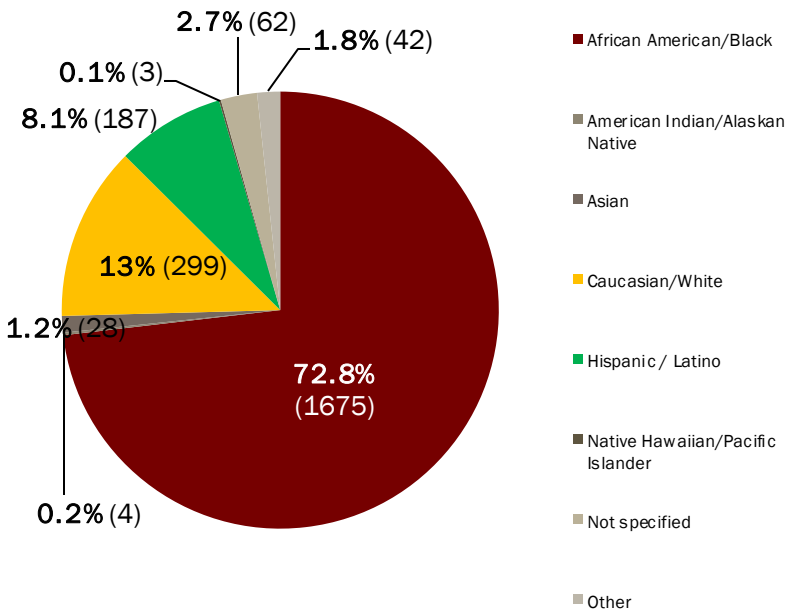
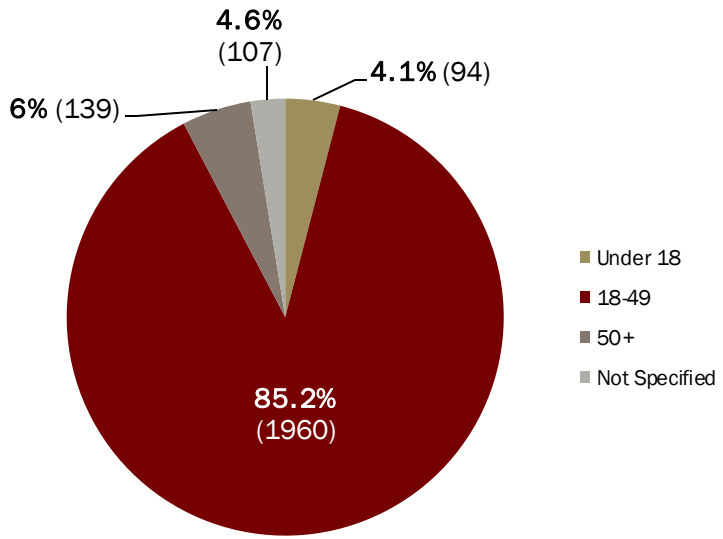
SURVEILLANCE

Race/Ethnicity of Individuals Screened in 6 Public Health Clinics (N=2300)



Age of Individuals Screened in 6 Public Health Clinics (N=2300)



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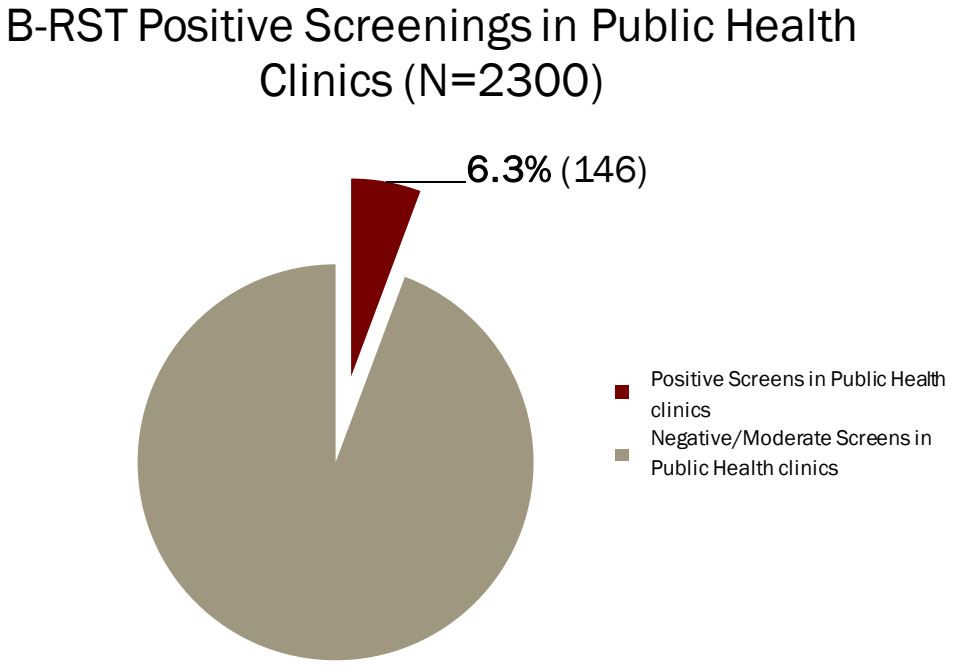

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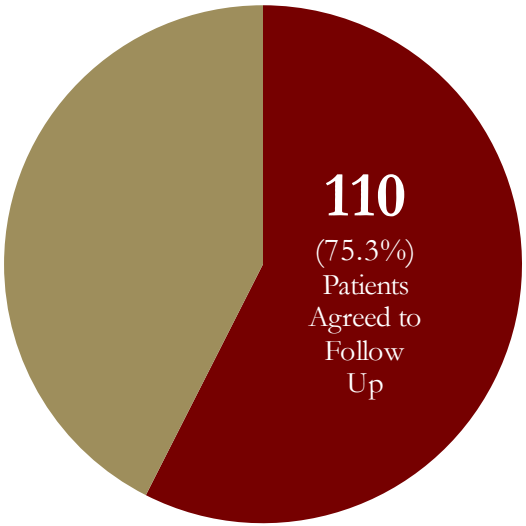
146 Positive Patients Screened



Patient Follow-Up	N (%)[1]
Successfully Contacted[2]	79 (71.8%)
Meet NCCN High Risk guidelines	53 (48.2%)
Tested	14 (12.7%)
Patients that reported findings[3]	20 (18.2%)
	N (%)[4]
Test Results	
BRCA 1/2 Positive	1 (7.1%)
Genetic Variant of Uncertain Significance[5]	1(7.1%)
Negative	12 (85.7%)

1. Total number divided by number of patients that agreed to follow-up
2. Obtained detailed family history and provided resource(s)/counseling/education.
3. Reported breast concern as reason for coming to public health clinic.
4. Patient test results divided by number of patients tested.
5. Not enough information about genetic change to know whether or not it results in increased risk of cancer.

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SUMMARY

- Challenges & Successes

  - Limited access to genetic counselors
    - Phone genetic counseling
    - Contract with genetic health professional
  - Implementation of ACA
    - Coverage for everyone (USPSTF)…eventually
  - Implications of patent challenge
    - Testing cost
  - Collaboration

THANK YOU

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