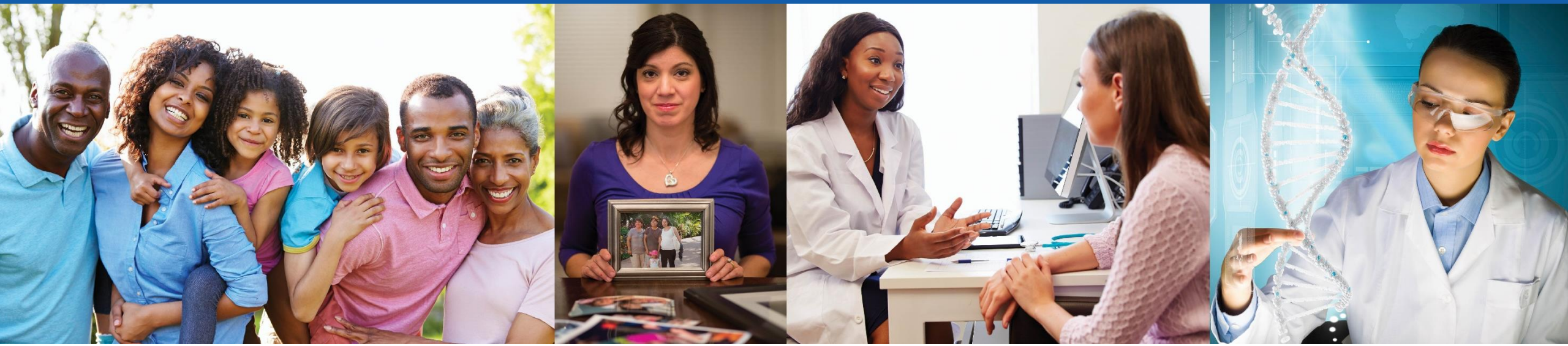
Public Health Genomics and Cancer: Family History and Burden



Lisa Richardson, MD, MPH

Director

Division of Cancer Prevention and Control

National Center for Chronic Disease Prevention and Health Promotion

Accessible Version: https://youtu.be/nw0almfi-ew

U.S. Department of Health and Human Services
Centers for Disease Control and Prevention

Leading Causes of Cancer Incidence in the United States, 2012

Males

Cancer Type	Incidence Rate
Prostate	105.3
Lung & Bronchus	71.6
Colon & Rectum	44.8
Urinary Bladder	35.4
Melanomas of the Skin	25.5
Non-Hodgkin Lymphoma	22.3
Kidney & Renal Pelvis	21.3
Leukemias	16.9
Oral Cavity & Pharynx	16.8
Pancreas	14.1

Females

Cancer Type	Incidence Rate
Breast	122.2
Lung & Bronchus	52.1
Colon & Rectum	34.1
Uterine Corpus	25.7
Thyroid	21.3
Melanomas of the Skin	15.9
Non-Hodgkin Lymphoma	15.5
Ovary	11.3
Kidney & Renal Pelvis	11.2
Pancreas	10.9

Incidence Rate per 100,000 population

United States Cancer Statistics. 1999-2012 Incidence and Mortality Web-based Report. www.cdc.gov/uscs

Leading Causes of Cancer Mortality in the United States, 2012

Males

Cancer Type	Mortality Rate
Lung & Bronchus	56.2
Prostate	19.6
Colon & Rectum	17.6
Pancreas	12.7
Liver & Intrahepatic Bile Duct	9.4
Leukemias	9.2
Urinary Bladder	7.6
Non-Hodgkin Lymphoma	7.6
Esophagus	7.3
Kidney & Renal Pelvis	5.6

Females

Cancer Type	Mortality Rate
Lung & Bronchus	36.4
Breast	21.3
Colon & Rectum	12.4
Pancreas	9.6
Ovary	7.4
Leukemias	5.1
Non-Hodgkin Lymphoma	4.6
Uterine Corpus	4.5
Liver & Intrahepatic Bile Duct	3.8
Brain & other Nervous System	3.5

Mortality Rate per 100,000 population

United States Cancer Statistics. 1999-2012 Incidence and Mortality Web-based Report. www.cdc.gov/uscs

Risk Factors for Different Types of Cancers

- **Common risk factors for cancer**
  - Age
  - Alcohol
  - Cancer-causing substances
  - Diet
  - Family history
  - Genetic mutations
  - Hormones
  - Infectious agents
  - Obesity
  - Radiation
  - Tobacco use

Genomics

Genomics refers to the functions and interactions of all the genes in the genome. It is a more comprehensive field than genetics, which refers to the study of heredity as well as the function and composition of individual genes.

- **Inherited genetic mutations play a major role in about 5%–10% of all cancers**

Doll R, Peto R. *J Natl Cancer Inst.* 1981;66(6):1191–308.
Blot WJ, Tarone RE. *J Natl Cancer Inst.* 2015 Mar 3;107(4).

Hereditary Breast and Ovarian Cancer (HBOC) Syndrome

- **Involves mainly mutations to tumor suppressor genes**
  - *BRCA1* or *BRCA2*
- **Associated with increased risks for breast, ovarian, prostate, and pancreatic cancers**
- **About 1 in every 500 women in the U.S. has a mutation in either the *BRCA1* or *BRCA2* genes**

Tumor suppressor genes

The *BRCA1* and *BRCA2* genes produce a protein that repairs damaged DNA. Mutations in these genes lead to the accumulation of genetic defects that can allow cells to grow and divide too fast or in an uncontrollable way.

Petrucelli N, Daly MB, Feldman GL. In: Pagon RA, Adam MP, Ardinger HH, et al., Eds. *GeneReviews*. ncbi.nlm.nih.gov/books/NBK1247/
U.S. National Library of Medicine. *Breast cancer*. ghr.nlm.nih.gov/condition/breast-cancer

Lynch Syndrome

- **Involves mutations in mismatch repair genes, which lead to tumors with microsatellite instability**
- **Associated with increased risks for colorectal, endometrial, ovarian, stomach cancers and other types of cancer**

Mismatch repair genes

When DNA is replicated during cell division, errors or mismatches may occur. Mutations in the *MLH1, MLH2, MSH6,* and *PMS2* genes prevent repairs from being properly made.

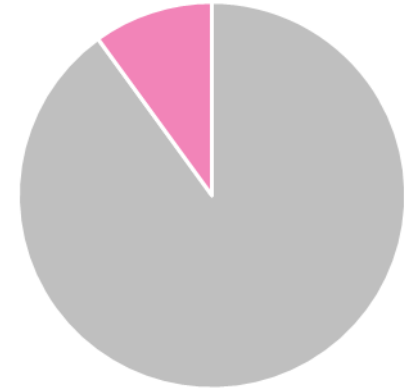
Microsatellite instability

Microsatellites are stretches of DNA with a repetitive sequence, which are susceptible to replication errors. Compared to normal tissue, cells with defective mismatch repair genes show a different number of microsatellite repeats.

Kohlmann W, Gruber SB. In: Pagon RA, Adam MP, Ardinger HH, et al., Eds. *GeneReviews*. ncbi.nlm.nih.gov/books/NBK1211/
U.S. National Library of Medicine. Lynch Syndrome. ghr.nlm.nih.gov/condition/lynch-syndrome

Cancers Associated with Hereditary Cancer Syndromes

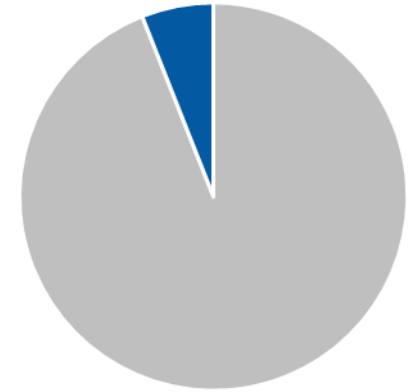
HBOC Syndrome



Up to 10% or approximately 22,000 cases of breast cancer each year

15% or approximately 3,000 cases of ovarian cancer each year

Lynch Syndrome



Up to 3% or approximately 4,000 cases of colorectal cancer each year

Campeau PM, Foulkes WD, Tischkowitz MD. *Human Genetics.* 2008; 124(1):31–42.
Pal T, Permuth-Wey J, Betts JA, et al. *Cancer.* 2005; 104(12):2807–16.
Kaz AM, Brentnall TA. *Nat Clin Pract Gastr* (2006); 3:670-679.

Women with HBOC Syndrome Have Increased Risk for Breast Cancer

Out of 100 women in the U.S. general population:

About 12 will get breast cancer by age 70

Out of 100 women with HBOC Syndrome:

About 65 will get breast cancer by age 70

Antoniou A , Pharoah PDP, Narod S, et al. *Am J Human Genetics.* 2003 May; 72(5): 1117–1130.
Chen S, Parmigiani G. *J Clin Oncol.* 2007; 25(11):1329–1333

Women with HBOC Syndrome Have Increased Risk for Ovarian Cancer

Out of 100 women in the U.S. general population:

About 1 will get ovarian cancer by age 70

Out of 100 women with HBOC Syndrome:

About 39 will get ovarian cancer by age 70

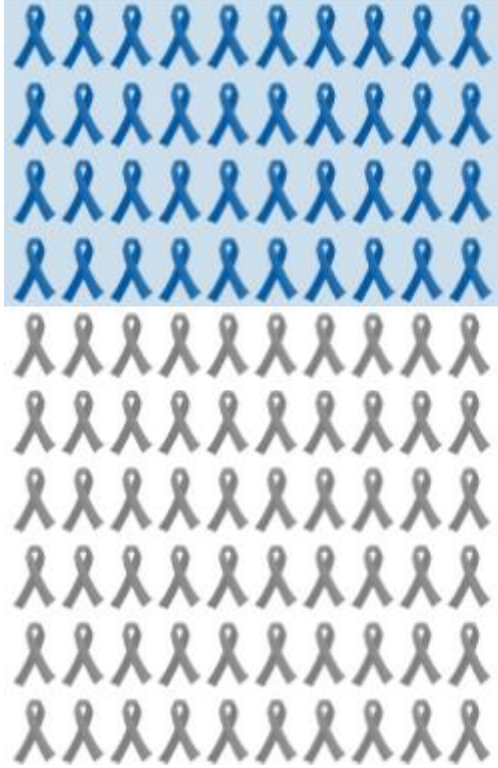
Chen S, Parmigiani G. *J Clin Oncol.* 2007; 25(11):1329–133.

Individuals with Lynch Syndrome Have Increased Risk for Colorectal Cancer

Out of 100 individuals in the U.S. general population:

About 4 will get colorectal cancer by age 70

Out of 100 individuals with Lynch Syndrome:



About 40 will get colorectal cancer by age 70

Evaluation of Genomic Applications in Practice and Prevention Working Group. *Genet Med* 2009;11(1):35-41.

Family History Associated with HBOC Syndrome

- Younger age at diagnosis
- Cancer in both breasts or in conjunction with ovarian cancer
- Breast cancer at any age in men
- Known BRCA mutations in relatives
- Ashkenazi Jewish ancestry

Petrucelli N, Daly MB, Feldman GL. In: Pagon RA, Adam MP, Ardinger HH, et al., Eds. *GeneReviews*. ncbi.nlm.nih.gov/books/NBK1247/

Family History Associated with Lynch Syndrome

- Younger age at diagnosis
- Colorectal cancer in conjunction with, or separately from, another Lynch syndrome-related cancer
- Relatives with Lynch syndrome-related cancers
- Known Lynch syndrome-related mutations in relatives

Kohlmann W, Gruber SB. In: Pagon RA, Adam MP, Ardinger HH, et al., Eds. *GeneReviews*. ncbi.nlm.nih.gov/books/NBK1211/

Genomic Recommendations for Cancer Screening

- **U.S. Preventive Services Task Force recommendation on *BRCA*-related cancer: Risk assessment, genetic counseling and genetic testing**
  - Screening to identify family history associated with *BRCA1* or *BRCA2*
  - If positive, receive genetic counseling and if indicated after counseling, then *BRCA* testing
- **Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group recommendation for people with newly diagnosed colorectal cancer**
  - Access to genetic testing to identify Lynch syndrome to prevent cancer in their close relatives

Moyer VA. *Ann Intern Med.* 2014; 160(4):271–281.
Evaluation of Genomic Applications in Practice and Prevention Working Group. *Genet Med*. 2009 Jan;11(1):35–41.

Mitigating Risk among Individuals with Hereditary Cancer Syndromes

- **Provider and patient determine best course of action to reduce risk**
- **Possible interventions for HBOC Syndrome**
  - Risk-reducing medications for breast cancer (e.g., tamoxifen or raloxifene)
  - Mammography (when to start, frequency, and when to combine with breast MRI)
  - Bilateral mastectomy and bilateral salpingo-oophorectomy
- **Possible interventions for Lynch Syndrome**
  - Colonoscopy starting at age 25 or earlier
  - Screen based only on colonoscopy and not other colorectal cancer screening tests (e.g., sigmoidoscopy, fecal occult blood test)

HBOC: Hereditary Breast and Ovarian Cancer

American Society of Clinical Oncology. Breast Cancer: Risk Factors (http://www.cancer.net/cancer-types/breast-cancer/risk-factors)
American Society of Clinical Oncology. Ovarian Cancer: Risk Factors and Prevention (http://www.cancer.net/cancer-types/ovarian-cancer/risk-factors-and-prevention)

Genomic Activities in Public Health

Know:BRCA

An interactive web resource with unique areas for consumers and healthcare providers

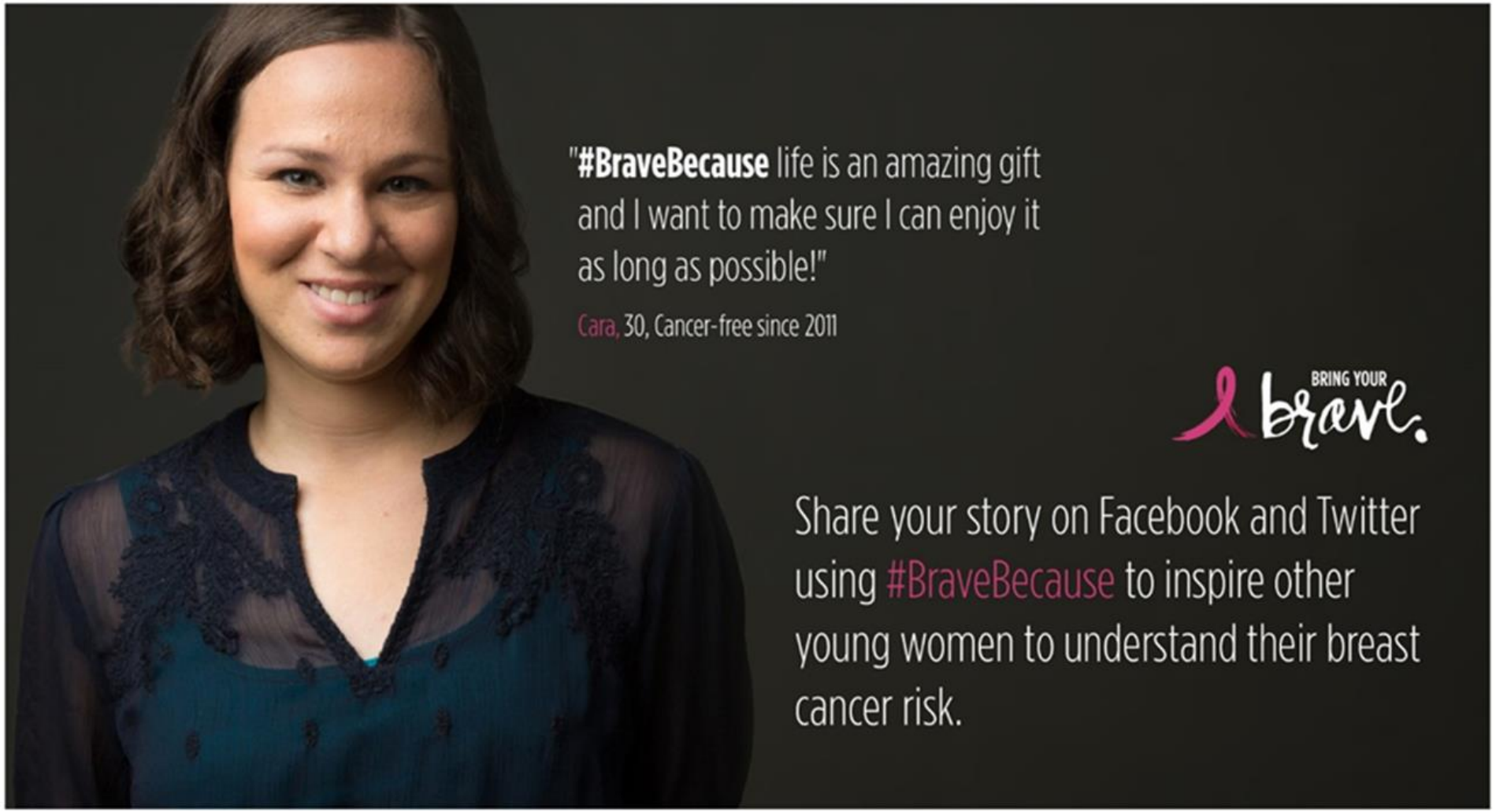
Launched on May 8, 2014

Know:BRCA

KNOWING YOUR BRCA GENE MUTATION RISK CAN SAVE YOUR LIFE

www.knowbrca.org

Bring Your Brave



CDC Public Health Cancer Genomics Program

- **Provides leadership and builds capacity for cancer genomics activities in state public health departments**
  - Implement education, surveillance, and policy or systems change activities that will translate and implement national recommendations for cancer genomics
- **Funded programs currently in five U.S. states**
  - Colorado
  - Connecticut
  - Michigan
  - Oregon
  - Utah

Trivers KF, Rodriguez JL, Cox SL, et al. *Healthcare*. 2015, 3(4), 948–963.
CDC Awards Funding to Support Cancer Genomics. cdc.gov/cancer/breast/what_cdc_is_doing/genomics_foa.htm

CDC Public Health Cancer Genomics Program

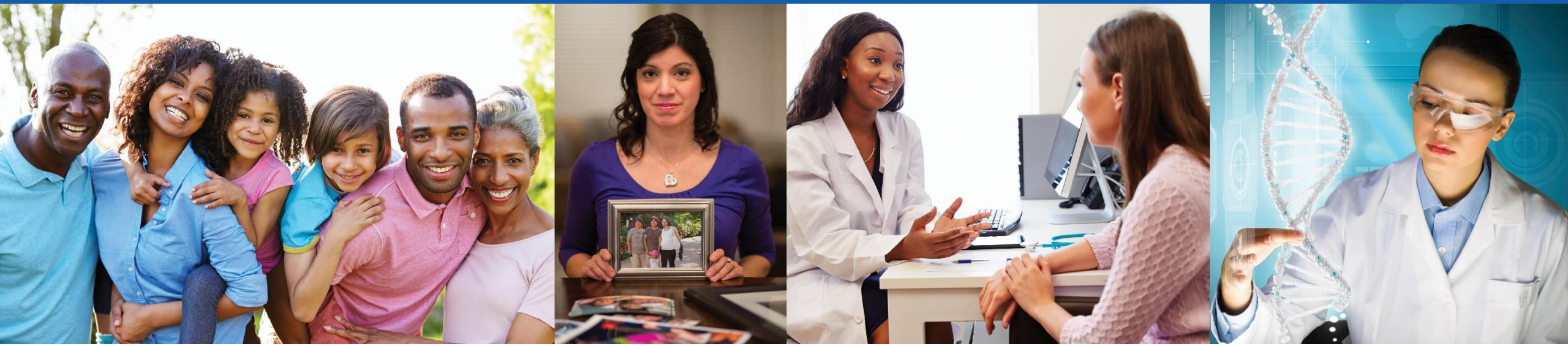
- **Due to current state of the science, current efforts focused on the hereditary cancer syndromes for which screening and intervention are recommended**

<u>What CDC is doing:</u>

- **Educating public and providers**
- **Developing or expanding surveillance systems**
- **Assessing system- or policy-level barriers**

CDC Awards Funding to Support Cancer Genomics. cdc.gov/cancer/breast/what_cdc_is_doing/genomics_foa.htm

A State Health Department Approach to Cancer Genomics Surveillance, Education and Policy

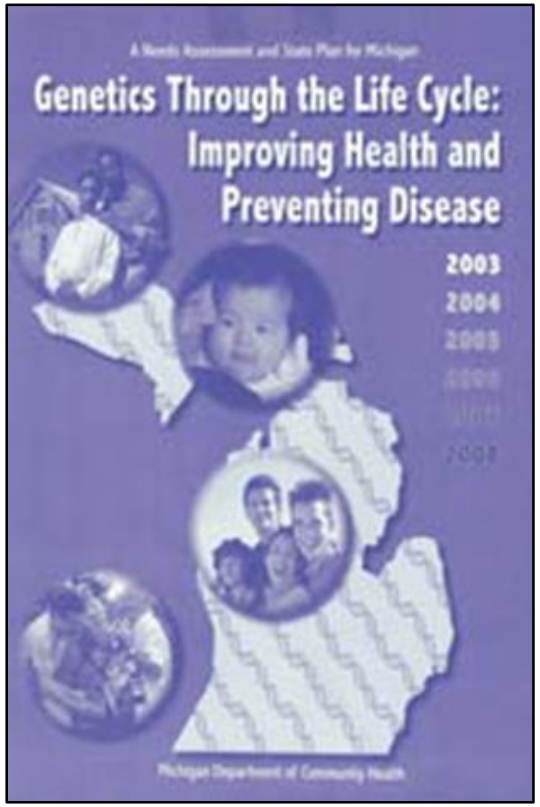


Debra Duquette, MS, CGC
Genomics Coordinator
Michigan Department of Health and Human Services (MDHHS)

MDHHS Michigan Department of Health & Human Services

U.S. Department of Health and Human Services
Centers for Disease Control and Prevention

Michigan Genetics: A Vision for the Role of Cancer Genetics in Public Health



michigan.gov/genomics

- **Improved health outcomes and an enhanced quality of life for the people of Michigan through appropriate use of genetic information, technology, and services**

Funding for the Michigan genetics needs assessment and state plan provided by the Maternal and Child Health Bureau, Health Resources and Services Administration (HRSA), Department of Health and Human Services, 2000–2006.

Michigan Cancer Genomics Goals and Objectives

- Increasing genetic literacy related to cancer
- Assessing the public health impact of hereditary cancer and the utilization of clinical genetics services
- Enhancing communications with cancer genetic service providers and promoting partnerships
- Reducing morbidity and mortality related to hereditary cancer

Comprehensive Cancer Control Plan for Michigan, 2009–2015

michigancancer.org/

Genomics Goal

- Increase availability of cancer-related genetic information to the Michigan public and decrease barriers to risk-appropriate services

Ovarian Cancer Goal

- Improve understanding of, and access to, genetic counseling services for women who may be at high risk for developing ovarian cancer

Promoting Cancer Genomics Best Practices through Surveillance, Education, and Policy Change, 2008–2011

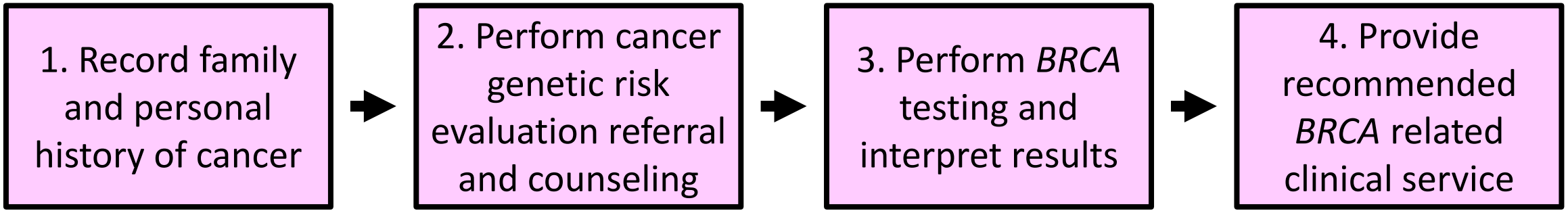
- **Purpose**
  - Move human genome applications into health practice to maximize health benefits and minimize harm through non-research activities
- **Awarded from CDC to four grantees**
- **Goals**
  - Develop and implement surveillance model of inherited cancer and use of relevant tests
  - Identify model provider education programs to increase use of appropriate screening, counseling and evidence-based genetic tests
  - Identify model health insurance policy for *BRCA1/2* genetic counseling and testing

Enhancing Breast Cancer Genomics Best Practices and Policies, 2011–2014

Goals

- Promote adoption of health plan policies to increase coverage of *BRCA* clinical services
- Increase healthcare provider knowledge and use of *BRCA* clinical practices
- Expand surveillance of *BRCA* clinical practices

BRCA counseling, testing, and clinical services



Utilizing State Cancer Registry Data for Cancer Genomics Surveillance

MDHHS Cancer Genomics and Michigan Cancer Surveillance Program (MCSP)

- **Examined potential cases at risk for HBOC or Lynch syndrome**
  - Multiple primary sites of cancer
  - Early onset breast cancer (age 50 or younger)
  - Triple negative breast cancer
    - Does not express the genes for estrogen receptor, progesterone receptor, or HER2/neu
  - Male breast cancer
  - Ovarian cancer
  - Early onset endometrial cancer (before age 50)
  - Colorectal cancer
- **Disseminated data to healthcare systems and providers to reinforce educational messages**

MDHHS: Michigan Department of Health and Human Services
HBOC: Hereditary Breast and Ovarian Cancer
HER2/neu: Human epidermal growth factor receptor 2

Use of Cancer Genetic Services among Young Breast Cancer Survivors

- **Black women were less likely than white/other women to use cancer genetic services**

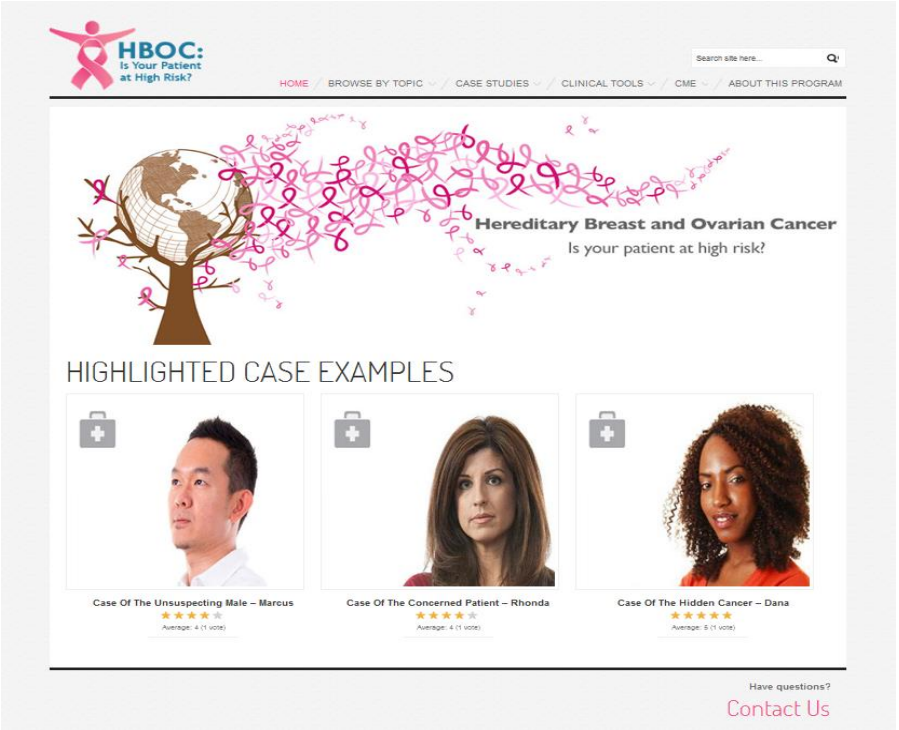
Use of cancer genetic services	Total (n=828)	Black (n=317)	Other (n=511)
Had genetic counseling*	32.9%	26.6%	37.1%
Had genetic testing*	28.5%	19.9%	33.7%
Had genetic counseling and testing*	27.5%	18.3%	32.9%

* Significant at the 0.001 level for black vs. other

Katapodi MC, Northouse LL, Schafenacker AM, et al. *BMC Cancer*. 2013 Mar 1;13:97.

Online CME Course
HBOC: Is Your Patient at High Risk?

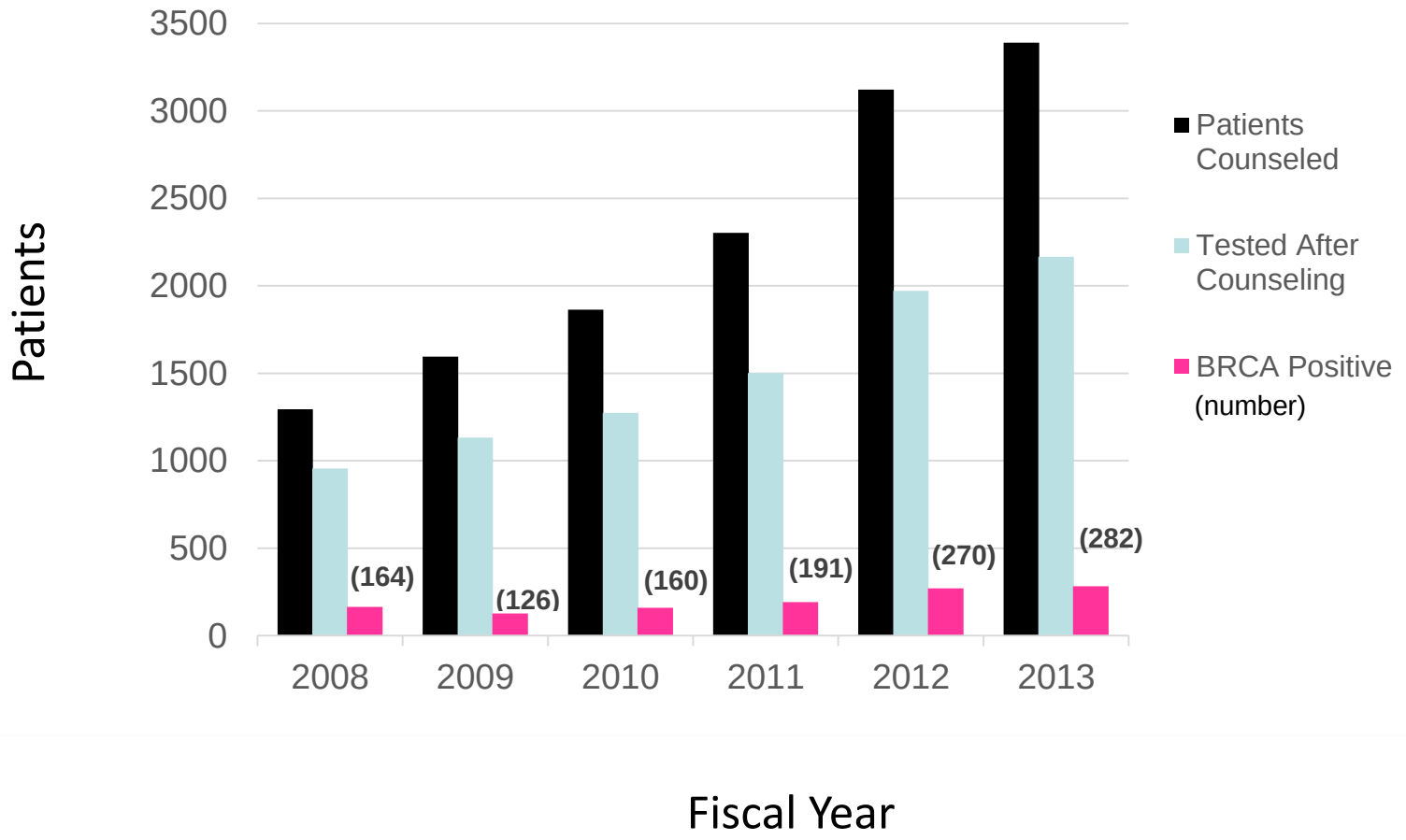
- **MDHHS provided in-person interactive case-based cancer genetics presentations since 2009**
- **Based on the success of case-based approach, in 2012–2013 MDHHS collaborated with federal, state, and local partners to develop online hereditary breast and ovarian cancer genomics module**
- **Michigan State University approved 2.0 CMEs until October 2016**
  - Available at no cost to participants
  - Over 4,400 sessions since launch in February 2014



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

HBOC: Hereditary Breast and Ovarian Cancer
MDHHS: Michigan Department of Health and Human Services
CME: Continuing Medical Education

Michigan Successes Related to *BRCA* Counseling Access and Referrals



Michigan Department of Health and Human Services, unpublished data.

Promoting Evidence-based Cancer Screening Polices Among Insurers

BREAST CANCER GENOMICS BEST PRACTICES

for Michigan Health Plan Partners

BRCA Policy Dashboard

*This dashboard was created for as an update on progress toward developing written policies related to all four areas of cancer genetic services **(Figure 1)**. For more information on policy development or for technical assistance from MDCH Cancer Genomics Program staff call 1-866-852-1247 or email genetics@michigan.gov. If this scorecard is not accurate, please contact us immediately. We would greatly appreciate up-to-date information from all health plans in Michigan.*

Figure 1. Spectrum of Cancer Genetic Services

= policy is consistent with project standards

= policy is not consistent with project standards

U = policy is unavailable/unknown if consistent with project standards

Your health plan has written policies related to *BRCA that…*	
1. include coverage for the following individuals:	
• Adults with a personal history of breast and/or ovarian cancer.[1]	👍
• Adults with a family history of breast and/or ovarian cancer.[1,2]	👍
2. require or strongly recommend genetic counseling ***prior*** to *BRCA* genetic testing.	👍
3. encourage providers to obtain written informed consent (as is required by Michigan law) ***prior*** to ordering *BRCA* genetic testing.	👍
4. cover *BRCA*-related clinical services for positive patients (policies would contain coverage information for the following services)[1]	
• **Mammography**	👍
• **MRI of the Breast**	👍
• **Prophylactic Mastectomy**	👍
• **Prophylactic Oophorectomy**	👍
• **Breast Reconstruction / Prostheses**	👍

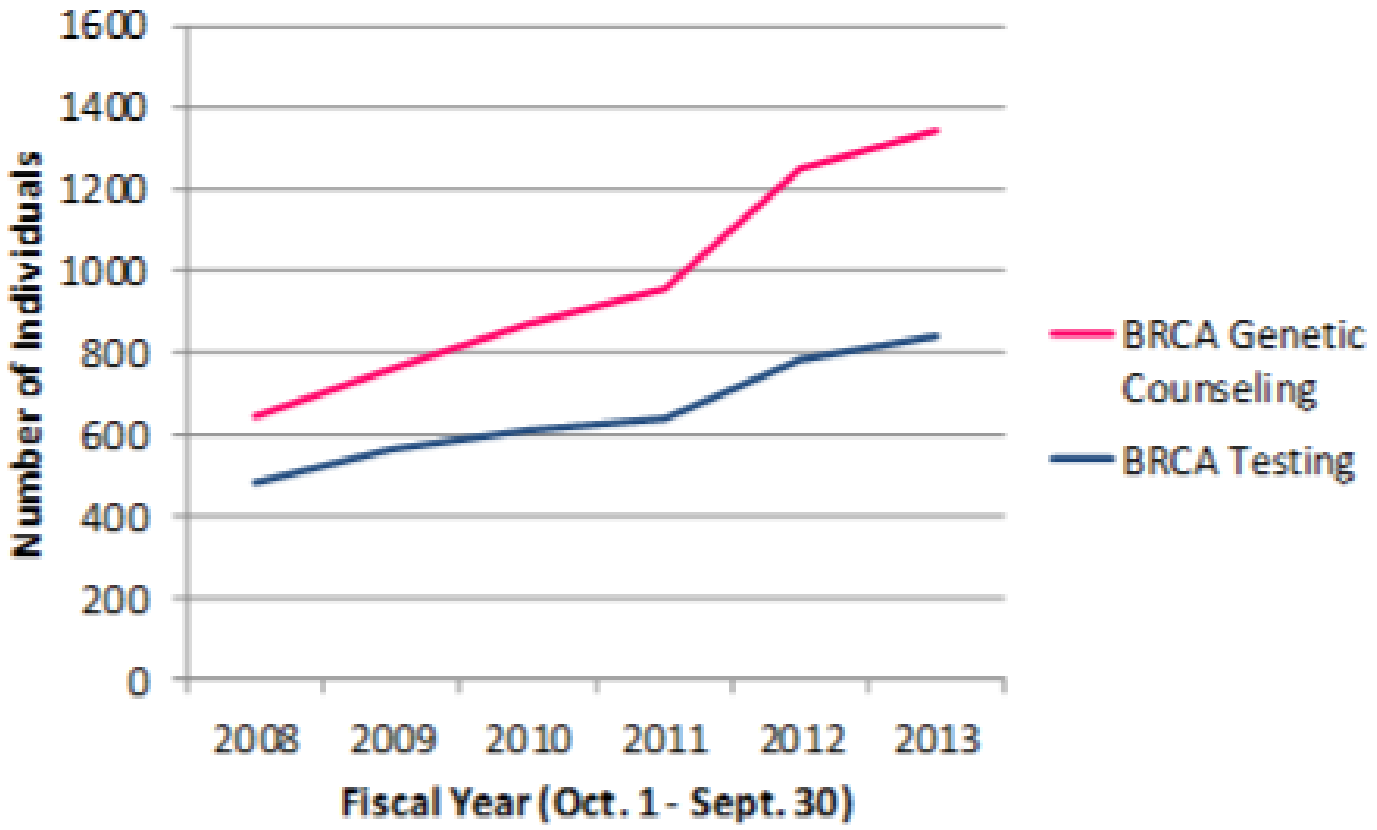
1. NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Genetic/Familial Risk Assessment: Breast and Ovarian V.3.2013.© National Comprehensive Cancer Network, Inc. 2013. All rights reserved. Accessed July 1, 2013. To view the most recent and complete version of the guideline, go online to www.nccn.org. NATIONAL COMPREHENSIVE CANCER NETWORK®, NCCN®, NCCN GUIDELINES®, and all other NCCN Content are trademarks owned by the National Comprehensive Cancer Network, Inc. 2. U.S. Preventive Services Task Force: Genetic risk assessment and *BRCA* mutation testing for breast and ovarian cancer susceptibility: recommendation statement. Ann Intern Med 2005; 143: 355–361.

Michigan Department of Community Health

MDCH

Promoting Evidence-based Cancer Screening Policies Among Insurers

Members from specific health plan receiving *BRCA* counseling and testing, October 1, 2007–September 30, 2013



Michigan Department of Health and Human Services, unpublished data

Lynch Syndrome Screening Network (LSSN)

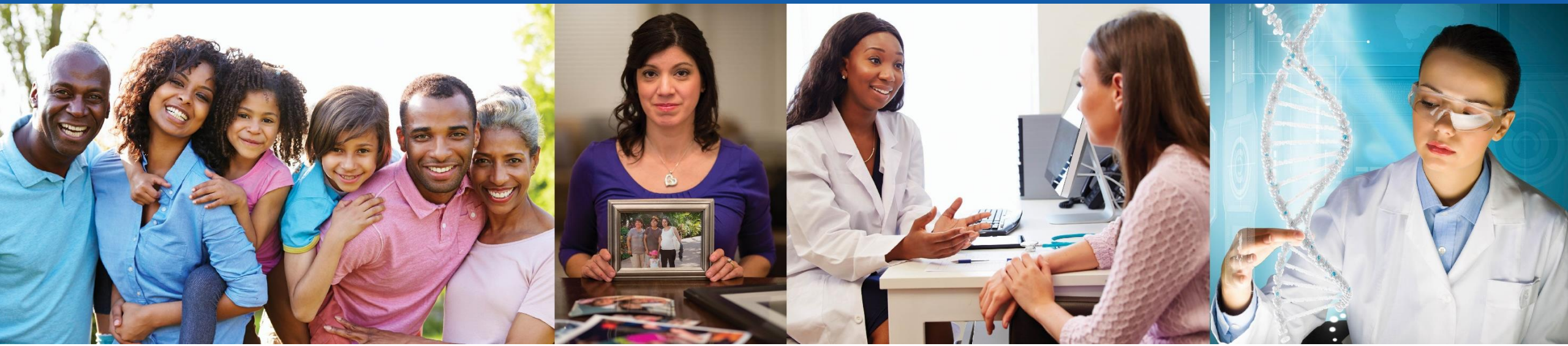
- **LSSN Mission**
  - Promote Lynch syndrome screening on all newly diagnosed colorectal and endometrial cancers
  - Facilitate the ability of institutions to implement appropriate screening through network collaboration
  - Investigate universal screening for other Lynch syndrome-related malignancies
- **Institutional membership**
  - Over 120 leading cancer institutions are members
  - Over 20,000 newly diagnosed cancers screened
  - Membership data assisting to measure Healthy People 2020 Lynch syndrome objective

http://www.lynchscreening.net/

Summary

- **State health department infrastructure can be used to integrate cancer genomics into public health programs**
  - Comprehensive Cancer Control Program State Plans
  - State Cancer Registry Data
- **In Michigan, cancer genetic counseling and testing is increasing, but appropriate use of these services remains low overall, and racial disparities are present**
- **Partnerships, dedicated staff, and use of core public health functions, e.g., assessment, policy, and assurance, are needed to advance cancer genomics activities**

Raising Awareness About Understanding Family Risk for Cancer



Lindsay Avner
Founder and CEO
Bright Pink

bright pink

U.S. Department of Health and Human Services
Centers for Disease Control and Prevention

Inspired By My Past To Take Action For My Future


bright pink®

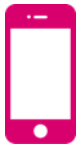
We Are Bright Pink

- **We save women's lives from being cut short by breast and ovarian cancer**
- **Empowering them to live proactively at a young age**
  - Our focus is health, not just cancer
- **Actions to prevent and detect breast and ovarian cancer begin with knowledge**
- **Conversations can save lives**
  - Within families, friends, healthcare providers

Awareness Doesn’t Save Lives, Action Does

We reach 18–45 Year Old Women  and Health Providers Nationwide with

 Evidence-Based Breast and Ovarian Health Content 

Where they Work, Connect  and Engage

Inspiring Them to Put Awareness in Action 

 With Education Programs focused on Scale and Behavior Change

A New Approach to Breast and Ovarian Health

Empowered Women

+

Proactive Providers

OUR COMMUNITIES

150

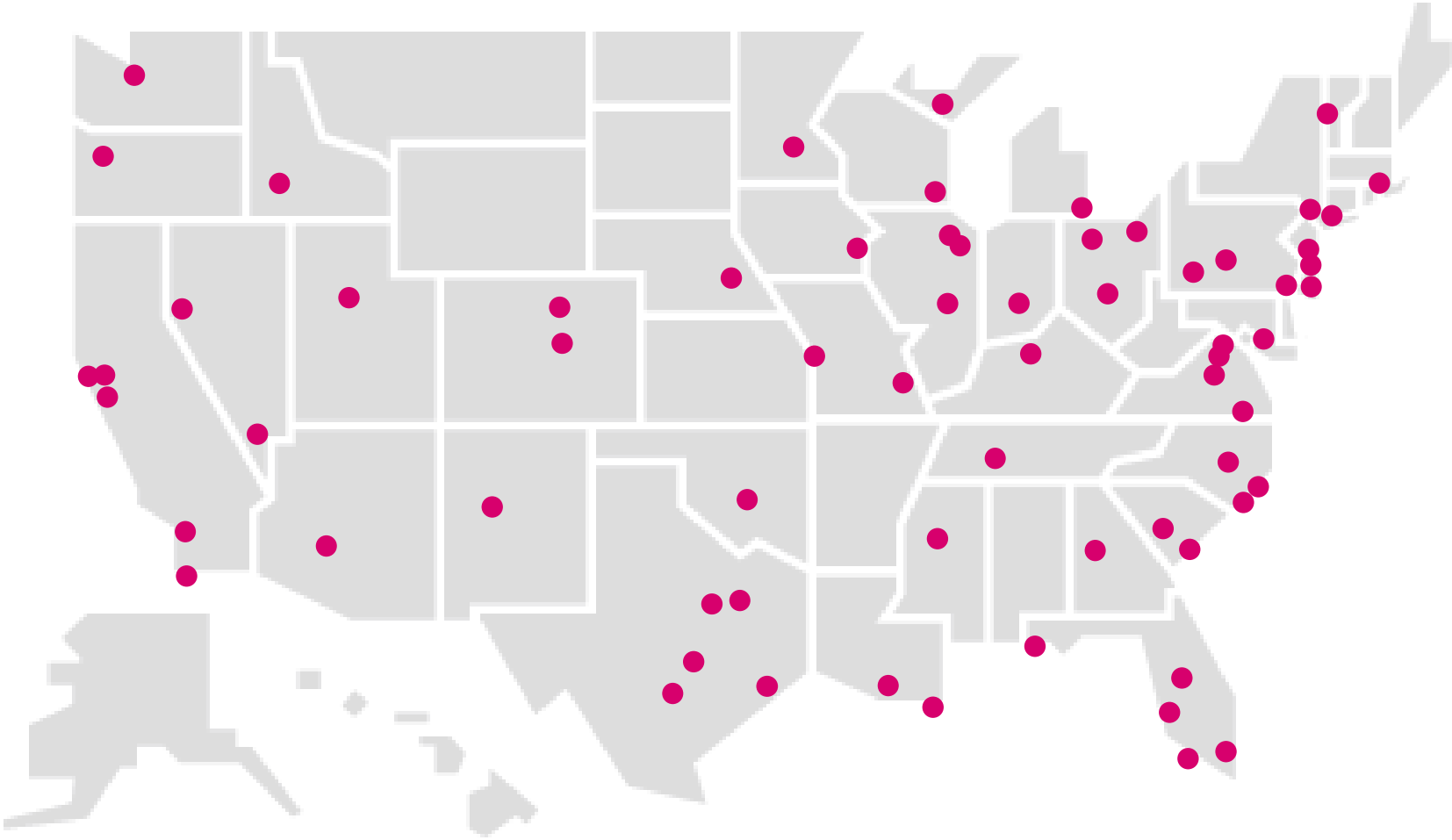
Trained ambassadors in key communities nationwide

39

States with active Bright Pink programs

1000+

Volunteers nationwide



Education and Tools	2015	Growth in 2016
Brighten Up® Educational Workshops	35,000 educated	70,000 educated
AssessYourRisk.org	120K completions	335K completions

High Risk Support	2015	Growth in 2016
PinkPal® One-on-One	1,000 matches*	1,300 matches*
Outreach Groups	16 communities	22 communities
Digital Connections	275 members	700 members

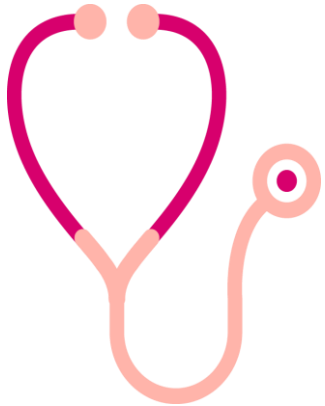
*Coming later this year: Genetic Testing Info One-Stop Shop website

“After this talk I was approached by a sweet young woman, who shared with me her striking family history of breast and ovarian cancer. She cried as she told me she was so scared and didn't know where to start. She was terrified for her mother. The breast cancer in her family started at age 31. I encouraged her to reach out to Bright Pink … she was relieved that there was genetic testing for her family.”

— Heather, Bright Pink Education Ambassador

“Thank you for providing accurate and evidence-based information on cancer risk. I am blown away by how effective Bright Pink has been in raising the visibility of genetic links to cancer. I already knew about my *BRCA2* mutation … but the information you provided helped me decide to undergo a prophylactic double mastectomy 6 weeks ago. I am now a success story. Bright Pink can provide you the information you need to determine whether you should seek genetic testing and … manage your risk.”

— Katie, Takoma Park, Maryland

Provider Education	2015	Growth in 2016
Women's Health Provider Education Initiative 	**4,000** providers educated at more than 80 institutions **14.4 million** women primed for better care	**6,000** providers educated at more than 100 institutions **21.6 million** women primed for better care

Coming later this year: Digital Complement

“Thanks for an interesting and practical Grand Rounds this morning. Already starting to change the way I think about screening some of my patients.”

— Michael Rabovsky, M.D., Chairman, Family Medicine Vice Chair, Medicine Institute Cleveland Clinic

89%

Providers indicated knowing how to better classify risk for young, female patients as a result of the program

“This lecture gives the right perspective – it breaks it down, it gives us an option to say “I can do something more than say your mammogram is abnormal. I know how to have a conversation with the average person who comes to our office.” It’s more than studying for the boards … It’s the actual heart of practicing.”

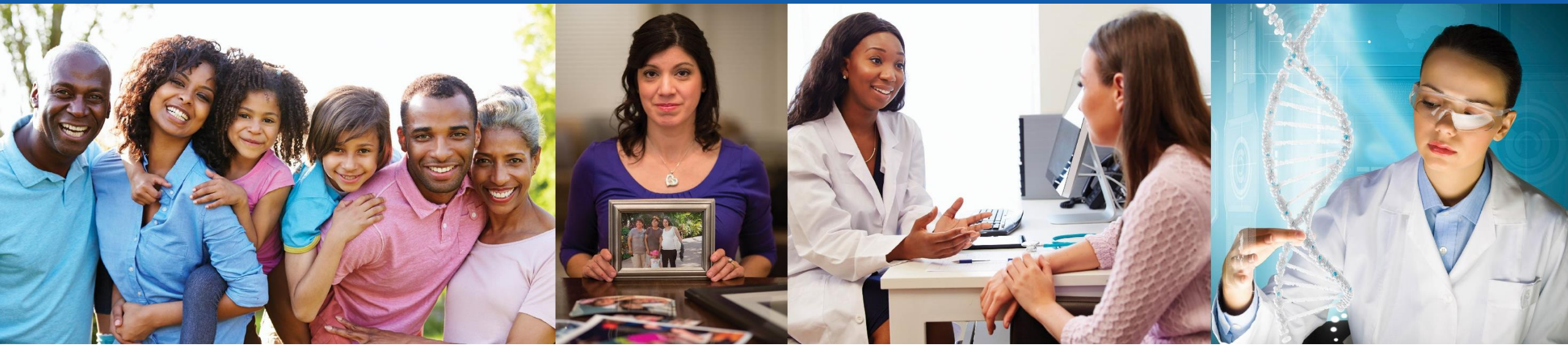
— Dr. Aisha Redmond, Kaiser Permanente Town Park

84%

Providers indicated increased knowledge of breast/ovarian cancer risk reduction and early detection options for young women as a result of the program

Visit BrightPink.org and AssessYourRisk.org for more information

The Role of Genomics in Public Health



Muin J. Khoury, MD, PhD
Director
Office of Public Health Genomics
Division of Public Health Information Dissemination
Center for Surveillance, Epidemiology, and Laboratory Services

U.S. Department of Health and Human Services
Centers for Disease Control and Prevention

The CDC Office of Public Health Genomics

- **Effective and responsible translation of genome-based discoveries into disease prevention and population health**
  - Identify evidence-based genomic applications
  - Inform and communicate
  - Integrate into practice and programs

cdc.gov/genomics/

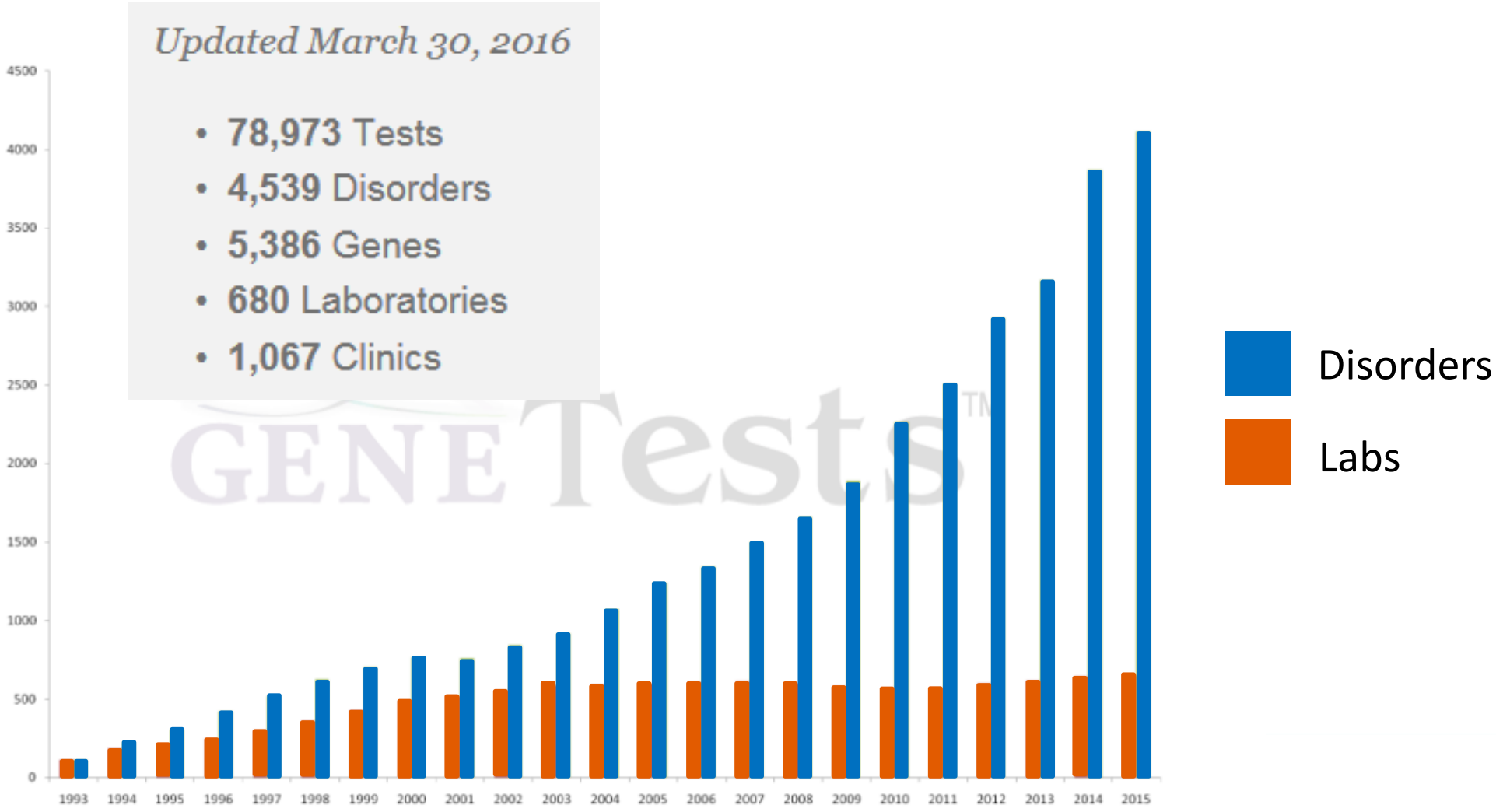
A Crucial Public Health Role is to Identify Population Health Impact of Genomics

- **Identifying genomic tests and family health history applications that are supported by synthesized evidence for their use**

- **Estimating the potential population health impact of these applications**
  - Lives saved
  - Disease prevented or detected earlier
  - Healthcare costs and savings

- **Promoting appropriate and equitable use**

Numerous Genetic Tests are Available



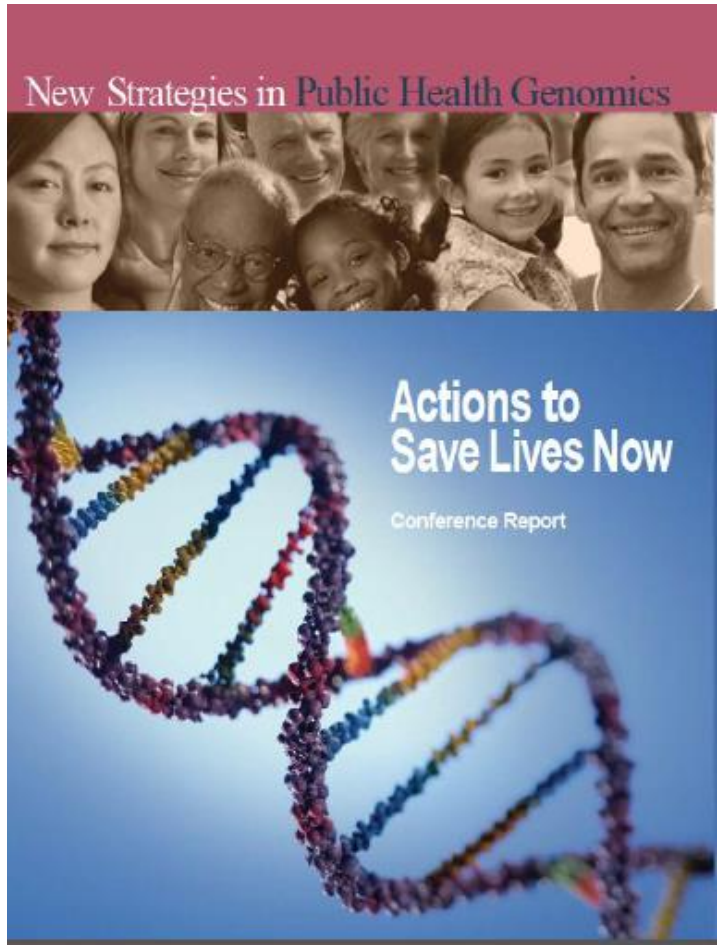
CDC Evidence-based Classification of Genomic Tests

Tier 1	Supported by a base of synthesized evidence for implementation in practice	e.g., HBOC, Lynch syndrome, newborn screening
Tier 2	Synthesized evidence is insufficient to support routine implementation in practice; may provide information for informed decision making	e.g., many pharmacogenomic tests
Tier 3	Evidence-based recommendations against use, or no relevant synthesized evidence identified; not ready for routine implementation in practice	e.g., direct-to-consumer personal genomic tests

Dotson WD, Douglas MP, Kolor K, et al. *Clin Pharmacol Ther*. 2014 Apr; 95(4): 394–402.

Tier 1 Genomic Tests can Save Lives and Many are Underused in Clinical Practice

- **>40 tests supported by evidence for use in practice**
  - >30 cancer-related tests
- **Many intended uses include**
  - Diagnosis
  - Prognosis
  - Risk prediction to inform prevention
  - Treatment, including choice of medication and dosage
  - Screening



Centers for Disease Control and Prevention. genomicsforum.org/editoruploads/ActionstoSaveLivesNowReport.pdf

Family Health History as a Tool for Public Health

- Family health history is a risk factor for many diseases
- Family health history offers opportunities for targeted screening and prevention
- The Surgeon General's Family Health History Initiative
- Thanksgiving is National Family History Day

familyhistory.hhs.gov/

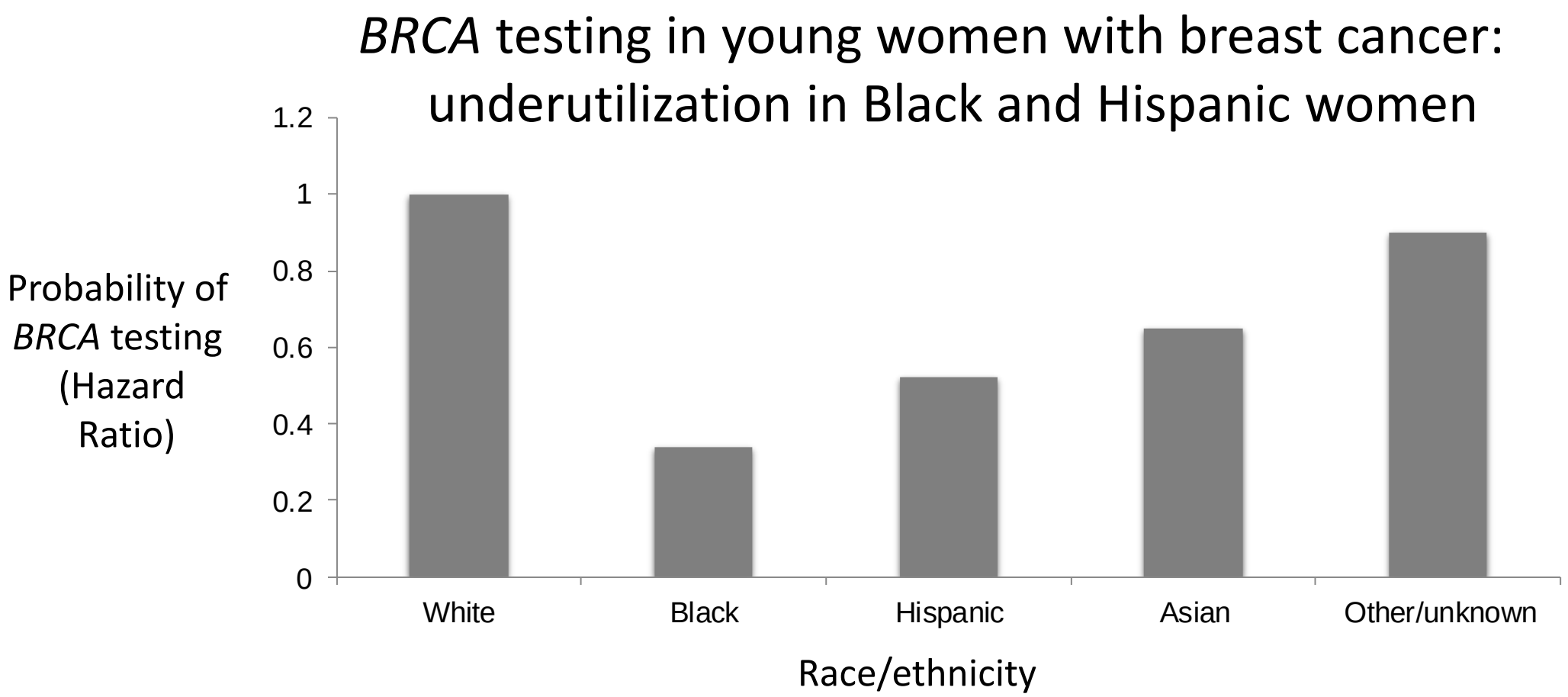
The Possibilities for Genomics to Improve Public Health are Growing Rapidly

- **More than a decade after the Human Genome Project was completed**
- **Whole-genome sequencing as tool in clinical and public health practice**
  - Human DNA sequencing (rare diseases, cancer, chronic diseases)
  - Pathogen DNA sequencing (CDC's Advanced Molecular Detection initiative role in surveillance and outbreak response)
- **Increased public awareness**
  - Especially with celebrity announcements
- **Proliferation of direct-to-consumer genetic tests**

Public Health is Needed to Address Challenges in Genomics Implementation

- Provider and public education
- Healthcare system limitations
- Evidence-based policy
- Population health impact data
- Laboratory quality
- Health disparities

Public Health Genomics and Health Disparities: Ensuring Benefits for All



Levy DE, Byfield SD, Comstock CB, et al. *Genet Med.* 2011 Apr;13(4):349–55.

Genomics-Related Policies and Legislation

- **Affordable Care Act**
  - Covers *BRCA* genetic services in accordance with the USPSTF grade B recommendation
- **Genetic Information Nondiscrimination Act of 2008**
  - Prohibits discrimination in both health insurance and employment based on genetic information
- **Current Procedural Terminology (CPT) code revisions**
  - Implemented specific billing codes for many genetic tests
- **Healthy People 2020**
  - New cancer genomics topic area, including objectives on hereditary breast and ovarian cancer and Lynch syndrome

USPSTF: United States Preventive Services Task Force

Genomic Activities in Public Health

The U.S. Precision Medicine Initiative

- **Launched in 2015, includes two components:**
  - A focus on molecularly targeted treatment for cancer
  - A national cohort of at least 1 million people

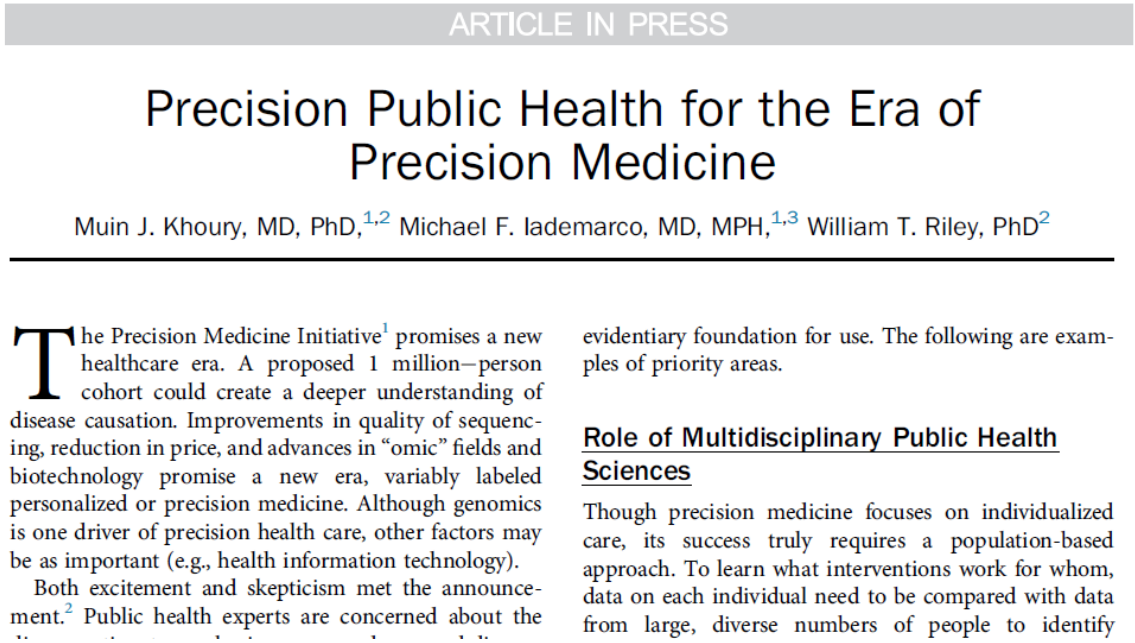
What is precision medicine?

"An emerging approach for disease prevention and treatment that takes into account people's individual variations in genes, environment, and lifestyle."

The White House. *The Precision Medicine Initiative*. whitehouse.gov/precision-medicine
Collins FS, Varmus, H. *N Engl J Med.* 2015; 372:793–795.

Success of Precision Medicine Requires Public Health Partnerships

- **Inclusion and generalizability**
- **Focus on prevention**
- **Implementing what we know**
- **A new era of precision public health**



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Precision Public Health for the Era of Precision Medicine

Muin J. Khoury, MD, PhD,[1,2] Michael F. Iademarco, MD, MPH,[1,3] William T. Riley, PhD[2]

The Precision Medicine Initiative[1] promises a new healthcare era. A proposed 1 million–person cohort could create a deeper understanding of disease causation. Improvements in quality of sequencing, reduction in price, and advances in "omic" fields and biotechnology promise a new era, variably labeled personalized or precision medicine. Although genomics is one driver of precision health care, other factors may be as important (e.g., health information technology).

Both excitement and skepticism met the announcement.[2] Public health experts are concerned about the evidentiary foundation for use. The following are examples of priority areas.

Role of Multidisciplinary Public Health Sciences

Though precision medicine focuses on individualized care, its success truly requires a population-based approach. To learn what interventions work for whom, data on each individual need to be compared with data from large, diverse numbers of people to identify

The White House. *The Precision Medicine Initiative*. whitehouse.gov/precision-medicine
Collins FS, Varmus, H. *N Engl J Med.* 2015; 372:793–795.
Khoury MJ, Iademarco MF, Riley WT. *Am J Prev Med*. 2016 Mar;50(3):398–401.