

# Human Genomics and Public Health: A Lot Has Happened in 20 Years



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Centers for Disease Control and Prevention

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

# Outline

- Emergence and Promise of Genomics and Precision Medicine
- Why We Need Public Health for Genomics and Precision Medicine to Succeed
- A Brief History of Public Health Genomics
- Human Genomics in Public Health Practice: What's Next?

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REVIEW | Genetics  
inMedicine

## From public health genomics to precision public health: a 20-year journey

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In this paper, we review the evolution of the field of public health genomics in the United States in the past two decades. Public health genomics focuses on effective and responsible translation of genomic science into population health benefits. We discuss the relationship of the field to the core public health functions and essential services, review its evidentiary foundation, and provide examples of current US public health priorities and applications. We cite examples of publications to illustrate how *Genetics in Medicine* reflected the evolution of the field. We also reflect on how

public-health genomics is contributing to the emergence of "precision public health" with near-term opportunities offered by the US Precision Medicine (AllotUs) Initiative.

*Genet Med* advance online publication 14 December 2017

**Key Words:** genomics; precision medicine; precision public health; public health

### INTRODUCTION

Over the 20-year history of *Genetics in Medicine* (GIM), public health has made important contributions toward integrating genomics into clinical practice and disease prevention. The Journal has published numerous public health and health services studies, systematic reviews, and guidelines and recommendations that use public health and epidemiological principles as a basis for clinical and population action in genomics. The Journal's editorial board has had a seat for "public health, epidemiology, and personalized medicine" for almost two decades.

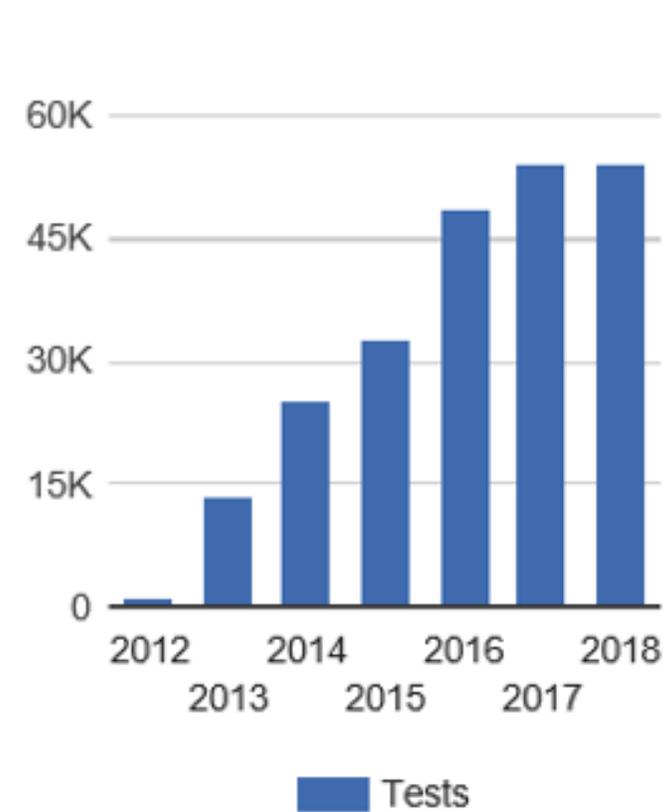
In this paper, we review the evolution of the field of public health genomics and its relation to frameworks of core public health functions and essential services in the United States,

responsible translation of genomic research into population health benefits.<sup>1</sup> Over the past two decades, the Centers for Disease Control and Prevention (CDC) and many other groups have developed and implemented public health genomics in the United States and globally. Examples of CDC initiatives for integrating genomics into public health are shown in **Table 1**.

Until recently, the role of public health in genomics has not been well defined. The mission of public health is to improve health on a population scale, and its unit of intervention is the "population."<sup>2</sup> Nevertheless, genomic medicine is practiced at multiple levels of intervention including patient-provider interactions, health-care organizations, families, communities, and state and federal policies and programs.<sup>3</sup> Also, as the use

# The Genomic Testing Landscape is Growing Rapidly

- **Increasing Number of genetic tests**
- **Whole-genome sequencing as tool in clinical and public health practice**
- **Increasing public awareness and interest**
- **Proliferation of direct-to-consumer genetic tests**



# The U.S. Precision Medicine Initiative Promises a New Era of Medicine and Public Health

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

## 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 future of health begins with **All of Us**

The *All of Us* Research Program is a historic effort to gather data from one million or more people living in the United States to accelerate research and improve health. By taking into account individual differences in lifestyle, environment, and biology, researchers will uncover paths toward delivering precision medicine.

# The Success of the Precision Medicine Initiative Requires Public Health Partnership

- Inclusion and generalizability
- Focus on prevention
- Implementing what we know
- Healthcare-public health partnerships

## A Public Health Perspective on a National Precision Medicine Cohort Balancing Long-term Knowledge Generation With Early Health Benefit

The new US precision medicine initiative<sup>1</sup> has been made possible by improvement and price reduction in genome sequencing, as well as advances in multiple sectors of biotechnology. The initiative includes 2 components: a focus on cancer intended to spur development of new targeted cancer treatments, and a proposal for establishing a national cohort of at least 1 million people to explore genetic and environmental determinants of health and disease. The success of this initiative requires a public health perspective to help ensure generalizability, assess methods of implementation, focus on prevention, and provide an appropriate balance between generation of long-term knowledge and short-term health gains.

For example, improving access to smoking cessation assistance is a component of the highly successful public health efforts that have resulted in reductions in smoking over the past few decades. Recent data suggest that using genetically informed biomarkers of the speed with which people metabolize nicotine<sup>2</sup> could lead to personalized smoking cessation. Another example of precision prevention is changes in recommended screening schedules for people at increased risk of cancer, identified either by acquisition of family health history or through detection of those individuals who carry pathogenic mutations in high-risk cancer genes.

The proposed long-term investment in precision medicine comes at a time of increasing fiscal restraint and

# Challenges in Genomics and Precision Medicine Implementation That Require Public Health Leadership

Opinion

- Population health impact
- Evidence-based policy
- Health system implementation issues
- Laboratory quality
- Provider and public education
- Health disparities

## VIEWPOINT

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Office of Public Health Genomics, Centers for Disease Control and Prevention, Atlanta, Georgia.

## No Shortcuts on the Long Road to Evidence-Based Genomic Medicine

**Rapid advances in genomics** have led to a new era of precision medicine, resulting in a substantial increase in the number of genetic tests available for research and clinical practice. As of April 27, 2017, the Genetic Testing Registry,<sup>1</sup> maintained and updated by the National Institutes of Health, contained information on 49 521 tests conducted at 492 laboratories for 10 733 disease conditions involving 16 223 genes. These tests cover a wide variety of diseases, rare and common, for different types of applications such as diagnosis, treatment, and prevention.

For 2 decades, there have been ongoing discussions of the importance of a strong evidentiary foundation for genetic testing. Several advisory groups, including the Task Force on Genetic Testing<sup>2</sup> and the Secretary's Advisory Committee on Genetic Testing,<sup>3</sup> made a number of recommendations to strengthen the evidence base for genomic medicine. The key element of the discussion is the need to have answers to a number of scientific questions that are relevant to establishing the analytic validity of genomic tests (the ability of tests to be accurate),

With the recent proliferation of direct-to-consumer genetic testing, the need for evidence in genomic medicine is more important than ever.

along with clinical validity (showing an association with disease end points) and clinical utility (showing effectiveness in improving health outcomes).<sup>2</sup>

### What Is the Status of the Evidence Base in Genomic Medicine?

ence in genomic medicine. In 2014, 283 published articles evaluated implementation of genomic medicine. Most studies described uptake of genomic tests or preferences for use by clinicians and patients. Key study design elements, such as the racial/ethnic composition of study populations, were underreported in studies. Few studies incorporated implementation science theoretical frameworks, sustainability measures, or capacity-building measures. Most studies focused on patient factors associated with implementation rather than macro-level factors (eg, health systems, policies, education, financing). Only a few studies attempted to develop and evaluate evidence-based strategies that can improve implementation of genomic medicine. The authors concluded that "the current knowledge base around implementation science to turn the promise of genomic medicine into reality is severely limited."<sup>5</sup>

### Moving Forward: A New Evidence Framework?

In March 2017, the National Academies of Sciences, Engineering, and Medicine released a study report titled "An Evidence Framework for Genetic Testing."<sup>6</sup> A special committee composed of a multidisciplinary group of experts examined the scientific literature to evaluate the evidence base for different types of genetic tests and "to develop a framework for decision making regarding the use of genetic tests in clinical care."<sup>6</sup> The committee focused on clinical applications and utility of genetic tests and examined how evidence is generated, evaluated, and synthesized. The committee reviewed several available methods for assessing the analytic validity, clinical validity, and clinical utility of genetic tests. These included the

# A Crucial Public Health Role is to Assess and Assure Population Health Impact of Genomics and Precision Medicine

- **Identifying applications that are supported by evidence for their use**
- **Assessing the population health impact of genomics and precision medicine**
  - Quantifying burden of disease
  - Assessing Impact of interventions in terms of lives saved, disease prevented or detected earlier
  - Quantifying and modeling healthcare costs and savings
  - Assessing barriers and facilitators to implementation
  - Documenting and addressing health disparities
  - Assessing Laboratory practice

# Public Health Genomics at CDC: The Office of Public Health Genomics

- **Mission: Enable and promote CDC efforts in integrating genomics and precision medicine applications into public health actions that prevent disease, save health care costs and reduce health disparities.**
  - Identify evidence-based genomic applications
  - Inform and communicate
  - Integrate into practice and programs

Public Health Genomics

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Top 10 Hits for 2017!

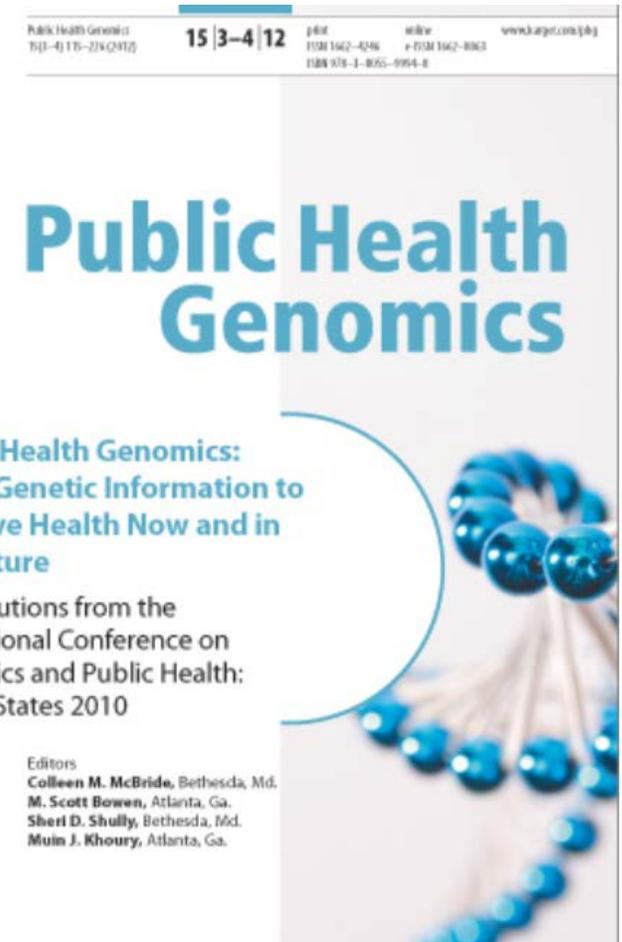
#1: The Public Health Genomics Knowledge Base: A continuously updated, searchable database of genomics and health impact information

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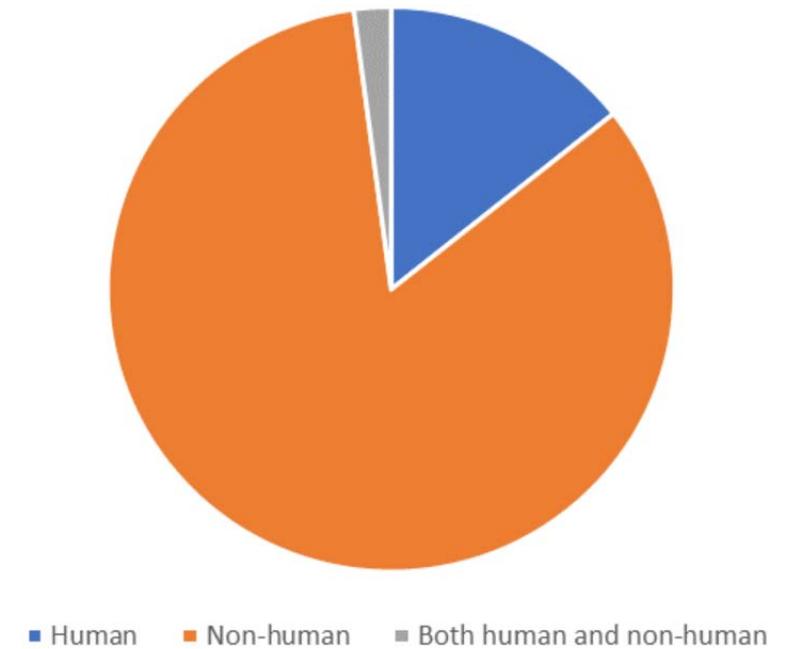
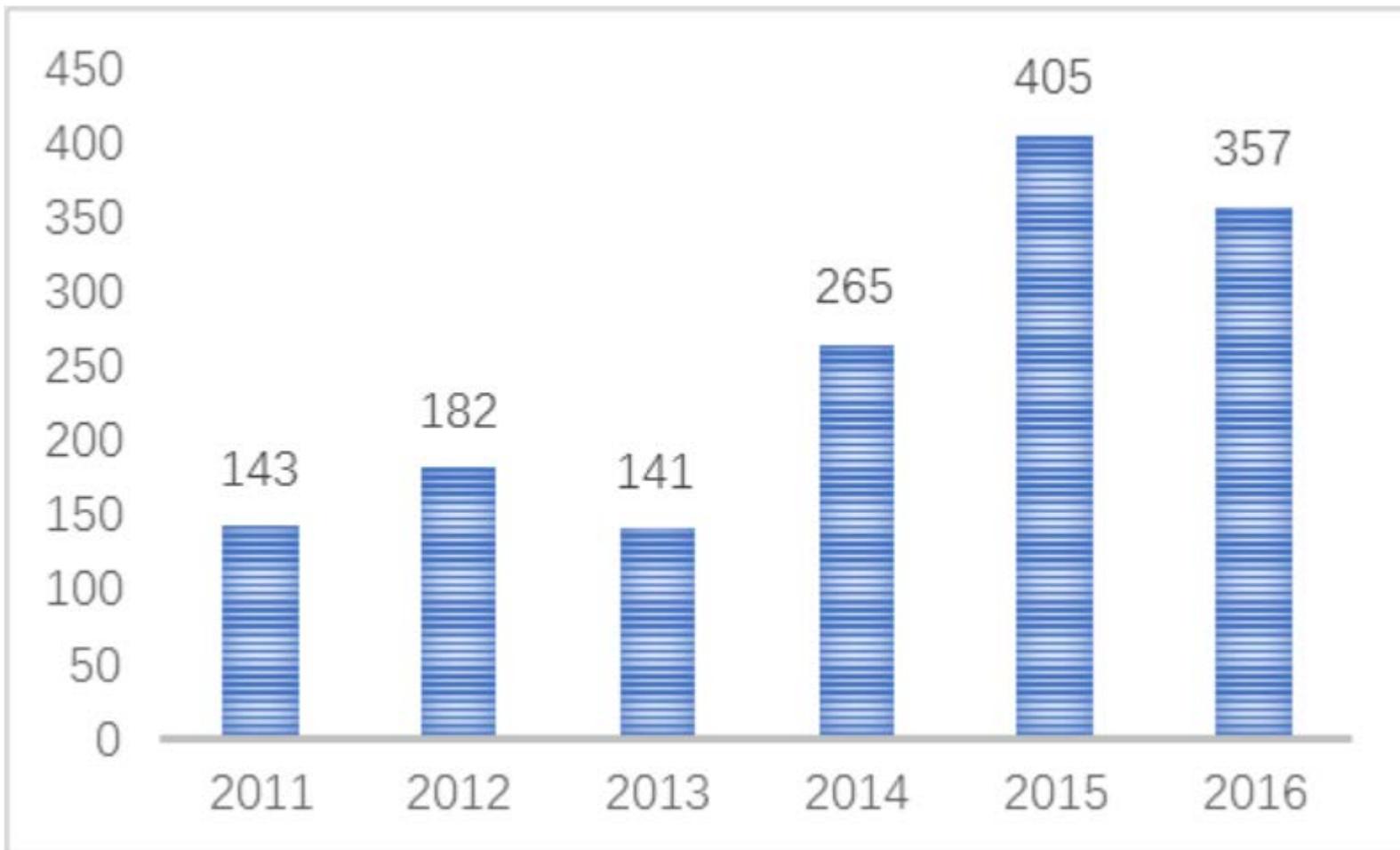
<b>What is Public Health Genomics?</b> PUBLIC HEALTH GENOMICS AT -A-GLANCE	 PRECISION PUBLIC HEALTH AND PRECISION MEDICINE	 ROLE OF PUBLIC HEALTH IN IMPLEMENTING GENOMICS	 PATHOGEN GENOMICS AND PUBLIC HEALTH
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# Public Health Genomics at CDC: A Brief History

- 1997: CDC Strategic Plan/Creation of OPHG
- 1998: First of 4 National Conferences
- 2001: Genomic Competencies for Public Health
- 2003: Model State Programs
- 2004: Family Health History Initiative
- 2004: EGAPP Initiative
- 2006: Seed Funding to CDC Programs
- 2012: New Plan with focus on 3 Conditions
- 2014: Advanced Molecular Detection
- 2014: Toolkit for Public Health, PHGKB
- 2016: Concept of “Precision Public Health”

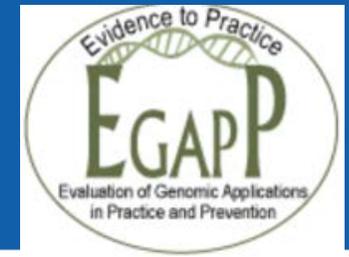


# Public Health Genomics at CDC: Publications 2012-2016



Source: <https://blogs.cdc.gov/genomics/2018/01/02/trends-in-cdc-publications/>

# EGAPP and Public Health Genomics: No Shortcuts to Evidence-based Genomics !



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## The EGAPP initiative: lessons learned

Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group\*

The Evaluation of Genomic Applications in Practice and Prevention Working Group was first convened in 2005 to develop and test evidence-based methods for the evaluation of genomic tests in transition from research to clinical and public health practice. Over the ensuing years, the Working Group has met 26 times, publishing eight recommendation statements, two methods papers, and one outcomes paper, as well as planning and serving as technical experts on numerous associated systematic reviews. Evaluation of Genomic Applications in Practice and Prevention methods have evolved to address implications of the proliferation of genome-wide association studies and are currently expanding to face challenges expected from clinical implementation of whole-genome

sequencing tests. In this article, we review the work of the Evaluation of Genomic Applications in Practice and Prevention Working Group over the first 8 years of its existence with an emphasis on lessons learned throughout the process. It is hoped that in addition to the published methods of the Working Group, the lessons we have learned along the way will be informative to others who are producers and consumers of evidence-based guidelines in the field of genomic medicine.

*Genet Med* advance online publication 8 August 2013

**Key Words:** evidence-based medicine; genetics; guideline development; public health genomics; systematic review methods

The completion of the human genome project was heralded as the dawn of the era of genomic-based personalized medicine. Numerous factors, however, have complicated the translation of scientific findings into clinical genomic testing with measurable health outcomes. The responsible integration of genomic technologies into medical care poses challenges to health-care providers, consumers, and other stakeholders. These emerging genomic applications to health care have been discovered by a

of their predisposition to diseases or response to treatment remains an inspiring goal of genomic medicine, and novel genomic diagnostics are allowing molecular targeting of therapies. As in any translation of new technology to health care, critical issues in genomic testing are now being defined. Little consensus exists among key stakeholders regarding the framework for developing, implementing, and evaluating genomic testing, and there are often sparse clinical data supporting the

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# CDC Evidence-based Classification of Genomics and Precision Medicine Applications

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

# Evidence-based Genomic Tests Are Available in Practice and Can Save Lives Now!

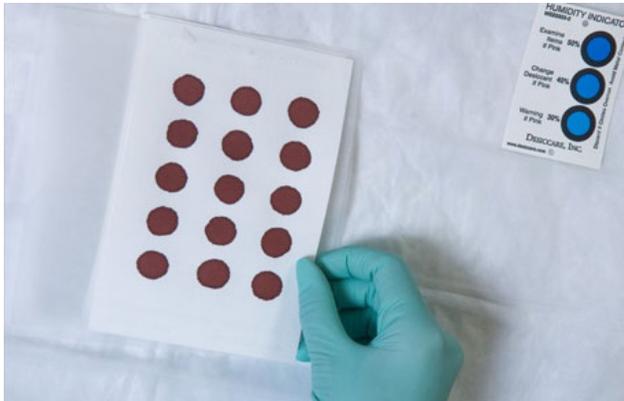
- 68 Tier 1 tests, more than half are cancer related
- 107 Tier 2 tests, many pharmacogenomics
- Information on guidelines, programs, publications and tools can be searched using the Public Health Genomics Knowledge Base (PHGKB)
- Intended uses across the lifespan include screening, diagnosis, treatment, prognosis and risk assessment
- Weekly Update reaches ~70,000 subscribers



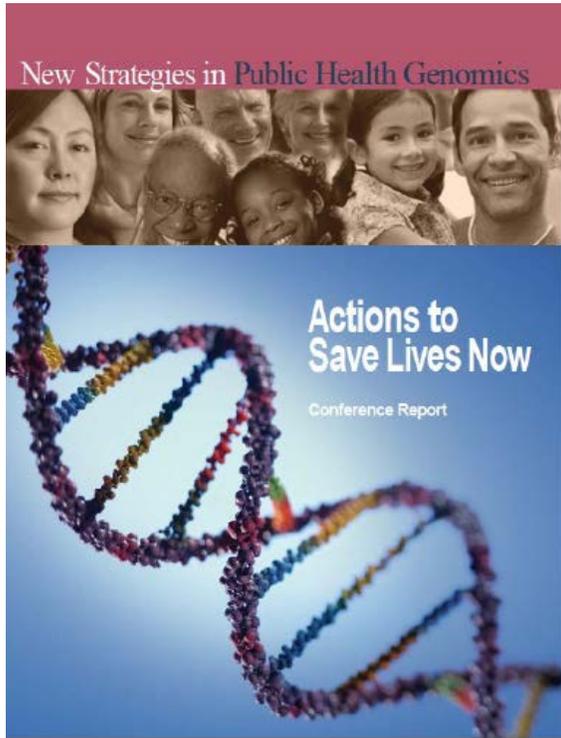
# Newborn Screening Remains the Largest Public Health Genetics Program in the World



- More than 5 decades in the US started with PKU
- State run public health program that screens 4 million newborns every year
- Identifies more than 10,000 babies with 30+ genetic, metabolic & other disorders
- Complex system & policy issues (public health, healthcare, laboratories, costs, etc...)
- Residual blood spots have been used for other purposes



# Selected Tier 1 Genomic Applications Beyond Newborn Screening



2012

- Hereditary Breast and Ovarian Cancer (*BRCA1/2*)
- Hereditary Nonpolyposis Colorectal Cancer (Lynch Syndrome)
- Familial Hypercholesterolemia
- Collectively Affect ~2 Million People in US and Most Don't know it.
- Implementation of existing evidence-based guidelines can prevent cancer & heart disease, & save thousands of lives every year!
- Toolkit for public health departments
- Working with CDC programs and external partners

# Selected Cancers Associated with Hereditary Cancer Syndromes

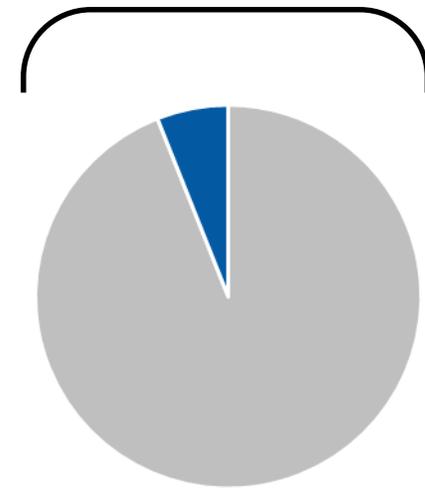
## HBOC Syndrome



**5%** or approximately  
**22,000** cases of  
breast cancer each year

**10%** or approximately  
**2,000** cases of  
ovarian cancer each year

## Lynch Syndrome



**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. *Nature Clinical Practice Gastroenterology & Hepatology* (2006); 3:670-679.

# Evidence-based Recommendations for Selected Hereditary Cancers

- **U.S. Preventive Services Task Force recommendation on *BRCA*-related cancer:**
  - Screening to identify family history associated with *BRCA1* or *BRCA2*, genetic counseling and *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



Georgia Hurst, Lynch syndrome patient advocate and her son

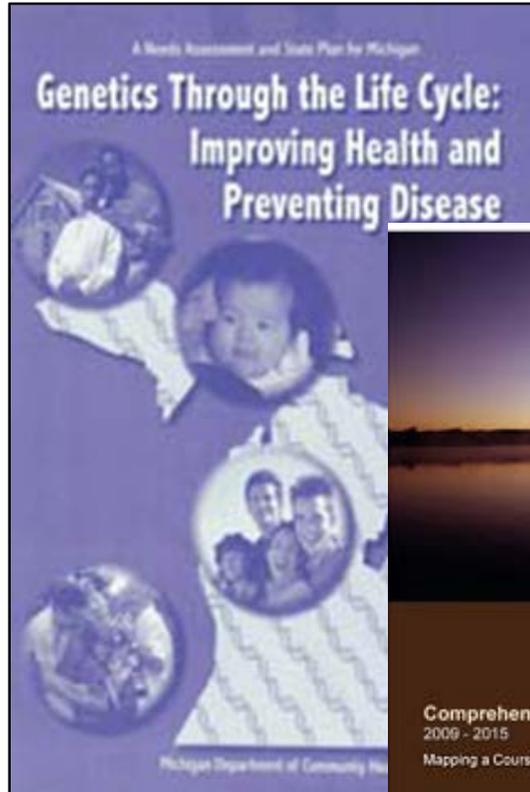
# CDC Public Health Cancer Genomics Program

- **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.

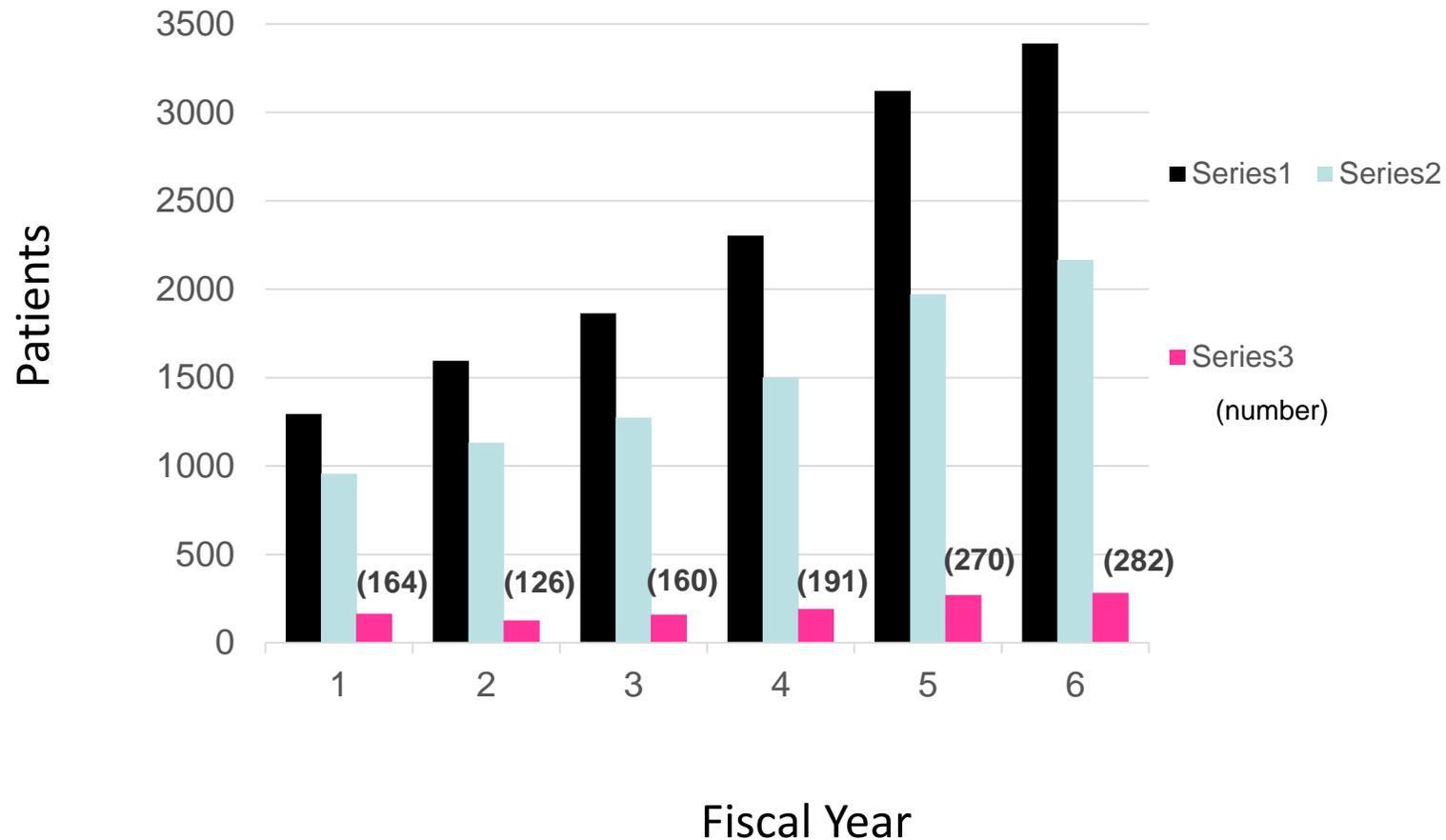
CDC Awards Funding to Support Cancer Genomics. [cdc.gov/cancer/breast/what\\_cdc\\_is\\_doing/genomics\\_foa.htm](https://www.cdc.gov/cancer/breast/what_cdc_is_doing/genomics_foa.htm)

# Michigan Illustrates State Public Health Activities in Cancer Genomics



- Improved health outcomes and an enhanced quality of life for the people of Michigan through appropriate use of genetic information, technology, and services
- Increase availability of cancer-related genetic information to the Michigan public and decrease barriers to risk-appropriate services

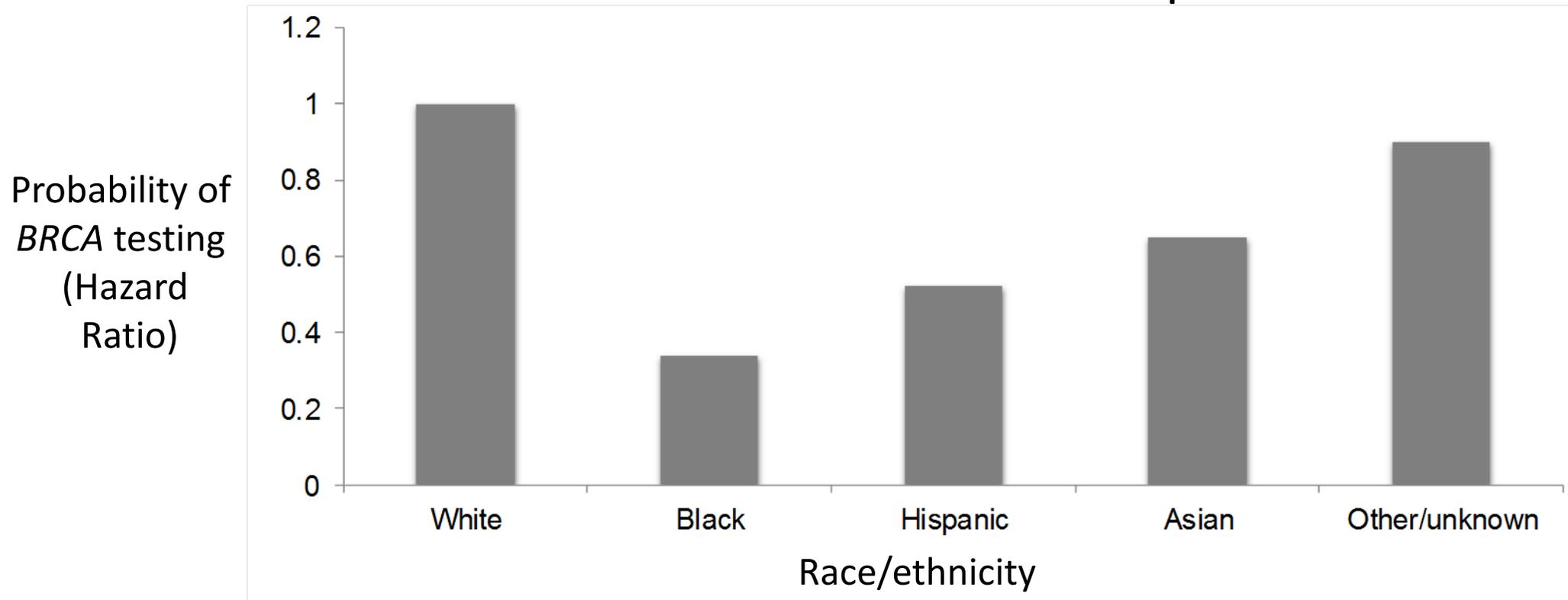
# Michigan Successes Related to *BRCA* Counseling Access and Referrals



Racial/ethnic and geographic disparities were present

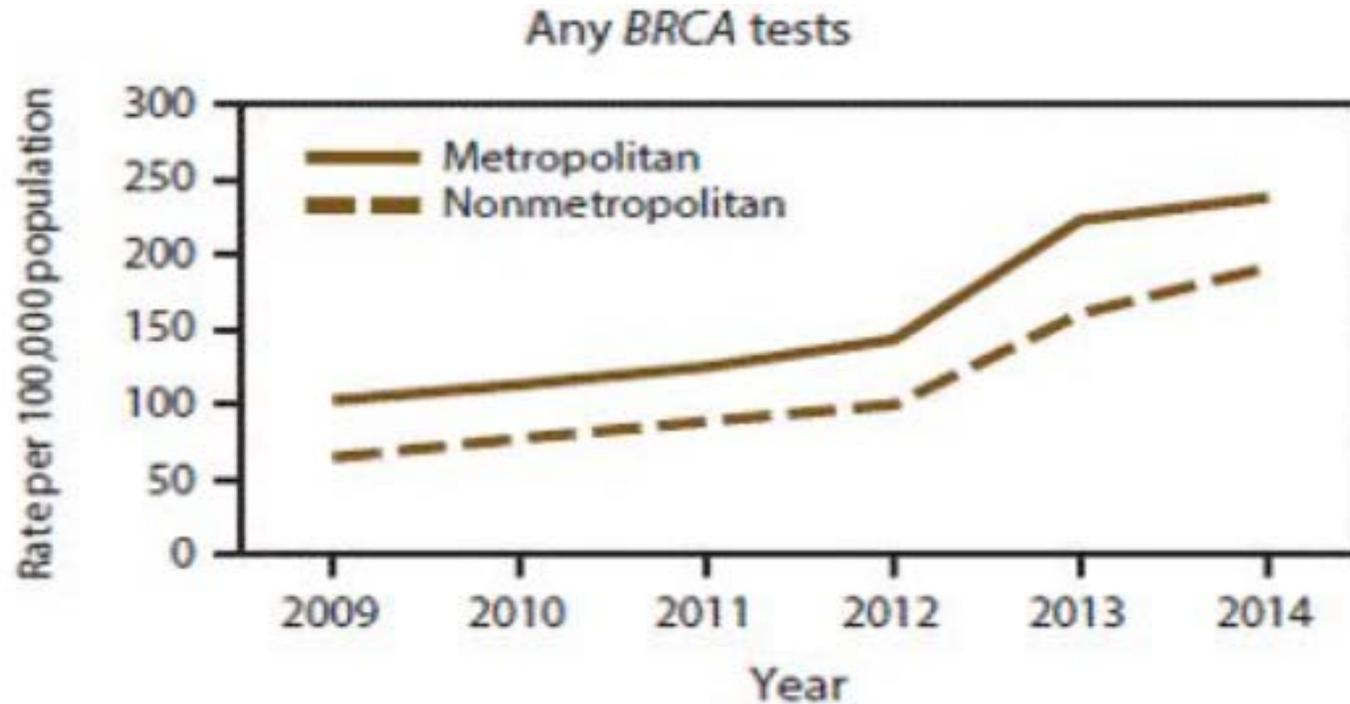
# Public Health Genomics and Health Disparities: Disparities by Race/Ethnicity

*BRCA* testing in young women with breast cancer:  
underutilization in Black and Hispanic women



# Public Health Genomics and Health Disparities: Rural-Urban Disparities

Probability of  
*BRCA* testing  
(Hazard  
Ratio)



**BRCA Genetic Testing Among Women Aged 18-64 Years with Employer-Sponsored Health Insurance in Nonmetropolitan and Metropolitan Areas - United States, 2009-2014.**

# Familial Hypercholesterolemia: A Missed Opportunity for Preventing Early Heart Attacks

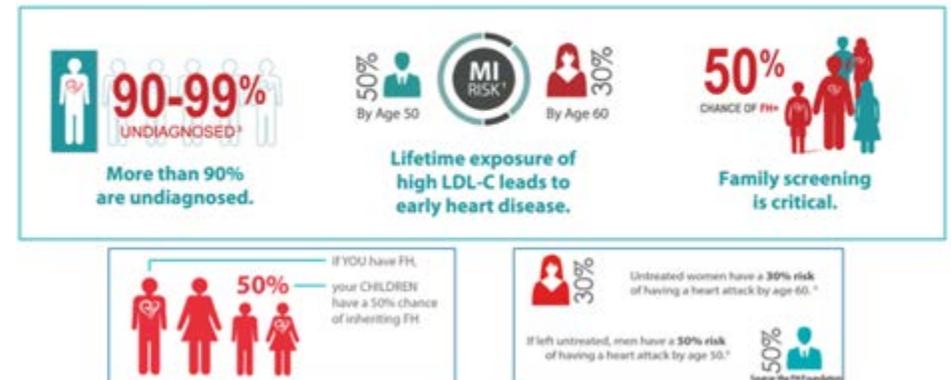
- Common autosomal dominant condition (1/250) associated with premature death from heart disease
- Evidence-based recommendation for aggressive cholesterol reduction and cascade screening in relatives
- Highly underdiagnosed and undertreated
- Racial and ethnic disparities in diagnosis and management
- Missed opportunities for public health-health care partnerships



Katherine Wilemon,  
President of  
The FH Foundation  
and her daughter

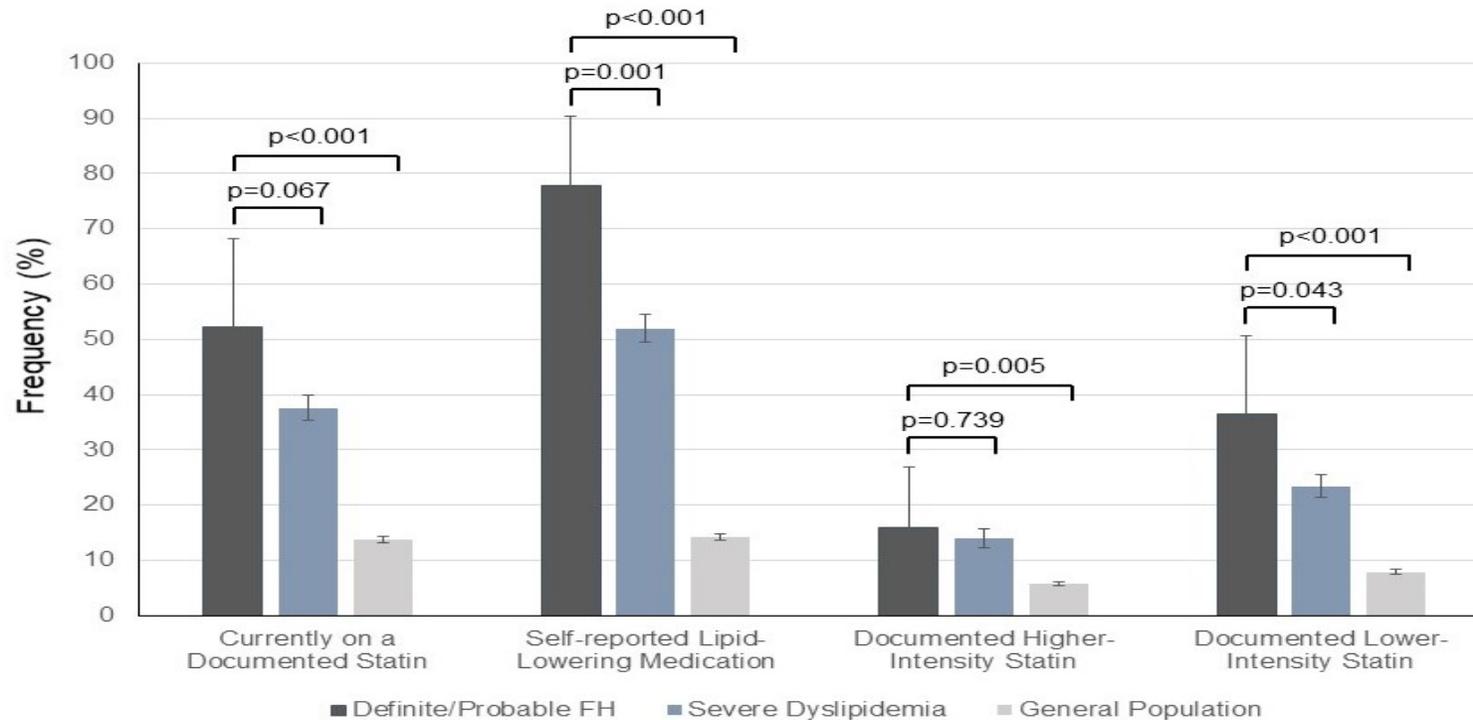


## Under-Detection and Under-Treatment of FH is High



# Familial Hypercholesterolemia is Common and Undertreated in the United States

## *Prevalence of documented statin and self-reported lipid lowering medication use*



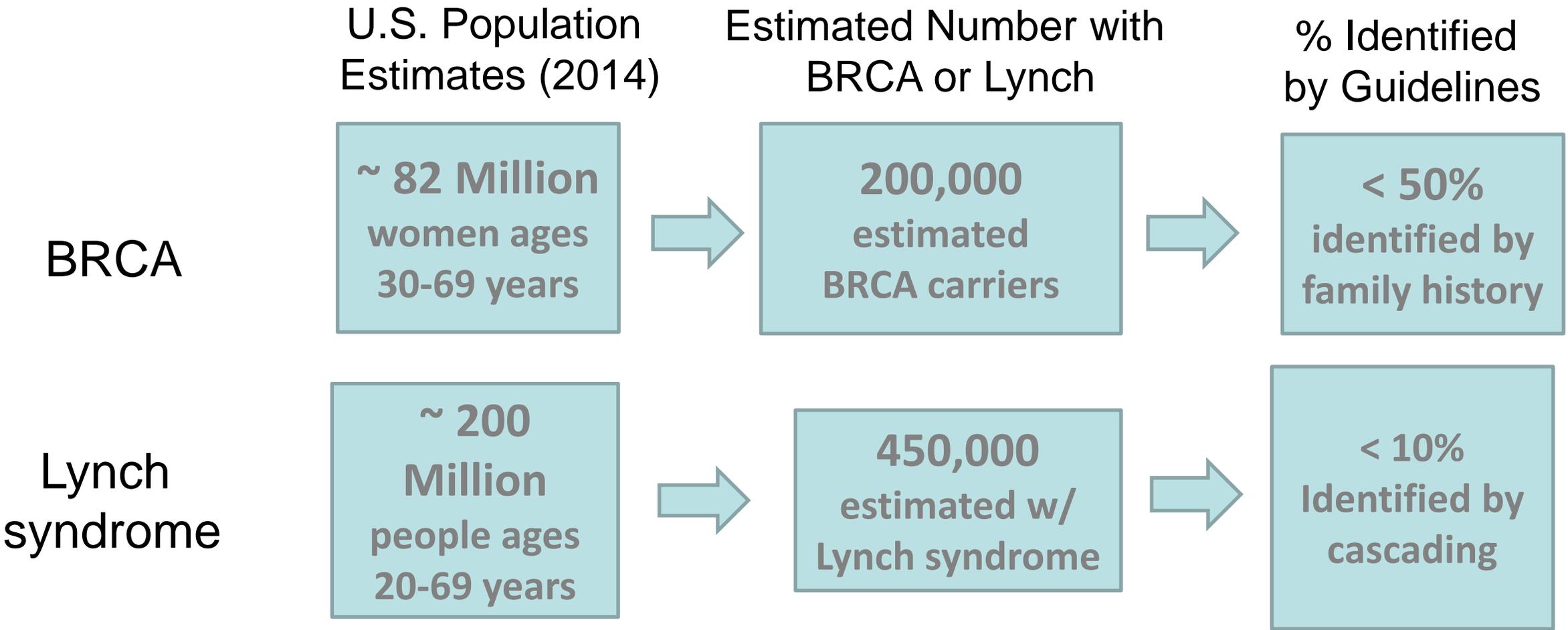
Young and uninsured patients are at the highest risk for under treatment

# Priority Areas For Human Genomics in Public Health Practice for the Coming 5 Years

- Expanding and integrating the CDC **tier 1 List**
- Developing an evidence-based approach to **genome sequencing** in health systems
- Exploring **pharmacogenomics** as a tool for public health
- Exploring genomics as a tool for **risk assessment and stratification** to enhance screening and prevention for a wide variety of common diseases
- Developing a public health approach to **rare diseases**
- **Preparing** the workforce and the public



# Ascertaining Persons with BRCA mutations and Lynch Syndrome using Current Evidence-based Guidelines Will Miss Most Cases Even with Cascade Testing



# Health Systems Partnerships for Appropriate Use of Population Wide Genome Sequencing and Measuring its Public Health Impact

## We screen newborns, don't we?: realizing the promise of public health genomics

James P. Evans, MD, PhD<sup>1</sup>, Jonathan S. Berg, MD, PhD<sup>1</sup>, Andrew F. Olshan, PhD<sup>2</sup>, Terry Magnuson, PhD<sup>1</sup> and Barbara K. Rimer, DrPH<sup>3</sup>

Genomics and public health have been uneasy bedfellows for some time. Most efforts to improve population health through genomic approaches have focused on the assessment of risks for common diseases, with the aim of tailoring interventions and screening.<sup>1</sup> However, the improvement of population health through such an approach has remained elusive.<sup>2</sup> Now, rapid progress in affordable, robust DNA sequencing offers a promising opportunity. By expanding the field's focus from common to rare diseases, it may be possible to realize the promise of public health genomics by identifying those millions of individuals who unknowingly carry mutations that confer a dramatic predisposition to preventable diseases.

population health risks;<sup>4</sup> medical interventions are usually most beneficial when identified disease risks and potential benefits often are justified by the hope that simply informing individuals of their genetic risks for disease will induce beneficial behavioral changes.<sup>5</sup> Thus far, this notion is largely contradicted by available evidence.<sup>5,6</sup> Although we already know how to lower risks for most common diseases, getting populations to eat properly, exercise, and give up unhealthy behaviors, especially without major policy changes, is challenging, and there is little evidence to suggest that genetic tweaking of risk will meaningfully augment these efforts.<sup>7,8</sup>

## MyCode<sup>®</sup> results returned

533 patient-participants have received results\*



For the latest results, see [go.geisinger.org/results](http://go.geisinger.org/results).

December 1, 2017

Risk condition	Patients per risk condition	Gene	Patients per gene
<i>CDC tier 1 conditions (click link)</i>			
<b>Hereditary breast and ovarian cancer</b> (early breast, ovarian, prostate and other cancers)	<b>203</b>	<b>BRCA1</b> <b>BRCA2</b>	<b>68</b> <b>135</b>
<b>Familial hypercholesterolemia</b> (early heart attacks and strokes)	<b>86</b>	<b>APOB</b> <b>LDLR</b>	<b>31</b> <b>55</b>
<b>Lynch syndrome</b> (early colon, uterine and other cancers)	<b>50</b>	<b>PMS2</b> <b>MSH6</b> <b>MSH2</b> <b>MLH1</b>	<b>18</b> <b>23</b> <b>6</b> <b>3</b>
<b>Cardiovascular risk</b>			
<b>Cardiomyopathy</b> (diseases of the heart muscle with dangerous complications)	<b>52</b>	<b>MYH7</b> <b>MYBPC3</b> <b>TPM1</b> <b>TNNI3</b> <b>TNNT2</b> <b>MYL3</b> <b>LMNA</b>	<b>8</b> <b>29</b> <b>2</b> <b>3</b> <b>5</b> <b>4</b> <b>1</b>

# Progress in Pharmacogenomics for Public Health Impact: Pockets of Success but Not Ready for Large Scale Implementation



Contents lists available at ScienceDirect

**Journal of Pharmaceutical Sciences**

journal homepage: [www.jpharmsci.org](http://www.jpharmsci.org)

ELSEVIER

Review

**Clinical Implementation of Pharmacogenomics for Personalized Precision Medicine: Barriers and Solutions**

Michelle E. Klein<sup>1</sup>, Md Masud Parvez<sup>1</sup>, Jae-Gook Shin<sup>1,2,\*</sup>

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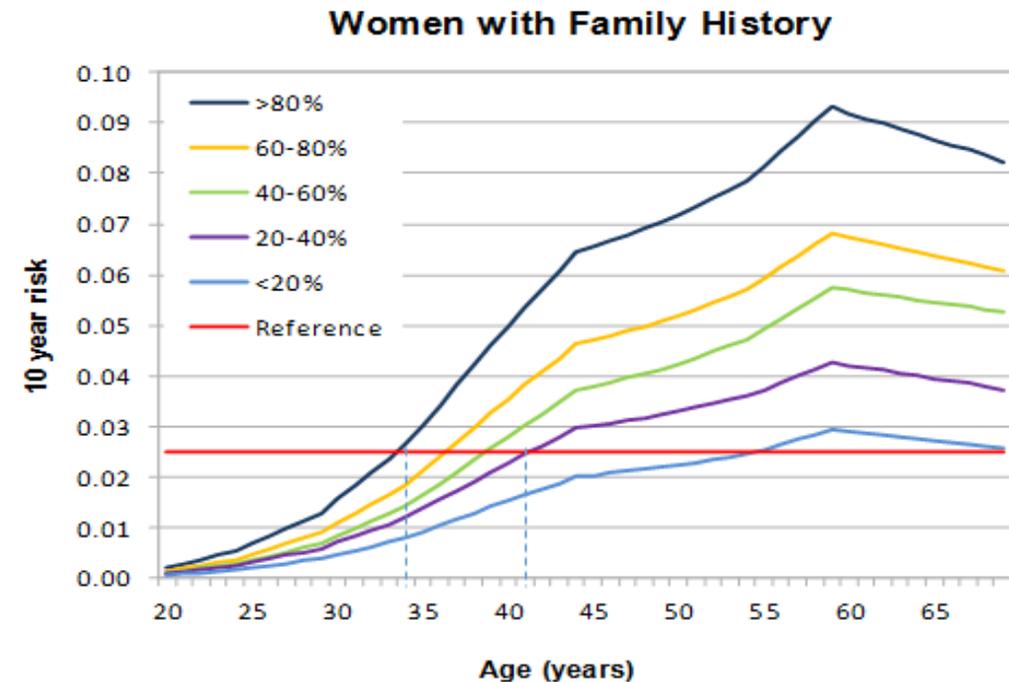
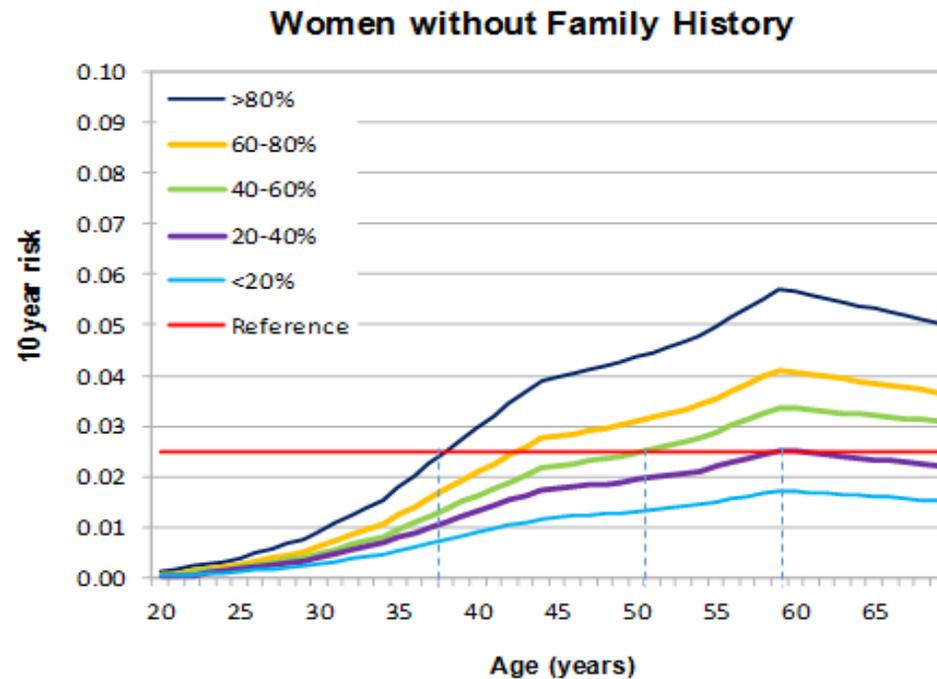
**ABSTRACT**

Clinical implementation of pharmacogenomics (PGx) leads to personalized medicine, which improves the efficacy, safety, and cost-effectiveness of treatments. Although PGx-based research has been conducted for more than a decade, several barriers have slowed down its widespread implementation in clinical practice. Globally, there is an imbalance in programs and solutions required to empower the clinical implementation of PGx between countries. Therefore, we aimed to review these issues comprehensively, determine the major barriers, and find the best solutions. Through an extensive review of ongoing clinical implementation programs, scientific, educational, ethical, legal, and social issues, information technology, and reimbursement were identified as the key barriers. The pace of global implementation of genomic medicine coincided with the resource limitations of each country. The key solutions identified for the earlier mentioned barriers are as follows: building of secure and suitable information technology infrastructure with integrated clinical decision support systems along with increasing PGx evidence, more regulations, reimbursement strategies for stakeholder's acceptance, incorporation of PGx education in all institutions and clinics, and PGx promotion to all health care professionals and patients. In conclusion, this review will be helpful for the better understanding of common barriers and solutions pertaining to the clinical application of PGx.

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# Example of Genetic Risk Stratification: Can We Use Age and “Genetic Risk Scores” in Breast Cancer Screening?

10-year absolute risk of developing breast cancer for women with and without family history by polygenic risk percentiles



# Rare Diseases, Genomics and Public Health: A Growing Intersection

- A rare disease affects < 200,000 people
- > 7000 rare diseases; 25 million people in the US
- 4300 “genetic” conditions with known molecular basis
- The “diagnostic odyssey”: Genome sequencing yield 25-50%
- More and more therapies found
- Assessing population burden of disease
- Assurance of health care and prevention needs

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COMMENTARY | Genetics  
inMedicine

## The need for a next-generation public health response to rare diseases

Rodolfo Valdez, MSc, PhD<sup>1</sup>, Scott D. Grosse, PhD<sup>1</sup> and Muin J. Khoury, MD, PhD<sup>2</sup>

Few public health research activities trigger stronger calls to public health action than research into the burden of disease. This research uses standard measures to quantify actual or potential losses that populations may experience due to the presence of diseases and injuries. Standard measures range from simple (e.g., mortality) to complex (e.g., disability-adjusted life years). Despite certain deficiencies in quantity and quality of data at a global scale, the burden of disease has been estimated for approximately 300 conditions that affect millions of people around the world, with the intent of informing the design of health systems and development of public health policy.<sup>1</sup> However, such efforts have been dedicated mostly to relatively common conditions and include only a handful of rare diseases under the label “congenital anomalies.”

There is no standard definition of rare disease, but, overall, a disease is considered rare when it affects fewer than 7 people out of 10,000 in a given population.<sup>2</sup> Between 5,000 and 8,000 rare diseases have been identified in the world, and approximately 80% of them have a genetic origin<sup>2</sup> (more on that later). Despite the suspected large number of adults and children affected by

refer to relatively benign conditions, such as heterozygosity for the hemoglobin S variant of the  $\beta$ -globin gene (i.e., sickle cell trait), and other codes refer to transient conditions (e.g., certain congenital heart abnormalities) that are particularly common among infants born preterm.

Walker et al.<sup>3</sup> also calculated the impact of rare diseases on the health-care system in Western Australia; hospital stays related to rare disease were, on average, approximately 3 days longer than stays for the general population. The 2% of the population affected by rare diseases accounted for 4.6 to 10.5% of total hospitalization expenditures.<sup>3</sup> These results affirm previous findings. Using codes that largely overlap with the codes used by Walker et al.,<sup>3</sup> Yoon et al.<sup>5</sup> reported that, in a pediatric population from two states in the United States comprising patients with hospital codes for birth defects and genetic diseases, approximately 2.5% accounted for 9 to 12% of pediatric hospital admissions and 16 to 28% of total costs. Rare diseases may account for a greater share of hospital admissions and costs for children than for adults. The data from Walker et al.<sup>3</sup> enable investigators to test this hypothesis.

# Preparedness, Preparedness, Preparedness!!

## Genomic Workforce Competencies 2001

Genomic competencies for the public health workforce at any level in any program

A public health worker is able to:

- Demonstrate basic knowledge of the role that genomics plays in the development of disease
- Identify the limits of his/her genomic expertise
- Make appropriate referrals to those with more genomic expertise

# THANK YOU!

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### WEEKLY UPDATE

Weekly summary of genomics and health impact information

### GENETICS 101

Genetics basics explained including a glossary of genetic terms

### PHGKB

Online searchable knowledge base on genomics and health impact information

### FAMILY HEALTH HISTORY

Family health history is known to be a risk factor for most diseases

### REPORTS AND PUBLICATIONS

Scientific reports and publications in public health genomics

### GENOMICS AND DISEASES

Genomics is important for many diseases of public health significance

### GENOMICS & HEALTH IMPACT BLOG

A blog devoted to genomic issues in research, policy and practice

### GENETIC COUNSELING

Helping to inform individuals and families about genetic risks, testing and interventions

### REAL STORIES

The power of personal stories for communicating genomic information

### GENOMIC TESTING

Genomic tests are available for many diseases across the lifespan

### EVENTS AND MULTIMEDIA

Podcasts, videos, slides and other CDC events information relevant to genomics and public health

### EPIDEMIOLOGY

Epidemiology is a scientific foundation for public health genomics