



# **Implementing Public Health Genomics in Your State: Resources from the CDC Office of Public Health Genomics**

**Muin J. Khoury, Dave Dotson, Ridgely Fisk Green, and  
Marta Gwinn**

**Office of Public Health Genomics**

Genetic Alliance and APHA Genomics Forum Webinar  
November 15, 2016

# Organizing Information for Public Health Genomics: “4 Phases of Translation”

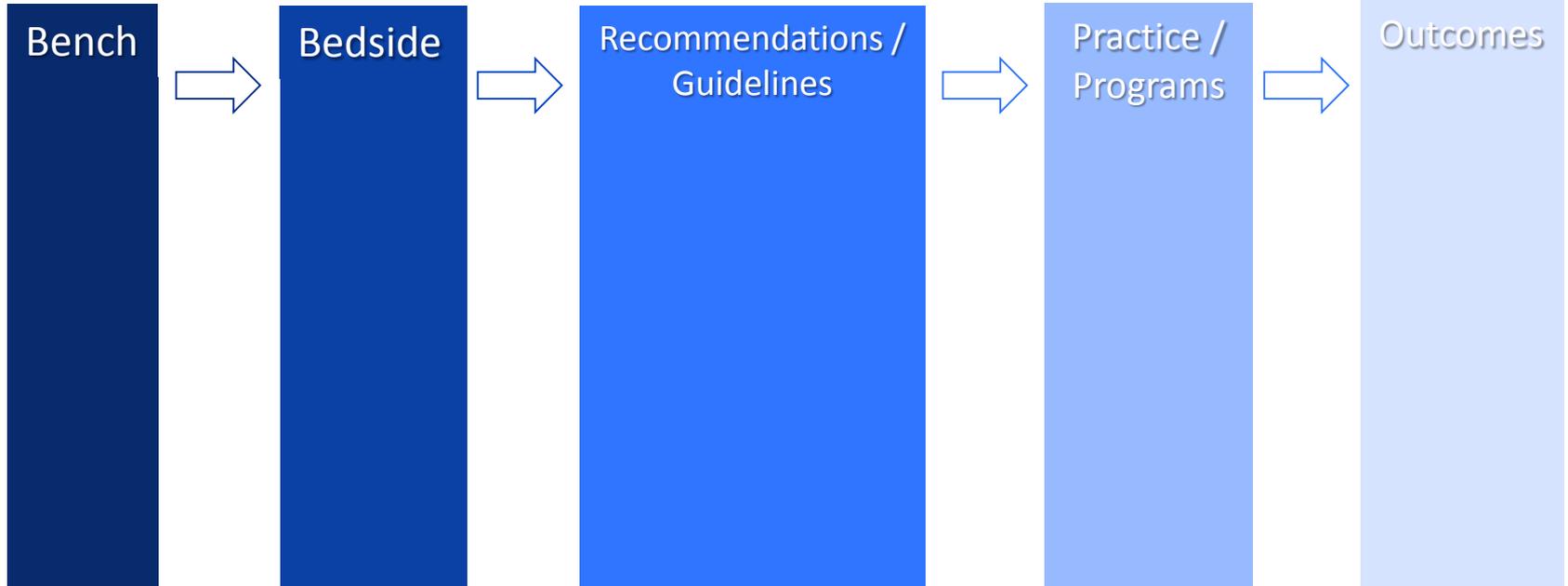
**T0**

**T1**

**T2**

**T3**

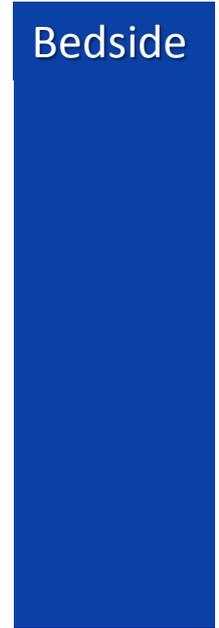
**T4**



# Organizing Information for Public Health Genomics: “4 Phases of Translation”

**T0**

**T1**



Research and development

*Drugs*

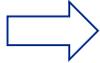
*Devices (Tests)*

# Organizing Information for Public Health Genomics: “4 Phases of Translation”

T0

T1

T2



Testing, observation, and evaluation

*Clinical trials*

*Epidemiologic studies*

*Evidence review*



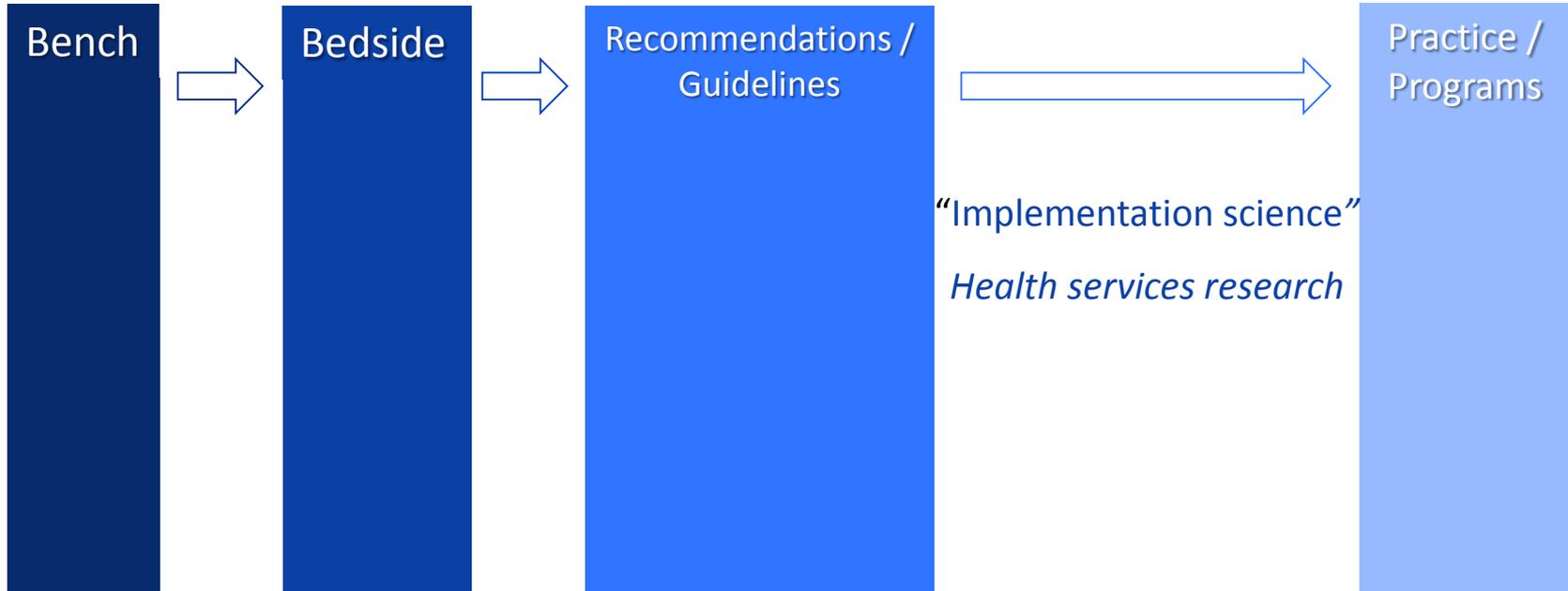
# Organizing Information for Public Health Genomics: “4 Phases of Translation”

**T0**

**T1**

**T2**

**T3**



# Organizing Information for Public Health Genomics: “4 Phases of Translation”

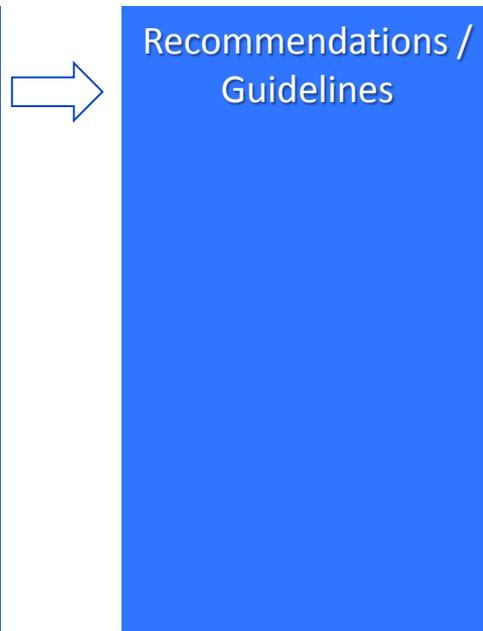
**T0**



**T1**



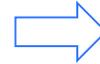
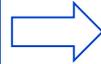
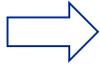
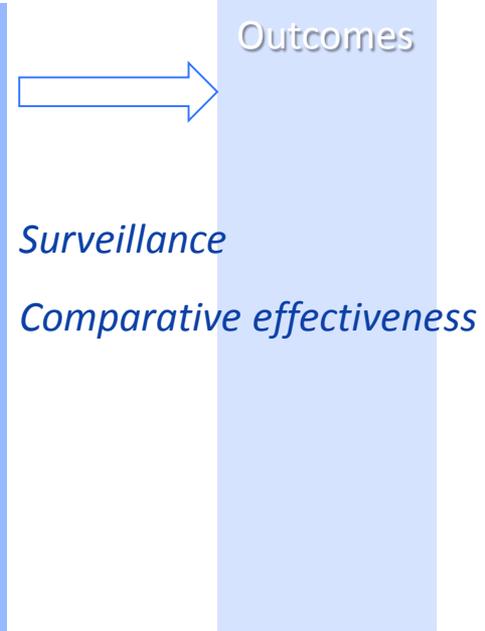
**T2**



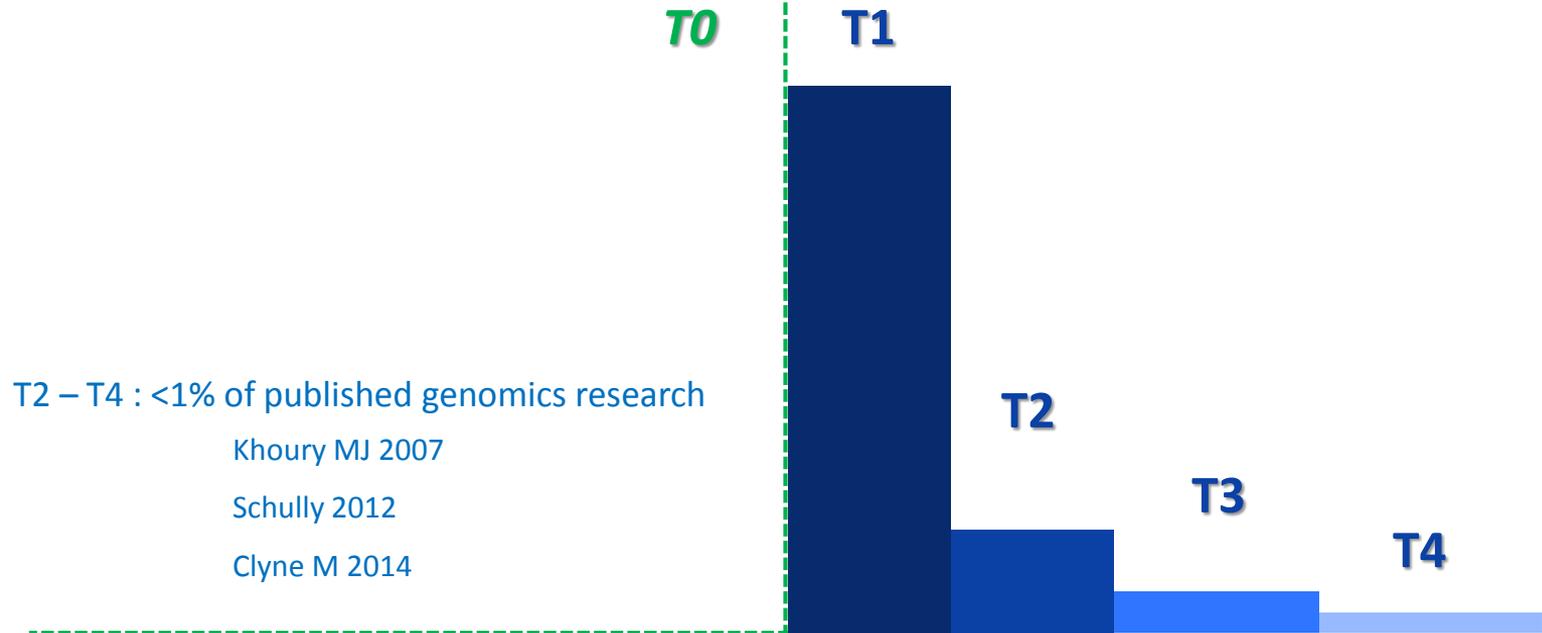
**T3**



**T4**



# Organizing Information for Public Health Genomics: “4 Phases of Translation”



## Public Health Genomics



# Implementing Public Health Genomics in Your State!



**Cancer  
Moonshot**

CANCER MOONSHOT



**Health History**

Classes/Contacts  
Neurological  
Psychological  
**Seizures/Epilepsy**  
Allergic Reactions  
Allergies

EPILEPSY



PRECISION PUBLIC HEALTH



SUDDEN DEATH

## WHAT'S NEW

Weekly summary of genomics and health impact information

## PHGKB

Online searchable knowledge base on genomics and health impact information

## GENOMICS AND DISEASES

Genomics is important for many diseases of public health significance

## AMD CLIPS

Weekly news and publications on pathogen genomics and bioinformatics

## FAMILY HEALTH HISTORY

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

## IMPLEMENTATION

What public health can do now to save lives using genomics

## PATHOGEN GENOMICS

New tools are changing the landscape in the fight against infectious diseases

## BLOG

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

## GENOMIC TESTING

Genomic tests are used in many diseases

## REPORTS AND PUBLICATIONS

CDC reports and publications in genomics

## EPIDEMIOLOGY

Epidemiology is a scientific foundation for public health genomics

## PODCASTS AND VIDEOCASTS

Podcasts on genetic testing, diseases, and family health history



## Dr Khoury's Tweets

@DrKhouryCDC

Using pathogen #genomics to track polio.  
<https://t.co/T15hVqyhQW> #WorldPolioDay  
<https://t.co/C5gRQqdHwH>

What does polio eradication have to do with genetic diseases? CDC blog post  
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From genetic counseling of individuals to cascade screening in populations: We can do this! <https://t.co/GneKEX9LEQ>  
<https://t.co/eUI9HaF5ol>

What is Hypertrophic Cardiomyopathy & what's the role of genetics & genetic testing? #PHGKB <https://t.co/4wlq8Z1t5s>  
<https://t.co/tbKxPvkG3R>

Is there a role for genomics in investigating sudden death? Get the latest info. #PHGKB  
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### About

[Genomics \(A-Z\)](#)

[Office of Public Health Genomics](#)

[State Implementation Map](#)

[Advanced Molecular Detection Weekly Clips \(Current Edition\)](#)

[Relevant Resources](#)

### All Databases

[CDC Information Database \(717\)](#)

[CDC-Authored Genomics Publication Database \(1471\)](#)

[Genomics & Health Impact Scan Database \(7477\)](#)

[Guideline Database \(257\)](#)

[Tier Table Database \(159\)](#)

[Implementation Database \(240\)](#)



## About PHGKB

The CDC Public Health Genomics Knowledge Base is an online, continuously updated, searchable database of published scientific literature, CDC resources, and other materials that address the translation of genomic discoveries into improved health care and disease prevention. The Knowledge Base, cosponsored by the Division of Cancer Control and Population Sciences at the National Cancer Institute, is curated by CDC staff and is regularly updated to reflect ongoing developments in the field. This compendium of databases can be searched for genomics-related information on any specific topic. We will continue to add additional features to the knowledge base and are interested in your feedback via email.

[Scientific publications related to PHGKB](#)

## Database Content (Last Updated: *Nov-10-2016 11AM*)

### CDC Resources



- CDC Information (717)
- CDC-Authored Pub (1471)

### Selected Insights & Reviews



- Human (3530)
- Pathogen(3021)

### Epidemiology



- Human (122833)
- Pathogen (604)

All Databases

CDC Information Database (666)

CDC-Authored Genomics Publication Database (1348)

Genomics & Health Impact Scan Database (6544)

Guideline Database (257)

Tier Table Database (159)

Implementation Database (230)

Advanced Molecular Detection Clips Database (4869)

HuGE Navigator

Last Updated: *Jul-15-2016 2PM*



66  
(1348)

### Selected Insights & Reviews



- Human (3007)
- Pathogen(2559)

### Epidemiology



- Human (119632)
- Pathogen (543)



### Evidence Synthesis



- Guidelines (257)
- Tier Table (159)
- Synthesis (823)

### Practice & Implementation



- Human (230)
- Pathogen (64)

Relevant Resources

All Databases -

CDC Information  
Database (666)

CDC-Authored  
Genomics Publication  
Database (1348)

Genomics & Health  
Impact Scan Database  
(6544)

Guideline Database  
(257)

Tier Table Database  
(159)

Implementation  
Database (230)

Advanced Molecular  
Detection Clips  
Database (4869)

HuGE Navigator +

# Public Health Genomics Knowledge Base

- **What** are the different databases?
- **Why** did we build these databases?
- **Where** do we find content for each database?
- **How** can you use PHGKB?

All Databases -

CDC Information Database (666)

CDC-Authored Genomics Publication Database (1348)

Genomics & Health Impact Scan Database (6544)

Guideline Database (257)

Tier Table Database (159)

Implementation Database (230)

Advanced Molecular Detection Clips Database (4869)

HuGE Navigator +

## What are the different databases?

- CDC Information – *web pages*
- CDC-Authored Genomics Publications – *journal articles*
- Genomics and Health Impact Scan Database
- Guideline Database
- Tier Table
- Implementation Database
- Advanced Molecular Detection Clips
- HuGE Navigator – *genetic association studies (PubMed)*

- Genomics
- About Us +
- What's New**
- PHGKB
- Family Health History
- Genomic Testing +
- Implementation +
- Genomics and Diseases +
- Publications
- Genomic Blog
- Pathogen Genomics
- Podcasts and Videocasts
- Epidemiology
- Resources

[Genomics](#) > [What's New](#)

## What's New



November 10, 2016

### Spotlight



#### Familial Hypercholesterolemia: New CDC Blog

This week, we feature a new blog post entitled: "[what gets measured gets done: public health progress in familial hypercholesterolemia](#)". For more information on public health implementation tool kit in FH, [click here](#).

For latest information on FH, [check our Public Health Genomics Knowledge Base](#).

# Genomics and Health Impact Scan Database

## Why did we build it?

- **Challenge:** *Keeping up with genomics and family health history developments relevant to public health*
- **Opportunity:** Identify the latest publications and other resources on population-based applications of genomic discoveries
- **Challenge:** *Addressing misconception that genomics applies only to research or clinical practice*
- **Opportunity:** Highlight public health applications of genomics—and the role of public health at the health care interface

# Genomics and Health Impact Scan Database

## *Where do we find the information?*

### Horizon scan

- Monitor Google Alerts, PubMed queries, key websites, and other sources
- Select news stories, blog posts, scientific articles, reports, websites
- Publish online in Weekly Update
- Add to searchable database

# Genomics and Health Impact Scan Database

- Indexed by category and “translation phase”

		T0/T1	T2	T3/T4
		Discovery, characterization, and development	Evaluation of tests and interventions	Implementation in practice and programs
A	Original studies	GWAS, biomarkers, and proposed new applications	Clinical trials, clinical cohorts, and new data on analytic or clinical validity	Studies generating new process or outcome data from clinical populations; surveillance
B	Research synthesis/modeling/meta-analysis/systematic reviews/narrative reviews	Meta-analysis and systematic reviews of gene–disease associations	Evidence reports	Cost-effectiveness analyses and national program evaluation
C	Guidelines/policies/recommendations	New nomenclature, data sharing, and publication standards	Clinical practice and professional guidelines	Electronic health standards, reporting requirements, and ethical standards
D	Tools/methods/training/education/decision support	Research road maps, databases, software, and training tools	Modeling methods, databases, and methods for systematic review	Clinical algorithms; provider and patient education materials

Clyne M, et al. Horizon scanning for translational genomic research beyond bench to bedside. Genet Med. 2014 Jul;16(7):535-8.

# Advanced Molecular Detection (AMD) Clips

Public Health Genomics Knowledge Base (v1.2)

PHGKB

About

Genomics (A-Z)

Office of Public Health  
Genomics

State Implementation Map

**Advanced Molecular  
Detection Weekly Clips  
(Current Edition)**

Relevant Resources

All Databases +

Contact Us

Search Knowledge Base

## Advanced Molecular Detection Clips

Publication Date: Nov 10, 2016

 Recommend  Tweet  Share

### About Advanced Molecular Detection Clips

Clips are selected weekly from a variety of sources, including PubMed, journal tables of contents, and online media. Special emphasis is given to the use of next-generation genetic sequencing in infectious disease public health surveillance, investigation, and development of new diagnostics and interventions. The collection is not comprehensive but aims to capture highlights, while surveying a wide range of topics. CDC-authored articles are flagged. Items marked I&E address aspects of implementation or evaluation in clinical or public health practice.

Archived Editions

[Search AMD Clips database](#)

[Visit AMD website](#)

# Public Health Genomics Knowledge Base (PHGKB)

## Global search of all databases

### PHGKB

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All Databases



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### Search Knowledge Base

Enter a search term

Submit

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Search PHGKB:

Search



## Hot Topics of the Day

Last Posted: Nov-10-2016 11AM



### Gene Therapy

Snapshots of Life: Lighting up the Promise of Retinal Gene Therapy

Francis Collins, NIH Director, November 10, 2016

Gene therapy for blistering skin disease appears to enhance healing in clinical trial

Stanford Medicine, November 1, 2016

Gene therapy shows promise for treating Niemann-Pick disease type C1

NIH, October 26, 2016

[More information on this topic](#)

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A paper describing the Public Health Genomics Knowledge Base (PHGKB) is now

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Search PHGKB:



## What's New

Last Updated: Nov 04, 2016

- [Association of Polymorphisms in FCGR2A and FCGR3A With Degree of Trastuzumab Benefit in the Adjuvant Treatment of ERBB2/HER2Positive Breast Cancer](#)  
PG Gavin et al, JAMA Oncology, November 3, 2016
- [Predictive Value of FcR Polymorphisms A Further Step on the Long and Winding Road to Application](#)  
R Dolcetti, JAMA Oncology, November 3, 2016
- [Validation of an Efficient Screening Tool to Identify Low-Income Women at High Risk for Hereditary Breast Cancer.](#)  
Stewart Susan L et al. Public health genomics 2016 Oct
- [Evaluation of human epidermal growth factor receptor 2 \(HER2\) single nucleotide polymorphisms \(SNPs\) in normal and breast tumor tissues and their link with breast cancer prognostic factors.](#)  
Furrer Daniela, et al. Breast (Edinburgh, Scotland) 2016 10 191-196
- [GSTP1, GSTM1, and GSTT1 polymorphisms as predictors of response to chemotherapy in patients with breast cancer: a meta-analysis.](#)  
Kong Xiangzhen, et al. Cancer chemotherapy and pharmacology 2016 10

[more](#)

PHGKB

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Search Knowledge Base

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- CDC-Authored Pub (13)



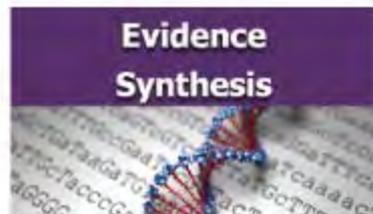
- Human (164)
- Pathogen (1)



- Human (5393)
- Pathogen (0)



- Human (498)
- Pathogen (0)



- Guidelines (23)
- Tier Table (19)
- Synthesis (53)



- Human (117)
- Pathogen (0)

## Relevant Resources

### All Databases -

CDC Information Database (666)

CDC-Authored Genomics Publication Database (1348)

Genomics & Health Impact Scan Database (6544)

Guideline Database (257)

Tier Table Database (159)

Implementation Database (230)

Advanced Molecular Detection Clips Database (4869)

HuGE Navigator +

Search result statistics are organized into following six modules below. (See description about each module.)

Click on the numbers to retrieve information records if they are hyperlinked.

### CDC Resources



- CDC Information (12)
- CDC-Authored Pub (13)

### Selected Insights & Reviews



- Human (164)
- Pathogen (1)

### Epidemiology



- Human (5393)
- Pathogen (0)

### Translational Research



- Human (498)
- Pathogen (0)

### Evidence Synthesis



- Guidelines (23)
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### Practice & Implementation



- Human (117)
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Original studies (A)

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Genetic associations, GxE, etc

### Translational Research



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### Translational Research



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### Practice & Implementation



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Research Synthesis / Modeling / Meta Analysis/ Systematic reviews (B)

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# Guidelines Database

## Why did we build it?

- **Challenge:** *Finding policies, guidelines, and recommendations that include genomics or family health history*
- **Opportunity:** Compile a centralized, searchable, publicly available database for policies, guidelines, and recommendations related to genomics or family health history
- **Challenge:** *Relevant policies, guidelines, and recommendations are developed independently by many different groups on different schedules*
- **Opportunity:** Keep updated with most recent guidelines, allow for easier cross-referencing

# Guidelines Database

Search Guidelines Database:

lung cancer

Search

Records 1-8 (of 8 Guidelines)

Download

**Query Trace:** lung cancer[original query]

[National Working Group Meeting on ALK diagnostics in lung cancer.](#)

Published 2014 (Pfizer-sponsored National Working Group Meeting on ALK Diagnostics in Lung Cancer)

[Systemic Therapy for Stage IV Non-Small-Cell Lung Cancer: American Society of Clinical Oncology Clinical Practice Guideline Update.](#)

Published 2015 (American Society of Clinical Oncology (ASCO))

[Final Recommendation Statement: Lung Cancer: Screening, December 2013](#)

Published 2013 (US Preventive Services Task Force)

[EGFR-TK mutation testing in adults with locally advanced or metastatic non-small-cell lung cancer](#)

Published 2013 (National Institute for Health and Care Excellence)

[Guideline Recommendations for EGFR Mutation Testing in Lung Cancer: Proposal of the Korean Cardiopulmonary Pathology Study Group.](#)

Published 2013 (Korean Cardiopulmonary Pathology Study Group)

# Tier Table Database

## *What it is:*

- Repository of genomic applications classified according to evidence
- Demonstration of a method for organizing horizon scanning results
- Potential aid to informed decision-making
- Scenario-based
- Systematic
- Subjective
- Context-dependent

# How it Works – Tier Level Criteria

Tier 1:

## Green

- FDA label requires use of test to inform choice or dose of a drug
- CMS covers testing
- Clinical practice guideline based on systematic review supports testing

Tier 2:

## Yellow

- FDA label mentions biomarker\*
- CMS coverage with evidence development
- Clinical practice guideline, not based on systematic review, supports use of test
- Clinical practice guideline finds insufficient evidence but does not discourage use of test
- Systematic review, without clinical practice guideline, supports use of test
- Systematic review finds insufficient evidence but does not discourage use of test
- Clinical practice guideline recommends dosage adjustment, but does not address testing

Tier 3:

## Red

- FDA label cautions against use
- CMS decision against coverage
- Clinical practice guideline recommends against use of test
- Clinical practice guideline finds insufficient evidence and discourages use of test
- Systematic review recommends against use
- Systematic review finds insufficient evidence and discourages use
- Evidence available only from published studies without systematic reviews, clinical practice guidelines, FDA label or CMS labels coverage decision

\*Can be reassigned to Green or Red if one or more conditions in these categories apply

# Tier Table Database

## *What it is **NOT**:*

- A substitute for informed decision-making
- An endorsement or recommendation for or against anything
- A comprehensive or complete assessment of tests or scenarios
- The final word in determining what is ready to implement

# Tier Table Database

## Why did we build it?

- **Challenge:** *The public and health care providers are bombarded with information on genomic tests, many with unproven utility*
- **Opportunity:** Educate providers and the public about potential benefits and harms of genomic tests and the need for evidence
- **Challenge:** *There is no widely agreed upon threshold level of evidence for determining whether genomic tests are ready for use*
- **Opportunity:** Develop flexible method(s) for classification of tests by level of evidence to aid in research/evaluation and help define which aspects of evidence should be considered in developing thresholds

# Tier Table Database

Last data update: Sep 09, 2016. (Total: 159 Documents)

Note: Simple Boolean operators are allowed, such as AND or OR

Search Tier Table Database:

159 records

Filtered by:

Tier



Use Filter to fine-tune your search

Records 1-100

Tier

Disease

Gene

Basis

Tier

Number of Records

Tier 1

46

Tier 2

105

Tier 3

9

# Examples of Tier 1 Genomic Applications

Disease/Disorder	Test to be Assessed	Intended Use	Tier Classified	Detail
31 core conditions	Newborn screening panel	Screening	Tier 1 <a href="#">?</a>	<a href="#">Detail</a>
Osteoporosis	Parental history of hip fracture	Estimate fracture risk to inform osteoporosis screening	Tier 1 <a href="#">?</a>	<a href="#">Detail</a>
Familial hypercholesterolemia (FH)	DNA testing and LDL-C concentration measurement	Cascade testing of relatives of people diagnosed with FH	Tier 1 <a href="#">?</a>	<a href="#">Detail</a>
BRCA-related cancer; hereditary breast and ovarian cancer	Family history	Risk prediction for referral for BRCA genetic counseling	Tier 1 <a href="#">?</a>	<a href="#">Detail</a>
Lynch syndrome	Various strategies	Screening, cascade testing of relatives	Tier 1 <a href="#">?</a>	<a href="#">Detail</a>

# Tier 1 Recommendation: *BRCA* Testing

## Genomic Application General Information

<b>Tier Classification:</b>	Tier 1
<b>Disease/Disorder:</b>	BRCA-related cancer; hereditary breast and ovarian cancer
<b>Test to be assessed:</b>	Family history
<b>Target Population:</b>	Women
<b>Intended Use:</b>	Risk prediction for referral for BRCA genetic counseling
<b>Application Type</b>	Family history
<b>Basis:</b>	Clinical Practice Guideline
<b>Entered Date:</b>	05/08/2015
<b>Last Updated Date:</b>	05/14/2015

## Relevant evidence and sources

### Primary Basis for Tier Classification

- USPSTF (2013) [\[link\]](#)

### Additional Synthesized Evidence Sources

• NCCN Clinical Practice Guidelines (2013) [\[link\]](#)

# Tier 1 Recommendation: Lynch Syndrome Testing

## Genomic Application General Information

<b>Tier Classification:</b>	Tier 1
<b>Disease/Disorder:</b>	Lynch syndrome
<b>Test to be assessed:</b>	Various strategies
<b>Target Population:</b>	Patients with newly diagnosed colorectal cancer
<b>Intended Use:</b>	Screening, cascade testing of relatives
<b>Application Type</b>	Screening
<b>Basis:</b>	Clinical Practice Guideline
<b>Entered Date:</b>	08/07/2015
<b>Last Updated Date:</b>	08/07/2015

## Relevant evidence and sources

### Primary Basis for Tier Classification

- EGAPP (2009)

# Tier 1 Recommendation: Familial Hypercholesterolemia

## Genomic Application General Information

<b>Tier Classification:</b>	Tier 1
<b>Disease/Disorder:</b>	Familial hypercholesterolemia (FH)
<b>Test to be assessed:</b>	DNA testing and LDL-C concentration measurement
<b>Target Population:</b>	
<b>Intended Use:</b>	Cascade testing of relatives of people diagnosed with FH
<b>Application Type</b>	Other
<b>Basis:</b>	Clinical Practice Guideline Systematic Review
<b>Entered Date:</b>	08/19/2015
<b>Last Updated Date:</b>	08/19/2015

## Relevant evidence and sources

Primary Basis for Tier Classification

- [NICE guidelines \[CG71\] \(2008\)](#)

# Example of a Tier 3 Genomic Application

## Genomics Application General Information

Tier Classification	Tier 3
Disease/Disorder:	Hereditary breast/ovarian cancer
Test to be assessed:	Routine BRCA genetic counseling, routine BRCA testing
Target Population:	Women whose family health history is not associated with an increased risk of BRCA mutations
Intended Use:	Population screening
Application Type	Screening / family history
Basis:	Clinical Practice Guidelines
Entered Date:	06/10/2015
Last Updated Date:	06/10/2015

## Relevant evidence and sources

- Primary Basis for Tier Classification
- USPSTF recommendation statement <#>

## Links to other sources

[Mammary Neoplasms \[Disease\]](#) 

[BRCA1 \[Gene\]](#) 

[BRCA2 \[Gene\]](#) 

## WHAT'S NEW

Weekly summary of genomics and health impact information

## GENOMICS AND DISEASES

Genomics is important for many diseases of public health significance

## FAMILY HEALTH HISTORY

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

## PATHOGEN GENOMICS

New tools are changing the landscape in the fight against infectious diseases

## GENOMIC TESTING

Genomic tests are used in many diseases

## EPIDEMIOLOGY

Epidemiology is a scientific foundation for public health genomics

## Genomics and Diseases



Genomics and family health history play a role in many diseases such as cancer and heart disease. These diseases are partly the result of how your genes interact with your behaviors, such as your diet and physical activity, and your environment. If you have a family health history of a disease, you are more likely to get that disease yourself, but you can take steps to prevent disease or find it early.



BREAST & OVARIAN CANCER



COLORECTAL CANCER



HEART DISEASE



DIABETES



NEWBORN SCREENING



OSTEOPOROSIS

A medical condition that causes bones to become weak and more likely to break

Podcasts on genetic testing, diseases, and family health history

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## Hereditary Breast and Ovarian Cancer



[Hereditary Breast and Ovarian Cancer](#)



If you are a woman with a family health history of breast or ovarian cancer, you may be more likely to get these cancers yourself. Collecting your family health history and sharing this information with your doctor can help you find out if you're at higher risk. If so, you can take steps to lower your risk.



THE BASICS



GENETIC TESTING



BRCA1 AND BRCA2



MEDICAL OPTIONS



FAMILY HEALTH HISTORY



TALKING TO FAMILY



GENETIC COUNSELING



FAMILY STORIES



[The Bring Your Brave](#)

[campaign](#) provides information about breast cancer to women younger than age 45 by sharing real stories about young women whose lives have been affected by breast cancer.



[The KnowBRCA tool](#)

helps you assess your risk of having a BRCA gene mutation.

Podcasts on genetic testing, diseases, and family health history

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## Family Health History



### Family Health History



Knowing and acting on your family health history is an important way to protect your health. Collect your family health history and share it with your doctor at your next visit.

Your doctor can use it to develop a more complete picture of your health and your risk factors for disease. Together you can work on ways to reduce that risk.



THE BASICS



FAMILY HEALTH HISTORY & CHRONIC DISEASES



PLANNING FOR PREGNANCY



INFORMATION FOR HEALTH PROFESSIONALS



DURING PREGNANCY



INFORMATION FOR RESEARCHERS



FOR CHILDREN



TOOLS AND RESOURCES



FOR ADULTS



Send a family health history eCard.

## WHAT'S NEW

Weekly summary of genomics and health impact information

## PHGKB

Online searchable knowledge base on genomics and health impact information

## GENOMICS AND DISEASES

Genomics is important for many diseases of public health significance

## AMD CLIPS

Weekly news and publications on pathogen genomics and bioinformatics

## FAMILY HEALTH HISTORY

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

## IMPLEMENTATION

What public health can do now to save lives using genomics

## PATHOGEN GENOMICS

New tools are changing the landscape in the fight against infectious diseases

## BLOG

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

## GENOMIC TESTING

Genomic tests are used in many diseases

## REPORTS AND PUBLICATIONS

CDC reports and publications in genomics

## EPIDEMIOLOGY

Epidemiology is a scientific foundation for public health genomics

## PODCASTS AND VIDEOCASTS

Podcasts on genetic testing, diseases, and family health history



## Dr Khoury's Tweets

@DrKhouryCDC

Using pathogen #genomics to track polio.  
<https://t.co/T15hVqyhQW> #WorldPolioDay  
<https://t.co/C5gRQqdHwH>

What does polio eradication have to do with genetic diseases? CDC blog post  
<https://t.co/RX0bX1f0IB> #WorldPolioDay  
<https://t.co/xV5plBea1s>

From genetic counseling of individuals to cascade screening in populations: We can do this! <https://t.co/GneKEX9LEQ>  
<https://t.co/eUI9HaF5ol>

What is Hypertrophic Cardiomyopathy & what's the role of genetics & genetic testing? #PHGKB <https://t.co/4wLq8Z1t5s>  
<https://t.co/tbKxPvkG3R>

Is there a role for genomics in investigating sudden death? Get the latest info. #PHGKB  
<https://t.co/tD1W0q6UJ1>  
<https://t.co/gCGMZOrv2X>

## Genomics Implementation



[Fact Sheet: Implementing Evidence-based Genomic Tests and Family Health History](#) [PDF 1.04 MB]

### What public health can do now in human genomics to save lives and improve health

This page summarizes current information on genomic applications that are ready to be integrated into public health practice to save lives, improve health and quality of life. The focus at this time is on three Tier I applications: Hereditary Breast/Ovarian Cancer Syndrome, Lynch Syndrome and Familial Hypercholesterolemia, but information about other Tier I applications will be added in the near future. The page includes information about completed and ongoing projects spearheaded by state health departments and their partners, and recommendations from evidence based groups which can be applied at the state or local level. Please contact us at [genetics@cdc.gov](mailto:genetics@cdc.gov) if you are aware of a statewide or other project not currently included which should be highlighted on this page.

#### Resources

- [Classification of Genomic Applications by Levels of Evidence](#)
- [State Implementation Activities Clickable Map](#)
- [Public Health Genomic Tier 1 Tool Kit](#)
- [Healthy People 2020 Genomics Objectives](#)
- [Search the Public Health Genomics Knowledge Base for Implementation Resources and Information](#)

Tier 1

State & Local Health  
Departments

How to use the Toolkit

Updates

Lynch Syndrome +

Hereditary Breast &  
Ovarian Cancer +

Familial  
Hypercholesterolemia +

Videos

## Genomics Implementation



### Tier 1 Genomic Applications Toolkit for Public Health Departments

#### ▼ Tier 1 Genomic Applications and their Importance to Public Health

- Tier 1 Genomic Applications and their Importance to Public Health



# CDC's Tier 1 Genomic Applications Toolkit for Public Health Departments

- Goal: Assist state and local public health departments in implementing Tier 1 recommendations using strategies from model state programs
- Hereditary Breast and Ovarian Cancer
- Lynch Syndrome/HNPCC
- Familial Hypercholesterolemia

# CDC's Tier 1 Genomic Applications Toolkit for Public Health Departments

- Approaches
  - Implement bidirectional cancer registry reporting
  - Inform policy making
  - Develop surveillance indicators
  - Track Healthy People 2020 genomics objective
  - Provide education and outreach
  - Promote cascade screening

# Bidirectional Cancer Registry Reporting Tools

- Customizable written materials
- Information for providers
- Information for patients and families
- Reporting tools
- Useful for cascade screening and other applications

Tier 1

State & Local Health  
Departments

How to use the Toolkit

Updates

Lynch Syndrome -

Phase 1

Phase 2

Tools -

Educational Tools

Publications

Hereditary Breast &  
Ovarian Cancer +

Familial  
Hypercholesterolemia +

## Genomics Implementation



### Tier 1 Genomic Applications Toolkit for Public Health Departments

#### ▼ Tier 1 Genomic Applications and their Importance to Public Health

- Tier 1 Genomic Applications and their Importance to Public Health



## Genomic Application Toolkit

[Genomic Application Toolkit](#) > [Lynch Syndrome](#) > [Tools](#)

### Tier 1

State & Local Health  
Departments

How to use the Toolkit

Updates

Lynch Syndrome

Phase 1

Phase 2

### Tools

Educational Tools

Publications

Hereditary Breast & Ovarian  
Cancer

Familial  
Hypercholesterolemia

Videos

## Lynch Syndrome tools



### Tools for Bidirectional Cancer Registry Reporting to Identify Individuals at Risk for Lynch Syndrome

The following materials were developed to support state programs using bidirectional cancer registry reporting to identify individuals at risk for Lynch syndrome. State health departments are encouraged to customize the materials to meet their needs. Materials are categorized by those intended for patients and for healthcare providers, but materials may be suitable for multiple audiences. Please note that some materials will need to be filled out with state-specific information, as noted below.

#### Information for Patients

- [Lynch Syndrome: A Guide for Patients and Their Families](#)
- [Brochure on Talking to Your Family About Your Diagnosis of Lynch Syndrome](#)
- [Sample Letter for Informing Your Family Members about Your Lynch Syndrome Mutation](#)
- [List of Cancer Genetic Specialists for Your State or Region](#) (Please note that state programs will need to complete this form.)
- [What You Need to Know About Cancer Registries: Frequently Asked Questions for Patients and Their Families](#)

#### Information for Providers

- [Lynch Syndrome: Fact Sheet for Healthcare Professionals](#)
- [Evidence-based Practice Guidelines Supporting Genetic Susceptibility Testing for Lynch Syndrome](#)
- [Bidirectional cancer registry reporting to identify patients at high risk for hereditary cancer syndromes: what providers and institutions need to know](#) . Video for educational outreach to providers and institutions in states that have bidirectional cancer registry reporting programs in place.

#### Reporting Tools

- [Sample Hospital and Medical Center Cancer Genetics Data Report on Potential Lynch Syndrome-Related Cancers](#) (Please note that state programs will need to complete this form.)

# Information for patients and families

## Lynch Syndrome: A Guide for Patients and Their Families

Lynch syndrome (LS) is a genetic condition that increases a person's chance of getting colorectal cancer (cancer of the large bowel or rectum), endometrial cancer (cancer of the lining of the uterus), ovarian cancer, and other cancers. LS runs in families, and a genetic test can help determine if your personal and/or family health history of cancer was caused by LS. If you are found to have LS, there are interventions that can help prevent cancer or detect it early. If you are concerned about your personal or family history of cancer, talk to your doctor.

### CAUSES OF LYNCH SYNDROME

LS is caused by mutations (genetic changes) in one of 5 genes: *MLH1*, *MSH2*, *MSH6*, *PMS2*, and *EPCAM*. About 3 out of every 100 colorectal cancers are caused by LS.

### WHY IT IS IMPORTANT TO KNOW ABOUT LYNCH SYNDROME

If you have LS, you are much more likely to get certain cancers, including:

- Up to an 82% risk (about 8 in 10) for colorectal cancer by age 70
- Up to a 60% lifetime risk (6 in 10) for endometrial cancer in women
- Increased risks for cancers of the stomach, ovary, small bowel, hepatobiliary tract (liver, pancreas, gallbladder), urinary tract (kidney, bladder, ureters), brain, and skin

If you are found to have LS, steps can be taken to reduce your cancer risks associated with LS, including:

- Having earlier, more frequent, and/or additional screening for cancer
- Undergoing preventive surgery

### GENETIC COUNSELING AND TESTING FOR LYNCH SYNDROME

An expert panel\* recommends that every person with a new diagnosis of colorectal cancer be offered genetic screening for LS. This screen is done on a sample of the colorectal cancer tissue after surgery. If the screen shows that you might have LS, additional genetic counseling and testing will often be needed to find out if you definitely have LS. Genetic counseling and testing for LS is often, but not always, covered by insurance.

In addition, genetic counseling and testing for LS may be appropriate if you meet any of the following criteria:

- You were diagnosed with colorectal cancer in the past
- You have been diagnosed with endometrial cancer (especially before age 50)
- You have several family members with colorectal or other cancers associated with LS
- You have a family member with a known Lynch syndrome mutation



### FOR MORE INFORMATION

First, talk with your doctor or other health care provider. You can also find more information on LS at:

Centers for Disease Control and Prevention. Genetic Testing for Lynch Syndrome  
<http://1.usa.gov/sJEgXS>  
<http://www.cdc.gov/Features/LynchSyndrome/>

Genetics Home Reference. National Library of Medicine  
<http://tiny.cc/1lqsww>

National Society of Genetic Counselors  
Find a Genetic Counselor Directory  
<http://nsgc.org/p/cm/ld/fid=164>

[ADD STATE SPECIFIC RESOURCES & SUPPORT GROUPS]

## Hereditary Breast and Ovarian Cancer Syndrome: A Guide for Patients and Their Families

Hereditary Breast and Ovarian Cancer syndrome (HBOC) is a genetic condition that makes it more likely that a person will get breast, ovarian, and other cancers. HBOC is hereditary, meaning that it is caused by a mutation (genetic change) that can be passed down in families. A genetic test can help determine if your personal or family history of cancer was caused by HBOC. If you are found to have HBOC, there are interventions that can help prevent cancer or detect it early. To learn whether you are more likely to have HBOC, collect your personal and family history of cancer and share this information with your healthcare provider. You can use the *KnowBRCA* tool to collect this information and assess your risk of having a *BRCA* mutation in preparation for your visit to your doctor.

### CAUSES OF HBOC

HBOC is most often caused by mutations (genetic changes) in the *breast cancer 1 (BRCA1)* and *breast cancer 2 (BRCA2)* genes. Less common mutations in other genes have also been associated with HBOC. However, most breast and ovarian cancers are not related to HBOC. In fact, only about 3 of every 100 breast cancers and 10 of every 100 ovarian cancers are caused by *BRCA1* and *BRCA2* mutations.

### WHY IT IS IMPORTANT TO KNOW ABOUT HBOC

If you have a *BRCA* mutation, you are much more likely to get certain cancers:

- Up to a 65% risk (about 6 out of 10) for breast cancer by age 70
- Up to a 39% risk (about 4 out of 10) for ovarian cancer by age 70
- Increased risks for other cancers including prostate, pancreatic, and male breast cancers

If you are found to have HBOC, steps can be taken to reduce your cancer risks, including:

- Having earlier, more frequent, and/or additional cancer screenings
- Taking medications that can decrease the risk of cancer
- Undergoing preventive surgery to remove your breasts, ovaries, and fallopian tubes

### SIGNS THAT HBOC MAY RUN IN YOUR FAMILY

HBOC can be passed down from either side of your family. An expert panel<sup>1</sup> recommends that doctors screen women who have family members with breast, ovarian, tubal, or peritoneal cancer with one of several screening tools designed to identify families that are more likely to have a *BRCA* mutation. You should let your doctor know if you have a personal or family history of any of the following:

- Breast cancer at age 45 or younger in women
- Breast cancer at age 46–50 in women and at least one close blood relative with breast cancer at any age or limited family history
- Triple negative\*\* breast cancer at age 60 or younger in women
- Breast cancer at any age in men
- Ovarian, fallopian tube, or primary peritoneal cancer
- Cancer in both breasts
- Pancreatic cancer or prostate cancer with Gleason score  $\geq 7$  (Gleason score is a measure of the grade of the cancer)
- Breast, ovarian, pancreatic, or prostate cancer among multiple blood relatives
- Ashkenazi (Eastern European) Jewish ancestry
- A known *BRCA* mutation in the family

### GENETIC COUNSELING AND TESTING FOR HBOC

If you are concerned about your personal or family history of cancer, talk to your doctor. Your doctor may refer you to a genetic counselor or other healthcare professional to discuss the benefits and risks of genetic testing for HBOC. *BRCA* genetic counseling and testing is often, but not always, covered without cost sharing by many health plans under the Affordable Care

### FOR MORE INFORMATION

First, talk with your health care provider. You can also find more information on HBOC at:

Centers for Disease Control and Prevention:

[http://www.cdc.gov/cancer/breast/young\\_women/bringyourbravehereditary\\_breast\\_cancer/index.htm](http://www.cdc.gov/cancer/breast/young_women/bringyourbravehereditary_breast_cancer/index.htm)

Know-*BRCA*

[http://www.cdc.gov/cancer/breast/young\\_women/knownbrca.htm](http://www.cdc.gov/cancer/breast/young_women/knownbrca.htm)

National Society of Genetic Counselors: Find a Genetic Counselor Directory  
<http://nsgc.org/p/cm/ld/fid=164>

[ADD STATE RESOURCES]



# Information for patients and families

## Cancer Genetic Specialists for [Your State or Region]

Institution name Address Phone: Fax: Website: Counselor(s):	

YOUR LOGO HERE

## What You Need to Know About Cancer Registries: Frequently Asked Questions for Patients and Their Families

### What is a cancer registry?

A cancer registry is an information system established to collect, store, and use information about patients with a cancer diagnosis to help improve cancer treatment and prevention. This information is stored in secure electronic databases and used by the state and healthcare professionals for public health purposes.

### How is my information given to the registry? Does the state need my consent to put my information in the registry?

Your doctor and hospital are required by law to report information about your cancer, treatment, and limited background information (like your age and race) to the cancer registry. The state does not require your consent for this process because cancer is legally defined as a "reportable" condition, which means that collecting this information serves a significant and useful purpose for patients, the community, and overall public health.

### What kinds of information do the registries collect?

Specific information that is collected varies across states, but most of the information will be related to your cancer diagnosis and treatment, as well as some background information. Basic examples include:

- Cancer type and stage
- Treatments (such as what type of surgery, chemotherapy, and radiation you had, if any)
- Age, race, and sex
- Height and weight
- Address

### What are cancer registries used for?

Cancer registries are used to collect and analyze data on cancers in the region or state. State and healthcare professionals look for patterns and trends that address questions such as: *Are more people getting cancer than in the past? Are certain locations having more or less cases of cancer? Do certain cancers occur more in one group of people than others? Are cancer treatments improving? Are certain treatments not working?* The results are used to help improve the health of the community and patients overall.

### What about privacy? Who can see my data?

Your information is kept secure. Only people who prove to the state that they will keep your information confidential and use it only for important public health work will be given access to your data. Who can access this information and steps required to access it vary by [state](#). If you have questions or want more information please contact your state's cancer registry.

[{Add Contact Information Specific to Your State Here}](#)

### Where can I find more information?

Visit [our website](#) for additional information: [http://www.cancerregistry.org](#)

# Information for patients and families

*Instructions: Please find below suggested wording for a letter to your family members. If possible, you might consider talking to your relatives about your BRCA testing first and then sharing the letter with them to help them remember what you discussed. Please read through this letter, fill in missing information, and make any additional changes you feel are needed. If possible, include a copy of your genetic testing results or informational sheet on your particular genetic change (mutation) with the letter. These results will be important for your relatives when they speak with their healthcare providers. This letter only applies to blood relatives and not "in-laws." Blood relatives include your parents, grandparents, children, siblings, aunts, uncles, nieces, nephews, and cousins.*

Dear [xx]

I'm writing to let you know that I have been diagnosed with an inherited condition called Lynch syndrome, or Hereditary Non-Polyposis Colorectal Cancer (HNPCC). People with Lynch syndrome are much more likely to get colorectal cancer and other types of cancer. Lynch syndrome runs in families and is due to errors in certain genes (known as mutations).

Because you are my blood relative, you are more likely to have Lynch syndrome and could benefit from genetic counseling and possibly genetic testing for Lynch syndrome.

If you find out that you have Lynch syndrome, you can take steps to lower your chances of getting cancer and to find cancer earlier if you do get it. These steps include preventive surgery and earlier, more frequent, and additional cancer screening. It is important to note if you have Lynch syndrome, it does not mean that you will definitely get cancer.

People with Lynch syndrome are more likely to get certain types of cancers, including colorectal cancer, endometrial (uterine) cancer, ovarian cancer, biliary tract cancer, sebaceous skin tumors, and urinary tract cancer. The attached document shows the specific mutation I have and this mutation is the one for which my family members should be tested. My parents

[brothers/sisters/children (include any of these that you have)] have a 50% (1 in 2) chance of having Lynch Syndrome. My other blood relatives (aunts, uncles, nieces, nephews, and cousins) might also have Lynch Syndrome. Please note that genetic testing for Lynch syndrome is not recommended for children under 18 years old, but can be considered when they reach adulthood.

The first step is to discuss this with your doctor who can provide you with more information about genetic testing for Lynch syndrome. Your doctor may refer you to a genetic counselor. You can find the genetic counselor closest to you at [www.mssk.org](http://www.mssk.org).

For more information about Lynch syndrome, here are some helpful resources:

- [www.cancer.net/cancer-types/lynch-syndrome](http://www.cancer.net/cancer-types/lynch-syndrome)
- [www.cdc.gov/Features/LynchSyndrome/](http://www.cdc.gov/Features/LynchSyndrome/)
- [www.ctr.ucla.nih.gov/condition/lynch-syndrome](http://www.ctr.ucla.nih.gov/condition/lynch-syndrome)

*Instructions: Please find below suggested wording for a letter to your family members. If possible, you might consider talking to your relatives about your BRCA testing first and then sharing the letter with them to help them remember what you discussed. Please read through this letter and make any changes you feel are needed. If possible, include a copy of your genetic testing results or informational sheet on your particular genetic change (mutation) with the letter. These results will be important for your relatives when they speak with their healthcare providers. This letter only applies to blood relatives and not "in-laws." Blood relatives include your parents, grandparents, children, siblings, aunts, uncles, nieces, nephews, and cousins.*

Dear [xx]

I recently had genetic testing for inherited mutations (changes) in the *breast cancer 1 (BRCA1)* and *breast cancer 2 (BRCA2)* genes. These are the genes most commonly affected in breast and ovarian cancer. My test found a [BRCA1/BRCA2] mutation that causes an increased risk for breast, ovarian and other cancers.

As one of my blood relatives, you could have the same BRCA1 or BRCA2 mutation and could benefit from genetic counseling and possibly genetic testing for this mutation.

If you find out that you have the mutation, you can take steps to reduce your risk of cancer and to find cancer earlier if you do get it. These steps include preventive surgery, medications, and earlier, more frequent, and additional cancer screening. It is important to note that if you have a BRCA1 or BRCA2 mutation, it does not mean that you will definitely get breast or ovarian cancer.

People who inherit a mutation in the BRCA1 or BRCA2 gene are more likely to get breast, ovarian, tubal, peritoneal, prostate, and pancreatic cancer. In general, women with a BRCA mutation are more likely to get breast or ovarian cancer before age 50 than women without a BRCA mutation. Men with BRCA1 or BRCA2 mutations also have a higher chance of getting breast cancer.

The attached document shows the specific mutation that I have. This mutation is the one for which my family members should be tested. People can inherit BRCA1 or BRCA2 mutations from their mother or father. Since I have the mutation, my parents [brothers/sisters/children (include any of these that you have)] have a 50% (1 in 2) chance of having it. My other blood relatives (aunts, uncles, nieces, nephews, and cousins) also have an increased chance of having the mutation. Please note that genetic testing for BRCA1 or BRCA2 mutations is not recommended for children under 18 years old, but can

## Why Talk to My Family?

Your family members can benefit from knowing about your diagnosis of Lynch syndrome. Talk to your family members about Lynch syndrome, so that they will know that:

- Lynch syndrome is passed through families.
- A person with Lynch syndrome is more likely to get colorectal, endometrial (uterine), ovarian, and other cancers.
- Genetic counseling and testing for Lynch syndrome can provide information about their risk.
- If they choose to be tested, they should be tested for the same mutation that you have.
- Steps can be taken to prevent colorectal and other cancers or find them earlier.

*IT'S NOT EASY...*

*...but talking about Lynch syndrome is one of the most important things you can do to protect your family.*



*When you share information with people, sometimes they feel helpless and don't want to address it. I used to have that mentality, too. After my genetic testing, the genetic counselor asked if I wanted to come in to talk about my test results or if I would rather have them over the phone. I wanted it quick like a band-aid. At that moment, my life changed: I was positive for Lynch syndrome. I could not ignore it anymore. Now there's always the question, where do you go from here? Because it's not something you get better from. That is the most difficult part of explaining to people what Lynch syndrome is and what it means for the future.*

Insert Your  
Logo/Organization  
Info Here



## Talking to Your Family About Your Diagnosis of Lynch Syndrome

## How Do I Talk to My Family About My Lynch Syndrome Diagnosis?

*WHO:* Your parents, siblings, and children are the family members who are most likely to have Lynch syndrome. Other blood relatives, such as aunts, uncles, nieces, nephews and cousins, are also more likely to have Lynch syndrome. Your healthcare provider or genetic counselor can help you figure out who in your family might have Lynch syndrome and thus would benefit from knowing about your diagnosis.

*WHAT:* You can share test results, letters from your doctor or genetic counselor, or other information you received about your diagnosis with your family. Giving family members information about your specific genetic mutation helps their healthcare providers know exactly which test to use and might possibly save your family money.

*HOW:* If you need extra support talking to your family, bring a friend. You can also ask a family member to attend your next medical appointment with you. The website <http://kintalk.org> can help you let your relatives know about your diagnosis and provides resources to help them learn more about Lynch syndrome. A sample letter that you can fill out and send to your family is available at [www.cdc.gov/genomics/restoflink](http://www.cdc.gov/genomics/restoflink)



## How Do I Talk to My Children?

If you have Lynch syndrome, each of your children has a 50% (1 in 2) chance of having Lynch syndrome. Genetic testing for Lynch syndrome is typically not recommended for children younger than 18, but can be considered when your children reach adulthood.

Younger children might not be able to understand what your diagnosis means for you or for them. Children differ in the age at which they are ready to learn about this information. Answer the questions they ask. They will ask more complex questions as they grow and are ready to learn more.

Know that your children may have fears about the risk both to themselves and to you. Just as you need time and support to cope with the information and accept it, so will your children.

## What if My Family Does Not Want to Talk?

Talking to some family members about Lynch syndrome might not be easy. Some might not understand why they need to know this information. Others might be nervous about receiving a diagnosis of Lynch syndrome. Remember that family members need to make their own choices about getting tested, whether or not you agree with their decisions. If family members don't want to talk about Lynch syndrome, respect their wishes. Let them know you are available to talk if they have questions, and give them places to find information.

When family members do not want to talk about Lynch syndrome, you might feel upset or alone. Seek support from friends, healthcare providers, other family members, or people you know with Lynch syndrome.

## Where Can I Find More Information?

You can find more information on Lynch syndrome at:

[www.ghr.nlm.nih.gov/condition/lynch-syndrome](http://www.ghr.nlm.nih.gov/condition/lynch-syndrome)

You can find information on support groups for Lynch syndrome at: <http://www.diseaseinfosearch.org/Lynch+syndrome/3371>

# Information for providers

## Lynch Syndrome Fact Sheet for Healthcare Professionals



Patients with Lynch syndrome are at increased risk of developing colorectal cancer, endometrial (uterine) cancer, ovarian cancer, and other cancers. Lynch syndrome is caused by mutations in the mismatch repair genes, *MLH1*, *MSH2*, *MSH6*, and *PMS2*, and the *EPCAM* gene. About 3% of patients with colorectal cancer have Lynch syndrome. Identification of patients and their relatives with Lynch syndrome is important to allow them to take advantage of interventions that can significantly reduce their risk of cancer in the future and allow for early detection of cancer if it develops.

### Cancer Risks Associated with Lynch Syndrome:

- 25- 82% risk of colorectal cancer by age 70<sup>1</sup>
- 20-60% risk of endometrial cancer by age 70<sup>2</sup>
- 11-19% risk of stomach cancer by age 70<sup>2</sup>
- 9-12% risk of ovarian cancer by age 70<sup>2</sup>
- 2-7% risk of cancers of the hepatobiliary tract by age 70<sup>2</sup>
- 4-5% risk of cancers of the urinary tract by age 70<sup>2</sup>
- Increased risks for cancers of small bowel, brain, and skin

### Evidence-Based Clinical Recommendation for Identifying Patients at Risk

*The Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Recommendation (2009)*<sup>1</sup>

The EGAPP Working Group found sufficient evidence to recommend offering tumor screening and genetic testing for Lynch syndrome to individuals with newly diagnosed colorectal cancer to reduce morbidity and mortality in relatives.

Screening for Lynch syndrome is done on the colorectal cancer tissue after surgery using immunohistochemistry (IHC) and/or microsatellite instability (MSI) testing. For those patients who screen positive for Lynch syndrome, genetic testing can then be performed to diagnose Lynch syndrome by identifying the patient's specific mutation. Other family members then can be tested for that mutation.

Other recommendations exist to help identify *unaffected* individuals who may be at risk for Lynch syndrome, including those from the National Comprehensive Cancer Network (NCCN)<sup>2</sup> and the American College of Medical Genetics (ACMG)/National Society of Genetic Counselors (NSGC).<sup>3</sup>

Genetic counseling and testing for Lynch syndrome may be appropriate for patients who meet any of the following criteria:

- Diagnosed with colorectal cancer in the past
- Diagnosed with endometrial cancer (especially before age 50)
- Several family members with colorectal, endometrial, or other cancers associated with Lynch syndrome

### Genetic Counseling and Testing for Lynch Syndrome

#### Genetic Counseling

Genetic counseling helps patients and their families better understand their risk for hereditary cancer so that they can make informed decisions about tumor screening, genetic testing, and follow-up care by:

- Reviewing an individual's personal and family medical history
- Reviewing risk for Lynch syndrome and the chance of finding a mutation through tumor screening and genetic testing

## Hereditary Breast and Ovarian Cancer Syndrome Fact Sheet for Healthcare Professionals

Add Your  
Logo Here

Hereditary Breast and Ovarian Cancer (HBOC) syndrome is associated with an increased risk for breast, ovarian, and other cancers. HBOC is usually caused by mutations in the *BRCA1* and *BRCA2* genes. *BRCA* mutations are responsible for approximately 3% of all breast cancers and 10% of all ovarian cancers. Identification of individuals with *BRCA* mutations is important to allow them to take advantage of interventions that can significantly reduce their risk of cancer and allow for early detection of cancer if it develops.

### Cancer Risks Associated with *BRCA1* and *BRCA2* Mutations:

- 45-65% risk of breast cancer by age 70 for women with *BRCA1* or *BRCA2* mutations, compared with a 12-13% risk for women in the general population.
- 10-39% risk of ovarian cancer by age 70 for women with *BRCA1* or *BRCA2* mutations, compared with a 1-2% risk for women in the general population
- Increased risk for tubal, peritoneal, prostate, pancreatic, and male breast cancers
- Increased risk for early onset breast or ovarian cancer (before age 50)

### Individuals Are More Likely to Have a *BRCA1* or *BRCA2* Mutation if They Have a Personal or Family Health History of Any of the Following:

- Breast cancer diagnosed at age 50 or younger in women
- Triple negative breast cancer diagnosed at age 60 or younger in women
- Epithelial ovarian, fallopian tube, or primary peritoneal cancer
- Two diagnoses of breast cancer or two types of *BRCA*-related cancer in the same person
- Breast cancer at any age in men
- Pancreatic cancer or prostate cancer with Gleason score =7
- Breast, ovarian, pancreatic, or prostate cancer among multiple blood relatives
- Ashkenazi (Eastern European) Jewish ancestry
- A known *BRCA1* or *BRCA2* mutation in the family

**Note:** The Centers for Medicare and Medicaid Services (CMS) Local Coverage Determination (LCD) on *BRCA1* and *BRCA2* Genetic Testing allows for regional coverage of *BRCA* genetic counseling and testing for individuals with personal histories of breast, ovarian, and other cancers that fit specific criteria for increased risk for a *BRCA* mutation. If this LCD applies to your state, the list above ("Individuals Are More Likely to Have a *BRCA1* or *BRCA2* Mutation if They Have a Personal or Family Health History of Any of the Following:") can be replaced with the list in the **Appendix**, which contains the specific criteria for referrals from the LCD.

### Evidence-Based Clinical Recommendations for Identifying Patients at Risk Because of Family Health History Who Should Be Referred for Genetic Services

*Recommendation for BRCA Mutation Testing for Breast and Ovarian Cancer Susceptibility from the U.S. Preventive Services Task Force (USPSTF) (2014)*<sup>1</sup>

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

# Information for providers

## Evidence-based Practice Guidelines on Genetic Susceptibility Testing for Lynch Syndrome

### Identifying Colorectal Cancer Patients at Risk for Lynch Syndrome:

#### Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Recommendation (2009)<sup>1</sup>

- The EGAPP Working Group found sufficient evidence to recommend offering screening or genetic testing for Lynch syndrome (LS) to all individuals with newly diagnosed colorectal cancer

### Identifying Patients at Risk for Lynch Syndrome Who Do Not Have Colorectal Cancer

- National Comprehensive Cancer Network (NCCN) (2014) Recommendations<sup>3</sup>
  - Referral for genetic counseling for Lynch syndrome is recommended for:
    - Women with endometrial cancer diagnosed before age 50
    - Individuals in families with known Lynch syndrome
  - Amsterdam Criteria should be used to identify patients at risk due to family health history:
    - At least three relatives diagnosed with a cancer associated with Lynch syndrome (colorectal, endometrial, small bowel, ureter, renal-pelvis) and
      - One must be a first-degree relative of the other two
      - At least 2 successive generations must be affected
      - At least one relative with cancer associated with Lynch syndrome should be diagnosed before age 50
    - Familial Adenomatous Polyposis (FAP) should be excluded in the colorectal cancer cases and reported family health history should be verified when possible
- American College of Medical Genetics (ACMG) and National Society of Genetic Counselors (NSGC) Recommendations (2015)<sup>2</sup>
  - Referral for genetic counseling for Lynch syndrome is recommended if any of the following are present in a personal or family health history:
    - Colorectal or endometrial cancer diagnosed before age 50
    - Colorectal or endometrial cancer diagnosed at 50 or older and a first-degree relative with colorectal or endometrial cancer at any age
    - Synchronous or metachronous colorectal or endometrial cancers in the same person
    - Sebaceous adenoma or carcinoma and one or more additional case of any Lynch syndrome associated cancer\*\*
    - Colorectal cancer showing mismatch repair deficiency on tumor screening<sup>4</sup>
    - 3 or more family members with Lynch syndrome associated cancers\*\*

\*\*Lynch syndrome associated cancers include: colorectal cancer, endometrial (uterine) cancer, urothelial cancers (ureter and renal collecting ducts), gastric cancer, ovarian cancer, small bowel cancer, glioblastoma, sebaceous adenocarcinoma, biliary tract cancer, and pancreatic cancer

## Evidence-based Practice Guidelines Supporting Genetic Susceptibility Testing for Hereditary Breast and Ovarian Cancer Syndrome

### United States Preventive Services Task Force (USPSTF) Recommendations (2015)<sup>1</sup>

The USPSTF provides guidelines for risk assessment, genetic counseling, and genetic testing for *BRCA*-related cancer in women who have not been diagnosed with a *BRCA*-related cancer:

- “Women who have a family history of breast, ovarian, tubal, or peritoneal cancer should be screened with 1 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*).”
- Family history screening tools include:
  - Ontario Family History Assessment Tool, Manchester Scoring System, Referral Screening Tool, Pedigree Assessment Tool and the FHS-7<sup>2</sup>
- “Women with positive screening results should receive genetic counseling and, if indicated after counseling, *BRCA* testing.”
- A family health history of any of the following are associated with an increased risk for *BRCA*-related Hereditary Breast and Ovarian Cancer (HBOC) syndrome:
  - A relative with breast cancer diagnosed before age 50
  - A relative with bilateral breast cancer or two primary types of *BRCA*-related cancer
  - Multiple blood relatives with breast and/or ovarian cancer
  - A male relative with breast cancer
  - Ashkenazi (Eastern European) Jewish ancestry
  - A relative with a known genetic mutation in the *BRCA1* or *BRCA2* gene

### Guidelines for Individuals Who Have Been Diagnosed with a *BRCA*-Related Cancer

- Additional guidelines have been developed for individuals previously diagnosed with cancer by several organizations including the National Comprehensive Cancer Network (NCCN)<sup>3</sup> and the American College of Medical Genetics/National Society of Genetic Counselors (ACMG/NSGC)<sup>4</sup>
- Referral to genetic counseling should be considered for any individual with a personal history of:
  - Breast cancer diagnosed  $\leq$  age 50
  - Triple negative breast cancer diagnosed  $\leq$  age 60
  - Two diagnoses of primary breast cancer
  - Epithelial ovarian cancer
  - Male breast cancer
  - Breast cancer diagnosed at any age, and
    - At least 1 close blood relative with breast cancer diagnosed  $\leq$  age 50
    - At least 2 close blood relatives with breast, pancreatic, and/or prostate cancer (Gleason score  $\geq$  7) at any age on the same side of the family
    - 1 close blood relative with epithelial ovarian cancer or male breast cancer
    - From a population of high risk (e.g., Ashkenazi Jewish ancestry)

# Information for providers

Bi-directional Cancer Registry Reporting



**Gregory Feero, MD, PhD**  
Research Director, Maine  
Dartmouth Family Medicine Residency

BRCA gene-associated mutations can undergo

4:15 / 9:55

# Reporting Tools

## [SAMPLE] Hospital and Medical Center Cancer Genetics Data Report [year(s) of data reported] on Potential Lynch Syndrome-Related Cancers

This report prepared by [ ] provides information on the number of patients at your facility who may be at risk for Lynch syndrome (LS), also called Hereditary Non-Polyposis Colorectal Cancer (HNPCC), based on data that were reported to the central cancer registry from your institution during the time period mm/yyyy to mm/yyyy. **All patients with colorectal cancer should be considered for tumor screening for Lynch syndrome.**<sup>1</sup> Some institutions test newly diagnosed endometrial cancers, especially those in women younger than 50.

How many patients were identified at [reporting institution] and statewide?

Number of cancers diagnosed during [Timeframe] in [Location] patients who could have Lynch syndrome (also called Hereditary Nonpolyposis Colorectal Cancer)		
Cancer Site	Your institution	Entire State
Colon or Rectum (All Ages)	# of cases	# of cases
Endometrium (< Age 50)	# of cases	# of cases

Source: [insert data source]

*Patient names associated with the reported diagnoses can be sent to a designated person in your facility upon request. If requested, the names will be disclosed to your facility using current confidentiality rules.*

**What is Lynch syndrome?** Lynch syndrome is a genetic condition that significantly increases an individual's risk for colorectal, endometrial, and other cancers.

**What are the benefits of identifying individuals with Lynch syndrome?** Identifying patients with Lynch syndrome is important because steps can be taken to detect cancer earlier if it develops and reduce cancer risks in the future for patients and their relatives. Patients with Lynch syndrome have a higher risk for developing future cancers and can benefit from closer monitoring or special medical management. Family members of these patients might have Lynch syndrome and should consider genetic counseling and testing for the same mutation identified in the patient.

**How can cancer registry data help identify individuals with Lynch syndrome?**

Patients with colorectal cancer at any age or with endometrial cancer (especially before age 50) are more likely to have Lynch syndrome and can be readily identified from

## [SAMPLE] Hospital and Medical Center Cancer Genetics Data Report [year(s) of data reported] on Potential Hereditary Breast and Ovarian Cancer Syndrome-Related Cancers

This report prepared by [ ] provides information on the number of patients at your facility and statewide who may be at risk for Hereditary Breast and Ovarian Cancer syndrome (HBOC), based on data that were reported to the central cancer registry from your institution during the time period mm/yyyy to mm/yyyy. **The cases listed below are cancers in patients who might benefit from further evaluation for referral to genetic counseling but are not confirmed to be HBOC-associated.**<sup>1</sup>

How many patients were identified at [reporting institution] and statewide?

Number of cancers diagnosed during [Timeframe] in [Location] patients who could have Hereditary Breast and Ovarian Cancer syndrome (HBOC)		
Cancer Site	Sample (Facility specific) <sup>1</sup>	Entire State
Female Breast (< Age 45)*	# of cases	# of cases
Female Breast (< Age 50)*	# of cases	# of cases
Triple Negative Female Breast (< Age 60)	# of cases	# of cases
Ovary, Fallopian Tube, or Peritoneum (All Ages)	# of cases	# of cases
Male Breast (All Ages)	# of cases	# of cases
Multiple Primary Breast Tumors	# of cases	# of cases

Source: [insert data source]

*<sup>1</sup>Patient names associated with the reported diagnoses can be sent to a designated person in your facility upon request. If requested, the names will be disclosed to your facility using current confidentiality rules.*

*\*Breast cancer at age 45 or younger is sufficient for referral to genetic counseling based on some recommendations, unlike breast cancer at age 50 or younger which requires that certain family health history criteria be met. Thus, female breast cancer at age 45 or younger can be reported separately from female breast cancer at age 50 or younger.*

# Reporting Tools

- Reporting tools
  - Sample data report
  - PowerPoint slide set for outreach to providers and institutions (in preparation)
    - In some states, genetic counselors have presented at Grand Rounds to educate providers about hereditary cancers

# Informing evidence-based policy making: approaches

- Educate payers about USPSTF, NCCN, and EGAPP recommendations
- Assess which health plans have policies consistent with recommendations
- Acknowledge health plans with policies that are consistent with current recommendations

# Informing evidence-based policy making: approaches

- Meet with health insurance plan medical directors
- Conduct key informant interviews with health plan administrators to address barriers and facilitators to having evidence-based coverage policies
- Developing and disseminating policy guidance documents for insurers

# Informing evidence-based policy making: approaches

- Incorporate genomics activities into the state cancer control plan, state cardiovascular disease prevention plan, and stroke prevention plan
- Create a list of alternative payment modalities for un/underinsured women
- Participate in Lynch syndrome screening network (LSSN)

# Surveillance indicators and Healthy People 2020 objective tracking

- Behavioral Risk Factor Surveillance System (BRFSS)
- Claims data
- Cancer registry data
- Genetics services data
- Surveys of healthcare providers
- State specific data systems

# Education and outreach

## Examples from states

- Board-certified genetic counselors provided in-service trainings to providers
- Clinical decision support tool containing referral criteria for HBOC
- CME materials
- Partnerships between states to create and disseminate materials

# Education and outreach

## Examples from states

- Survey providers to identify knowledge gaps
- Develop community-specific outreach to target at-risk populations
- Awareness Days
- Communicate via in-person conferences, displays at health fairs, online modules and webinars, provider newsletters, and information at clinician's offices

## Cascade screening

- Active process to find relatives of index patient at a pre-symptomatic stage
- Inform relatives about available testing and interventions

# Implementation Database

## Why did we build it?

- **Challenge:** *State, local, and territorial health departments need practical information that they can use to integrate genomics and family health history into their activities*
- **Opportunity:** Provide a searchable database of available resources categorized by resource type, disease, and state so that health departments can find new resources and learn from other states
- **Challenge:** *State, local, and territorial public health departments and policymakers want to know about genomic and family health history activities in their state and communities*
- **Opportunity:** Activities can be searched by state and can also be identified through the clickable map

# Implementation Database

Search Implementation Database:

Records 1-29 (of 29 Records)

Filtered by:   [Use Filter to fine-tune your search](#)



**Query Trace:** diabetes[original query]

[IGNITE: Implementing Genomics in Practice](#)

[**Disease:** Multiple Diseases; **Type:** Education|Tools; **State:** Multiple States]

[WISEWOMAN](#)

[**Disease:** Stroke|Familial Hypercholesterolemia|Heart Disease; **Type:** Program; **State:** Multiple States]

[Statewide Screening of Fifth Graders Leads to Identification and Treatment of Those With Genetic Predisposition to Early-Onset Heart Disease](#)

[**Disease:** Heart Disease|Familial Hypercholesterolemia|Stroke; **Type:** Data|Program; **State:** West Virginia]

[The Ohio Plan to Prevent Heart Disease and Stroke 2008-2012](#)

[**Disease:** Heart Disease|Stroke|Familial Hypercholesterolemia; **Type:** Data|Policy|Program; **State:** Ohio]

[Ohio Department of Health Heart Disease and Stroke Prevention Program Take Heart: Know Your Heart Disease and Stroke Family Health History](#)

[**Disease:** Stroke|Heart Disease|Familial Hypercholesterolemia; **Type:** Education|Tools; **State:** Ohio]

# Implementation Database

*Filtered by Location, Resource Type, or Disease*

State	Number
<input type="checkbox"/> Alabama	42
<input type="checkbox"/> Alaska	40
<input type="checkbox"/> Arizona	40
<input type="checkbox"/> Arkansas	41
<input type="checkbox"/> California	47
<input type="checkbox"/> Colorado	45
<input type="checkbox"/> Connecticut	49
<input type="checkbox"/> Delaware	41
<input type="checkbox"/> District of Columbia	39

Resource Type	Number
<input type="checkbox"/> Data	49
<input type="checkbox"/> Policy	16
<input type="checkbox"/> Education	50
<input type="checkbox"/> Tools	29
<input type="checkbox"/> Program	147
<input type="checkbox"/> General Info	23
<input type="checkbox"/> Others	1

Disease	Number
<input type="checkbox"/> Birth defects	16
<input type="checkbox"/> Breast Cancer	117
<input type="checkbox"/> Cancer	30
<input type="checkbox"/> Diabetes	12
<input type="checkbox"/> Education, Medical	2
<input type="checkbox"/> Familial Hypercholesterolemia	51
<input type="checkbox"/> Family Health History	1
<input type="checkbox"/> Genetic Counseling	4
<input type="checkbox"/> Heart Disease	49
<input type="checkbox"/> Lynch syndrome	78

# Implementation Database

## Public Health Genomics Knowledge Base (v1.0)

### PHGKB

About

Genomics & Health Impact  
Weekly Scan  
(Current Edition)

Advanced Molecular  
Detection Weekly Clips  
(Current Edition)

Genomics (A-Z)

Office of Public Health  
Genomics

**State Implementation Map**

Relevant Resources

All Databases

Contact Us

Search Knowledge Base

Enter a search term

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### State Genomics Implementation Map

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Click on a state to find information on public health genomics activities in that state. States have implemented genomics applications for Hereditary Breast and Ovarian Cancer syndrome, Lynch Syndrome, Familial Hypercholesterolemia, newborn screening, and more. Find relevant information from your own state or learn about what's been accomplished in other states. You can filter results by condition and resource type (data, programs, education, policy, tools, and general information). Please let us know about new efforts so that we can keep the information current!

#### State Implementation Map



Select a State



<https://phgkb.cdc.gov>

[genetics@cdc.gov](mailto:genetics@cdc.gov)

For more information, contact CDC  
1-800-CDC-INFO (232-4636)  
TTY: 1-888-232-6348 [www.cdc.gov](http://www.cdc.gov)

The findings and conclusions in this report are those of the authors and do not necessarily represent the official position of the Centers for Disease Control and Prevention.

Center for Surveillance, Epidemiology, and Laboratory Services  
Division of Public Health Information Dissemination

