



# Resources in Genomics and Precision Health to Enhance Public Health Impact of New Technologies

**Dave Dotson, Ridgely Fisk Green, Marta Gwinn, Muin J. Khoury**  
**Office of Genomics and Precision Public Health**

March 25, 2021

# About the CDC Office of Genomics and Precision Public Health

## MISSION

Facilitate, evaluate, and promote CDC efforts in integrating genomics and precision health technologies into public health research and actions that prevent disease and reduce health disparities.

## VISION

Healthier people as a result of appropriate use of genomics and precision health technologies.

**1. Identify**  
evidence-based  
applications

**2. Inform**  
and communicate

**3. Integrate**  
into practice & programs



Data science:  
New Blog Post



Pathogen Genomics:  
AMD Program



Mendelian  
Randomization:  
What's New



Diabetes and Ethnicity:  
CDC Information

Hot Topics of the Day

Family Health History

Weekly Update

Genetic Counseling & Testing



# Genomics and Precision Health: What are we dealing with?

- Genomics: The applications of genome-based technologies, including human and pathogens, to health care and disease prevention
- Precision Health: The applications of big data, data science, machine learning and artificial intelligence to health care and disease prevention
- Precision medicine and precision public health as two peas in a pod!

# Organizing Information on Genomics and Precision Health: “4 Phases of Translation”

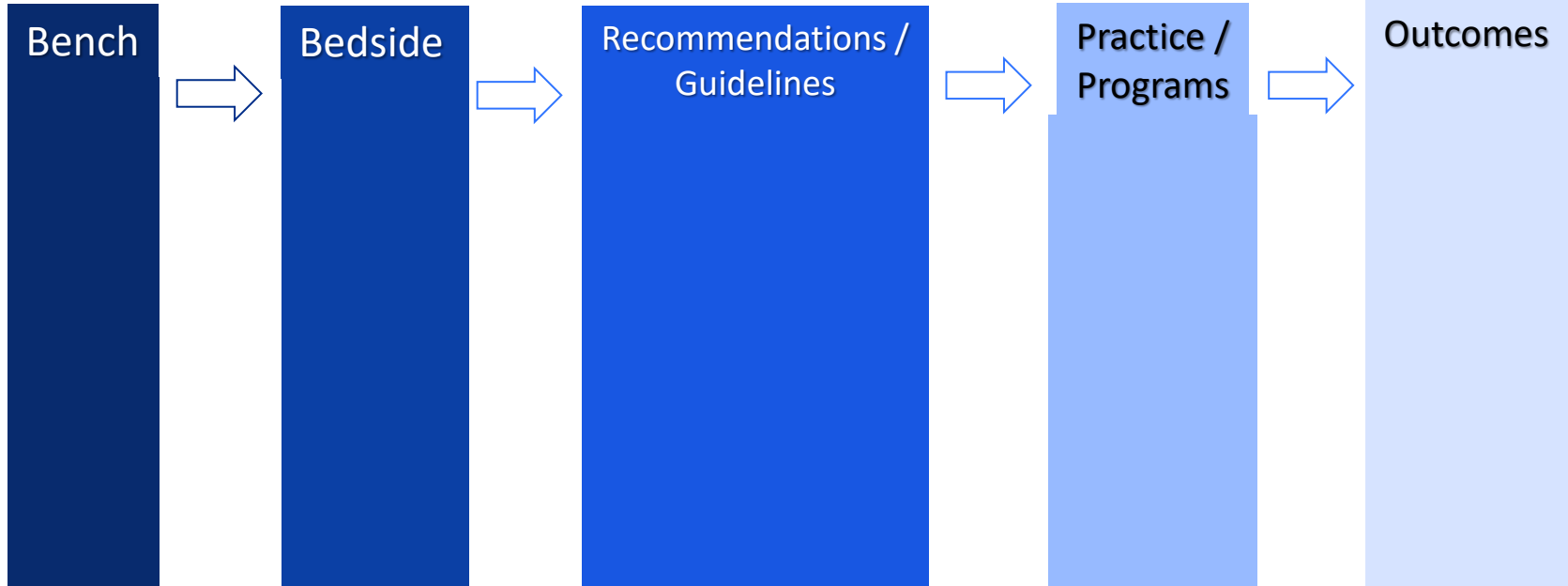
**T0**

**T1**

**T2**

**T3**

**T4**



# Organizing Information on Genomics and Precision Health: “4 Phases of Translation”

**T0**

**T1**



Research and development

*Drugs*

*Devices (Tests)*

# Organizing Information on Genomics and Precision Health: “4 Phases of Translation”

T0

T1

T2



Testing, observation, and evaluation

*Clinical trials*

*Epidemiologic studies*

*Evidence review*



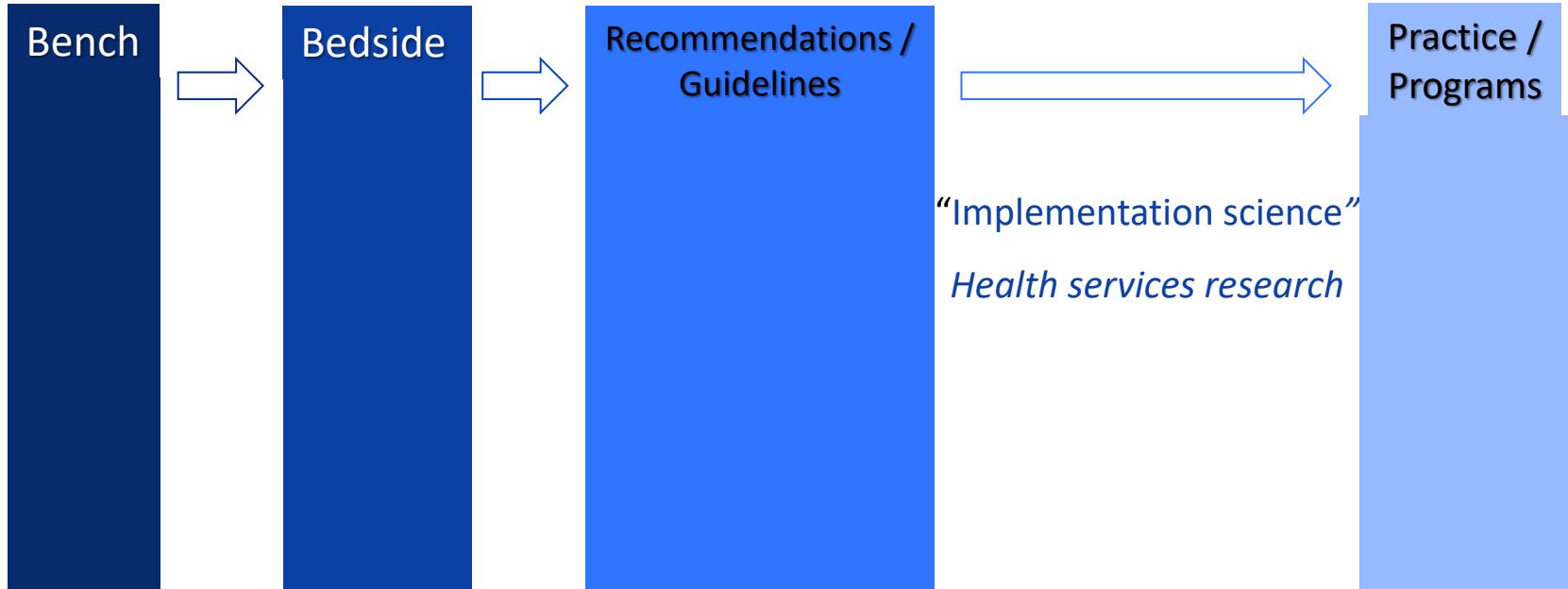
# Organizing Information on Genomics and Precision Health: “4 Phases of Translation”

**T0**

**T1**

**T2**

**T3**



# Organizing Information on Genomics and Precision Health: “4 Phases of Translation”

**T0**



**T1**



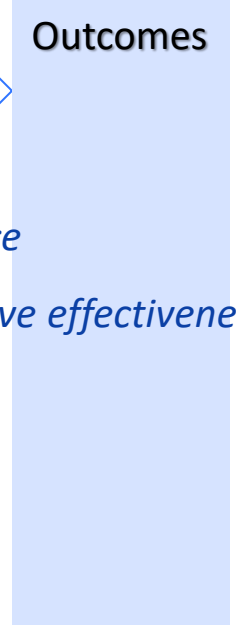
**T2**



**T3**



**T4**





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

*T0*

**T1**

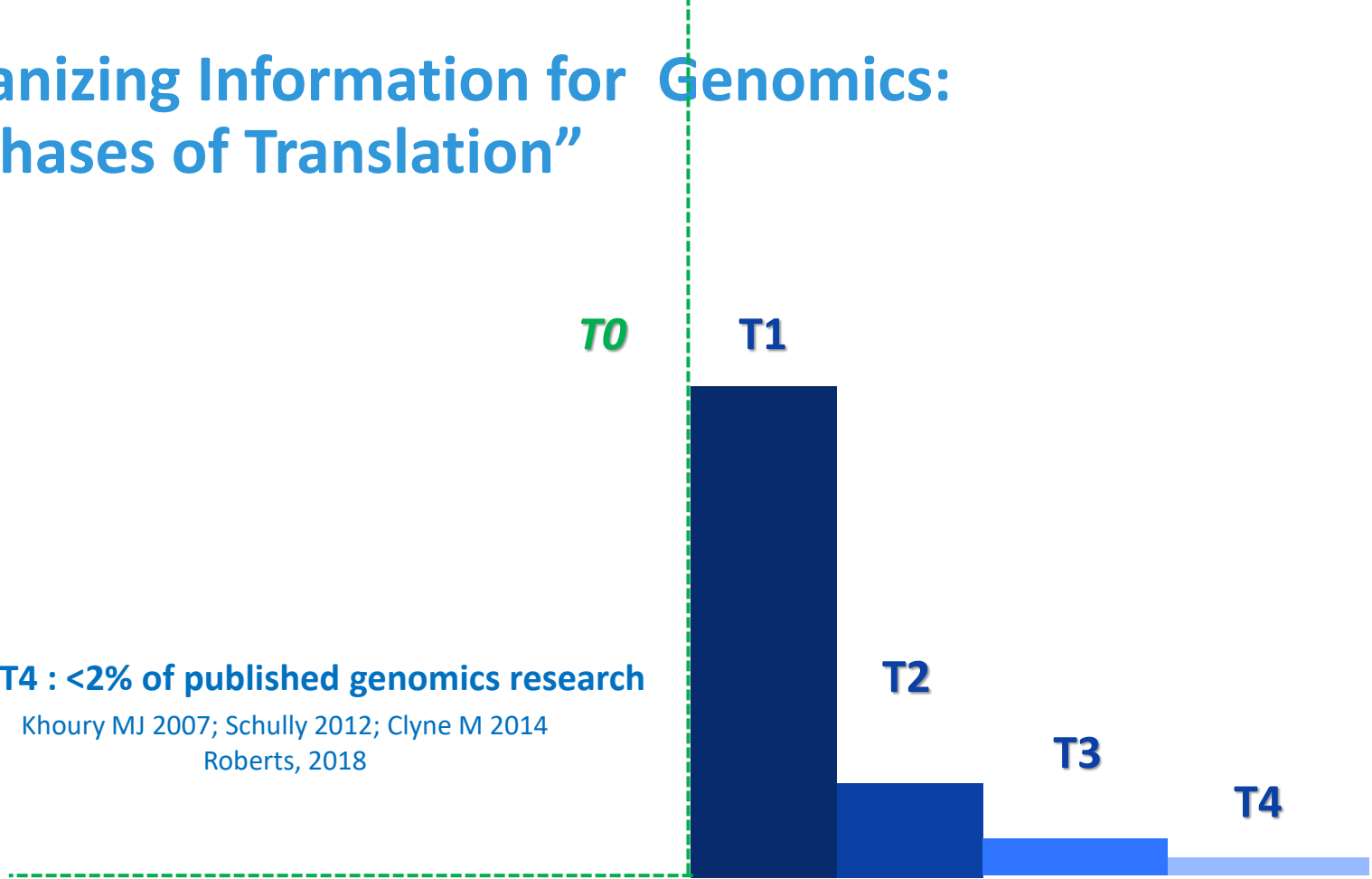
**T2**

**T3**

**T4**

**T2 – T4 : <2% of published genomics research**

Khoury MJ 2007; Schully 2012; Clyne M 2014  
Roberts, 2018



## Genomics & Precision Health



CDC March 25 Webinar:  
Resources in Genomics  
and Precision Public  
Health

Hot Topics of the Day

Weekly Update

PHGKB Database

Reports and Publications

Genomics & Precision Health Blog

Family Health History

Genetic Counseling & Testing

Genomics and Precision Health Topics

Events and Multimedia

About Us



**My Family Health Portrait**

- Record your family health history
- Learn about your risk for conditions
- Print & save your family health history

[VISIT My Family Health Portrait](#)



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and Precision Health  
Tweets

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PHGKB

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Genomics Precision Health Weekly Scan (Current Edition)

Advanced Molecular

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## Hot Topics of the Day

Last Posted: *Mar-23-2021 12:11:07*



Coronavirus

What is my covid risk?

Finnikin Samuel et al. BMJ (Clinical research ed.) 2021 3 n637



## PHGKB News

- [A New Specialized PHGKB - Pharmacogenomics PHGKB \(03/03/2021\)](#)

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Search PHGKB:

## What's New

Last Posted: *Mar 23, 2021*

- [A two-stage modeling approach for breast cancer survivability prediction.](#)  
Sedighi-Maman Zahra et al. International journal of medical informatics 2021 149104438
- [Mainstream genetic testing for breast cancer patients: early experiences from the Parkville Familial Cancer Centre.](#)  
Beard Catherine et al. European journal of human genetics : EJHG 2021

## Search Result Summary

- CDC Information (20)
- NIH Information (16)
- CDC Publications (10)
- Human Genome Epidemiologic Studies (7598)
- GWAS Studies (100)
- Human Genomics Translation/Implementation Studies (1764)
- Genomic Tests Evidence Synthesis (156)

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## Hot Topics of the Day

Last Posted: Mar-23-2021 12:11:07



What is my covid risk?

Finnikin Samuel et al. BMJ (Clinical research ed.) 2021 3 n637

Risk calculators can give an estimate of the risk of dying from contracting covid-19 given an individual's characteristics, but the figures the tool provides will need to be given context before they can be a meaningful part of a decision. Consider an individual's risk from covid-19 in two parts; the risk of catching it and the risk of poor outcome if they do. Both can usually be modified to some extent.

Routine asymptomatic testing strategies for airline travel during the COVID-19 pandemic: a simulation study

MV Kiang et al. Lancet Infectious Diseases, March 22, 2021

Routine asymptomatic testing for SARS-CoV-2 before travel can be an effective strategy to reduce passenger risk of infection during travel, although abbreviated quarantine with post-travel testing is probably needed to reduce population-level transmission due to importation of infection when travelling from a high to low incidence setting.

v-safe COVID-19 Vaccine Pregnancy Registry

CDC, March 2021 [CDC](#)

If you are pregnant, you might choose to be vaccinated when it's available to you.

### COVID-19 GPH

## PHGKB News

- A New Specialized PHGKB - Pharmacogenomics PHGKB (03/03/2021) **New**
- PHGKB launched a new version (v7.2) (03/03/2021) **New**
- PHGKB launched a new version (v7.1) (02/10/2021)

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## Special Topics

- Cancer
- Diabetes
- Infectious Diseases
- Heart, Lung, Blood and Sleep Diseases
- Rare Diseases
- Health Equity
- Family Health History
- Reproductive and Child Health
- Pharmacogenomics

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COVID-19

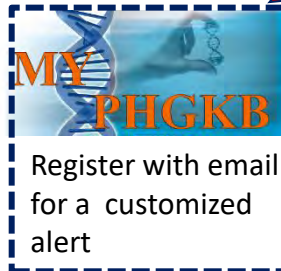
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COVID-19 GPH (22574)

CDC/NIH Web Information  
Database (25068)

CDC-Authored Genomics  
and Precision Health  
Publications Database  
(3186)

Precision Health Database  
(39250)

Tier-Classified Guidelines  
Database (413)

State Public Health  
Genomics Programs  
Database (324)

Pathogen Advanced  
Molecular Detection  
Database (17223)

HuGE Literature Finder  
(181062)

# PHGKB: Public Health Genomics and Precision Health Knowledge Base

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

COVID-19 GPH (22574)

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HuGE Literature Finder (181062)

## What are the different resources?

- CDC-Authored Genomics Publications – *journal articles*
- Precision Health Database: Genomics, non-Genomics
- Advanced Molecular Detection Database
- COVID-19 Genomics and Precision Health Portal
- Tier-Classified Guidelines Database
- My Family Health Portrait: Surgeon General Tool
- CDC Information – *web pages*
- State Public Health Genomics Programs Database
- HuGE Navigator – *genetic association studies (PubMed)*

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## All Databases

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# Precision Health Database

## Precision Health Database

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Last data update: Mar 23, 2021. (Total: 39541 Documents since 2012)



(since 2019)



## About Precision Health Database

This database includes published scientific literature on evidence-based translation of genomic and precision health discoveries into improved health care and population health, featuring information on topics that include reproductive health, birth defects, newborn screening, chronic diseases such as cancer and diabetes, pharmacogenomics, and family health history, guidelines and recommendations. [View Data Selection Criteria](#)

**Genomics Precision Health (GPH):** includes published scientific literature on evidence-based translation of genomic discoveries into improved health care and population health.

**Non-Genomics Precision Health (non-GPH):** includes published scientific literature on the translation of big data, data science and machine learning methods into improved health care and population health.

# Precision Health Database

## Why did we build it?

- **Challenge:** *Keeping up with developments in genomics and data science relevant to public health*
- **Opportunity:** Identify the latest information on population-based applications of discoveries in genomics and precision health
- **Challenge:** *Addressing the 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
- **Challenge:** *Monitoring development of data science relevant to public health*
- **Opportunity:** Highlight public health applications of machine learning and other “big data” techniques to precision public health

# Precision Health 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, in-house curation
- Publish online in Weekly Update
- Add to searchable database

# Precision Health 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	
<b>A</b>	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>B</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
<b>C</b>	Guidelines/policies/ recommendations	New nomenclature, data sharing, and publication standards	Clinical practice and professional guidelines	Electronic health standards, reporting requirements, and ethical standards
<b>D</b>	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.

## Genomics Precision Health Weekly Scan

The latest information and publications on the impact of human genomics and family history across the lifespan.

## Non-Genomics Precision Health Weekly Scan

The latest information and publications on the impact of big data science, machine learning, and predictive analytics on health.

## Advanced Molecular Detection Clips



The latest information and publications on the impact of pathogen genomics on public health.

## CDC-Authored Publications Update

The latest CDC publications in human and pathogen genomics , advanced molecular detection, data science, and precision health.





## All Databases

COVID-19 GPH (22574)

CDC/NIH Web Information Database (25068)

CDC-Authored Genomics and Precision Health Publications Database (3186)

Precision Health Database (39250)

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Pathogen Advanced Molecular Detection Database (17223)

HuGE Literature Finder (181062)



# Advanced Molecular Detection Database

## Pathogen Advanced Molecular Detection Database

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Last data update: Mar 4, 2021. (Total: 17223 Documents since 2015)

Enter a search term  Search  dataset  CDC Author Only

### This Week's New Publications

Evaluation of a SARS-CoV-2 Vaccine NVX-CoV2373 in Younger and Older Adults  
Formica N, et al. medRxiv, Mar 1, 2021.  
[Similar articles in PubMed](#)

Why Virus Variants Have Such Weird Names  
Mandavilli A, Mueller B. New York Times, Mar 2, 2021.  
[Similar articles in PubMed](#)

Travel from the United Kingdom to the United States by a Symptomatic Patient Infected with the SARS-CoV-2 B.1.1.7 Variant - Texas, January 2021  
Ojelade M, et al. MMWR, Mar 3, 2021.  
[Similar articles in PubMed](#)

Persistent SARS-CoV-2 RNA Shedding without Evidence of Infectiousness: A Cohort Study of Individuals with COVID-19.  
Owusu Daniel et al. The Journal of infectious diseases 2021 3  
[Similar articles in PubMed](#)

### This Week's CDC Authored Publications

The State of Microbiome Science at the Intersection of Infectious Diseases and Antimicrobial Resistance. [🔗](#)  
Ranallo Ryan T et al. The Journal of infectious diseases 2021 3  
[CDC Author](#)

Whole-Genome Enrichment and Sequencing of Chlamydia trachomatis Directly from Patient Clinical Vaginal and Rectal Swabs. [🔗](#)  
Bowden Katherine E et al. mSphere 2021 3 (2) [CDC Author](#)

Complete and Circularized Bacterial Genome Sequence of [🔗](#)  
Gulvik Christopher A et al. Microbiology resource announcements 2021 3 (9) [CDC Author](#)

mRNA COVID-19 Vaccines: An Incredible Feat of Genomic Technology  
(blog post) Cono J, et al. Office of Genomics and Precision Public Health/OS/CDC, Mar 5, 2021. [CDC Author](#)

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# Advanced Molecular Detection Clips

- Methods / Tools
- Evolution / Ecology / Populations
- Pathogenicity / Antimicrobial Resistance
- Detection / Diagnosis
- Epidemiology / Outbreaks / Transmission
- Host-Microbe Interactions

All Databases

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# COVID-19 GPH Portal (since April 2020)



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Enter a search term

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All

dataset  All  GPH  Non-GPH

Last Posted: Mar-22-2021 09:01:39



Spotlight

What is GPH/Non-GPH?

COVID-19 GPH Weekly Update

- Genomics, molecular, and other precision health tools (machine learning) in the investigation and control of COVID-19
- PubMed records via an automated PubMed search algorithm
- Preprint records from NIH iCite
- Links to contents from our curated PHGKB databases

All Databases

COVID-19 GPH (22574)

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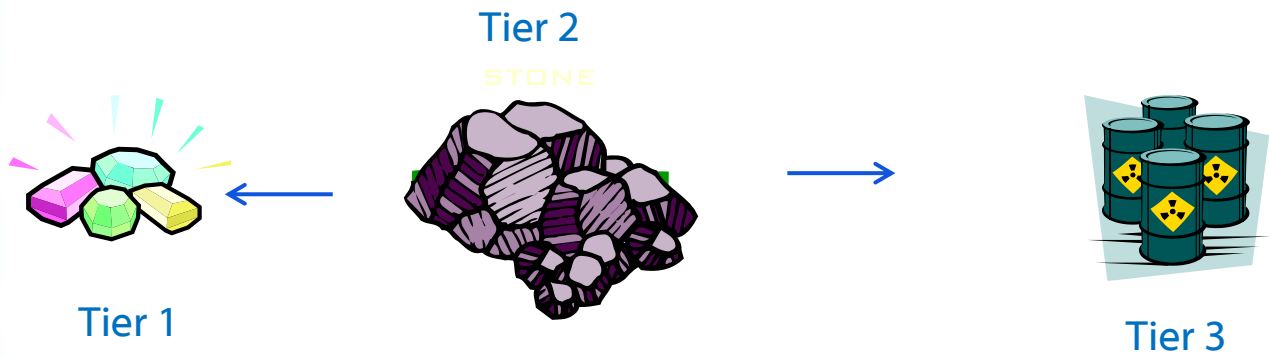
State Public Health Genomics Programs Database (324)

Pathogen Advanced Molecular Detection Database (17223)

HuGE Literature Finder (181062)

# The Genomic Gold Rush

*or, All that Glitters is Not Gold*



# Tier-Classified Guidelines Database (TCGD)

## 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:** *Finding policies, guidelines, and recommendations that include evidence on implementation of genomics or family health history applications*
- **Opportunity:** Compile a centralized, searchable, publicly available database for policies, guidelines, and recommendations related to genomics or family health history

# Tier-Classified Guidelines Database (TCGD)

## *What it is:*

- Repository of genomic guidelines classified according to evidence
- Potential aid to informed decision-making
- Scenario-based
- Systematic
- Subjective
- Context-dependent

# Tier-Classified Guidelines Database (TCGD)

## *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-Classified Guidelines Database (TCGD)

## *How it works:*

- Guidelines identified in weekly horizon scan are cataloged in PHGKB
- Two reviewers assess these guidelines for inclusion in TCGD component of PHGKB
- Guidelines that address clinical scenario(s) involving genetic testing are included in TCGD
  - Decision rules applied to determine Tier level of clinical scenarios within the guidelines
  - Included guideline documents are assigned the highest Tier level applicable to any recommendation they contain



## Tier 1

- FDA label requires use of test to inform choice or dose of a drug
- FDA cleared or approved companion diagnostic device
- CMS covers testing
- Clinical practice guidelines based on systematic review supports testing

## Tier 2

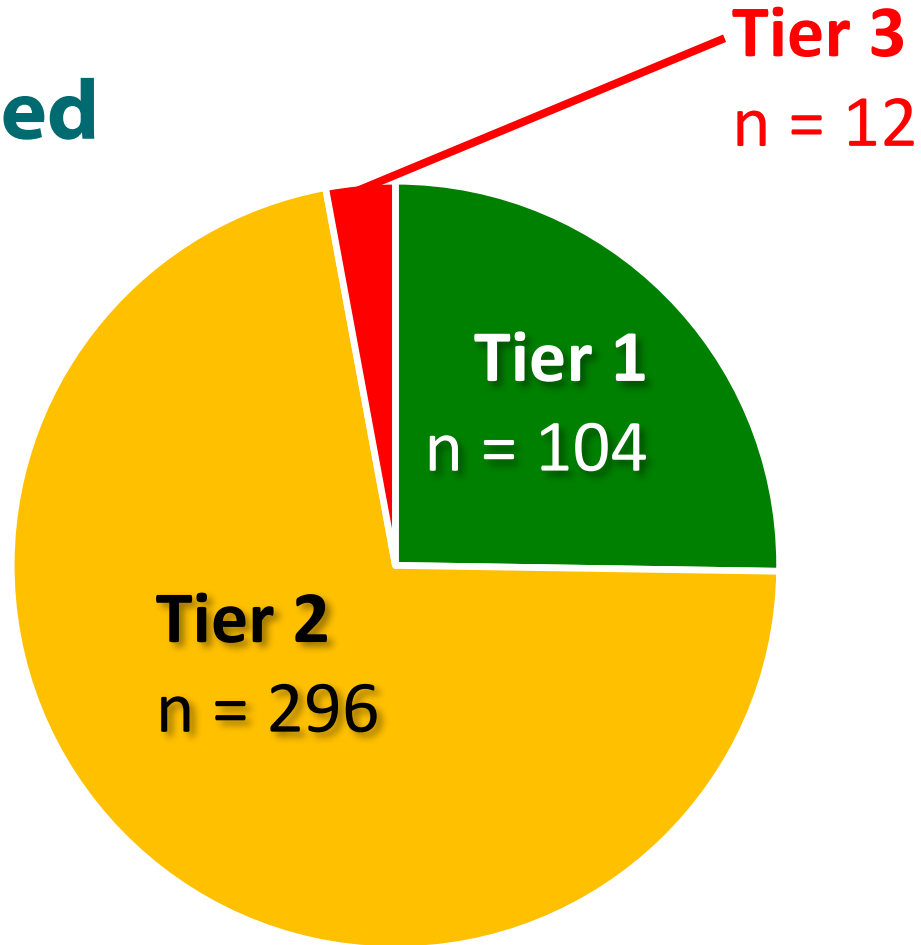
- FDA label mentions biomarkers
- FDA premarket approval (PMA)
- FDA 510(k) substantially equivalent decision
- CMS coverage with evidence development
- Clinical practice guideline, not based on systematic review, supports use of test
- Clinical practice guideline finds insufficient evidence but does not discourage use of test
- Clinical practice guideline recommends dosage adjustment, but does not address testing

## Tier 3

- 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

# Tier-Classified Guidelines

N = 412



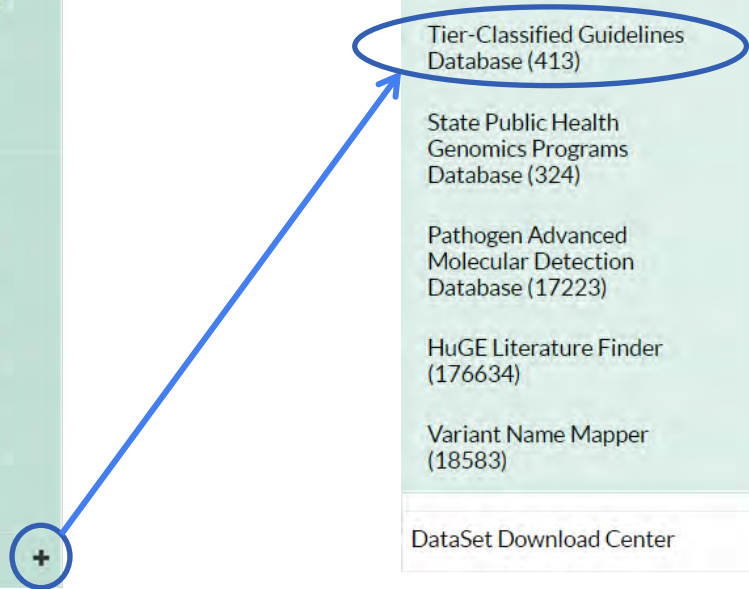
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- CDC-authored Publications Update
- COVID-19 Precision Health Weekly Update (Current Edition)
- All Databases **+**

All Databases **-**

- COVID-19 GPH (22493)
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- HuGE Literature Finder (176634)
- Variant Name Mapper (18583)

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## Tier-Classified Guidelines Database

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Search  dataset

- All
- All Tier 1
- All Tier 2
- All Tier 3

### Recent Uploaded Publications

ASH ISTH NHF WFH 2021 guidelines on the diagnosis of von Willebrand disease.  
James Paula D et al. Blood advances 2021 Jan 5(1) 280-300  
[Similar articles in PubMed](#)

### ABOUT

This database contains guidelines from [the Genomics and Precision Health Database](#), sorted according to the highest evidence tier level that could be assigned to recommendations from each guideline. The sorting process is based on our previous

**Publication****Tier**

ASH ISTH NHF WFH 2021 guidelines on the diagnosis of von Willebrand disease.

James Paula D et al. Blood advances 2021 Jan 5(1) 280-300

Similar articles in PubMed [↗](#)

**2**

**Organization:** [The American Society of Hematology](#) [The International Society on Thrombosis and Haemostasis \(ISTH\)](#) [National Hemophilia Foundation \(NHF\)](#) [World Federation of Hemophilia \(WFH\)](#) [↗](#)

Focused Revision: ACMG practice resource: Genetic evaluation of short stature.

Mintz Cassie S et al. Genetics in medicine : official journal of the American College of Medical Genetics 2021 Jan

Similar articles in PubMed [↗](#)

**2**

**Organization:** [The American College of Medical Genetics and Genomics \(ACMG\)](#) [↗](#)

Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic, Version 2.2021, NCCN Clinical Practice Guidelines in Oncology.

Daly Mary B et al. Journal of the National Comprehensive Cancer Network : JNCCN 2021 Jan 19(1) 77-102

Similar articles in PubMed [↗](#)

**1**

**Organization:** [The National Comprehensive Cancer Network \(NCCN\)](#) [↗](#)

Consumer Testing for Disease Risk: ACOG Committee Opinion, Number 816.

et al. Obstetrics and gynecology 2021 Jan 137(1) e1-e6

Similar articles in PubMed [↗](#)

**2**

**Organization:** [The American College of Obstetricians and Gynecologists \(ACOG\)](#) [↗](#)

Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for CYP2D6, OPRM1, and COMT genotype and select opioid therapy.

Crews Kristine R et al. Clinical pharmacology and therapeutics 2021 Jan

Similar articles in PubMed [↗](#)

**3**

**Organization:** [The Clinical Pharmacogenetics Implementation Consortium \(CPIC\)](#) [↗](#)

# Tier-Classified Guidelines Database

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Last data update: Feb 26, 2021. (Total: 413 Documents since 2012)

Search  dataset

Filtered By:

- Select to fine-tune your search
- Tier
- Year
- Organization

Records 1-

Query Trace: lynch syndrome[original query]

Filtered By:

Records 1-20 (of 20 Record(s))

**Reminder!**  
The tier level is assigned to the guideline, NOT to the specific search terms. For example, a Tier 1 guideline has at least one recommendation that meets criteria for Tier 1, but also could include tier 2 or 3 recommendations. **DO** read the guidelines to find out about specific search terms. **DON'T** rely on the overall tier classification of the guideline for the specified search terms. See More FAQ

Query Trace: lynch syndrome[original query]

Publication	Tier
Guidelines for the management of hereditary colorectal cancer from the British Society of Gastroenterology (BSG)/Association of Coloproctology of Great Britain and Ireland (ACPGBI)/United Kingdom Cancer Genetics Group (UKCGG). Monahan Kevin J et al. Gut 2019 Nov <a href="#">Similar articles in PubMed</a>	2

Publications are indexed with the following 3 tier(s).

Click Continue button after making selection.

Tier	Number of Publications
<input checked="" type="checkbox"/> 1	3
<input type="checkbox"/> 2	16
<input checked="" type="checkbox"/> 3	1

Query Trace: lynch syndrome[original query]>>1, 3[Tier]

Publication

Tier

Australasian Gastrointestinal Pathology Society (AGPS) consensus guidelines for universal defective mismatch repair testing in colorectal carcinoma.

1

Yozu Masato et al. Pathology 2019 Mar

Similar articles in PubMed [↗](#)

**Organization:** *The Australasian Gastrointestinal Pathology Society (AGPS)* [↗](#)

Lynch Syndrome: A Primer for Urologists and Panel Recommendations.

3

Similar articles in PubMed [↗](#)

**Organization:** *Expert Panel*

American Gastroenterological Association Institute Guideline on the Diagnosis and Management of Lynch Syndrome.

1

Rubenstein Joel H et al. Gastroenterology 2015 Jul 27.

Similar articles in PubMed [↗](#)

**Organization:** *The American Gastroenterological Association (AGA)* [↗](#)

Guidelines on genetic evaluation and management of Lynch syndrome: a consensus statement by the US Multi-society Task Force on colorectal cancer.

1

Similar articles in PubMed [↗](#)

# Lynch Syndrome



# Records down

Records 1-4 (of 4 Record(s))

Download

	A	B	C	D	E	F	G
1	ID	Title	Url	Published	Pubmed ID	Tier	Organization (Title, Url)
2	1149	Australasian Gastrointestinal Pathology Society (AGPS) consensus guidelines for universal defective mismatch repair testing in colorectal carcinoma.	<a href="https://pubmed">https://pubmed</a>	2019	30851981	1	<a href="http://www.agps.org.au/">http://www.agps.org.au/</a> <a href="#">[The Australasian Gastrointestinal Pathology Society (AGPS)]</a>
3	1199	Guidelines on genetic evaluation and management of Lynch syndrome: a consensus statement by the US Multi-society Task Force on colorectal cancer.	<a href="https://pubmed">https://pubmed</a>	2014	25070057	1	[Expert Panel]
4	1200	American Gastroenterological Association Institute Guideline on the Diagnosis and Management of Lynch Syndrome.	<a href="https://pubmed">https://pubmed</a>	2015	26226577	1	<a href="https://www.gastro.org/">https://www.gastro.org/</a> <a href="#">[The American Gastroenterological Association (AGA)]</a>
5	1406	Lynch Syndrome: A Primer for Urologists and Panel Recommendations.	<a href="https://pubmed">https://pubmed</a>	2015	25711197	3	[Expert Panel]

# Tier-Classified Guidelines Database (TCGD)

## *Where to learn more:*

- About The Tier Classified Guidelines Database & FAQs  
<https://phgkb.cdc.gov/PHGKB/tierFinder.action?Mysubmit=about>
- Introducing the CDC Tier-Classified Guidelines Database (2019)  
<https://blogs.cdc.gov/genomics/2019/07/16/introducing-the-cdc-tier/>
- Frequently Asked Questions about the CDC Tier-Classified Guidelines Database  
<https://blogs.cdc.gov/genomics/2019/07/16/frequently-asked-questions/>
- Prioritizing Genomic Applications for Action by Level of Evidence: A Horizon-Scanning Method (2014)  
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4689130/>

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Family Health History

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**My Family Health Portrait**

- Record your family health history
- Learn about your risk for conditions
- Print & save your family health history

[VISIT My Family Health Portrait](#)



Latest CDC Genomics  
and Precision Health  
Tweets

More Resources

# My Family Health Portrait

- Free, online family health history collection tool
- Information on several chronic conditions
- Information saved on user's computer
  - Never saved on server (by design)

*My Family Health Portrait*  
A tool from the Surgeon General

Get Help

Using My Family Health Portrait you can:

- Enter your family health history.
- Learn about your risk for conditions that can run in families.
- Print your family health history to share with family or your health care provider
- Save your family health history so you can update it over time.

Talking with your health care provider about your family health history can help you stay healthy!

[Learn more about My Family Health Portrait](#)

Create a Family Health History      Use a Saved History



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Created By [NIH](#) | [NHGRI](#) | Maintained By [CDC](#) - [OGPPH](#)

# My Family Health Portrait

- Developed by Surgeon General and NHGRI/NIH, first released 11/2004
- Hosted online by NHGRI/NIH from 11/2005 - 9/2018
- Hosted by CDC since 9/2018
- Almost 1.5 million page views annually

## My Family Health Portrait

A tool from the Surgeon General

Get Help

Using My Family Health Portrait you can:

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[Learn more about My Family Health Portrait](#)

Create a Family Health History

Use a Saved History



[Glossary](#) | [FAQ](#) | [Accessibility](#) | [Privacy & Security Policy](#) | [About This Site](#) | [Contact Us](#) | [Site Updates](#)

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[Help](#)

Disclaimer: The Surgeon General's My Family Health Portrait tool does NOT keep a government record of the information you fill in nor make your health information available to anyone else but you. It only provides the software for organizing your information. By accessing the tool on the web, you make use of that software. But the information you fill in is not transmitted back to our servers, and never available to anyone else, unless you choose to share or disclose it. After you fill in your information, it is available only to you for downloading. After that, it's up to you whether you want to share the information with other family members or provide it to your health care practitioner. The Surgeon General's tool helps gather information that will be useful for you and your health care practitioner, but it does not provide medical advice. You should consult with a health professional about advice based on your family health history information.

### Your Personal Information

We start the family health history with you. Enter the required personal information and your health history information. At the bottom of the page (you may need to scroll), press the 'Next' button. You will then be asked to tell the system which family members you would like to add to the health history.

\* Indicates required information.

\*Name:

\*Sex assigned at birth:  Male  Female

\*Date of Birth  (mm/dd/yyyy)

Were you born a twin?  No  Yes - Identical (Same)  Yes - Not Identical (Fraternal)

Were you adopted?  Yes

Height  Feet  Inches -OR-  Centimeters

Weight  lbs

### Your Health Information

In the list below, select a **Disease or Condition** (if any) from the dropdown box. Then select the **Age at Diagnosis** and press the **Add** button. You may repeat this process as necessary.

Disease or Condition	Age at Diagnosis	Action
<input type="text" value="Please Select a Disease"/>	<input type="text" value="Select Age at Diagnosis"/>	<input type="button" value="Add"/>

### Your Family Background Information

Check here if your parents are related to each other in any way other than marriage.

Multiple races and ethnicities may be selected.

Race:

- American Indian or Alaska Native  Asian  Black or African-American  
 Native Hawaiian or Other Pacific Islander  White

Ethnicity:

- Hispanic or Latino  Ashkenazi Jewish  Not Hispanic or Latino

[Why are we asking about Ashkenazi Jewish heritage?](#)

## Disease or Condition

Please Select a Disease

- Cancer (more options...)
- Clotting Disorder (more options...)
- Dementia/Alzheimers
- Diabetes (more options...)
- Gastrointestinal Disorder (more options...)
- Heart Disease (more options...)
- High Cholesterol
- Hypertension
- Kidney Disease (more options...)
- Lung Disease (more options...)
- Osteoporosis
- Psychological Disorder (more options...)
- Septicemia
- Stroke/Brain Attack
- Sudden Infant Death Syndrome
- Unknown Disease
- Other - Add New

Select

any way other than marriage.

ive  Asian  Black or African

ic Islander  White

azi Jewish  Not Hispanic or Latin

## Disease or Condition

Heart Disease (more options...)



Please Select a Specific Subtype

Angina

Coronary Artery Disease

Heart Attack

Heart Disease

Familial Hypercholesterolemia

Unknown Heart Disease

tives.(related to €



## Disease or Condition

Please Select a Disease

- Cancer (more options...)
- Clotting Disorder (more options...)
- Dementia/Alzheimers
- Diabetes (more options...)
- Gastrointestinal Disorder (more options...)
- Heart Disease (more options...)
- High Cholesterol
- Hypertension
- Kidney Disease (more options...)
- Lung Disease (more options...)
- Osteoporosis
- Psychological Disorder (more options...)
- Septicemia
- Stroke/Brain Attack
- Sudden Infant Death Syndrome
- Unknown Disease
- Other - Add New

Select

any way other than marriage.

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## Disease or Condition

Please Select a Disease

- Cancer (more options...)
- Clotting Disorder (more options...)
- Dementia/Alzheimers
- Diabetes (more options...)
- Gastrointestinal Disorder (more options...)
- Heart Disease (more options...)
- High Cholesterol
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- Kidney Disease (more options...)
- Lung Disease (more options...)
- Osteoporosis
- Psychological Disorder (more options...)
- Septicemia
- Stroke/Brain Attack
- Sudden Infant Death Syndrome
- Unknown Disease
- Other - Add New

Select

any way other than marriage.

ive  Asian  Black or African

ic Islander  White

azi Jewish  Not Hispanic or Latin

## Add Family Members



Help

Now we will build your family.

- We automatically add your parents and grandparents for you.
- Tell us about your immediate family, including your brothers, sisters, children, aunts, and uncles.
- You can add more family members like cousins, nieces, nephews, half siblings, and grandchildren later.
- We collect information only for blood relatives, not household members, not step-relatives, nor spouses (unless related by blood).

How many brothers do you have?

How many sisters do you have?

How many sons do you have?

How many daughters do you have?

How many brothers does your mother have? (your uncles)
























How many sisters does your mother have? (your aunts)

How many brothers does your father have? (your uncles)

How many sisters does your father have? (your aunts)


Cancel

Next

Name	Relationship To Me:	Still Living	Update History	Remove Relative
<b>My Family</b>				
Test	Self	Yes		
	Father			
	Mother			
	Brother			
	Sister			
	Son			
	Daughter			
<b>My Father's Side of the Family</b>				
	Paternal Grandfather			
	Paternal Grandmother			
	Paternal Uncle			
	Paternal Aunt			
<b>My Mother's Side of the Family</b>				
	Maternal Grandfather			
	Maternal Grandmother			
	Maternal Uncle			
	Maternal Aunt			
<b>Recently Added Family Members</b>				

## Enter Family Member's Health History

[Help](#)

### Personal Information for your Father

Enter required personal information and health history information for this family member. Background information may also be entered. At the bottom of the page (you may need to scroll), press the 'Save' button to save this person's information.

\* Indicates required information.

Relationship:

Name:

\*Sex assigned at birth:  Male  Female

Is this person still alive:

Was this person born a twin?  No  Yes - Identical (Same)  Yes - Not Identical (Fraternal)

Was this person adopted?  Yes

### Health Information for your Father

In the list below, select a **Disease or Condition** (if any) from the dropdown box. Then select the **Age at Diagnosis** and press the **Add** button. You may repeat this process as necessary.

Disease or Condition	Age at Diagnosis	Action
<input type="text" value="Please Select a Disease"/>	<input type="text" value="Select Age at Diagnosis"/>	<input type="button" value="Add"/>

### Your Family Background Information

Multiple races and ethnicities may be selected.

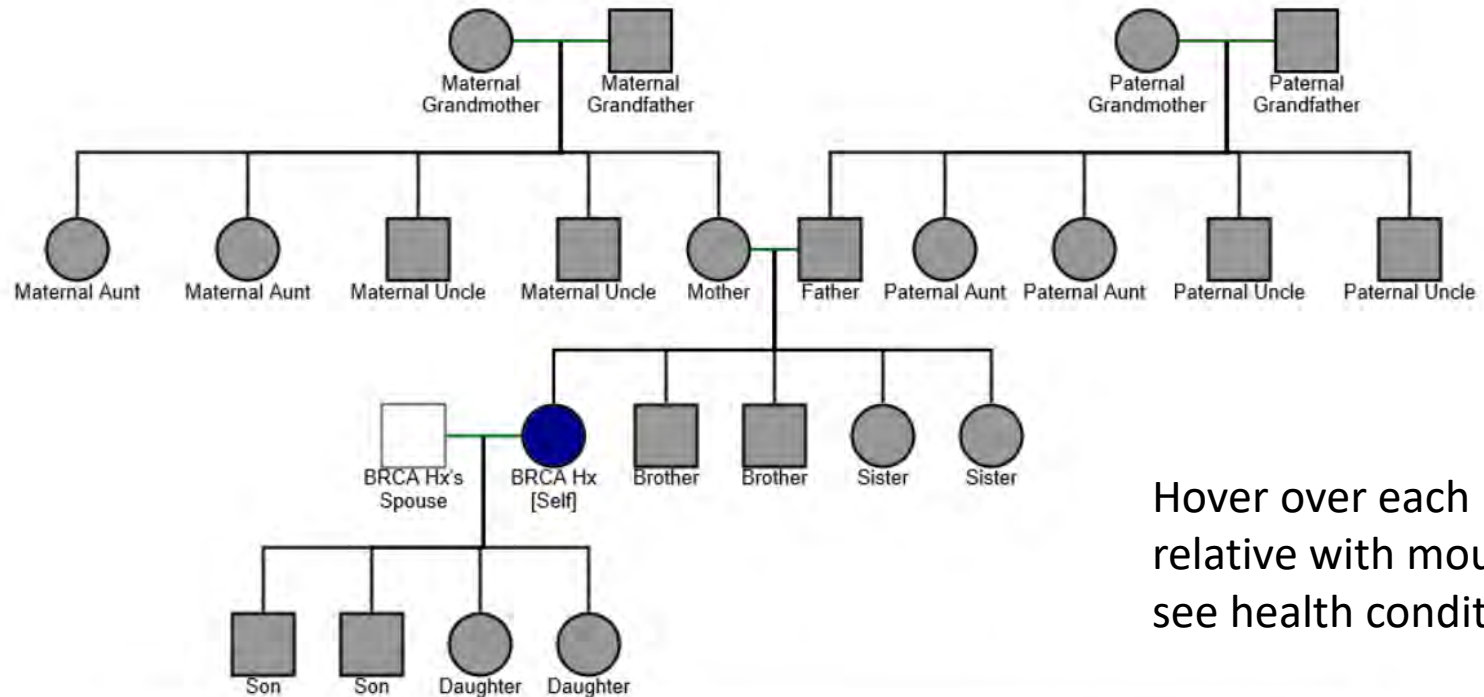
Race:

American Indian or Alaska Native  Asian  Black or African-American  
 Native Hawaiian or Other Pacific Islander  White

Ethnicity:

Hispanic or Latino  Ashkenazi Jewish  Not Hispanic or Latino

[Why are we asking about Ashkenazi Jewish heritage?](#)







Hover over each relative with mouse to see health conditions








## Disease Familial Risk


Check your familial risks for the following diseases:

Disease	Overall Risk Assessment	Assessment Detail
Colorectal Cancer		
Diabetes		

: possible risk increase based on the current information.

: average risk based on the current information.

: not assessed due to lack of required information.

Click on the Assessment Detail icon  to perform the assessment.

Note: Overall Risk Assessment result may be changed after more information provided during the assessment.

Pedigree Drawing

Family Health Table














## Your Colorectal Cancer Familiar Risk Information

On this screen you can:

- Get your risk for colorectal cancer based on your personal and family history information
- Learn which risk factors you may have for colorectal cancer

This tool will run a series of tests, one at a time, to determine your risk for colorectal cancer.

Test	Reason
	You have had pancreatic cancer in the past.
	You have never had any polyps or Familial adenomatous polyposis (FAP).
	You have never had inflammatory bowel disease (either ulcerative colitis or crohn's disease).
	None of your family members have had Lynch Syndrome/Hereditary non-polyposis colorectal cancer (HNPCC) or Familial adenomatous polyposis (FAP)
	None of your immediate relatives (father, mother, brothers, sisters, children) have had colon, colorectal, rectal, or gastric cancer.
	None of your immediate relatives (father, mother, brothers, sisters, children) have had polyps.
	None of your secondary relatives (aunts, uncles, grandparents, grandchildren, halvesiblings) have had colon, colorectal, or rectal cancer. (There should be two or more to trigger this test.)
	None of your secondary relatives (aunts, uncles, grandparents, grandchildren, halvesiblings) have had colon, colorectal, or rectal cancer.
	None of your primary or secondary relatives (mother, father, sisters, brothers, sons, daughters, aunts, uncles, grandparents, grandchildren, halvesiblings) have had uterine cancer before the age of 50.
	None of your secondary relatives (aunts, uncles, grandparents, grandchildren, halvesiblings) have had uterine cancer. (There should be two or more to trigger this test.)
	You have at least one of the above risk factors. Based on this your risk of colorectal cancer is increased.

Get Increased  
Risk Letter

Get Increased  
Risk Provider Letter

## Disease Familial Risk



Please check that the information you have added about you and your relatives is complete and correct

In order to calculate your diabetes risk, we need some additional information

Have you ever had Gestational Diabetes?  ▼

Are you physically active\*:  ▼

Do you have High Blood Pressure (Hypertension):  ▼

[Continue](#)

\* Physical activity is defined as 150 minutes of moderate exercise per week

## Your Type 2 Diabetes Risk Information

On this screen you can:

- Get your risk for type 2 diabetes based on your personal and family history information
- Learn which risk factors you may have for type 2 diabetes
- Read and print sharable letters for you and your health care provider explaining your type 2 diabetes risk

1. How old are you

2

Less than 40 years (0 points) 40-49 years (1 point)  
50-59 years (2 points) 60 years or older (3 points)

2. Are you a man or a woman

0

Man (1 point) Woman (0 points)

3. If you are a woman, have you ever been diagnosed with gestational diabetes?

1

Yes (1 point) No (0 points)

4. Do you have a mother, father, sister or brother with diabetes?

1

Yes (1 point) No (0 points)

5. Have you ever been diagnosed with high blood pressure?

1

Yes (1 point) No (0 points)

6. Are you physically active?

1

Yes (0 point) No (1 points)

7. What is your Body Mass Index? (see chart at right)

0

Height:

5 feet 5 inches

Weight:

130 pounds

Total Points

6

0 - 4 points: Risk not increased

5+ points: Risk Increased

Height	Weight			
	130 lbs			
	Normal	Overweight	Obese	Morbidly Obese
BMI	25 or less	25 - 30	30 - 35	35 or more
4'10"	119 -	120-143	144-167	168 +
4'11"	123 -	124-148	149-173	174 +
5'0"	128 -	129-153	154-179	180 +
5'1"	132 -	133-158	159-185	186 +
5'2"	136 -	137-164	165-191	192 +
5'3"	141 -	142-169	170-197	198 +
5'4"	145 -	146-174	175-203	204 +
5'5"	150 -	151-180	181-210	211 +
5'6"	154 -	155-185	186-216	217 +
5'7"	159 -	160-191	192-223	224 +
5'8"	164 -	165-197	198-230	231 +
5'9"	169 -	170-203	204-237	238 +
5'10"	174 -	175-209	210-243	244 +
5'11"	179 -	180-215	216-250	251 +
6'0"	184 -	185-221	222-258	259 +
6'1"	189 -	190-227	228-265	266 +
6'2"	194 -	195-233	234-272	273 +
6'3"	200 -	201-240	241-280	281 +
6'4"	205 -	206-246	247-287	288 +
	0 points	1 points	2 points	3 points

Get Personal Elevated Risk Letter

Get Provider Elevated Risk Letter

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- Record your family health history
- Learn about your risk for conditions
- Print & save your family health history

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Breast & Ovarian Cancer



Colorectal Cancer



Cystic Fibrosis



Hereditary Hemochromatosis



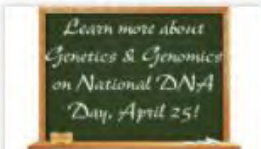
Muscular Dystrophy



Newborn Screening



Diabetes



DNA Day



Down Syndrome



Obesity



Osteoporosis



Pathogen Genomics



Epigenetics



Family Health History



Folic Acid



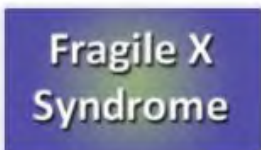
Pharmacogenomics



Precision Health



Primary Immunodeficiency



Fragile X Syndrome



Genetics 101



Genetic Counseling



Public Health Genomics Toolkit



Real Stories



Sickle Cell Disease



Genetic Testing



Heart Disease



Hemophilia



Stroke



Thalassemia



von Willebrand Disease



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

## My Family Health Portrait

- Record your family health history
- Learn about your risk for conditions
- Print & save your family health history

[VISIT My Family Health Portrait](#)



Planning for Pregnancy



Information for Health Professionals



During Pregnancy



Information for Researchers



For Children



Tools and Resources



For Adults

## Knowing is Not Enough—Act on Your Family Health History



For more information, visit the [Genomics and Precision Public Health home page](#).



Having a family health history of heart disease makes you more likely to develop heart disease yourself. In some cases, having family members with heart disease at a young age (age 50 or younger) can be a sign of familial hypercholesterolemia, a genetic disorder that causes high cholesterol. Take time to collect your family health history of heart disease and share this information with your doctor and other family members. Your doctor can help you take steps to lower your chances of having heart disease.

## Coronavirus Disease 2019 (COVID-19) and Familial Hypercholesterolemia (FH)

COVID-19 is a new disease and information on risk factors for severe disease is limited. Based on currently available information and clinical expertise, people who have serious heart disease are among those [more likely to have severe illness from COVID-19](#). If untreated, people with FH are up to 22 times more likely to have coronary heart disease than those without FH. Although not everyone with FH has heart disease, many do. Everyone is encouraged to take certain steps to protect themselves from getting sick with COVID-19. It is especially important that those with increased risks, such as serious heart disease, take these steps.

[Click here](#) to learn steps you can take to help protect yourself if you have serious heart disease. Be sure to keep taking prescribed FH medications including statins and PCSK9 inhibitors as directed by your healthcare provider. Contact your healthcare provider to ask about obtaining extra FH medications. Consider telemedicine appointments if you need to see your healthcare provider.



The Basics



Genetic Counseling for FH



Familial Hypercholesterolemia (FH)



Genetic Testing



Finding Family Members with FH



Medical Options



Family Health History of Heart Disease



Talking to Your Family about Your FH Diagnosis



Family Health History of FH



Personal Stories



Does Heart Disease Run In Your Family?

### My Family Health Portrait

- Record your family health history
- Learn about your risk for conditions
- Print & save your family health history

[VISIT My Family Health Portrait](#)

For more information, visit the [Genomics and Precision Public Health home page](#).

# 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



## Genomic Application Toolkit

Tier 1

State & Local Health Departments

How to use the Toolkit

Updates

Lynch Syndrome

Phase 1

Phase 2

Tools

Publications

Hereditary Breast & Ovarian Cancer

Familial Hypercholesterolemia

Videos

# Tier 1 Genomic Applications Toolkit for Public Health Departments

Tier 1 Genomic Applications and their Importance to Public Health



State and Local Public Health Departments Can Play Key Roles in Addressing Tier 1 Genomic Applications

How to use this Toolkit

Check Here for Recent Changes/Updates to the Toolkit

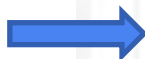
Lynch Syndrome (LS)

Hereditary Breast and Ovarian Cancer (HBOC)

Familial Hypercholesterolemia

Tier 1 Implementation Videos, Other Tools, and Resources Available to Help

Contact Us



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](#) [DOC 1.05 MB]
- [Brochure on Talking to Your Family About Your Diagnosis of Lynch Syndrome](#) [PPT 602 KB]
- [Sample Letter for Informing Your Family Members about Your Lynch Syndrome Mutation](#) [DOC 15.9 KB]
- [List of Cancer Genetic Specialists for Your State or Region](#) [DOC 23 KB] (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](#) [DOC 24.9 KB]

### Information for Providers

- [This slide set is intended to be a resource for state health departments to use when presenting to hospitals and other institutions.](#) [PPT 6.77 MB] States are welcome to select and modify slides to use and are not expected to use the entire set in a single presentation.
- [Lynch Syndrome: Fact Sheet for Healthcare Professionals](#) [DOC 31.2 KB]
- [Evidence-based Practice Guidelines Supporting Genetic Susceptibility Testing for Lynch Syndrome](#) [DOC 18 KB]
- [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.
- [Sample clinician's letter to provide your patients to help them let their family members know about their Lynch syndrome mutation.](#) [DOC 18.8 KB] [DOC - 19 KB]

### Reporting Tools

- [Sample Hospital and Medical Center Cancer Genetics Data Report on Potential Lynch Syndrome-Related Cancers](#) [DOC 23.5 KB] (Please note that state programs will need to complete this form.)

# State Public Health Genomics Program 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 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

# State Public Health Genomics Program Database

## Public Health Genomics and Precision Health Knowledge Base (v7.2)

### PHGKB

About

MyPHGKB

Specialized PHGKB

Genomics (A-Z)

Office of Genomics and Precision Public Health

My Family Health Portrait

State Public Health Genomics Programs Map

Genomics Precision Health Weekly Scan (Current Edition)

Advanced Molecular Detection Weekly Clips (Current Edition)

Non-Genomics Precision Health Weekly Scan (Current Edition)

CDC-authored Publications Update

COVID-19 Precision Health Weekly Update (Current Edition)

All Databases

COVID-19 GPH (23313)

CDC/NIH Web Information Database (25155)

CDC-Authored Genomics and Precision Health Publications Database (3198)

Precision Health Database (39434)

Tier-Classified Guidelines Database (413)

State Public Health Genomics Programs Database (324)

## State Public Health Genomics Programs Database

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Last data update: Dec 23, 2020. (Total: 324 Documents)

### About State Public Health Genomics Programs Database

The State Public Health Genomics Programs database has information about state public health programs and activities relevant to genomics. 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). You can also view the content of the database using [State Genomics Implementation Map](#).

### State Public Health Genomics Program Map



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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

