An Introduction to the CDC Public Health Genomics Knowledge Base

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Office of Public Health Genomics

CDC Office of Public Health Genomics (OPHG)

Effective and responsible translation of genome-based discoveries

➡ into disease prevention and population health

https://www.cdc.gov/genomics

1. Identify
evidence-based applications

2. Inform
and communicate

3. Integrate
into practice & programs

OPHG, DPHID, CSELS

Public Health Genomics

NIH-CDC Public Health Genomics Webinar

February 14, 2018

OBESITY AND GENOMICS: READ NEW CDC BLOG

INFLUENZA, GENOMICS AND PUBLIC HEALTH

WORLD CANCER DAY: PREVENTING HEREDITARY CANCERS

WEAR RED DAY ON FEBRUARY 2: KNOW YOUR FAMILY HISTORY

WEEKLY UPDATE
Weekly summary of genomics and health impact information

GENETICS 101
Genetics basics explained including a glossary of genetic terms

PHGKB
Online searchable knowledge base on genomics and health impact information

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

AMD CLIPS
Weekly news and publications on pathogen genomics and bioinformatics

GENOMICS AND DISEASES
Genomics is important for many diseases of public health significance

IMPLEMENTATION
What public health can do now to save lives using genomics

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

REPORTS AND PUBLICATIONS
CDC reports and publications in genomics

GENOMIC TESTING
Genomic tests are used in many diseases

GENOMICS & HEALTH IMPACT BLOG
A blog devoted to genomic issues in research, policy and practice

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

PODCASTS AND VIDEOCASTS

EPIDEMIOLOGY

The **CDC Office of Public Health Genomics** provides timely and credible information for the effective and responsible translation of genomics research into population health benefits.

About Us **At A Glance**

Genomics Across CDC

- Advanced Molecular Detection
- Birth Defects
- Blood Disorders
- Cancer Genomics
- Diabetes Prevention
- Laboratory Practice
- Newborn Screening

Dr Khoury's Tweets

@DrKhouryCDC
Check out the NEW customized education toolkit for physicians. Via @genome_gov. https://t.co/Ncu89vTuq4

Public Health Genomics Knowledge Base (v2.1)

PHGKB
About
MyPHGKB
Specialized PHGKB +
Genomics (A-Z)
Office of Public Health Genomics
State Public Health Genomics Programs Map
Genomics & Health Impact Weekly Scan (Current Edition)
Advanced Molecular Detection Weekly Clips (Current Edition)
All Databases +
DataSet Download Center
Release Note

Search PHGKB Search

Hot Topics of the Day

Last Posted: *Feb-04-2018 10AM*

Disparities

Integrating the Genetics of Race and Ethnicity Into Cancer Research: Trailing Jane and John Q. Public.
Newman Lisa A et al. JAMA surgery 2018 Jan

Study Finds Biological Differences in Lung Tumors of African Americans and Whites
NCI Blog, Jan 2018

Announcement

"Public health genomics information is now easily accessible online", a new blog post about PHGKB authored by Wei Yu and Muin Khoury. **Check it out!**

Content Summary

- CDC Information (886)

(Current Edition)
Advanced Molecular Detection Weekly Clips (Current Edition)
All Databases −
CDC Information Database (886)
CDC-Authored Genomics Publication Database (1920)
Genomics & Health Impact Scan Database (12166)
Tier Table Database (181)
State Public Health Genomics Programs Database (275)
Advanced Molecular Detection Clips Database (8379)
HuGE Navigator +
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Database Content (Last Updated:)

Genomics & Health Impact Weekly Scan

This weekly update features emerging roles of human genomics, testing and interventions in a wide variety of noncommunicable diseases across the life span, including, birth defects, newborn screening, reproductive health, childhood diseases, cancer, chronic diseases, pharmacogenomics, family health history, guidelines and recommendations. The weekly sweep also includes news, reviews, commentaries, tools and databases.

Advanced Molecular Detection Clips

Advanced Molecular Detection(AMD) 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 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.

CDC Information Database

This database includes general CDC public health information on specific diseases and health related topics. When available, the database displays genomic information from various CDC web pages. Users are also encouraged to conduct searches of CDC website for additional information.

CDC-Authored Genomics Publication Database

This database contains CDC-authored scientific publications on genomics-related topics, and includes articles on infectious diseases, reproductive health, newborn screening, birth defects, developmental disabilities, genetic testing, chronic diseases such as cancer and diabetes, environmental and occupational health, laboratory methods, bioinformatics, and statistical methods.

CDC Information Database
Why did we build it?

- ***Challenge:*** *Finding information about genomics- and family health history-related activities at CDC*
- **Opportunity:** Provide a centralized, searchable, publicly available database for CDC resources related to genomics and family health history

CDC-Authored Genomics Publications Database

Why did we build it?

- ***Challenge:*** *Finding CDC-authored publications on genomics and family health history*
- **Opportunity:** Provide a centralized, searchable, publicly available database for these CDC publications
- ***Challenge:*** *CDC's work in genomics and family health history is not well known*
- **Opportunity:** Showcase CDC publications to highlight work related to genomics and family health history

CDC-Authored Genomic Publications Database continued

- Scientific articles and reports with at least one CDC author
- CDC Science Clips: https://www.cdc.gov/library/sciclips/
- PubMed
- Scopus
- Author notifications

affiliation search

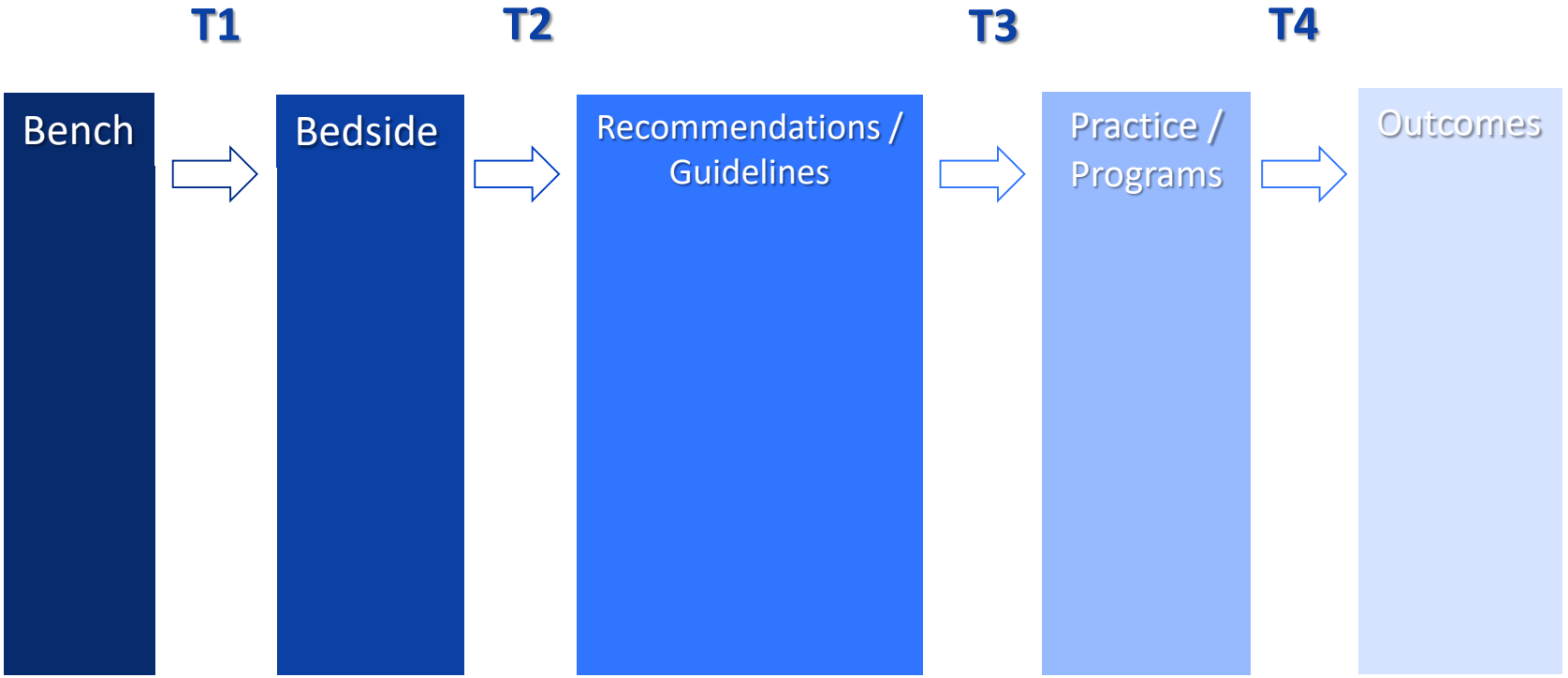
Genomics and Health Impact Scan Database
Why did we build it?

- ***Challenge:*** *Keeping up with the latest developments in genomics and family health history 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

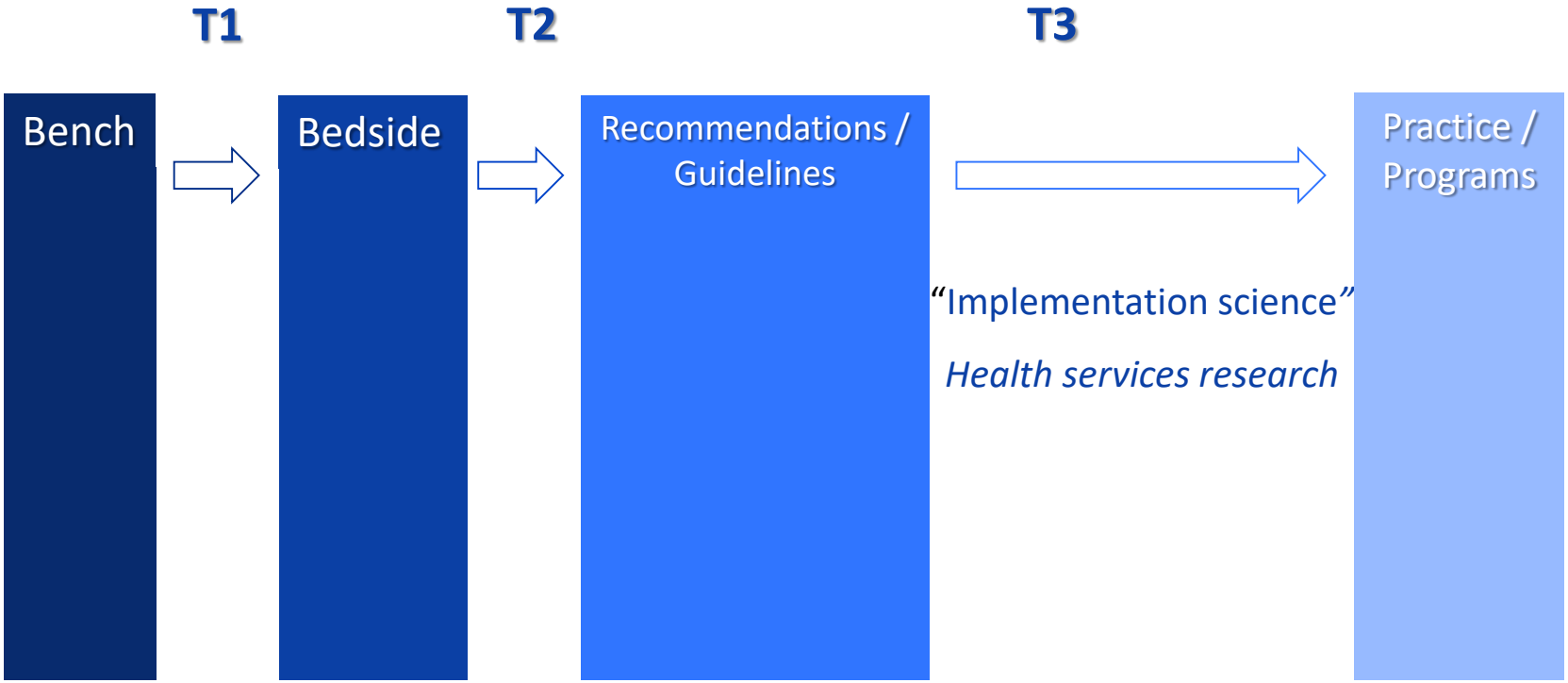
Genomics and Health Impact Scan Database

- Horizon Scan
  - Monitor Google Alerts, PubMed queries, key websites
  - Select news stories, blog posts, scientific articles, websites
  - Publish online in Weekly Update
- Categorized by
  - Translation and implementation studies
  - Evidence synthesis (systematic reviews, modeling)
  - Guidelines
  - Tools/Methods
  - Reviews/Commentaries

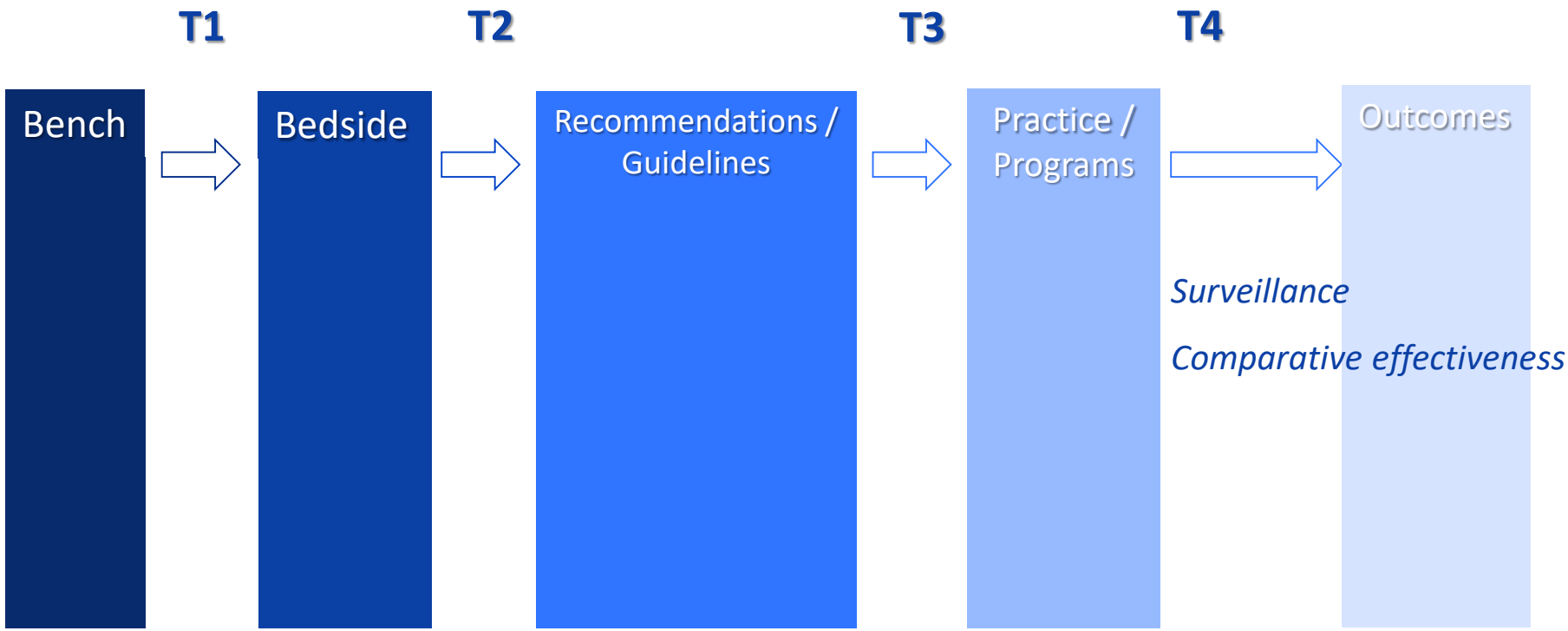
Organizing Information for Public Health Genomics: "4 Phases of Translation"



Organizing Information for Public Health Genomics: "4 Phases of Translation" continues



Organizing Information for Public Health Genomics: "4 Phases of Translation" continues…



Tier Table Database
Why did we build it?

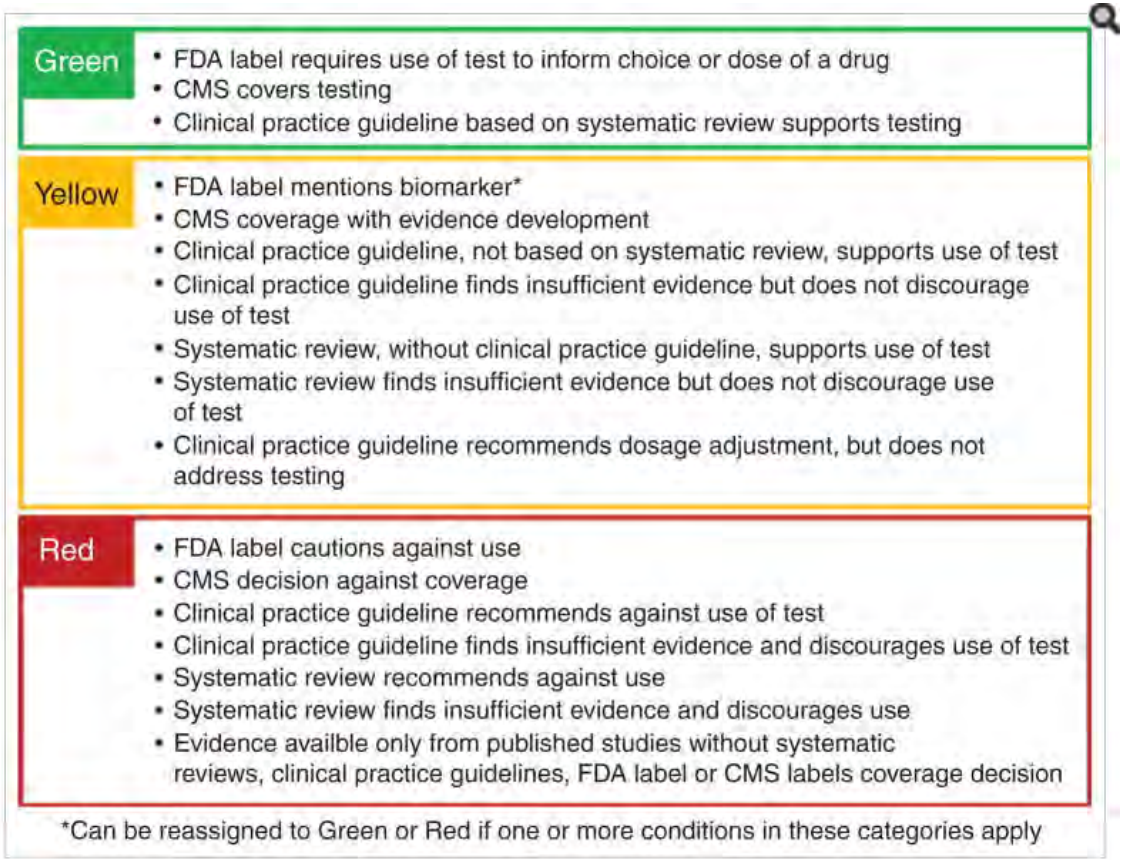
- ***Challenge:*** *The public and health care providers are bombarded with information on genomic tests, many with unproven utility*
- **Opportunity:** Educate providers and the public about potential benefits and harms of genomic tests and the need for evidence
- ***Challenge:*** *There is no widely 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

Tier 1:

Tier 2:

Tier 3:



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
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
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 availble 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

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	Detail
Osteoporosis	Parental history of hip fracture	Estimate fracture risk to inform osteoporosis screening	Tier 1	Detail
Familial hypercholesterolemia (FH)	DNA testing and LDL-C concentration measurement	Cascade testing of relatives of people diagnosed with FH	Tier 1	Detail
Hereditary breast and ovarian cancer	Family history of known breast/ovarian cancer with deleterious BRCA mutation	Risk prediction; referral to counseling for BRCA genetic testing	Tier 1	Detail
Lynch syndrome	Various strategies	Screening, cascade testing of relatives	Tier 1	Detail

State Public Health Genomics Programs 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

State Public Health Genomics Program Map

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 Public Health Genomics Program Map

Advanced Molecular Detection (AMD) Clips Database

Why did we build it?

- ***Challenge:*** *Genomic technology is transforming the work of CDC and other public health laboratories in infectious diseases.*
- **Opportunity:** Help public health labs keep abreast of this fast-moving field by providing a curated selection of scientific articles and resources.
- ***Challenge:*** *CDC laboratories are moving forward on many different fronts through the AMD Program.*
- **Opportunity:** Showcase work by CDC laboratories and allow quick look-up in a cumulative database.

HuGE Navigator
Why did we build it?

- ***Challenge:*** *The scientific literature on human genetic associations with disease has been growing rapidly. Epidemiologic data are key for evaluating clinical validity but are not easily identified by PubMed queries.*
- **Opportunity:** Use machine learning and automated indexing to maintain a searchable database of PubMed articles on gene-disease associations.
- ***Challenge:*** *Examine relationships from either genotype or phenotype (disease) perspective*
- **Opportunity:** Build navigation tools using PubMed structures and data.

Centers for Disease Control and Prevention
CDC 24/7: Saving Lives. Protecting People.™

SEARCH

CDC A-Z INDEX

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All Databases +
DataSet Download Center
Release Note

Recommend Tweet Share

Search PHGKB: Family history Search

What's New

Last Posted: *Feb 01, 2018*

- [Genetic counselling is relevant in familial as well as sporadic cases of amyotrophic lateral sclerosis].
  Lindquist Suzanne Granhøj et al. Ugeskrift for laeger 2014 Oct 176(43)
- Colorectal Cancer Screening and Surveillance in Individuals at Increased Risk.
  Wilkins Thad et al. American family physician 2018 Jan 97(2) 111-116
- Communicating with Daughters About Familial Risk of Breast Cancer: Individual, Family, and Provider Influences on Women's Knowledge of Cancer Risk.
  Peipins Lucy A et al. Journal of women's health (2002) 2018 Jan
- Genes and genetics in eye diseases: a genomic medicine approach for investigating hereditary and inflammatory ocular disorders.

Information in Specialized Databases

CDC Information (16) | NIH Resources (6) | CDC-Authored Publications (14) | State Public Health Genomics Programs (93)

Tier Table (4) | Epidemiologic Studies (3018) | Translation/Implementation Studies (681) | Evidence Synthesis (59) | Guidelines (25)

Reviews/Commentaries (113) | Tools/Methods (14) | Ethical/Legal and Social Issues (3) | GWAS Catalog (18)

From CDC Information Database

This database includes general CDC public health information on specific diseases and health related topics. When available, the database displays genomic information from various CDC web pages. Users are also encouraged to conduct searches of CDC website for additional information.

- Colorectal Cancer Family History
  CDC YouTube Video
- Six Tips for Healthy Aging Include Knowing Your Family History
- Family History and Other Characteristics That Increase Risk for High Cholesterol
- Preventing Suicide
  Risk factors include family history of suicide
- Don't Let Glaucoma Steal Your Sight!
  Family history is a risk factor

more

Information in Related Databases

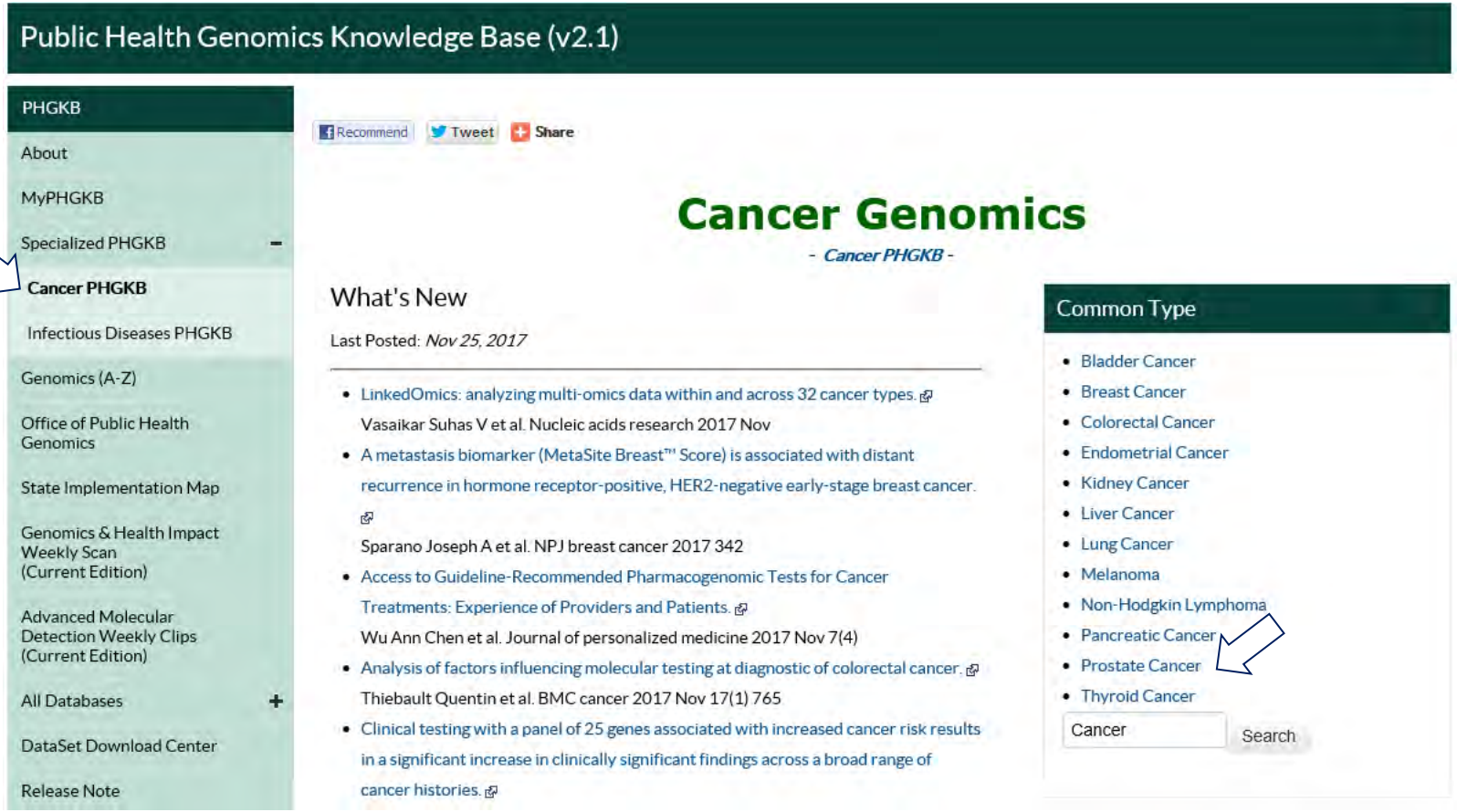
Genetic Testing (GTR) | Genetic Disease (OMIM) | PubMed Review | PubMed Clinical Queries | PharmGKB

From NIH The Genetic Testing Registry

The Genetic Testing Registry (GTR®) provides a central location for voluntary submission of genetic test information by providers. The scope includes the test's purpose, methodology, validity, evidence of the test's usefulness, and laboratory contacts and credentials. The overarching goal of the GTR is to advance the public health and research into the genetic basis of health and disease.

- Search the Genetic Testing Registry for "Family history"

Specialized PHGKB Databases: Cancer

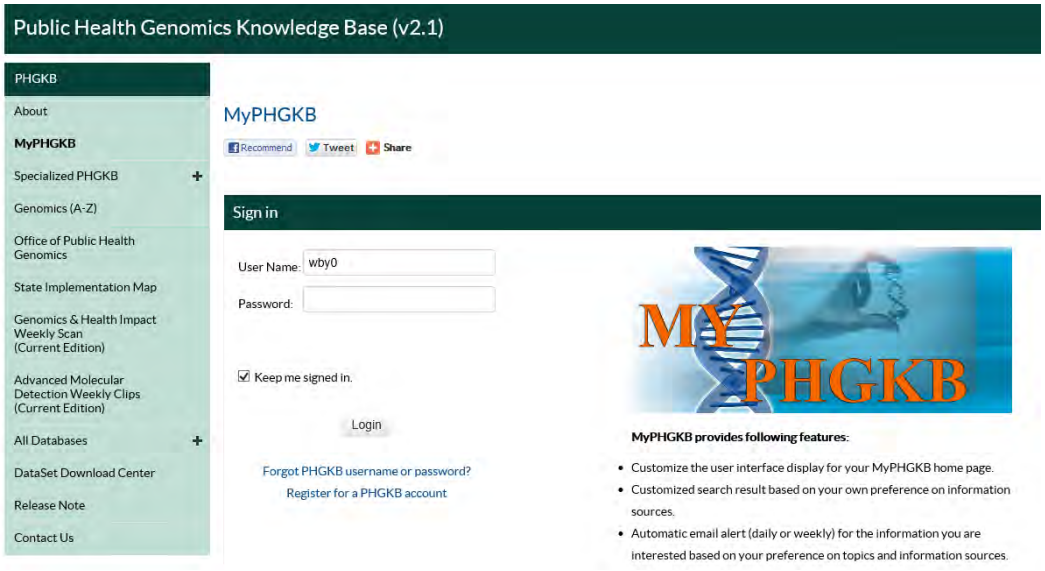


MyPHGKB – A Special Informatics Tool in PHGKB

- Customize the user interface display for your MyPHGKB home page.
- Customized search result based on your own preference on information sources.
- Automatic email alerts for the information you are interested based on your preference on topics and information sources.

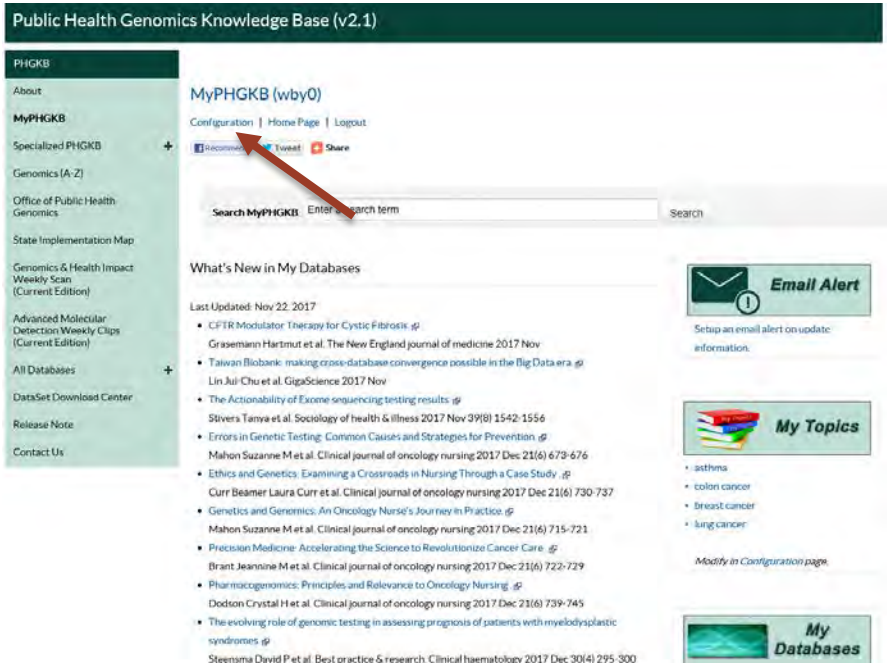
MyPHGKB – A Special Informatics Tool in PHGKB continues

- Create an account with your email address information only



MyPHGKB – A Special Informatics Tool in PHGKB continues ..

- Configuration



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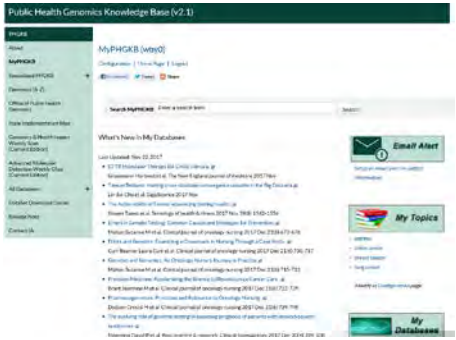
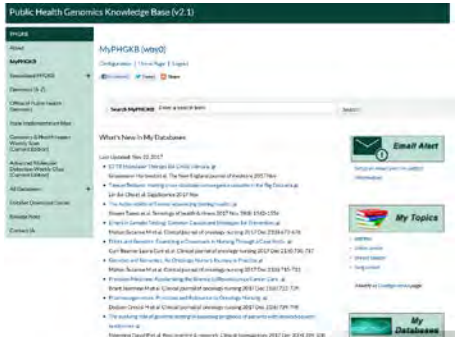


MyPHGKB – A Special Informatics Tool in PHGKB continues ….

- Automatic email alerts for the information you are interested based on your preference on topics and information sources.
  - Daily hot topics picked by experts
  - Daily or weekly alert based on your configuration.



MyPHGKB – A Special Informatics Tool in PHGKB continues …..

Mon 11/27/2017 10:00 PM

noreply@cdc.gov

Hot Topics of the Day from MyPHGKB (November 27, 2017)

To Yu, Wei (CDC/OPHSS/CSELS/DPHID)

Dear wby0:

The Hot Topics: Expert picks on public health and genomic insights into specific diseases and health related topics. Sources include published scientific literature, reviews, blogs and popular press articles on current newsworthy genomics discoveries and potential applications for policy and practice.

Personal Genomics

- Genome Culture: Welcoming in a New Era of Direct-to-Consumer Genetic Testing
  L Hercher, Genome Magazine, Nov 24, 2017

Alzheimer's Disease

- Alzheimer's Disease Fact Sheet
  Risk factors for Alzheimer's disease include genetics and family history

Chronic Obstructive Pulmonary Disease

- National COPD Awareness Month

Genome Editing

- The FDA says it's illegal to sell do-it-yourself kits to edit human genes. But what, exactly, does that mean?
  I Swetlitz, StatNews Plus, Nov 24, 2017

Autism

- Elevated polygenic burden for autism is associated with differential DNA methylation at birth.
  E Hannon et al, BioRXIV, Nov 26, 2017
- Common risk variants identified in autism spectrum disorder
  J Grove et al, BioRXIV, Nov 25, 2017

MyPHGKB – A Special Informatics Tool in PHGKB continues ……

noreply@cdc.gov Yu, Wei (CDC/OPHSS/CSELS/DPHID)

New Information Weekly Alert From MyPHGKB

Dear wby0:

Articles in this email alert were added to the Public Health Knowledge Base (PHGKB) this week and sent based on your preference setting on MyPHGKB.

Asthma

- Treatment response heterogeneity in asthma: the role of genetic variation.
  Vijverberg Susanne J H et al. Expert review of respiratory medicine 2017 Nov *(From Genomics & Health Impact Scan Database)*
- Pharmacogenetic and pharmacogenomic considerations of asthma treatment.
  Matera Maria Gabriella et al. Expert opinion on drug metabolism & toxicology 2017 Nov 13(11) 1159-1167 *(From Genomics & Health Impact Scan Database)*

Lynch syndrome

- Immunohistochemical Pitfalls: Common Mistakes in the Evaluation of Lynch Syndrome.
  Markow Michael et al. Surgical pathology clinics 2017 Dec 10(4) 977-1007 *(From Genomics & Health Impact Scan Database)*
- Universal determination of microsatellite instability using BAT26 as a single marker in an Argentine colorectal cancer cohort.
  González María Laura et al. Familial cancer 2017 Nov *(From Genomics & Health Impact Scan Database)*
- Mismatch Repair Deficiency Testing in Patients With Colorectal Cancer and Nonadherence to Testing Guidelines in Young Adults.
  Shaikh Talha et al. JAMA oncology 2017 Nov e173580 *(From Genomics & Health Impact Scan Database)*
- Risk of colorectal cancer for carriers of a germ-line mutation in POLE or POLD1.
  Buchanan Daniel D et al. Genetics in medicine : official journal of the American College of Medical Genetics 2017 Nov *(From Genomics & Health Impact Scan Database)*
- An NRG Oncology/GOG study of molecular classification for risk prediction in endometrioid endometrial cancer.
  Cosgrove Casey M et al. Gynecologic oncology 2017 Nov *(From Genomics & Health Impact Scan Database)*

Breast cancer

- Budget impact analysis of gene expression tests to aid therapy decisions for breast cancer patients in Germany.
  Lux M P et al. Breast (Edinburgh, Scotland) 2017 Nov 3789-98 *(From Genomics & Health Impact Scan Database)*
- Considerations in Testing for Inherited Breast Cancer Predisposition in the Era of Personalized Medicine.
  Powers Benjamin et al. Surgical oncology clinics of North America 2018 Jan 27(1) 1-22 *(From Genomics & Health Impact Scan Database)*
- Impact of ABCB1 and CYP2D6 polymorphisms on tamoxifen treatment outcomes and adverse events in breast cancer patients.

MyPHGKB – A Special Informatics Tool in PHGKB continues……

- User Management

For more Information

https://phgkb.cdc.gov

https://www.cdc.gov/genomics

genetics@cdc.gov

For more information, contact CDC
1-800-CDC-INFO (232-4636)
TTY: 1-888-232-6348 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.