An Introduction to CDC Information and Resources for Translation and Implementation in Genomic Medicine to Improve the Public's Health

Muin J. Khoury

CDC Office of Public Health Genomics

Outline

- What is PHGKB?
- What is GPHAC?
- Healthcare-Public Health Collaboration

Genetics inMedicine | BRIEF REPORT

A knowledge base for tracking the impact of genomics on population health

Wei Yu, MS, PhD[1], Marta Gwinn, MD, MPH[1,2], W. David Dotson, PhD[1], Ridgely Fisk Green, MS, PhD[1,3], Mindy Clyne, MHS[4,5], Anja Wulf, BA[1,6], Scott Bowen, MPH[1], Katherine Kolor, MS, PhD[1] and Muin J. Khoury, MD, PhD[1]

Purpose: We created an online knowledge base (the Public Health Genomics Knowledge Base (PHGKB)) to provide systematically curated and updated information that bridges population-based research on genomics with clinical and public health applications.

Methods: Weekly horizon scanning of a wide variety of online resources is used to retrieve relevant scientific publications, guidelines, and commentaries. After curation by domain experts, links are deposited into Web-based databases.

Results: PHGKB currently consists of nine component databases. Users can search the entire knowledge base or search one or more component databases directly and choose options for customizing the display of their search results.

Conclusion: PHGKB offers researchers, policy makers, practitioners, and the general public a way to find information they need to understand the complicated landscape of genomics and population health.

Genet Med advance online publication 9 June 2016

INTRODUCTION

Genomic information is increasingly finding its way into clini-

"lack of readily accessible information about the utility of most genomic applications and the lack of necessary knowledge by

Genomics and **P**opulation **H**ealth **A**ction **C**ollaborative
NASEM Roundtable on Genomics and Precision Health

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

Public Health Genomics

Routine Genomic Screening in Health Care? New CDC-NIH Paper and Blog Post

Events

2018 PUBLIC HEALTH GENOMICS WEBINARS

FAMILIAL HYPERCHOLESTEROLEMIA

WHAT IS PRECISION PUBLIC HEALTH?

PRENATAL TESTING AND GENOMICS

WEEKLY UPDATE
Weekly summary of genomics and health impact information

GENETICS 101
Genetics basics explained including a glossary of genetic terms

PHGKB
Online searchable knowledge base on genomics and health impact information

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

REPORTS AND PUBLICATIONS
Scientific reports and publications in public health genomics

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

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

MyPHGKB
provides personalized features based on your preferences of databases and topics
VISIT MyPHGKB

WEEKLY UPDATE
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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

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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

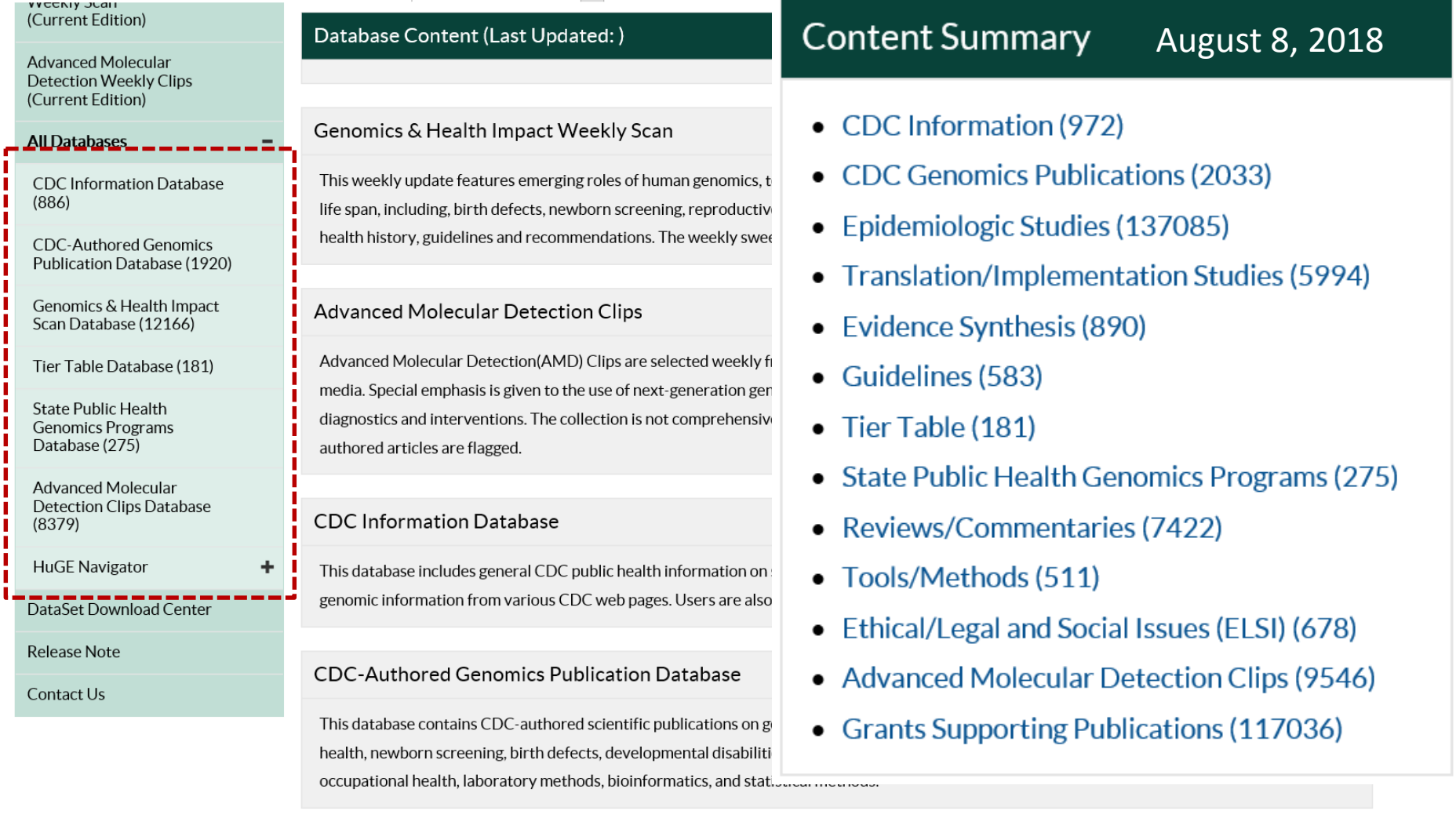
Study Finds Biological Differences in Lung Tumors of African Americans and Whites
NCI Blog, Jan 2018

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
DataSet Download Center
Release Note
Contact Us
Database Content (Last Updated:)
Genomics & Health Impact Weekly Scan
This weekly update features emerging roles of human genomics, t
life span, including, birth defects, newborn screening, reproductiv
health history, guidelines and recommendations. The weekly swe
Advanced Molecular Detection Clips
Advanced Molecular Detection(AMD) Clips are selected weekly f
media. Special emphasis is given to the use of next-generation ge
diagnostics and interventions. The collection is not comprehensiv
authored articles are flagged.
CDC Information Database
This database includes general CDC public health information on
genomic information from various CDC web pages. Users are also
CDC-Authored Genomics Publication Database
This database contains CDC-authored scientific publications on g
health, newborn screening, birth defects, developmental disabiliti
occupational health, laboratory methods, bioinformatics, and stat
Content Summary August 8, 2018
CDC Information (972)
CDC Genomics Publications (2033)
Epidemiologic Studies (137085)
Translation/Implementation Studies (5994)
Evidence Synthesis (890)
Guidelines (583)
Tier Table (181)
State Public Health Genomics Programs (275)
Reviews/Commentaries (7422)
Tools/Methods (511)
Ethical/Legal and Social Issues (ELSI) (678)
Advanced Molecular Detection Clips (9546)
Grants Supporting Publications (117036)

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?

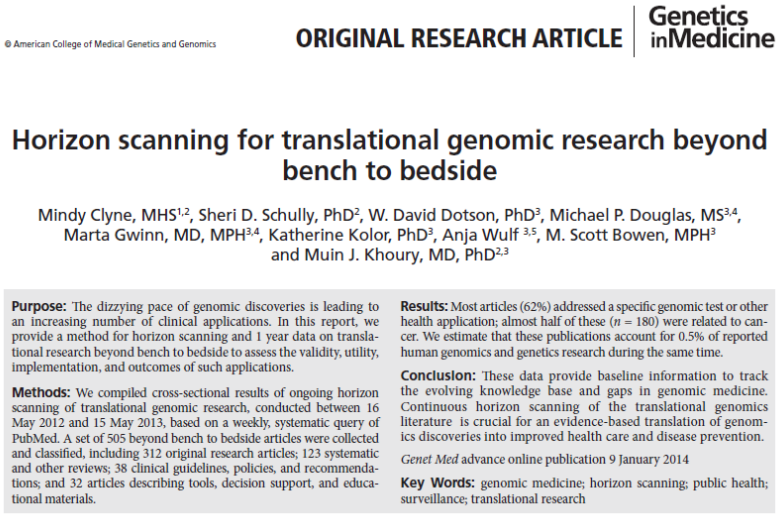
- ***Challenge:*** *Finding CDC-authored publications on genomics and family health history*
- **Opportunity:** Provide a centralized, searchable, publicly available database for these CDC publications
- ***Challenge:*** *CDC's work in genomics and family health history is not well known*
- **Opportunity:**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

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, etc)
  - Guidelines
  - Tools/Methods
  - Reviews/Commentaries



ORIGINAL RESEARCH ARTICLE | Genetics inMedicine

Horizon scanning for translational genomic research beyond bench to bedside

Mindy Clyne, MHS[1,2], Sheri D. Schully, PhD[2], W. David Dotson, PhD[3], Michael P. Douglas, MS[3,4], Marta Gwinn, MD, MPH[3,4], Katherine Kolor, PhD[3], Anja Wulf [3,5], M. Scott Bowen, MPH[3] and Muin J. Khoury, MD, PhD[2,3]

Purpose: The dizzying pace of genomic discoveries is leading to an increasing number of clinical applications. In this report, we provide a method for horizon scanning and 1 year data on translational research beyond bench to bedside to assess the validity, utility, implementation, and outcomes of such applications.

Methods: We compiled cross-sectional results of ongoing horizon scanning of translational genomic research, conducted between 16 May 2012 and 15 May 2013, based on a weekly, systematic query of PubMed. A set of 505 beyond bench to bedside articles were collected and classified, including 312 original research articles; 123 systematic and other reviews; 38 clinical guidelines, policies, and recommendations; and 32 articles describing tools, decision support, and educational materials.

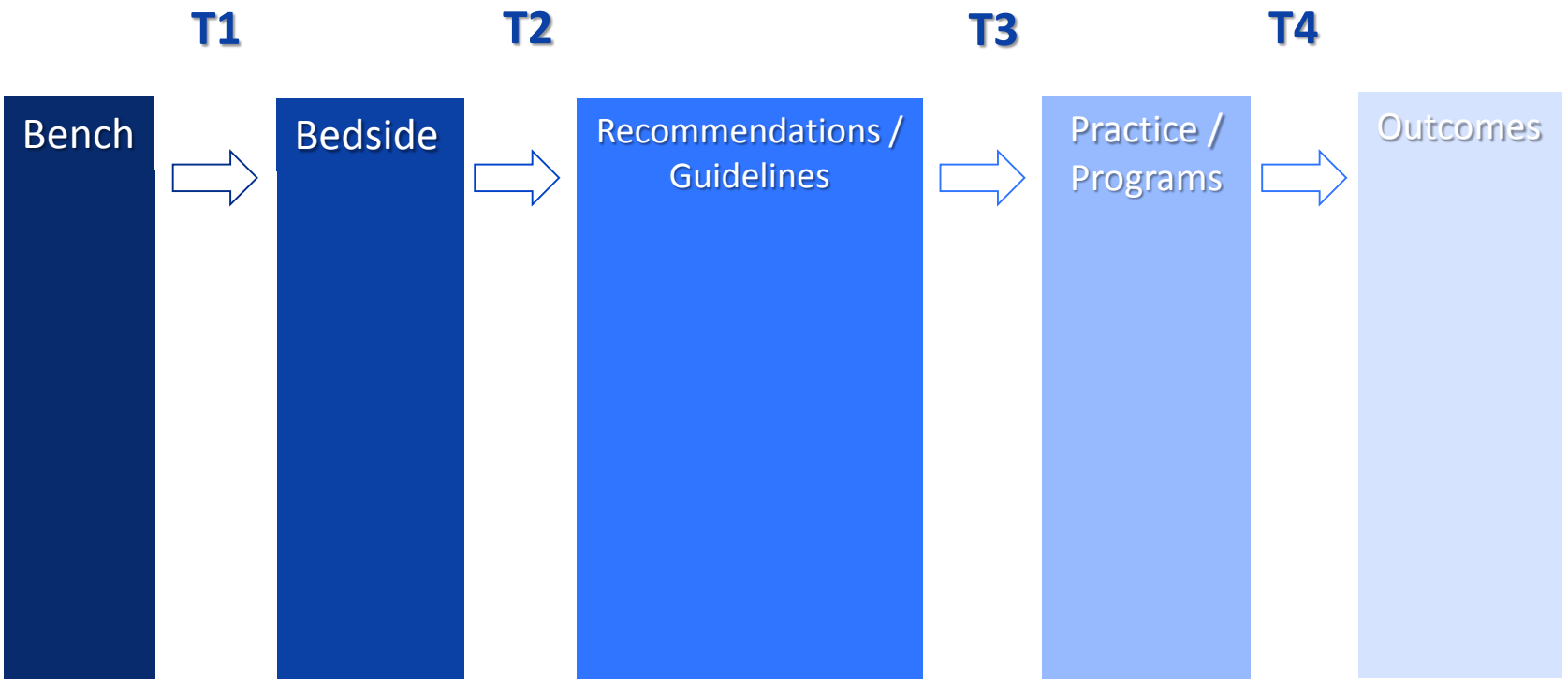
Results: Most articles (62%) addressed a specific genomic test or other health application; almost half of these ($n = 180$) were related to cancer. We estimate that these publications account for 0.5% of reported human genomics and genetics research during the same time.

Conclusion: These data provide baseline information to track the evolving knowledge base and gaps in genomic medicine. Continuous horizon scanning of the translational genomics literature is crucial for an evidence-based translation of genomics discoveries into improved health care and disease prevention.

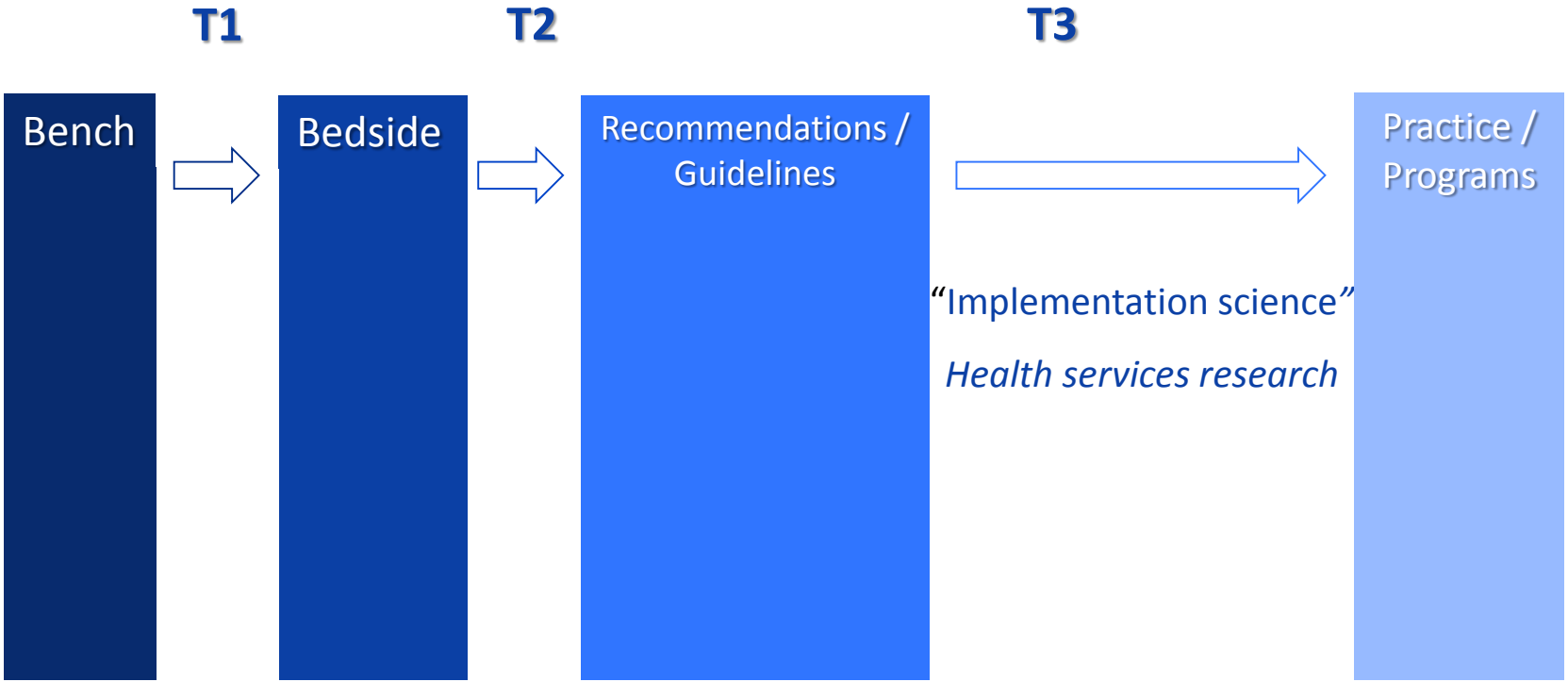
Genet Med advance online publication 9 January 2014

Key Words: genomic medicine; horizon scanning; public health; surveillance; translational research

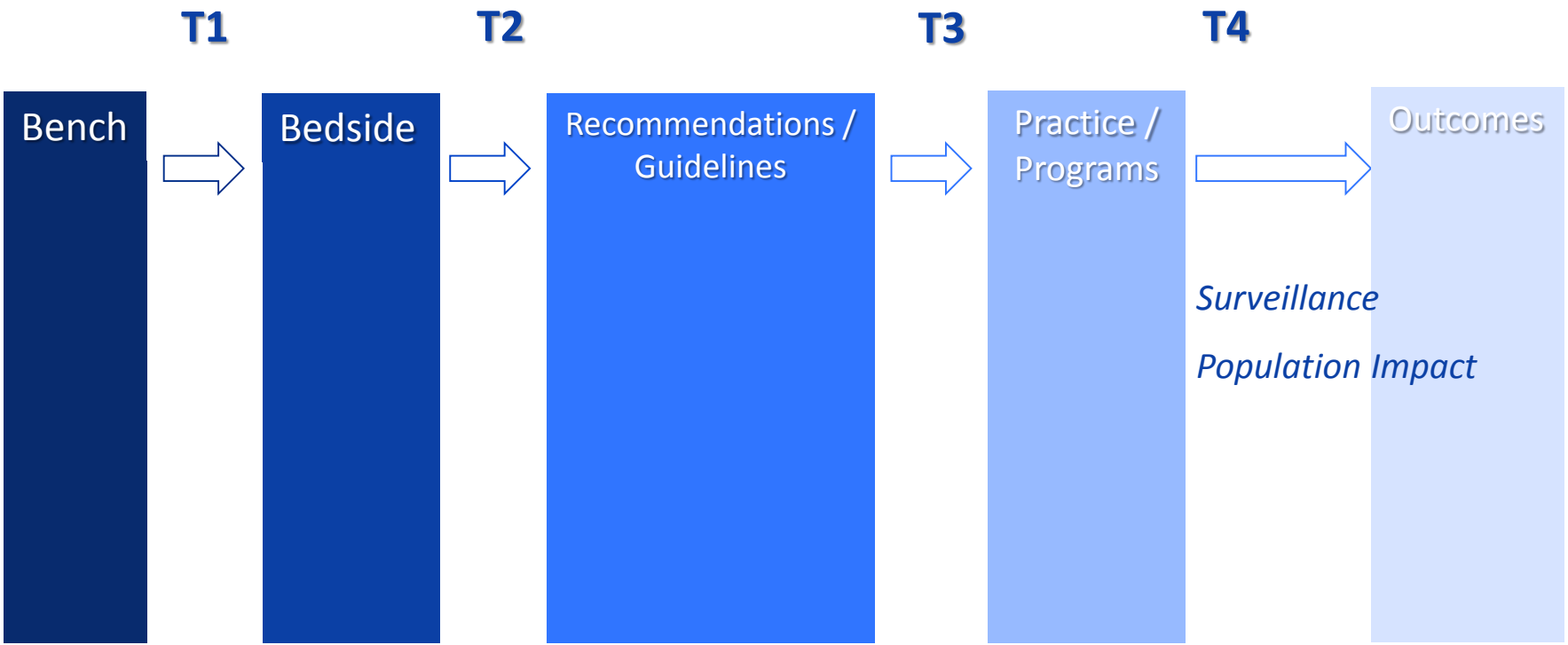
Organizing Information for Public Health Genomics: "4 Phases of Translation"



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



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



Tier Table Database
Why did we build it?

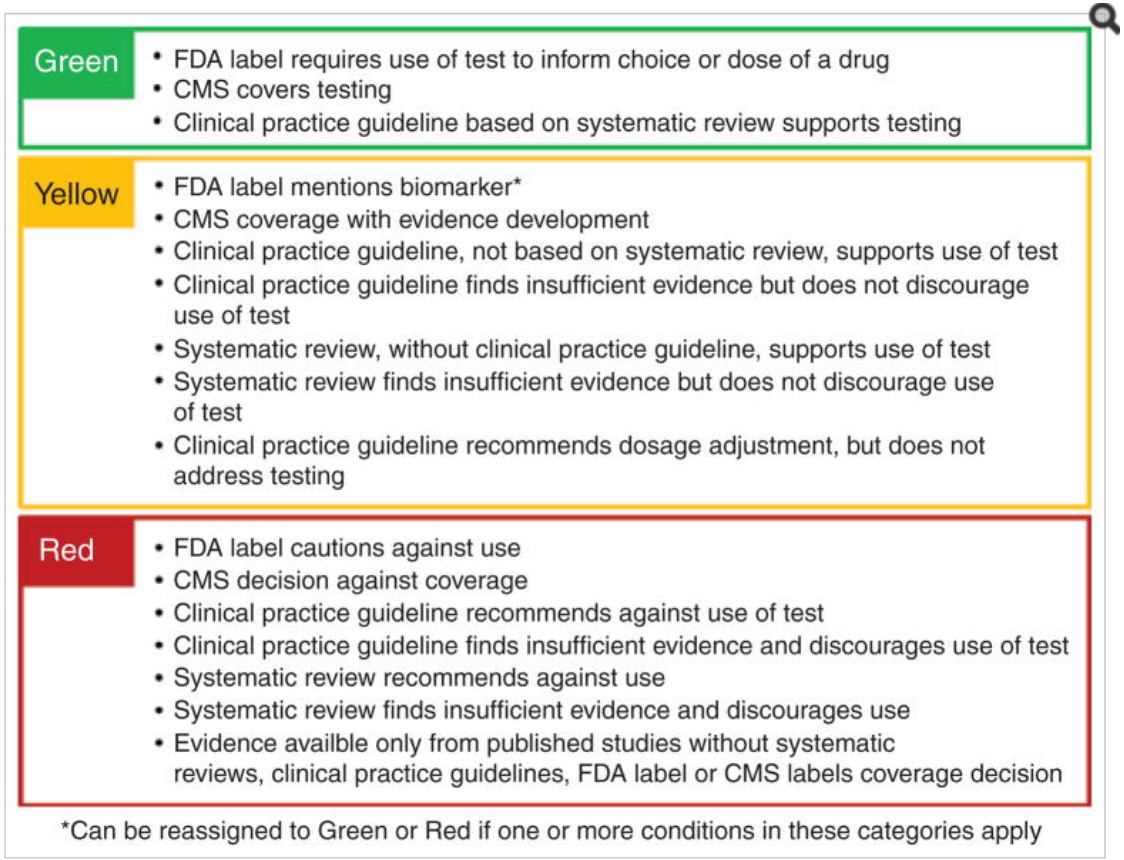
- ***Challenge:*** *The public and health care providers are bombarded with information on genomic tests, many with unproven utility*
- **Opportunity:** Educate providers and the public about potential benefits and harms of genomic tests and the need for evidence
- ***Challenge:*** *There is no widely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

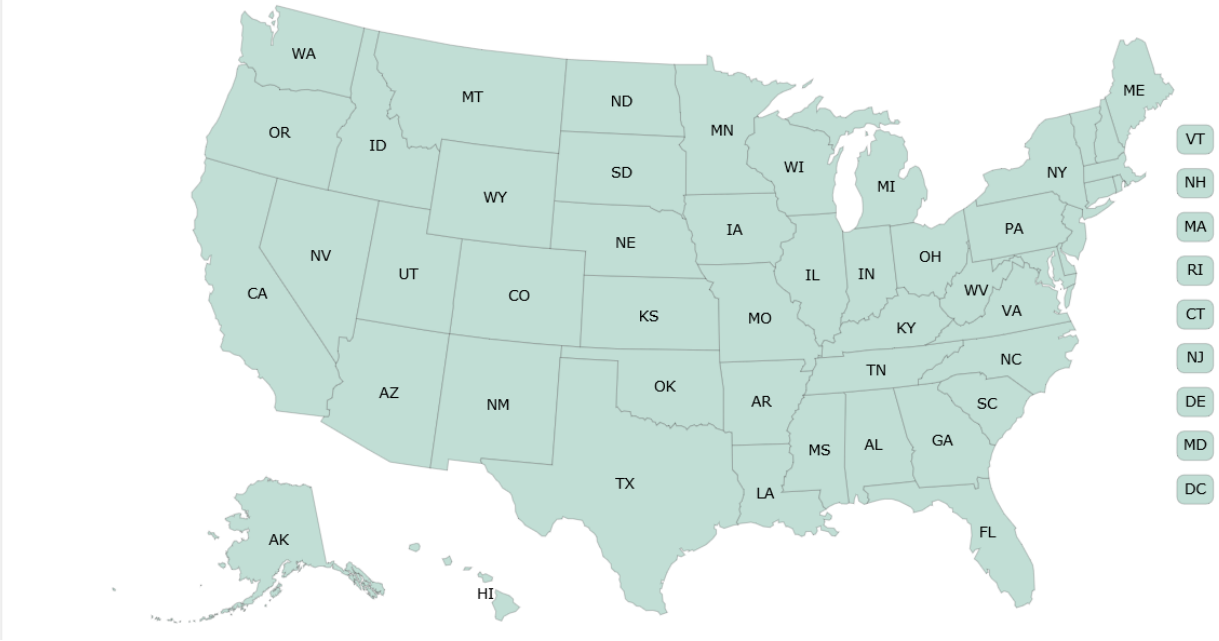
State Public Health Genomics Programs Database
Why did we build it?

- ***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

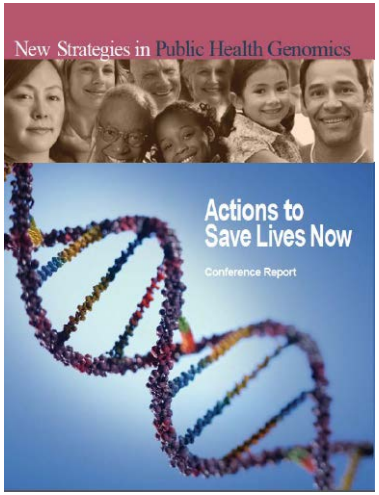


State Public Health Genomics Programs Database
CDC Funding to States Over Time

- 2003-2008: Michigan, Minnesota, Oregon, and Utah
- 2008-2011: Michigan and Oregon
- 2011-2014: Georgia, Michigan, and Oregon
- 2014-2019: Colorado, Connecticut, Michigan, Oregon, and Utah
  - Enhancing Cancer Genomic Best Practices through Education, Surveillance, and Policy
  - Goal: Provide leadership and build capacity for cancer genomics activities in state public health departments

State Public Health Genomics Programs Database

CDC Tier 1 Priorities and Toolkit

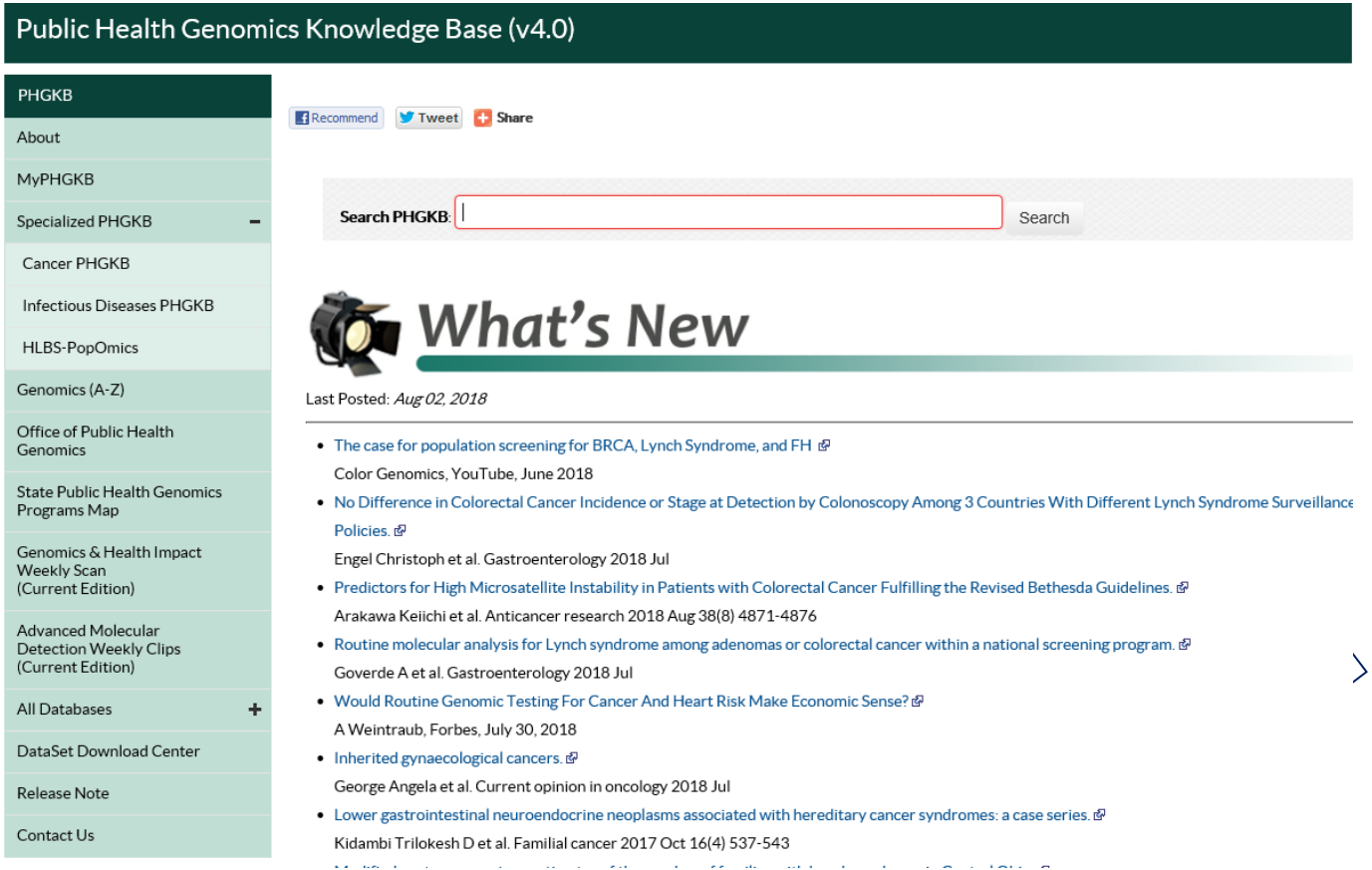


- Hereditary Breast and Ovarian Cancer (*BRCA1/2*)
- Hereditary Nonpolyposis Colorectal Cancer (Lynch Syndrome)
- Familial Hypercholesterolemia
- **Collectively Affect ~3 Million People in US and Most Don't know it.**
- Implementation of existing evidence-based guidelines can prevent cancer & heart disease, & save thousands of lives every year!
- Toolkit for public health departments

Public Health Genomics Grants Database
Why did we build it?

- ***Challenge: The translation pathway from genome discoveries to improved health requires funding. It is not clear what translational research and implementation science is been funded*** *and by whom?*
- **Opportunity:** Provide a searchable database of available grant information connected with PHGKB related publications. Grant information can be searched by disease/condition, environmental risk factors, or gene of study and other free text.
- Still work in progress- Currently connecting to NIH reporter and other sources

Specialized PHGKB Databases: Cancer, HLBS, Infectious Diseases and MyPHGKB



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.

Searching PHGKB: An Example

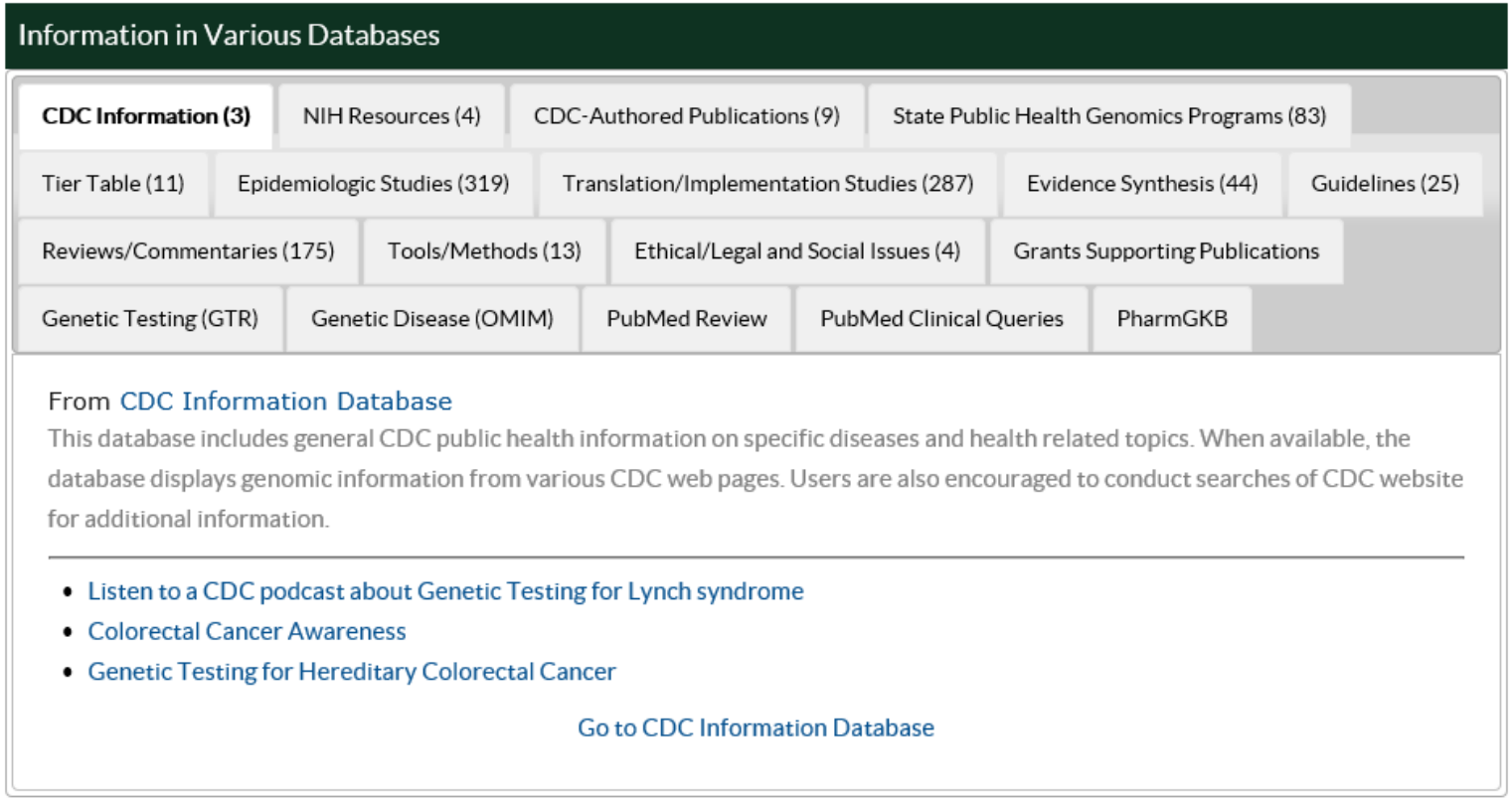
Search PHGKB: Lynch syndrome Search

What's New

Last Posted: *Aug 02, 2018*

- The case for population screening for BRCA, Lynch Syndrome, and FH
  Color Genomics, YouTube, June 2018
- No Difference in Colorectal Cancer Incidence or Stage at Detection by Colonoscopy Among 3 Countries With Different Lynch Syndrome Surveillance Policies.
  Engel Christoph et al. Gastroenterology 2018 Jul
- Predictors for High Microsatellite Instability in Patients with Colorectal Cancer Fulfilling the Revised Bethesda Guidelines.
  Arakawa Keiichi et al. Anticancer research 2018 Aug 38(8) 4871-4876
- Routine molecular analysis for Lynch syndrome among adenomas or colorectal cancer within a national screening program.
  Goverde A et al. Gastroenterology 2018 Jul
- Would Routine Genomic Testing For Cancer And Heart Risk Make Economic Sense?
  A Weintraub, Forbes, July 30, 2018
- Inherited gynaecological cancers.
  George Angela et al. Current opinion in oncology 2018 Jul
- Lower gastrointestinal neuroendocrine neoplasms associated with hereditary cancer syndromes: a case series.
  Kidambi Trilokesh D et al. Familial cancer 2017 Oct 16(4) 537-543
- Modified capture-recapture estimates of the number of families with Lynch syndrome in Central Ohio.
  Ranola John Michael O et al. Familial cancer 2018 Jul

Searching PHGKB: An Example:



An overview of grant information associated with 3 CDC tier 1 genomic applications, by country, agency and associated publications

* Analysis conducted using grants database on March 6, 2018

Condition	Familial Hyper-Cholesterolemia No (%)	BRCA (HBOC) No (%)	Lynch Syndrome No (%)
Number of Grants	239	796	421
Number of Funders	24	42	32
Country			
USA	115 (48%)	591 (74%)	322 (76%)
UK	94 (39%)	179 (23%)	89 (22%)
Other	30 (13%)	26 (3%)	10 (2%)
Funding			
NHLBI or NCI	73 (NHLBI)	472 (NCI)	239 (NCI)
Other NIH	29	85	48
Total NIH	102 (43%)	557 (70%)	287 (57%)
Non-NIH	137 (57%)	239 (30%)	134 (43%)
Associated Publications			
Total	55 (100%)	226 (100%)	161(100%)
Epidemiology	28 (48%)	140 (62%)	58 (36%)
Translation/ Implementation	14 (26%)	74 (33%)	69 (43%)
Evidence Synthesis/ Guidelines	7 (13%)	6 (2.5%)	13 (8%)
Other	5 (9%)	6 (2.5%)	20 (13%)

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Genomics and **P**opulation **H**ealth **A**ction **C**ollaborative
NASEM Roundtable on Genomics and Precision Health

National Academies of Science, Engineering & Medicine Genomics and Population Health Action Collaborative

Genomics and Population Health Action Collaborative[1]

Building the Evidence Base for Genomics in Public Health: Implications for Decision Making, Public Policy, and Population Health Planning

EVIDENCE WORKING GROUP MEMBERS

Chair: Ned Calonge, The Colorado Trust

- Naomi Aronson, Blue Cross Blue Shield Association
- Benjamin Djulbegovic, University of South Florida
- Dave Dotson, CDC Office of Public Health Genomics
- Jim Evans, University of North Carolina at Chapel Hill
- Heather Hampel, Ohio State University
- George Isham, HealthPartners
- Cecile Janssens, Emory University
- Muin Khoury, CDC Office of Public Health Genomics
- Roger Klein, Cleveland Clinic
- Jeanne Mandelblatt, Georgetown University
- Doug Campos Outcalt, Mercy Care Plan
- Sun Hee Rim, CDC Division of Cancer Prevention and Control
- Wendy Rubinstein, NIH Genetic Testing Registry
- Sheri Schully, NIH Office of Disease Prevention

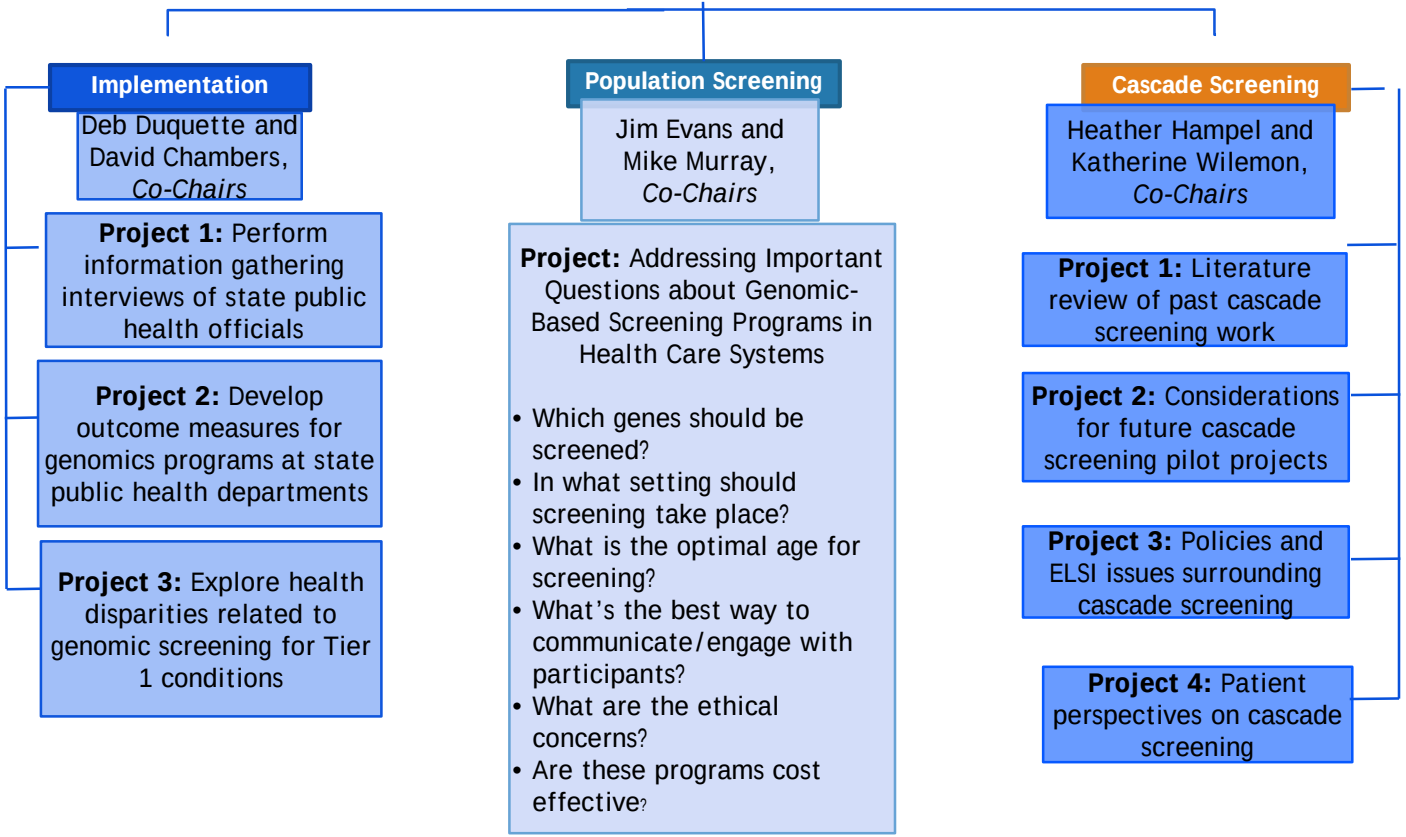
- Goal
  - Explore ways to integrate evidence-based genomic applications into population health programs at the health care/public health interface

Focus on Lynch syndrome, HBOC and FH as prototypes (CDC Tier 1s)

Output/Deliverables

- 2017: Report on Evidence-based Genomics in Public Health
- 2018: Add to PHGKB Online guide/toolkit for states interested in implementing PH genomics
- 2019: Extend collaborations to health systems

Genomics and Population Health Action Collaborative – 2018



GPHAC Public Health Genomics Implementation: Outcome Metrics

ORIGINAL RESEARCH ARTICLE | Genetics inMedicine

Proposed outcomes measures for state public health genomic programs

Debra Lochner Doyle, MS, LCGC[1], Mindy Clyne, MHS, CGC[2], Juan L. Rodriguez, MPH, MS[3], Deborah L. Cragun, PhD, MS[4], Laura Senier, MPH, PhD[5], Georgia Hurst[6], Kee Chan, PhD[7] and David A. Chambers, DPhil[2]

Purpose: To assess the implementation of evidence-based genomic medicine and its population-level impact on health outcomes and to promote public health genetics interventions, in 2015 the Roundtable on Genomics and Precision Health of the National Academies of Sciences, Engineering, and Medicine formed an action collaborative, the Genomics and Public Health Action Collaborative (GPHAC). This group engaged key stakeholders from public/population health agencies, along with experts in the fields of health disparities, health literacy, implementation science, medical genetics, and patient advocacy.

Methods: In this paper, we present the efforts to identify performance objectives and outcome metrics. Specific attention is placed on measures related to hereditary breast ovarian cancer (HBOC) syndrome and Lynch syndrome (LS), two conditions with existing evidence-based genomic applications that can have immediate impact on morbidity and mortality.

Results: Our assessment revealed few existing outcome measures. Therefore, using an implementation research framework, 38 outcome measures were crafted.

Conclusion: Evidence-based public health requires outcome metrics, yet few exist for genomics. Therefore, we have proposed performance objectives that states might use and provided examples of a few state-level activities already under way, which are designed to collect outcome measures for HBOC and LS.

Genet Med advance online publication 4 January 2018

Key Words: hereditary breast and ovarian cancer; implementation science; Lynch syndrome; outcome measures; public health genomics

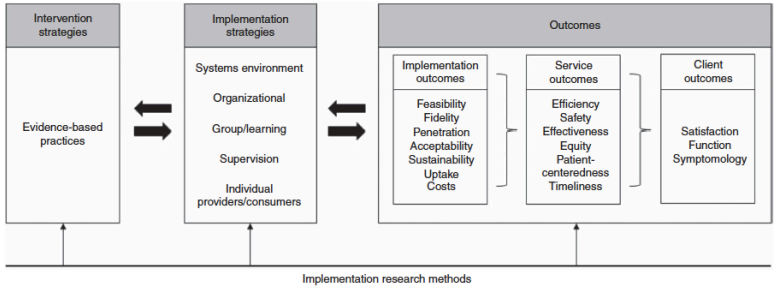


Figure 1 The implementation framework developed by Proctor et al.[2]

Genetics in Medicine, December, 2017

GPHAC Public Health Genomics Implementation: State Interviews

- State Semi-structured interviews
  - 11 states
  - States
  - with diverse funding arrangements

- ✓ Prior experiences
- ✓ Funding
- ✓ Legislation
- ✓ Staffing resources
- ✓ Influencers/champions (internal and external)
- ✓ Interest
- ✓ Training/education
- ✓ Tools/resources
- ✓ Evaluations
- ✓ Approaches/frameworks

GPHAC Public Health Genomics Implementation: Cascade Screening

By Megan Roberts, W. David Dotson, Christopher S. DeVore, Erica M. Bednar, Deborah J. Bowen, Theodore G. Ganiats, Ridgely Fisk Green, Georgia M. Hurst, Alisdair R. Philp, Charité N. Ricker, Amy C. Sturm, Angela M. Trepanier, Janet L. Williams, Heather A. Zierhut, Katherine A. Wilemon, and Heather Hampel

Delivery Of Cascade Screening For Hereditary Conditions: A Scoping Review Of The Literature

ABSTRACT Cascade screening is the process of contacting relatives of people who have been diagnosed with certain hereditary conditions. ~~The~~ purpose is to identify, inform, and manage those who are also at risk. We conducted a scoping review to obtain a broad overview of cascade screening interventions, facilitators and barriers to their use, relevant policy considerations, and future research needs. We searched for relevant peer-reviewed literature in the period 1990–2017 and reviewed 122 studies. Finally, we described 45 statutes and regulations related to the use and release of genetic information across the fifty states. We sought standardized best practices for optimizing cascade screening across various geographic and policy contexts, but we found none. Studies in which trained providers contacted relatives directly, rather than through probands (index patients), showed greater cascade screening uptake; however, policies in some states might limit this approach. Major barriers to cascade screening delivery include suboptimal communication between the proband and family and geographic barriers to obtaining genetic services. Few US studies examined interventions for cascade screening or used rigorous study designs such as randomized controlled trials. Moving forward, there remains an urgent need to conduct rigorous intervention studies on cascade screening in diverse US populations, while accounting for state policy considerations.

Health Affairs, May 2018

GPHAC Public Health Genomics Population Screening

Discussion Paper

A Guide for Considering Genomics-Based Screening Programs[1, 2]

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"The central idea of early disease detection and treatment is essentially simple. However, the path to its successful achievement (on the one hand, bringing to treatment those with previously undetected disease, and, on the other, avoiding harm to those persons not in need of treatment) is far from simple though sometimes it may appear deceptively easy." –Wilson and Jungner, Principles and Practice of Screening for Disease

Roundtable-GPHAC Inspired Essay on Population Screening, Implementation Science and Learning Health Systems

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ESSAY

A collaborative translational research framework for evaluating and implementing the appropriate use of human genome sequencing to improve health

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Citation: Khoury MJ, Feero WG, Chambers DA, Brody LE, Aziz N, Green RC, et al. (2018) A collaborative translational research framework for evaluating and implementing the appropriate use of

PLOS Medicine, August 2018

Outline

- What is PHGKB?
- What is GPHAC?
- Healthcare-Public Health Collaboration

-A natural partnership around translation and implementation
- Healthcare –public health interface
- Sharing tools and resources
- Collaborative analyses to explore gaps and needs
- Training and technical assistance

Last but Not Least,

Surgeon General's My Family Health Portrait is NOW Housed at CDC/PHGKB

https://phgkb.cdc.gov/FHH/html/index.html