An Introduction to the CDC
Public Health Genomics Knowledge Base

Marta Gwinn, Dave Dotson, Wei Yu & Ridgely Fisk Green
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
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
<table>
<thead>
<tr>
<th>WEEKLY UPDATE</th>
<th>GENETICS 101</th>
</tr>
</thead>
<tbody>
<tr>
<td>Weekly summary of genomics and health impact information</td>
<td>Genetics basics explained including a glossary of genetic terms</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>PHGKB</th>
<th>FAMILY HEALTH HISTORY</th>
</tr>
</thead>
<tbody>
<tr>
<td>Online searchable knowledge base on genomics and health impact information</td>
<td>Family health history is known to be a risk factor for most diseases</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>AMD CLIPS</th>
<th>GENOMICS AND DISEASES</th>
</tr>
</thead>
<tbody>
<tr>
<td>Weekly news and publications on pathogen genomics and bioinformatics</td>
<td>Genomics is important for many diseases of public health significance</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>IMPLEMENTATION</th>
<th>GENETIC COUNSELING</th>
</tr>
</thead>
<tbody>
<tr>
<td>What public health can do now to save lives using genomics</td>
<td>Helping to inform individuals and families about genetic risks, testing and interventions</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>REPORTS AND PUBLICATIONS</th>
<th>GENOMIC TESTING</th>
</tr>
</thead>
<tbody>
<tr>
<td>CDC reports and publications in genomics</td>
<td>Genomic tests are used in many diseases</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>GENOMICS &amp; HEALTH IMPACT BLOG</th>
<th>PATHOGEN GENOMICS</th>
</tr>
</thead>
<tbody>
<tr>
<td>A blog devoted to genomic issues in research, policy and practice</td>
<td>New tools are changing the landscape in the fight against infectious diseases</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>PODCASTS AND VIDEOCASTS</th>
<th>EPIDEMIOLOGY</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
</tr>
</tbody>
</table>
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-Authoried 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-Aauthored 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-Authorized Genomic Publications Database continued

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

T1: Bench → T2: Bedside → T3: Recommendations / Guidelines → Practice / Programs

“Implementation science”

Health services research
Organizing Information for Public Health Genomics:
“4 Phases of Translation” continues...

T1: Bench → T2: Bedside → T3: Recommendations / Guidelines → T4: Practice / Programs

- Surveillance
- Comparative effectiveness

Outcomes
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:
- **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 discourages use of test
  - Systematic review, without clinical practice guideline, supports use of test
  - Systematic review finds insufficient evidence but does not discourage use of test
  - Clinical practice guideline recommends dosage adjustment, but does not address testing

### Tier 2:
- **Red**
  - FDA label cautions against use
  - CMS decision against coverage
  - Clinical practice guideline recommends against use of test
  - Clinical practice guideline finds insufficient evidence and discourages use of test
  - Systematic review recommends against use
  - Systematic review finds insufficient evidence and discourages use
  - Evidence available only from published studies without systematic reviews, clinical practice guidelines, FDA label or CMS labels coverage decision

---

*Can be reassigned to Green or Red if one or more conditions in these categories apply*
## Examples of Tier 1 Genomic Applications

<table>
<thead>
<tr>
<th>Disease/Disorder</th>
<th>Test to be Assessed</th>
<th>Intended Use</th>
<th>Tier Classified</th>
<th>Detail</th>
</tr>
</thead>
<tbody>
<tr>
<td>31 core conditions</td>
<td>Newborn screening panel</td>
<td>Screening</td>
<td>Tier 1</td>
<td></td>
</tr>
<tr>
<td>Osteoporosis</td>
<td>Parental history of hip fracture</td>
<td>Estimate fracture risk to inform osteoporosis screening</td>
<td>Tier 1</td>
<td></td>
</tr>
<tr>
<td>Familial hypercholesterolemia (FH)</td>
<td>DNA testing and LDL-C concentration measurement</td>
<td>Cascade testing of relatives of people diagnosed with FH</td>
<td>Tier 1</td>
<td></td>
</tr>
<tr>
<td>Hereditary breast and ovarian cancer</td>
<td>Family history of known breast/ovarian cancer with deleterious BRCA mutation</td>
<td>Risk prediction; referral to counseling for BRCA genetic testing</td>
<td>Tier 1</td>
<td></td>
</tr>
<tr>
<td>Lynch syndrome</td>
<td>Various strategies</td>
<td>Screening, cascade testing of relatives</td>
<td>Tier 1</td>
<td></td>
</tr>
</tbody>
</table>
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!
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.
Public Health Genomics Knowledge Base (v2.1)

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 Granhoj et al. Ugeskrift for læger 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
- Genes and genetics in eye diseases: a genomic medicine approach for investigating hereditary and inflammatory ocular disorders.
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

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"

Disclaimer: Articles listed in the Public Health Knowledge Base are selected by the CDC Office of Public Health Genomics to provide current awareness of the literature and news. Inclusion in the update does not necessarily represent the views of the Centers for Disease Control and Prevention nor does it imply endorsement of the article's methods or findings. CDC and DHHS assume no responsibility for the factual accuracy of the items presented. The selection, omission, or content of items does not imply any endorsement or other position taken by CDC or DHHS. Opinion, findings and conclusions expressed by the original authors of items included in the update, or persons quoted therein, are strictly their own and are in no way meant to represent the opinion or views of CDC or DHHS. References to publications, news sources, and non-CDC Websites are provided solely for informational purposes and do not imply endorsement by CDC or DHHS.
Specialized PHGKB Databases: Cancer

Public Health Genomics Knowledge Base (v2.1)

Cancer Genomics

What's New

Last Posted: Nov 25, 2017

- LinkedOmics: analyzing multi-omics data within and across 32 cancer types.
  Vasakar Suhat V et al. Nucleic acids research 2017 Nov
- A metastasis biomarker (MetaSite Breast™ Score) is associated with distant recurrence in hormone receptor-positive, HER2-negative early-stage breast cancer.
  Sparano Joseph A et al. NPJ breast cancer 2017 342
  Wu Ann Chen et al. Journal of personalized medicine 2017 Nov 7(4)
- Analysis of factors influencing molecular testing at diagnosis of colorectal cancer.
  Thiebault Quentin et al. BMC cancer 2017 Nov 17(1) 765
- Clinical testing with a panel of 25 genes associated with increased cancer risk results in a significant increase in clinically significant findings across a broad range of cancer histories.

Common Type

- Bladder Cancer
- Breast Cancer
- Colorectal Cancer
- Endometrial Cancer
- Kidney Cancer
- Liver Cancer
- Lung Cancer
- Melanoma
- Non-Hodgkin Lymphoma
- Pancreatic Cancer
- Prostate Cancer
- Thyroid Cancer

Cancer

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

- Customize the user interface display for your MyPHGKB home page.
- Customized search result based on your own preference on information sources.
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.....

Dear [Name],

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 noteworthy genomic discovery and potential applications for policy and practice.

**Personal Genomics**
- **Genome Culture**: Welcoming in a New Era of Direct-to-Consumer Genetic Testing
  - Letter, 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?**
  - [Sweitz, 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
MyPHGKB – A Special Informatics Tool in PHGKB continues ......

Dear why0;

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**

- 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**

- 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 173580 (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 5789-98 (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)

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.