CDC Public Health Genomics Today -
Twenty Years in the Making…

Scott Bowen MPH
Office of Public Health Genomics

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# CDC Public Health Genomics – Twenty Years in the Making

- 1996 and the Human Genome Project: First steps and a vision for translation
- Evidence, EGAPP, and honest brokers
- Family history – early research and clear messaging
- States are where public health genomics happens
- Saving lives with PHG: the CDC PHGKB Tier Classification Table
- Today and precision public health is here

Newborn Screening
The Largest Public Health Genetics Program in the World

❑ More than 5 decades in the US started with PKU

❑ State run public health program that screens 4 million newborns every year

❑ Identifies more than 10,000 babies with 30+ genetic, metabolic & other disorders
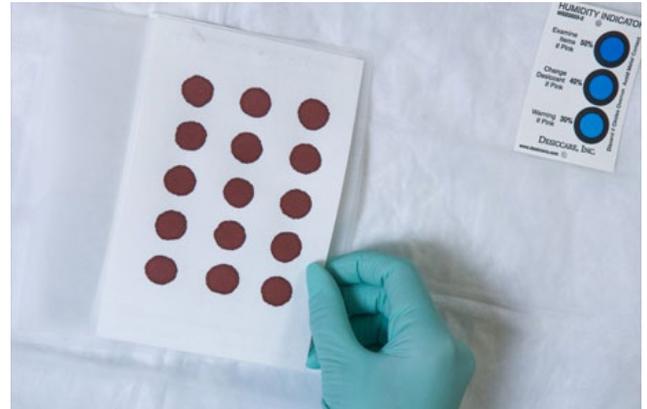
CDC Public Health Genomics: A Brief History

- 1997: CDC Strategic Plan/OPHG
- 1998: First National Conference
- 2001: PH Genomic Competencies
- 2003: CDC Model State Programs
- 2004: Family History Initiative
- 2004: EGAPP Initiative
- 2006: Seed Translation Research
    Funds to CDC Programs & Academia
- 2010: Fourth National Conference
- 2012: New CDC Strategic Plan
- 2016: CDC Concept of "Precision Public Health"

Public Health Policy Forum

From Genes to Public Health:
The Applications of Genetic
Technology in Disease Prevention

Muin J. Khoury, MD, PhD, and the Genetics Working Group

Introduction
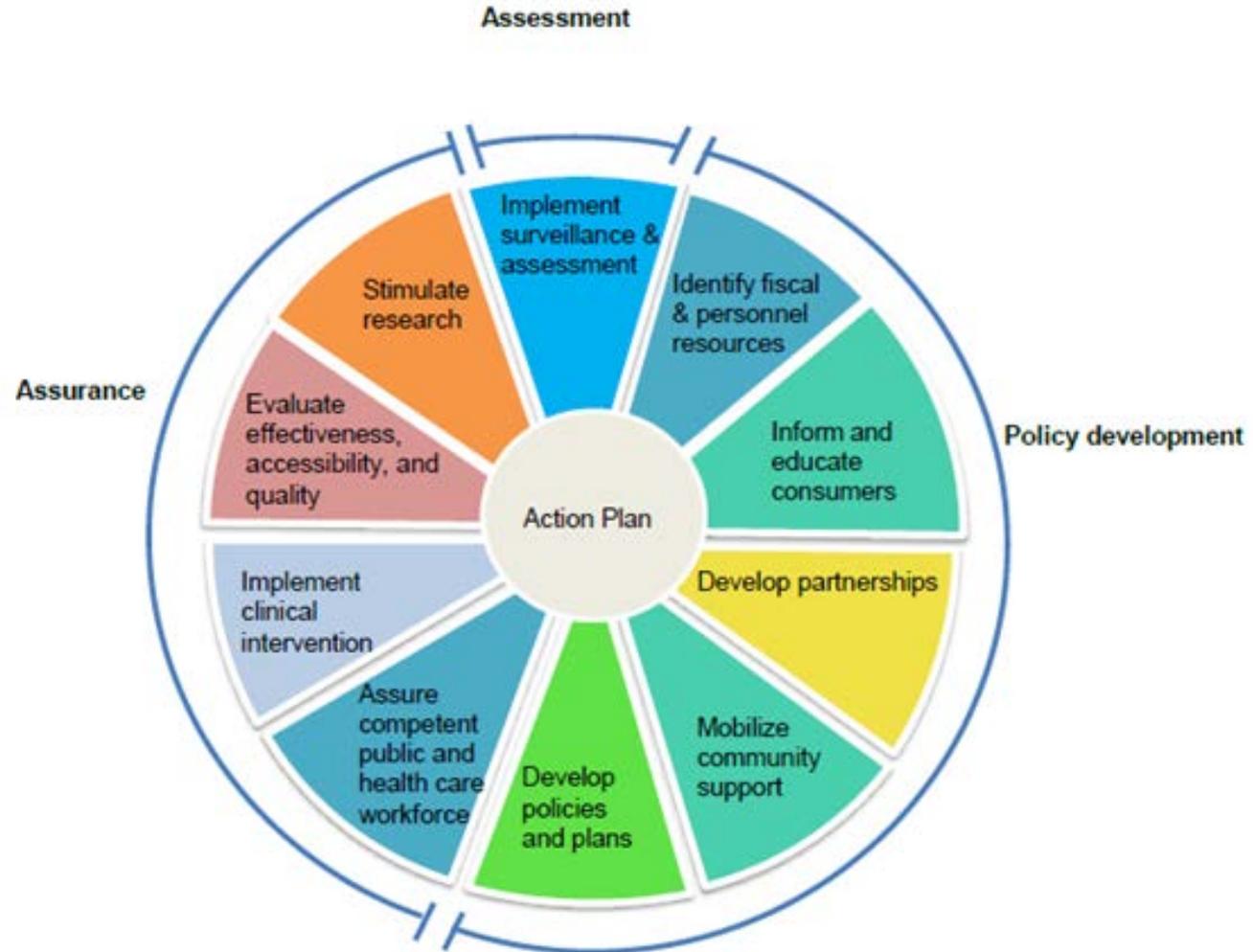
During the past decade, there have

Also, there are disease genes that account
for a small fraction of the more common
chronic diseases, such as α₁-antitrypsin

AJPH 1996

New Strategies in Public Health Genomics

Actions to
Save Lives Now
Conference Report
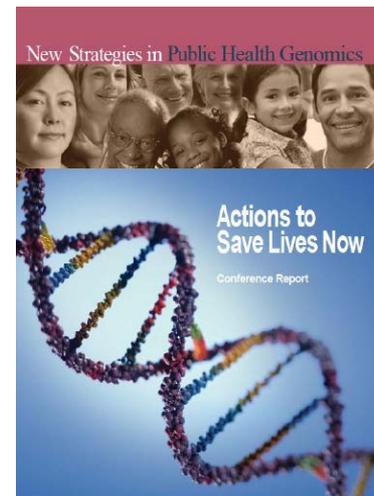
2012

What will it take?

Genomic Medicine and Public Health Genomics:

Public health is needed to fulfill the promise of genomic medicine

Medicine ↔ Public Health

Medicine → Genomic Medicine

Public Health → Public Health Genomics

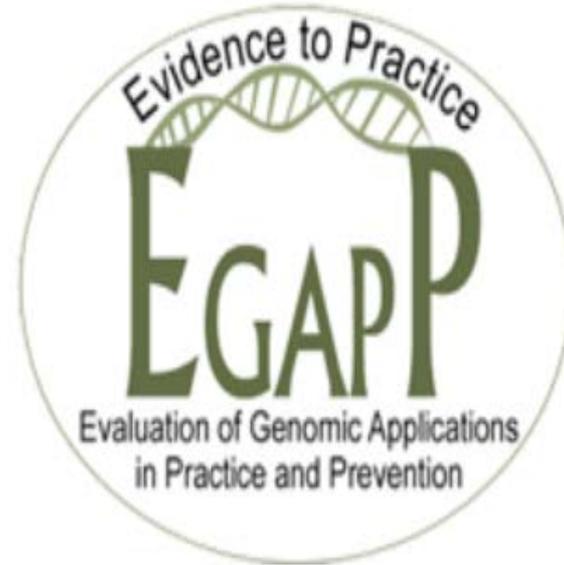Genomic Medicine ↔ Public Health Genomics
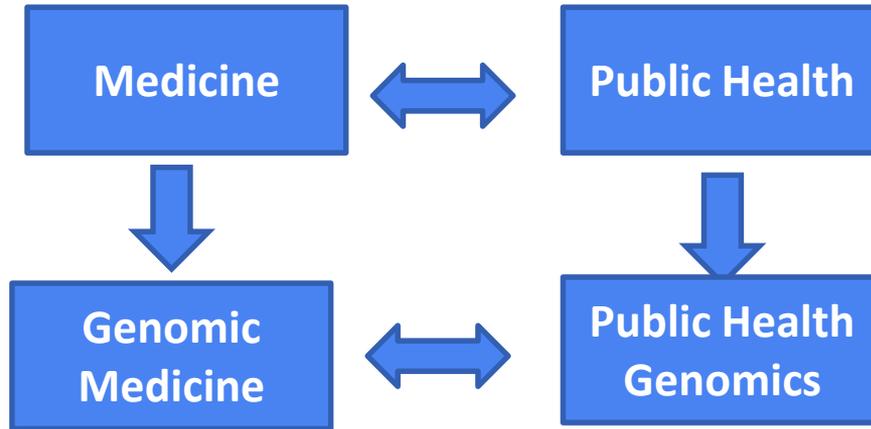
1990

Human Genome Project

1997

Public Health Response

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Evaluating Genomic Tests

G. Palomaki et al

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Family history is an important risk factor for chronic diseases

	Relative Risk
Heart disease	2.0 – 5.4
Breast cancer	2.1 – 3.9
Colorectal cancer	1.7 – 4.9
Prostate cancer	3.2 –11.0
Melanoma	2.7 – 4.3
Diabetes	2.4 – 4.0
Osteoporosis	2.0 – 2.4
Asthma	3.0 – 7.0

CDC's Family History Public Health Initiative

Evaluate the use of family history for assessing risk of common diseases and influencing early detection and prevention strategies

Components

- Assessment of existing strategies & research gaps

- Development of new tools and methods

- Research and evaluation

- Public awareness and provider education

https://familyhistory.hhs.gov/FHH/html/index.html

"This Thanksgiving Day, learn about your #familyhealthhistory, share it & act on it. You could save lives! http://thndr.me/UQ6EUe"

Dr. Muin Khoury   EMBED </>

SUPPORTERS
10 of **100**
10% of goal supported

SOCIAL REACH
149,525
People

TIME LEFT
21 days
Ends Nov 23, 12:00 PM EST

CDC's Thanksgiving Thunderclap Family Health History Message

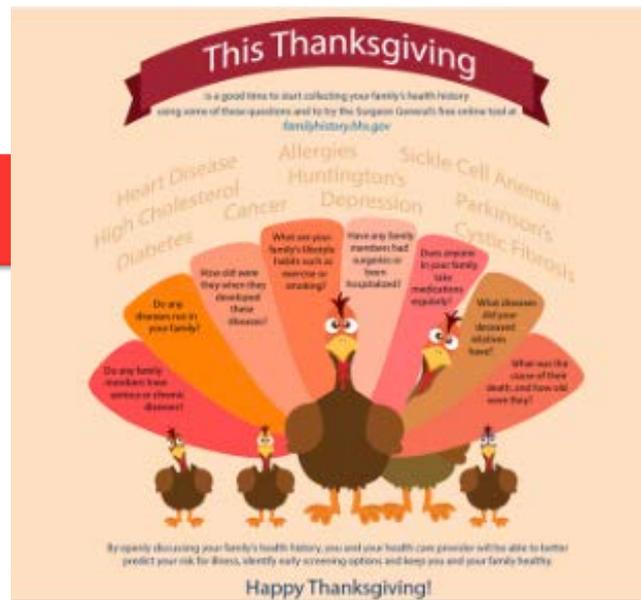Support **Dr. Muin Khoury** in sharing this message.

| support with **FACEBOOK** | support with **TWITTER** | support with **TUMBLR** |

We will post this one-time message to your account on
November 23 at 12:00PM EST. About Support & Privacy

Join with us today here:
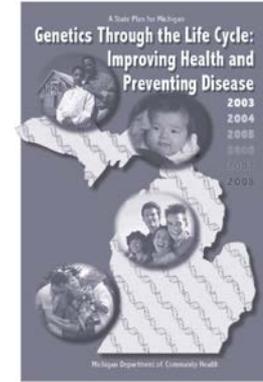https://www.thunderclap.it/projects/64131

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CDC State Cooperative Agreements in Cancer Genomics

- 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
- 2003-2008: Michigan, Minnesota, Oregon, and Utah
- 2008-2011: Michigan and Oregon
- 2011-2014: Georgia, Michigan, and Oregon
- 2011: Connecticut (Healthy People 2020 Action Award)
- 2014-2019: Colorado, Connecticut, Michigan, Oregon, and Utah

http://www.cdc.gov/cancer/breast/what_cdc_is_doing/genomics_foa.htm

CDC's Genomic Applications Toolkit for Public Health Departments

■ Goal: Assist state and local public health departments in implementing genomics

http://www.cdc.gov/genomics/implementation/toolkit/index.htm

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CDC Evidence-based Classification of Genomic Tests

|		
Tier 1	**Supported by a base of synthesized evidence for implementation in practice**	**e.g., Newborn screening**
Tier 2	**Synthesized evidence is insufficient to support routine implementation in practice; may provide information for informed decision making**	**e.g., many pharmacogenomics tests**
Tier 3	**Evidence-based recommendations against use, or no relevant synthesized evidence identified; not ready for routine implementation in practice**	**e.g., direct-to-consumer personal genomic tests**

# Selected Emerging Public Health Genomic Applications Beyond Newborn Screening
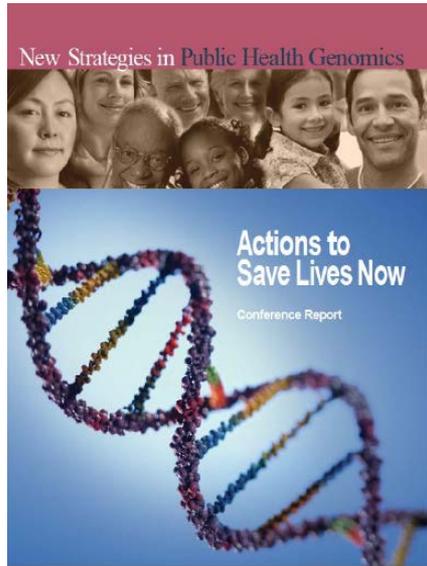
- Hereditary Breast and Ovarian Cancer (BRCA)
- Hereditary Nonpolyposis Colorectal Cancer (Lynch Syndrome)
- Familial Hypercholesterolemia
- Collectively Affect ~2 Million People in US and Most Don't know it.
- Implementation of specific guidelines can prevent cancer & heart disease, & save thousands of lives every year!

BRCA-associated Hereditary Breast and Ovarian Cancer Syndrome

- Caused by inherited changes in *BRCA1* and *BRCA2* genes
- Increased risk for breast, ovarian, and other types of cancer (high grade prostate, male breast, pancreatic)
- Certain ethnic groups are at increased risk for *BRCA* mutations
  - 1 in 40 Ashkenazi Jews
- Interventions can significantly reduce risk of cancers
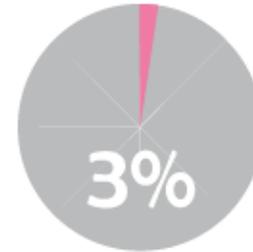
3% of breast cancers
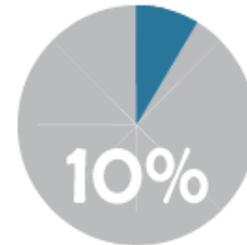
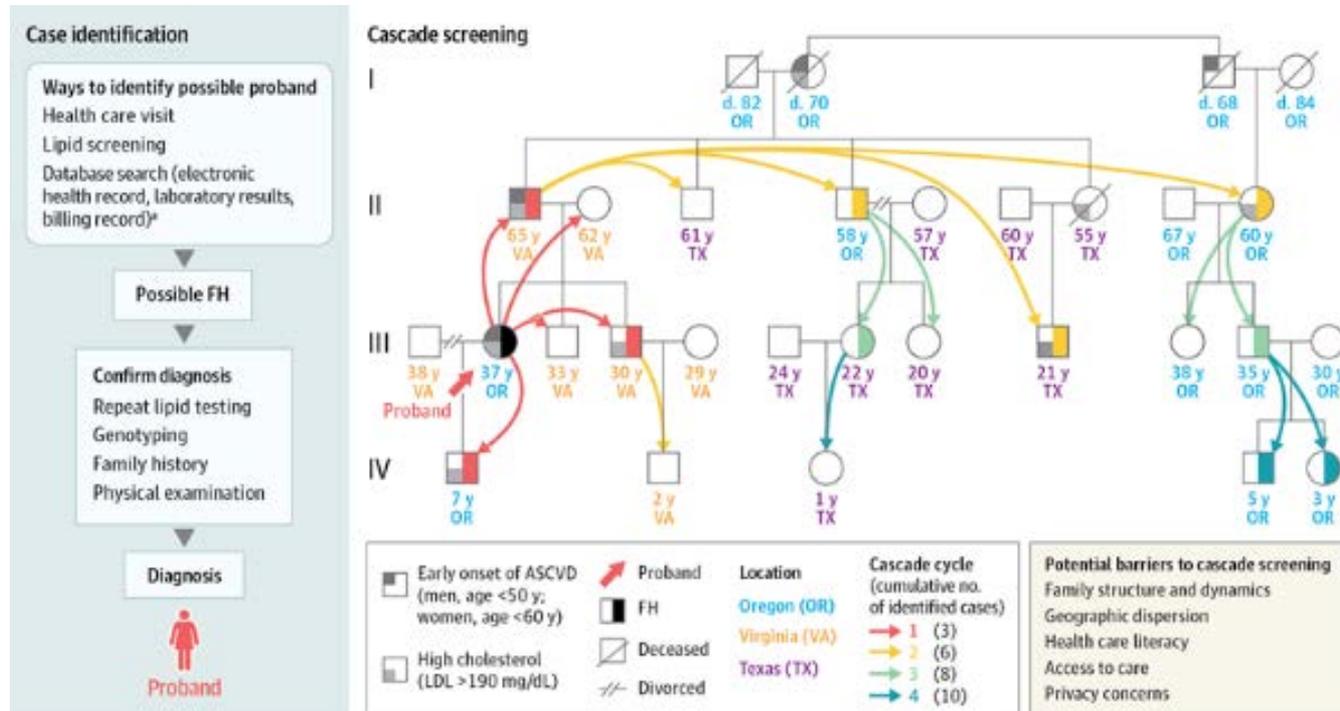10% of ovarian cancers

Lynch Syndrome

- Increased risk for certain cancers:

  – Colorectal

  – Endometrial (Uterine)

  – Ovarian

  – Bladder

  – Stomach

- Caused by inherited mutations in the *MLH1*, *MSH2, MSH6, PMS2,* and *EPCAM* genes that affect mismatch repair

- Interventions can significantly reduce risk of cancers

1 in **30** patients with colorectal cancer has Lynch Syndrome

H Hampel 2010

Cascade Screening for Familial Hypercholesterolemia: Recommendations from National Institute for Clinical Excellence (NICE, 2008)

J Knowles et al. JAMA 2017

Healthy People 2020 Genomics Objectives

- Increase the proportion of women with a family history of breast and/or ovarian cancer who receive genetic counseling.

- Increase the proportion of persons with newly diagnosed colorectal cancer who receive genetic testing to identify Lynch syndrome (or familial colorectal cancer syndromes).

Health disparities:
Importance of Public Health Approach

Black young breast cancer survivors less likely to have genetic counseling or testing for hereditary breast and ovarian cancer

- Most commonly reported reason: health care provider did not recommend genetic services

Jones, T. et al. 2016
Cragun, D. et al. 2017

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CDC Office of Public Health Genomics

1. Identify evidence-based applications

2. Inform & communicate

3. Integrate into practice & programs

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

SEARCH

CDC A-Z INDEX ∨

Public Health Genomics

MY PHGKB

Customized Tool for Genomics and Population Health Impact Information

GENOMICS AND HEALTH DISPARITIES

EPILEPSY, GENETICS AND FAMILY HISTORY

DIABETES, GENETICS AND FAMILY HISTORY

IMPLEMENTATION SCIENCE IN GENOMIC MEDICINE

What we do…

1. Identify opportunities for genomics to improve health & reduce health disparities by conducting horizon scanning and health impact analyses

K Kolor et al. MMWR 2017

What we do…

2. Inform stakeholders about impact of genomics on population health and health disparities

Precision Public Health

Can We Conduct Public Health Functions With More "Precision"?

The 3 Core Public Health Functions

- **Assessment**
  - More "precision" in measuring population health problems

- **Policy Development**
  - Developing the right intervention for the right population

- **Assurance**
  - More "precision" in delivering interventions & addressing health disparities

Precision Public Health for the Era of Precision Medicine

Muin J. Khoury, MD, PhD,[1,2] Michael F. Iademarco, MD, MPH,[1,3] William T. Riley, PhD[2]

The Precision Medicine Initiative[1] promises a new healthcare era. A proposed 1 million−person cohort could create a deeper understanding of disease causation. Improvements in quality of sequencing, reduction in price, and advances in "omic" fields and biotechnology promise a new era, variably labeled personalized or precision medicine. Although genomics is one driver of precision health care, other factors may be as important (e.g., health information technology).

Both excitement and skepticism met the announcement.[2] Public health experts are concerned about the disproportionate emphasis on genes, drugs, and disease, while neglecting strategies to address social determinants of health. A prime concern for public health is promoting health, preventing disease, and reducing health disparities by focusing on modifiable morbidity and mortality. In 2014, CDC estimated the annual number of poten-

evidentiary foundation for use. The following are examples of priority areas.

Role of Multidisciplinary Public Health Sciences

Though precision medicine focuses on individualized care, its success truly requires a population-based approach. To learn what interventions work for whom, data on each individual need to be compared with data from large, diverse numbers of people to identify population subgroups likely to respond differently to interventions. In addition, collecting information from large numbers of people is far more informative when diverse people are included from the underlying population. Using data from convenience samples alone (i.e.,

AJPM, 2016

Precision Medicine and Precision Public Health:
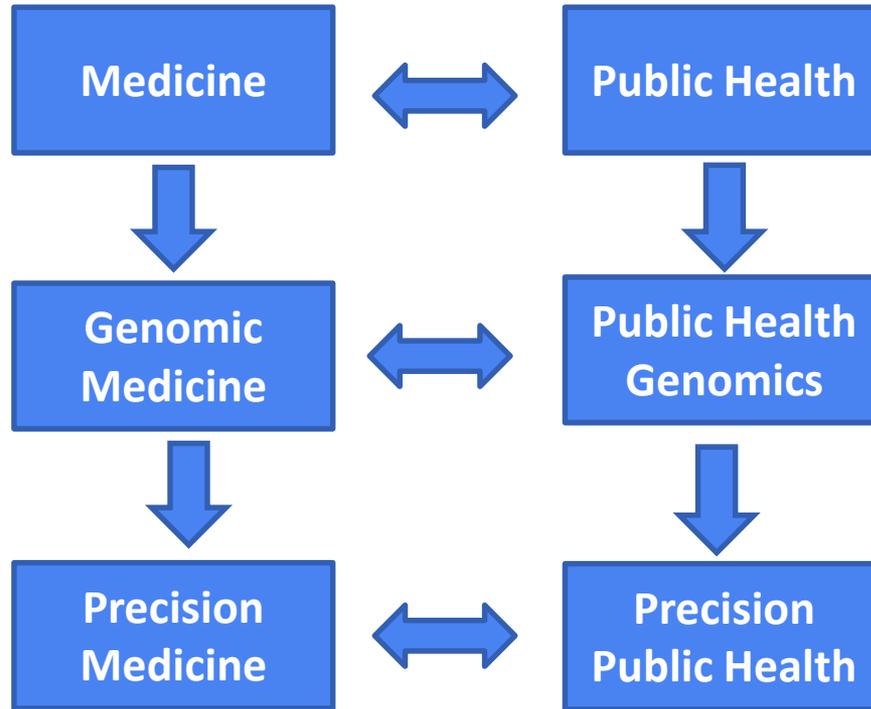
Thanksgiving Day Thunderclap

Join our #familyhealthhistory Thanksgiving Day Thunderclap! Encourage families to collect & act on their health histories.

https://www.thunderclap.it/projects/64131

Thank you!

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.