Reflections on Precision Public Health

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What is Precision Public Health?

Accompanied transcript of the slide set

1) Medicine alone cannot not improve the health of a population. We also need public health!

Medicine ⟷ **Public Health**

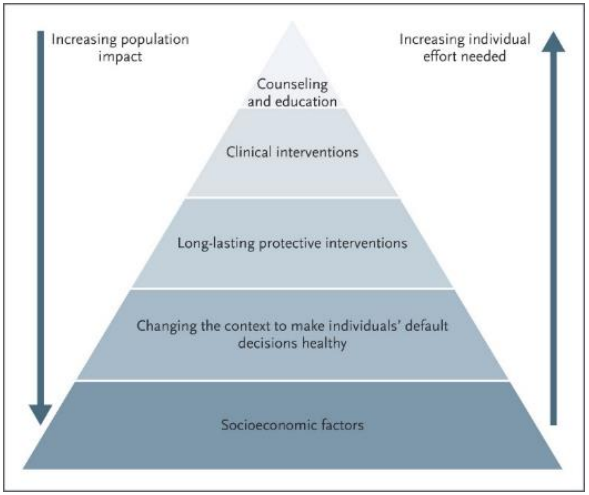
Individuals — Populations

Healthcare — Health

Clinical — Community

Treatment — Prevention/Control

Genomics/Biology — Social/Environmental



Frieden TR. The Health Impact Pyramid, NEJM (2015)

Population level activities have more impact than individual ones!

2) As medicine becomes more “precise”, we need public health to help implement what we know!

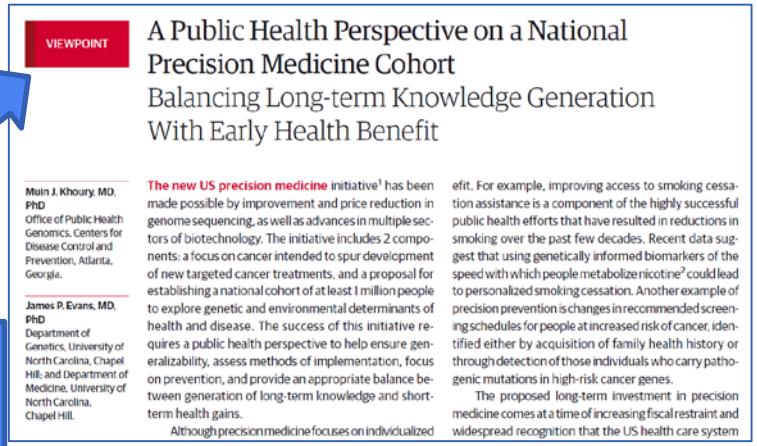
Medicine ⟷ **Public Health**

What is precision medicine?

“An emerging approach for disease prevention and treatment that takes into account variations in genes, environment and lifestyle”

NIH ALL of US

Precision Medicine



VIEWPOINT

A Public Health Perspective on a National Precision Medicine Cohort
Balancing Long-term Knowledge Generation With Early Health Benefit

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The new US precision medicine initiative[1] has been made possible by improvement and price reduction in genome sequencing, as well as advances in multiple sectors of biotechnology. The initiative includes 2 components: a focus on cancer intended to spur development of new targeted cancer treatments, and a proposal for establishing a national cohort of at least 1 million people to explore genetic and environmental determinants of health and disease. The success of this initiative requires a public health perspective to help ensure generalizability, assess methods of implementation, focus on prevention, and provide an appropriate balance between generation of long-term knowledge and short-term health gains.

Although precision medicine focuses on individualized efit. For example, improving access to smoking cessation assistance is a component of the highly successful public health efforts that have resulted in reductions in smoking over the past few decades. Recent data suggest that using genetically informed biomarkers of the speed with which people metabolize nicotine[2] could lead to personalized smoking cessation. Another example of precision prevention is changes in recommended screening schedules for people at increased risk of cancer, identified either by acquisition of family health history or through detection of those individuals who carry pathogenic mutations in high-risk cancer genes.

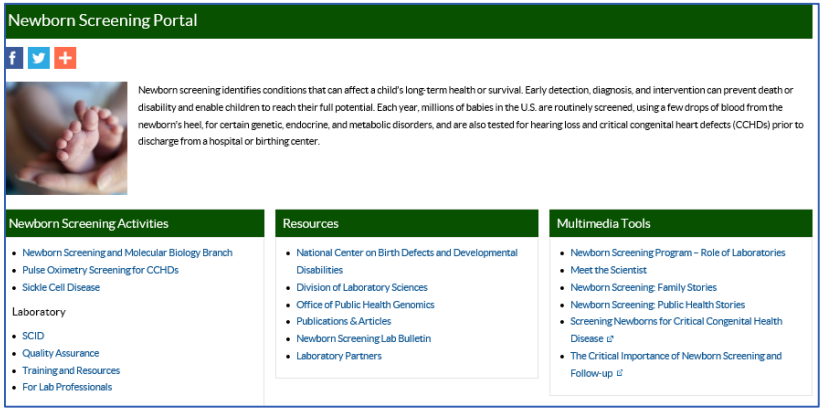
The proposed long-term investment in precision medicine comes at a time of increasing fiscal restraint and widespread recognition that the US health care system

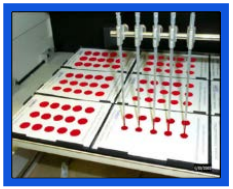
JAMA 2015

Newborn Screening

The Largest Precision Public Health Program in the World

- More than 5 decades, 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
- Potential for genome sequencing to change newborn screening and for adult genetic screening



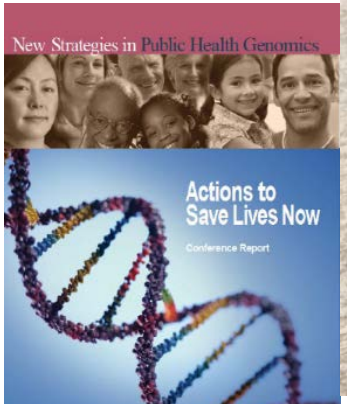


Cancer Genetics and Public Health

- Hereditary Breast and Ovarian Cancer (BRCA)
- (25,000 new cases/year)

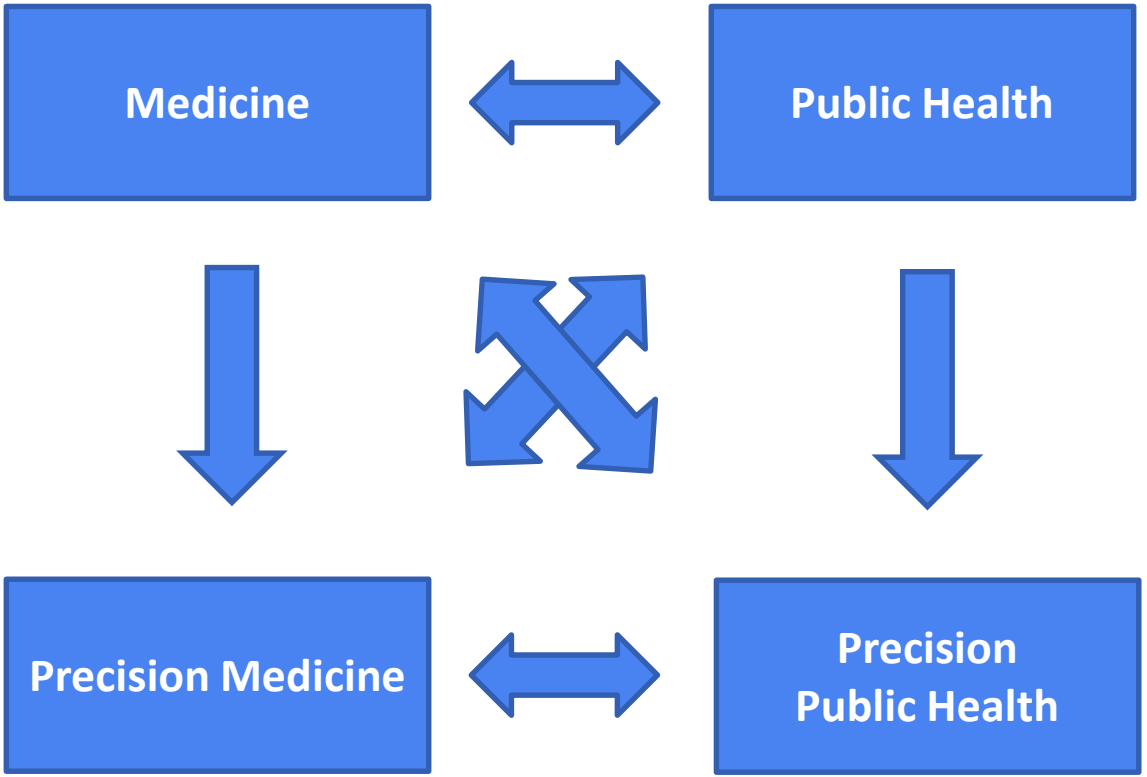
- Hereditary Nonpolyposis Colorectal Cancer (Lynch Syndrome) (4000 new cases/year

- Collectively Affect >1 Million People in US and Most Don't know it. "Precision" evidence-based action can prevent cancer & save lives!



- **Provider and public education**
- **Healthcare system limitations**
- **Evidence-based policy**
- **Population health impact data**
- **Laboratory quality**
- **Health disparities**

3) We are entering a new era of “precision” in public health activities beyond genomics



Conducting Public Health Functions With More "Precision"

- **Assessment**
  - More "precision" in measuring population health problems
- **Policy Development**
  - Developing the right intervention for the right population at the right time
- **Assurance**
  - More "precision" in delivering interventions & addressing health disparities

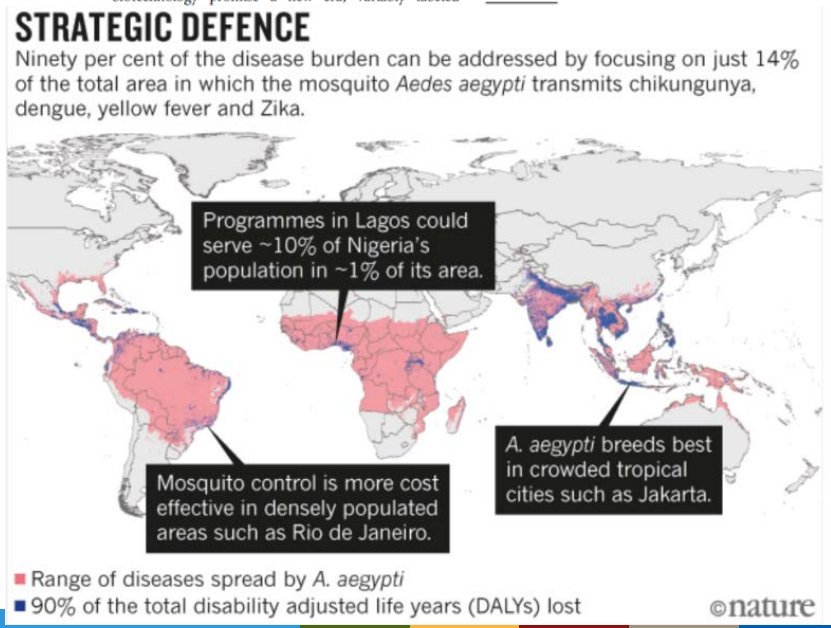
AJPM, 2016

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]

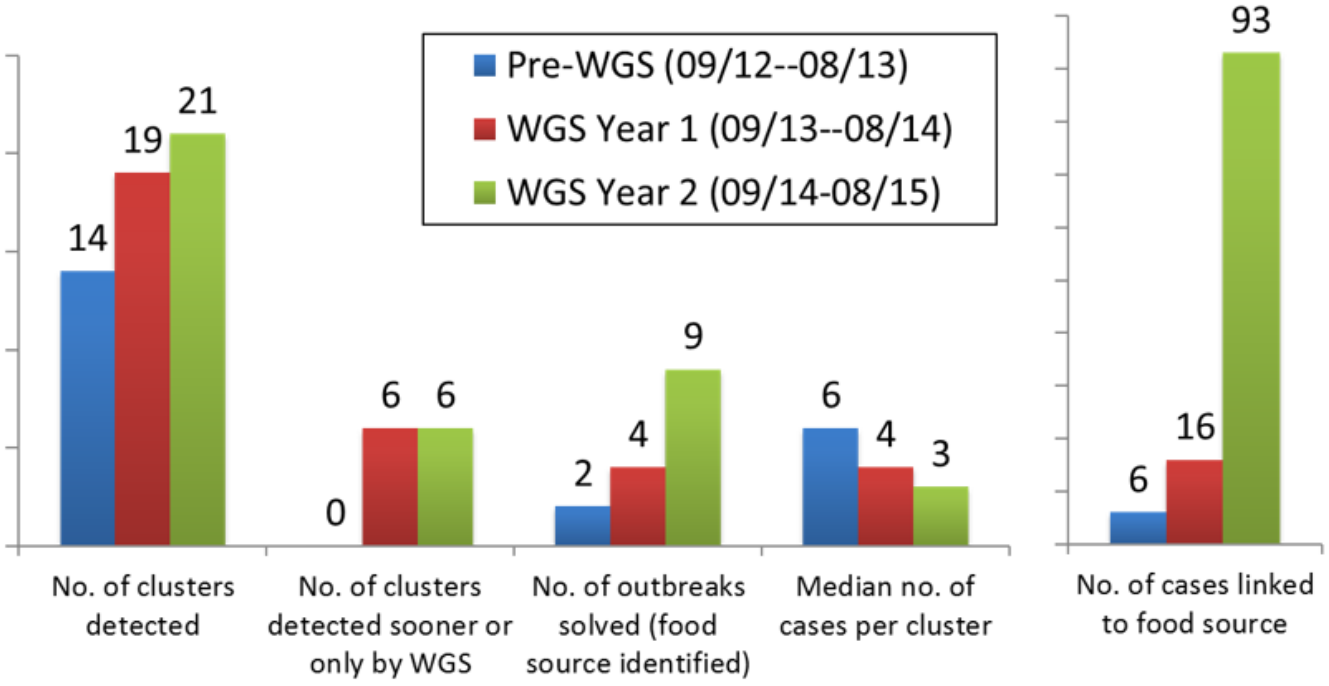
The Precision Medicine Initiative[1] promises a new healthcare era. A proposed 1 million–person cohort could create a deeper understanding of disease causation. Improvements in quality of sequencing, reduction in price, and advances in "omic" fields and biotechnology promise a new era, variably labeled evidentiary foundation for use. The following are examples of priority areas.

Role of Multidisciplinary Public Health Sciences



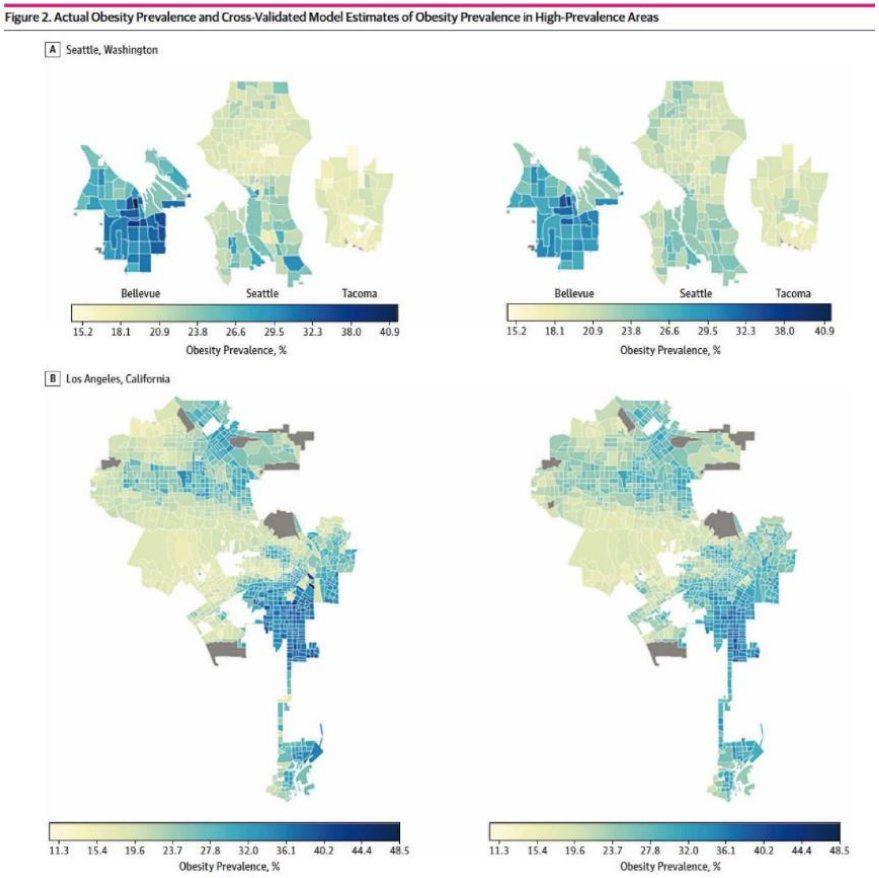
Nature, 2016

CDC Advanced Molecular Detection Initiative Using Whole Genome Sequencing in Tracking Listeria Outbreaks In the United States



M. Gwinn et al. J Clin Micro 2017

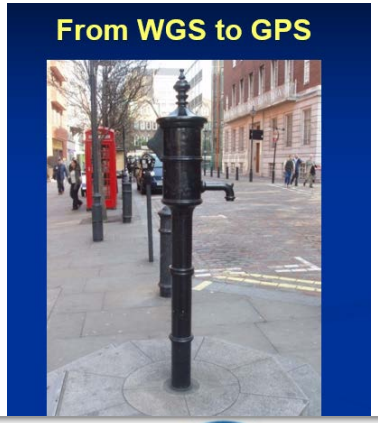
Artificial Intelligence Spots Obesity from Space



A Maharana et al, JAMA Network Open, August 31, 2018

In this study of 4 US urban areas, extraction of built environment information from images using convolutional neural networks to assess associations between the built environment and obesity prevalence showed that physical characteristics of a neighborhood (eg, the presence of parks, highways, green streets, crosswalks, diverse housing types) can be associated with variations in obesity prevalence across different neighborhoods.

Promises and Challenges of Big Data-Driven Public Health



MEDICINE

Big data meets public health

Human well-being could benefit from large-scale data if large-scale noise is minimized

By **Muin J. Khoury**[1,2] *and* **John P. A. Ioannidis**[3]

In 1854, as cholera swept through London, John Snow, the father of modern epidemiology, painstakingly recorded the locations of affected homes. After long, laborious work, he implicated the Broad Street water pump as the source of the outbreak, even without knowing that a *Vibrio* organism caused cholera. "Today, Snow might have crunched Global Positioning System information and disease prevalence data, solving the problem within hours" (*1*). What is the potential impact of "Big Data" on the public's health. But the promise of Big Data is also accompanied by claims that "the scientific method itself is becoming obsolete" (*2*), as next-generation computers, such as IBM's Watson (*3*), sift through the digital world to provide predictive models based on massive information. Separating the true signal from the gigantic amount of noise is neither easy nor straightforward, but it is a challenge that must be tackled if information is ever to be translated into societal well-being.

The term "Big Data" refers to volumes of large, complex, linkable information (*4*). Beyond genomics and other "omic" fields, Big Data includes medical, environmental, fi-

For nongenomic associations, false alarms due to confounding variables or other biases are possible even with very large-scale studies, extensive replication, and very strong signals (*9*). Big Data's strength is in finding associations, not in showing whether these associations have meaning. Finding a signal is only the first step.

Even John Snow needed to start with a plausible hypothesis to know where to look, i.e., choose what data to examine. If all he had was massive amounts of data, he might well have ended up with a correlation as spurious as the honey bee–marijuana connection. Crucially, Snow "did the experiment." He removed the handle from the water pump and dramatically reduced the spread of cholera, thus moving from correlation to causation and effective intervention.

How can we improve the potential for Big Data to improve health and prevent disease? One priority is that a stronger epidemiological foundation is needed. Big Data analysis is currently largely based on convenient samples of people or information available on the Internet. When associations are probed between perfectly measured data (e.g., a genome sequence) and poorly measured data (e.g., administrative claims health data), research accuracy is dictated by the weakest link. Big

In Summary

- We need both medicine and public health to improve population health
- As medicine becomes more "precise", public health is needed to implement it to save lives and ensure health equity
- We are entering a new era of "precision" in public health beyond applications of precision medicine that requires more evidence & evaluation
- Our collective challenge is to set up global, regional and local priorities, infrastructures, scientific and implementation strategies!