Integrating Genome Sequencing in Health Care Systems: Evaluation, Implementation and Population Health Impact

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Disclaimers

- I have no known conflicts of interest related to the materials presented.

- I speak for myself and not any of the organizations mentioned in this presentation, particularly NASEM.

Extramural Research Reports and Publications

1998 Five Year Plan: New Goals

ELSI Program Reports

Long-Range Planning: Reports and Publications

Workshop and Priority Setting Reports from ERP

Understanding Our Genetic Inheritance

The United States Human Genome Project
The First Five Years: Fiscal Years 1991-1995

Executive Summary

The Human Genome Initiative is a worldwide research effort that has the goal of analyzing the structure of human DNA and determining the location of the estimated 100,000 human genes. In parallel with this effort the DNA of a set of model organisms will be studied to provide the comparative information necessary for understanding the functioning of the human genome. The information generated by the human genome project is expected to be the source book for biomedical science in the 21st century and will be of immense benefit to the field of medicine. It will help us to understand and eventually treat many of the more than 4000 genetic diseases that afflict mankind, as well as the many multifactorial diseases in which genetic predisposition plays an important role.

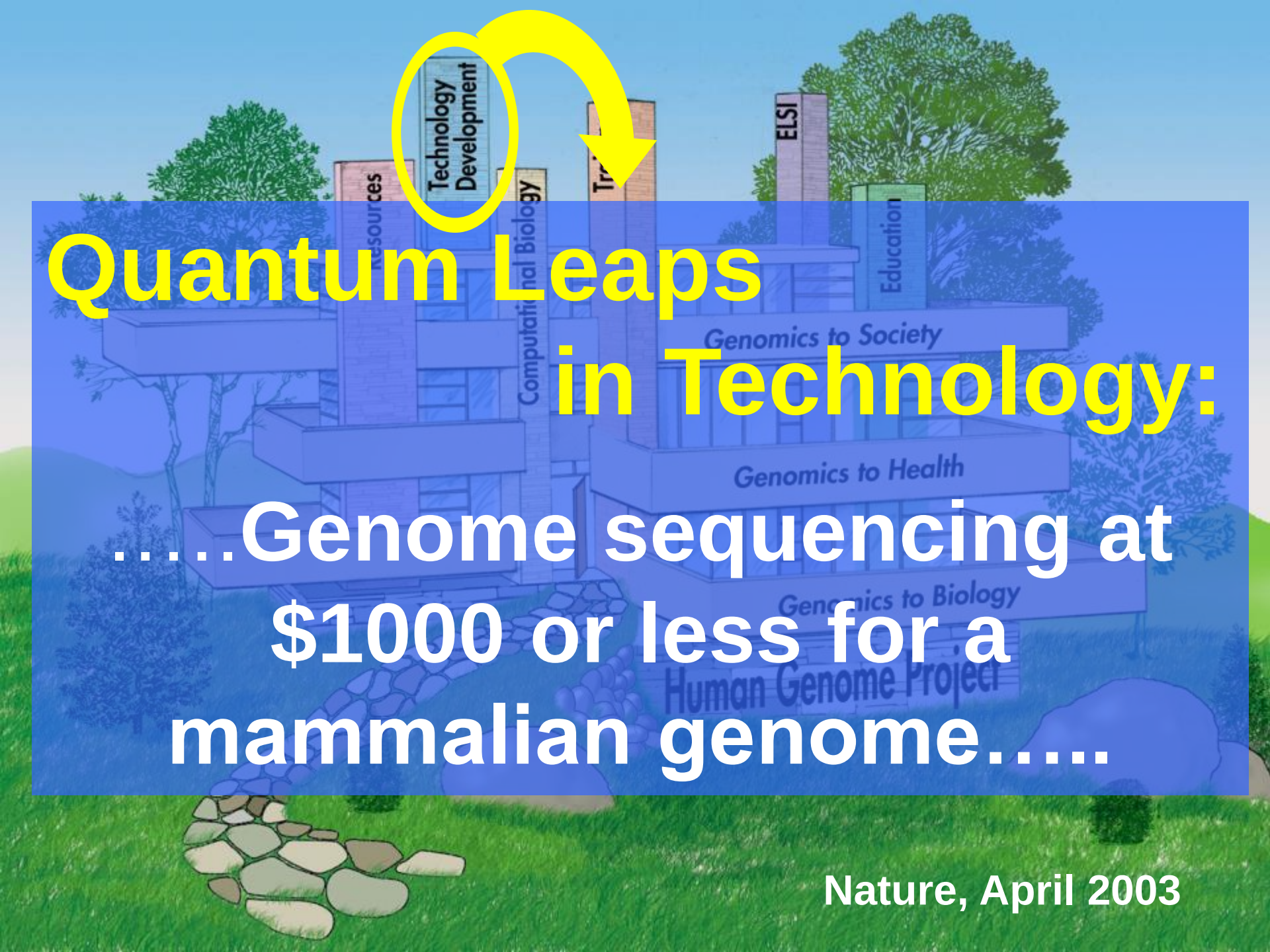
A centrally coordinated project focused on specific objectives is believed to be the most efficient and least expensive way of obtaining this information. In the course of the project much new technology will be developed that will facilitate biomedical and a broad range of biological research, bring down the cost of many experiments, and find application in numerous other fields. The basic data produced will be collected in electronic databases that will make the information readily accessible in convenient form to all who need it.

“5 YEAR GOAL: Improve current methods and/or develop new methods for DNA sequencing that will allow large scale sequencing of DNA at a cost of $0.50 per base pair.”

Human genome – 3 billion bases

A bargain at a cool:

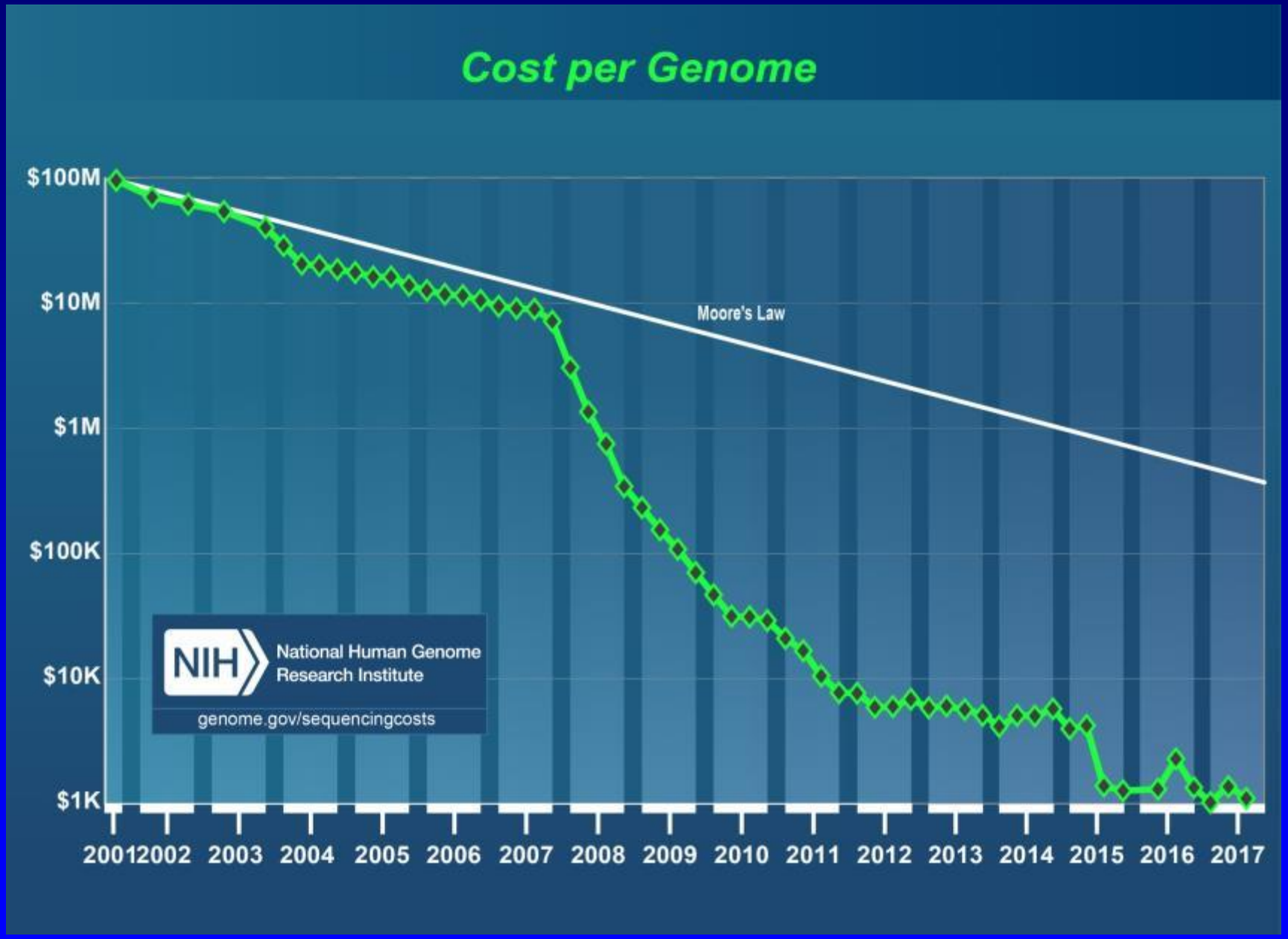
$500,000,000- $1B



Quantum Leaps in Technology:

…..Genome sequencing at $1000 or less for a mammalian genome…..

Nature, April 2003


Cost per Genome
$100M
$10M
$1M
$100K
$10K
$1K
Moore's Law
NIH
National Human Genome Research Institute
genome.gov/sequencingcosts
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The 21st Century Cures Act — A View from the NIH

Kathy L. Hudson, Ph.D., and Francis S. Collins, M.D., Ph.D.

The Cures Act, formally known as H.R. 34 or the 21st Century Cures Act,[1] passed overwhelmingly in the U.S. House of Representatives and Senate in the waning days of the 114th Congress and was signed into law by President Barack Obama on December 13, 2016. Weighing in at nearly 1000 pages, this bipartisan bill is the product of years of hard work by Republican and Democratic lawmakers, in collaboration with a broad array of diverse stakeholders. As with any landmark piece of legislation, these measures will cut bureaucratic red tape that slows the progress of science, enhance data sharing and privacy protections for research volunteers, improve support for the next generation of biomedical researchers, exhort agency with critical tools and resources to advance biomedical research across the spectrum from basic, curiosity-driven studies to advanced clinical trials of promising new therapies. Affecting everyone from researchers to research participants to patients suffering from numerous conditions, these measures will cut bureaucratic red tape that slows the progress of science, enhance data sharing and privacy protections for research volunteers, improve support for the next generation of biomedical researchers, exhort Paperwork Reduction Act,[2] was enacted when the Internet was nascent and paper still ruled. Its purpose was to limit government's ability to ask Americans to fill out endless forms, especially when those forms were required to receive government services or benefits. Minimizing needless paperwork and bureaucracy is an admirable goal. However, as applied to biomedical research, the law requires multiple levels of government review and public comment on any set of questions that NIH researchers

NEJM pub online 12/13/16

CANCER MOONSHOT℠

Blue Ribbon Panel Report +

Funding Opportunities

Milestones +

Cancer Moonshot℠

The Cancer Moonshot to accelerate cancer research aims to make more therapies available to more patients, while also improving our ability to prevent cancer and detect it at an early stage.

To ensure that the Cancer Moonshot's goals and approaches are grounded in the best science, a Cancer Moonshot Task Force consulted with external experts, including the presidentially appointed National Cancer Advisory Board (NCAB). A Blue Ribbon Panel of experts was established as a working group of the NCAB to assist the board in providing this advice. The panel's charge was to provide expert advice on the vision, proposed scientific goals, and implementation of the Cancer Moonshot.

Congress passed the 21st Century Cures Act in December 2016 authorizing $1.8 billion in funding for the Cancer Moonshot over 7 years.

Funding Opportunities to Support Cancer Moonshot Priorities

NCI is accepting applications for research grants that align with the goals of the Cancer Moonshot.

Get email updates from NCI on the Cancer Moonshot

NEWS Posted March 11 | Updated March 13

INCREASE FONT SIZE

Augusta's cancer center hosts genomics research initiative

The program, directed by two headed from the Jackson Laboratory, will allow oncologists throughout the state to share technology, information.

BY BETTY ADAMS STAFF WRITER

Share Comment

The Jackson Laboratory's name has been added to the sign, seen Wednesday, at the Harold Alfond Center for Cancer Care in Augusta. *Staff photo by Joe Phelan*

http://www.centralmaine.com/2017/03/11/augustas-cancer-center-hosts-genomics-research-component/

All of Us℠ Research Program

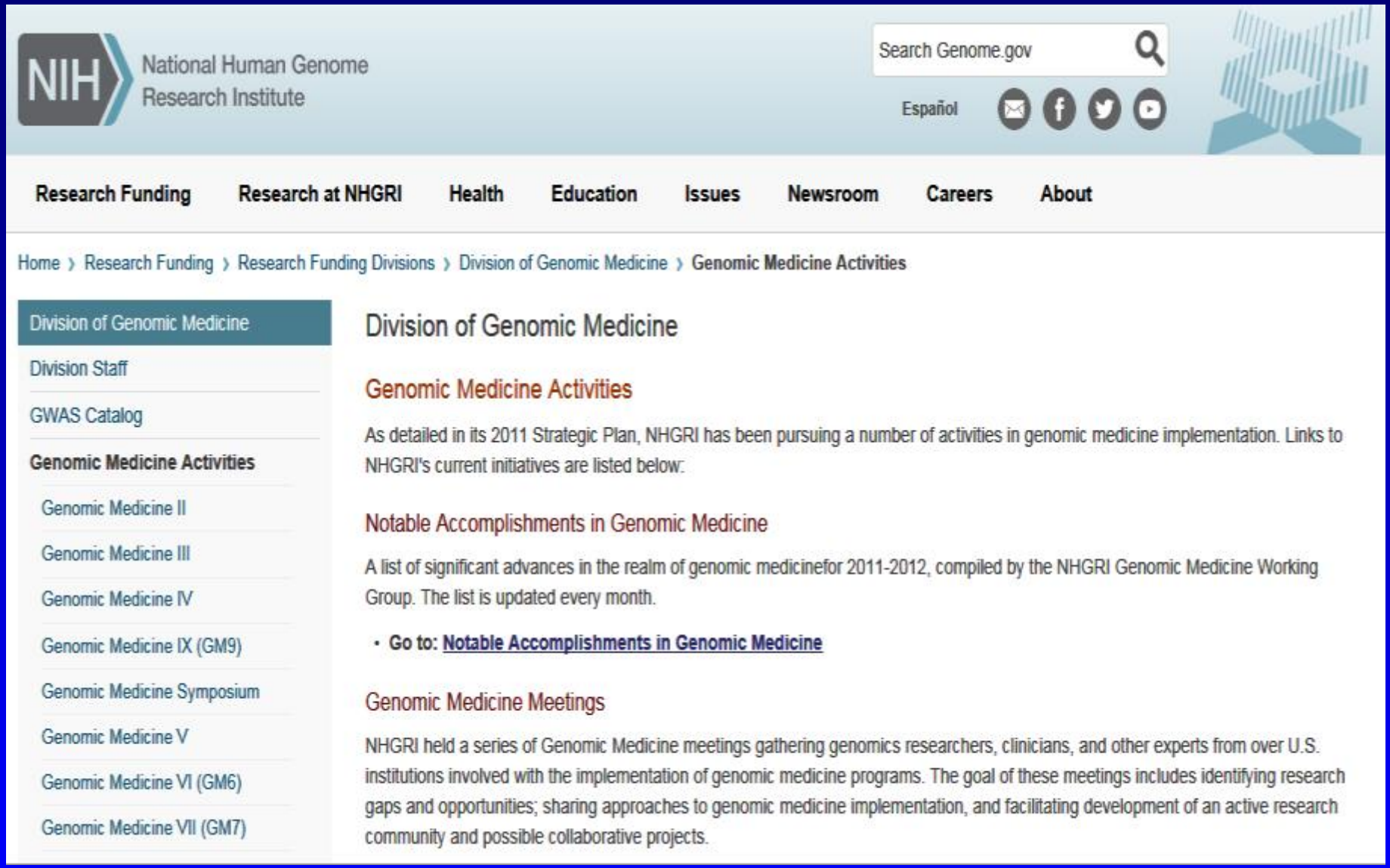
WHAT IS IT?

Precision medicine is a groundbreaking approach to disease prevention and treatment based on people's individual differences in environment, genes and lifestyle.

The *All of Us* Research Program will lay the foundation for using this approach in **clinical practice.**

Can genomic sequence data be used to improve the health of large populations?

Prediction, prevention, screening…



https://www.genome.gov/27549225/genomic-medicine-activities/

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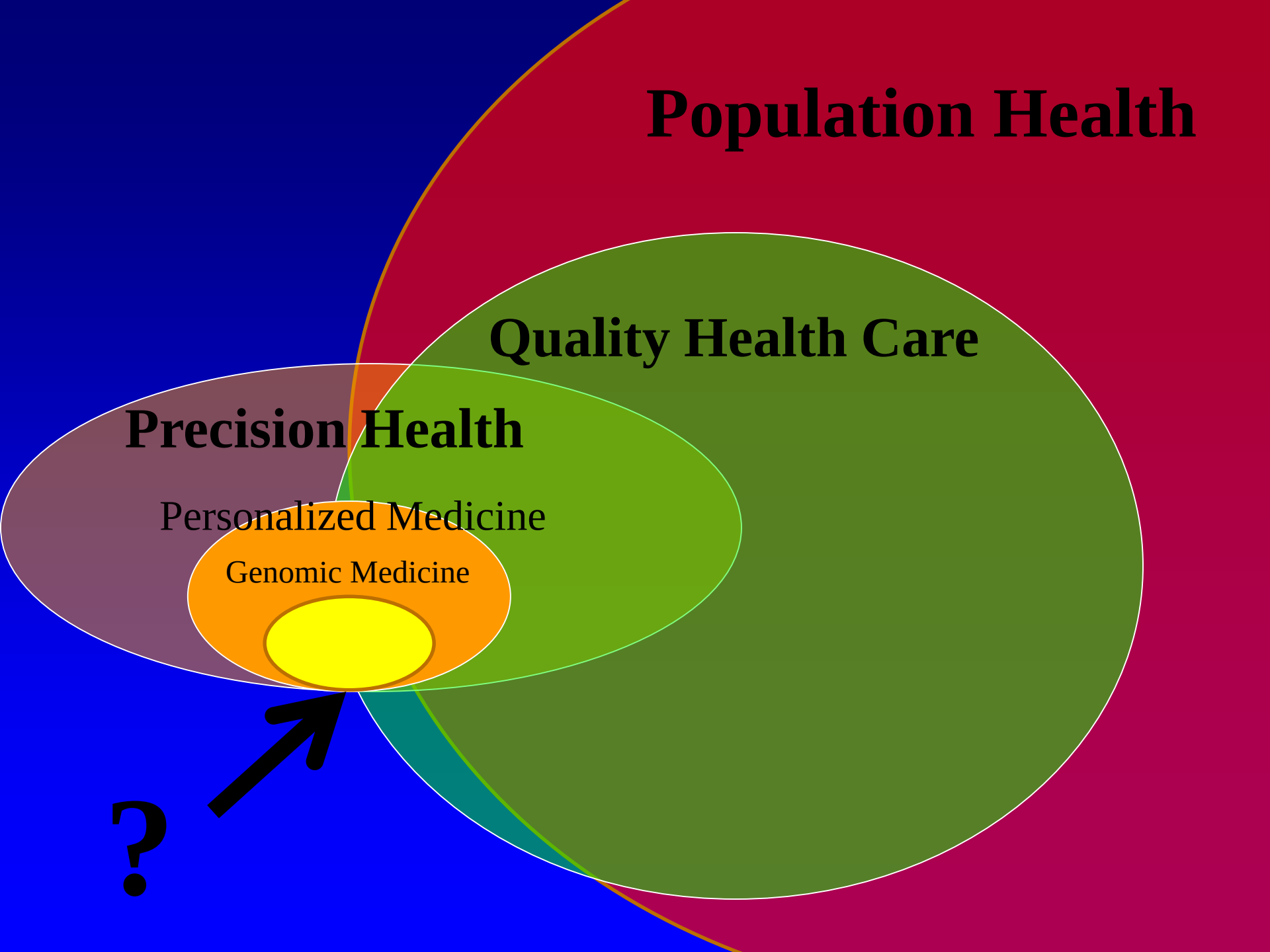
About Us | 100,000 Genomes Project | Taking Part | For Healthcare Professionals | Research | Industry Partnerships | News & Events

Genome BritishColumbia
Leading ▸ Investing ▸ Connecting

WHY GENOMICS? FUNDING PARTNER WITH US BLOG EDUCATION ABOUT CONNECT

Canadian patients to benefit from major investment in genomics and precision health research


Population Health
Quality Health Care
Precision Health
Personalized Medicine
Genomic Medicine
?

Activity

Roundtable on Genomics and Precision Health

Type: Roundtable

Topics: Biomedical and Health Research, Public Health

Board: Board on Health Sciences Policy

Activity Description

The Roundtable on Genomics and Precision Health (previously called the Roundtable on Translating Genomic-Based Research for Health) brings together leaders from government, academia, industry, foundations, associations, patient communities, and other stakeholder groups to meet and discuss global issues surrounding the translation of genomics and genetics research findings into medicine, public health, education, and policy. The primary purpose of the Roundtable is to foster dialogue across sectors and among interested parties and institutions, and to illuminate and scrutinize critical scientific and policy issues where Roundtable engagement and input will help further the field.

The Roundtable membership identifies scientific and policy issues where discussion and collaboration will help enable the translation of genomics into health care applications. Specific issues and agenda topics are

Publications

Enabling Precision Medicine: The Role of Genetics in Clinical Drug Development: Proceedings of a Workshop

Released: July 10, 2017

Deriving Drug Discovery Value from Large-Scale Genetic Bioresources: Proceedings of a Workshop

Released: September 9, 2016

View All Publications from this Activity

MEETING AGENDA

Workshop on Diffusion and Use of Genomic Innovations in Health and Medicine

8:30-8:40 Welcome and Overview of Workshop

WYLIE BURKE

Roundtable Chair

Professor and Chair

Department of Medical History and Ethics

University of Washington School of Medicine

8:40-10:00 Panel on Translation of Innovations

Other Meeting Resources

+ Presentations

Workshop Summary

Diffusion and Use of Genomic Innovations in Health and Medicine

Study Staff

- Sarah Beachy, Study Director

+ View Full Study Staff Roster

INSTITUTE OF MEDICINE
OF THE NATIONAL ACADEMIES

Board on Health Sciences Policy

Roundtable on Translating Genomic-Based Research for Health

Integrating Large-Scale Genomic Information into Clinical Practice

July 19, 2011

The Keck Center of the National Academies
500 Fifth St., N.W.
Washington, DC 20001

Workshop Objective:

- To highlight and identify the challenges and opportunities in integrating large-scale genomic information into clinical practice.

Workshop Assumptions:

- Sequencing technology will advance enough to produce clinically meaningful results.
- Whole genome sequencing (WGS) will be cost-effective and comparable to other diagnostic tests.

A workshop co-hosted by:

INSTITUTE OF MEDICINE
OF THE NATIONAL ACADEMIES

CMTP
Center for Medical Technology Policy

Board on Health Sciences Policy
Roundtable on Translating Genomic-Based Research for Health

Evidence for Clinical Utility of Molecular Diagnostics in Oncology: A Workshop

May 24, 2012

PEW DC Conference Center
901 E. St., N.W.
Washington, DC, 20004

Workshop Objectives:

- To assess the evidentiary requirements for clinical validity and clinical utility of molecular diagnostics which are used to guide treatment decisions for cancer

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Roundtable on Translating Genomic-Based Research for Health

Genomics-Enabled Learning Health Care Systems: Gathering and Using Genomic Information to Improve Patient Care and Research

A Workshop

December 8, 2014

The Keck Center of the National Academies, Room 100
500 Fifth Street, NW
Washington, DC 20001

MEETING OBJECTIVES

- TO EXPLORE HOW KEY PIECES OF GENETIC/GENOMIC INFORMATION CAN BE EFFECTIVELY AND EFFICIENTLY DELIVERED TO PATIENTS AND CLINICIANS FOR IMPROVING CARE.
- TO DISCUSS HOW BOTH THE HEALTH CARE SYSTEM AND GENOMIC DATA CAN BE USED FOR EVIDENCE GENERATION

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INSTITUTE OF MEDICINE
Board on Health Sciences Policy
Roundtable on Translating Genomic-Based Research for Health

Applying an Implementation Science Approach to Genomic Medicine: A Workshop

November 19, 2015

National Academy of Sciences Building
Lecture Room
2101 Constitution Avenue NW
Washington, DC 20418

MEETING OBJECTIVES

- TO ELUCIDATE OPTIONS FOR ACCELERATING THE PACE OF IMPLEMENTATION AND EVIDENCE GENERATION IN GENOMIC MEDICINE BY CONVENING MEDICAL IMPLEMENTATION SCIENCE EXPERTS WITH STAKEHOLDERS REPRESENTING THE CONTINUUM OF GENOMICS TRANSLATIONAL RESEARCH

Selected Filters

- Activity: Roundtable on Genomics and Precision Health [Remove]

Filter Meetings

By Keyword

Keyword Search Filter

By Topic

- Biomedical and Health Research (29)
- Public Health (28)

Meetings

Upcoming (0) Previous (29)

Displaying: 1 to 10 of 29 Meetings

1 2 3

Implementing and Evaluating Genomic Screening Programs in Health Care Systems: A Workshop

Date: November 1, 2017 (8:30 AM Eastern)

The Roundtable on Genomics and Precision Health will hold a public workshop on November 1, 2017 that will explore the challenges and opportunities associated with integrating genomics into large-scale health organizations. Case studies of large-scale genomics programs and collaborative learning networks may be highlighted during the workshop as a way to understand successes and lessons learned regarding economic considerations (e.g., clinical utility, value), policy environments (e.g., alleviating privacy and discrimination concerns for participants), and data sharing. Workshop discussions will be held with a broad array of stakeholders which may include health economists,

Observation:

Multiple health-care entities in the U.S. (and world-wide) have, or are, developing clinical programs that apply genome sequencing to large populations of individuals that they care for.

Two driving questions:

- Why and how are health care organizations making the decision to apply genomic technologies to large populations?
- Are there opportunities to foster collaborations among early-adopter organizations to study more distal translational research questions/implementation?

The National Academies of
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Board on Health Sciences Policy
Roundtable on Genomics and Precision Health

Implementing and Evaluating Genomic Screening Programs in Health Care Systems – A Workshop

November 1, 2017

Keck Building of the National Academies
500 Fifth Street NW
Room 100
Washington, DC 20001

Statement of Task:

An ad hoc committee will plan and conduct a one day public workshop to explore challenges and opportunities associated with integrating genomics into large-scale health organizations. These initiatives have a variety of goals such as providing information about clinically actionable genetic variants, seeking diagnoses for individuals suspected to have rare diseases, and/or advancing research on the genetic contributors to human illnesses. Case studies of large-scale genomics programs and collaborative learning networks may be highlighted during the workshop as a way to understand successes and lessons learned regarding (1) economic considerations (e.g., clinical utility, value), (2) policy environments (e.g., alleviating privacy and discrimination concerns for participants), and (3) data sharing. Workshop discussions will be held with a broad array of stakeholders which may include health economists, representatives from health care delivery systems, public health officials, bioethicists, implementation science researchers, clinicians, payers, and policy makers. The committee will develop the workshop agenda, select and invite speakers and discussants, and may moderate the discussions. Proceedings of the workshop will be prepared by a designated rapporteur in accordance with institutional policy and procedures.

Agenda

1. Stage setting
2. Evidence considerations for integration of genomics into health systems
3. Financial considerations for implementation
4. Data sharing
5. Ensuring the program meets the population's needs
6. Next steps

Key Conclusions

Evidence generation

- Still very much in the mode of evidence generation – important to make clear what is research and what is clinical.

- Numbers and diversity of participants is very important for multiple types of knowledge.

- Clinical utility data will be key to broader adoption and ongoing maintenance

Key Conclusions

Financial Considerations

- Multiple avenues to funding a program can work.
- Key to (clinical and economic) value is use of information with evidence based interventions.
- Financial sustainability remains a work in progress – currently organizational leadership buy-in is key.

Key Conclusions

Data sharing

- Data sharing is critical given needed sample sizes, but much infrastructure needs to be developed.
- Models for data sharing exist and might be adapted to purpose.
- Not entirely clear what to share (outcomes), or how.

Key Conclusions

Meeting the population's needs

- Engagement/inclusion of 'population' early and often.
- Active management of inclusiveness of work is key.
- Ignoring personal (dis)utility for all participants in this space risks misadventure .

Action steps? - Near

What data do we collect and share?

- Establishing a process for identification and development of common outcomes and metrics for data sharing that are agreed upon by researchers and participants.

Who participates?

- Convene group to identify and develop tools to ensure early engagement, entry, and long-term meaningful participation of typically under-included population groups in developing clinical genomics programs.

Action steps? - Far

Evidence needs

- Engage key decision makers (including employers?) in discussions of value and process of developing models that meet their decision making needs.

Infrastructure

- Authoritative multi-stakeholder organization providing guidance to field?
- Common data model, test coding and result

Incentives

- Work on process to help ensure that payment for testing requires data sharing/ deposition into ClinVar or similar
- Coverage with evidence development/risk-sharing agreements

ACTION COLLABORATIVES

Genomics and Population Health Action Collaborative

Issue

Integrating genomics at the population health level has the potential to increase our understanding of disease, improve public health, reduce health disparities, and promote genomic literacy. While many of the goals of precision medicine focus on long-term discovery efforts, current evidence for certain genomic applications suggests that many lives could be saved now if these were implemented in the recommended populations. A coordinated, collaborative effort to engage key stakeholders is needed to identify current evidence and determine best practices for widespread integration in population health programs.

Activity

The Genomics and Population Health Action Collaborative, is an *ad hoc* activity under the auspices of the Roundtable on Genomics and Precision Health at the National Academies of Sciences, Engineering, and Medicine (the National Academies). The products of the action collaborative do not necessarily represent the views of any one organization, the Roundtable, or the National Academies and have not been subjected to the review procedures of, nor are they a report or product of, the National Academies.

Action Collaboratives

- DIGITizE: Displaying and Integrating Genetic Information Through the EHR
- Global Genomic Medicine Collaborative (G2MC)

Conclusions

- Inexpensive sequencing has made its population level application for health-related purposes feasible.
- Early adopter organizations in the U.S. and abroad are rapidly exploring this new space. Expansion is inevitable.
- Opportunities abound to leverage ongoing work to answer pressing questions related to the use of genomic sequence data to improve population health.