

Value of a Coordinated US Federal Strategy for Genomic Medicine

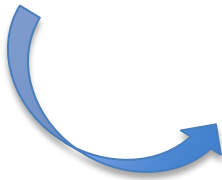
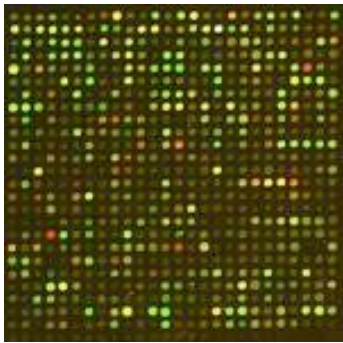
Geoffrey S Ginsburg MD PhD

May 28, 2013

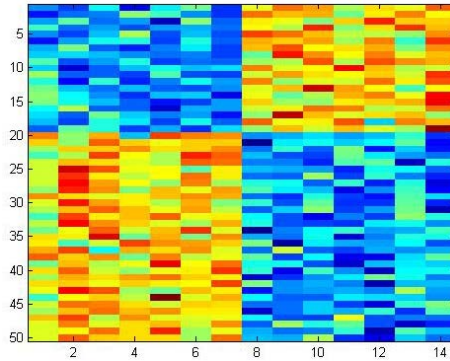
Genomic Medicine Working Group: Where we started – where we are going

- GM1: Barriers and solutions (home grown)
- GM2: Opportunities for demonstration projects
- GM3: Engagement of other stakeholders
(Payers, Diagnostics Labs)
- GM4: Engagement of professional societies
- GM5: Engagement of federal agencies
- GM6: International opportunities

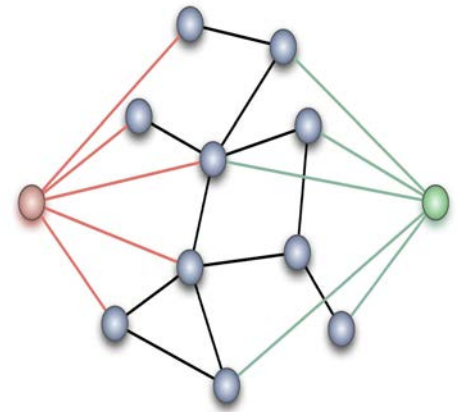
An 'Array' of Genomic Medicine Activities



GM1-4: Common GM Themes Mapped



A Network of Genomic Medicine Activities and Organizations



The Opportunity

- Accelerate the implementation of genomics research findings and technologies – *where appropriate* - to improve diagnosis, prognosis, drug safety, and overall quality of health care
- Standardize implementation and outcomes methods and measures across agencies (to enhance data sharing, accelerate the science)
- Coordinate implementation initiatives and catalyze cross-agency learning; reduce redundancy of efforts
- Develop a framework for enabling public-private partnerships
- Identify workforce and public education gaps and opportunities

The UK Human Genomics Strategy Group

- Monitor advances in genetic and genomics evaluate their benefit to healthcare services in the NHS
- Ensure successful translation of laboratory and academic research into quality care
- Develop infrastructure to enable access to high quality genomic and genetic testing services
- Develop the bioinformatics platform needed to underpin genomic and genetic testing
- Train the NHS and public health workforce
- Articulate the legal and ethical issues around the use of genomic data
- Raise public awareness of genomic technology and how it can be used to benefit the care of patients across the NHS

Is a National Strategy Possible?

The UK Has a Bold Vision

By 2020:

- *The NHS will be a world leader in the development and use of genomic technology in the areas of healthcare and public health*
- *Genomic information and clinical genetic testing will be used across the NHS*
- *Healthcare providers within the NHS will confidently use genomic information within their roles*
- *Effective public engagement will increase awareness of the role of genomic information in healthcare*

The UK Has Some Advantages

- The structure and scale of the (single payer) NHS
- Centralized and standardized data collection and increasing use of electronic health records
- A well developed network of NHS Regional Genetics Centers supported by molecular genetics and pathology laboratories
- A strong genomics research base: medical schools, universities and research institutions such as the Wellcome Trust Sanger Institute and the MRC Institute of Genetics and Molecular Medicine;
- Large research cohorts that offer unparalleled opportunities to explore the determinants of health throughout the life course.
- The UK Biobank

UK Biobank

- Initiated in 2006, £62M to initiate
- ~503,000 participants (2010)
- In depth phenotypes
- £10M for genome analysis of a subset
- Expansion in 2012 to include 8000 brain imaging
- ~ 20 projects underway

The screenshot shows the UK Biobank website homepage. At the top, the 'biobank^{uk}' logo is on the left, and the contact number 'Call Us On: 0800 0 276 276' is on the right, with a small icon of a person. Below the logo is a navigation menu with links: Home, About, Participants, Update Details, Resources, Scientists, Data Showcase, Register & apply, and Approved research. The main content area features a large image of a meeting with the text 'First research applications approved' and a 'Read more...' button. To the right of this image are two smaller news items: 'Genetics study targets serious disease: video' and 'First research applications approved'. Below the main image is a search bar for the FAQ, with an 'Ask now' button and a 'View all FAQs' link. At the bottom, there are two sections: 'Participants' with links for 'Update your contact details', 'Confirm or cancel appointment', 'Find out how the resource is being used', and 'Access Procedures', along with buttons for 'Activity monitor', 'Repeat assessment visit', and 'Look at the Data Showcase'; and a 'Poll' section asking 'Do you use your mobile phone to browse the Internet?' with 'Yes' and 'No' radio buttons and a 'Vote' button.

100,000 British Genomes

A new initiative lead by the UK's National Health Service aims to sequence the genomes of as many as 100,000 patients, a project that will cost £100 million.

By Jef Akst | December 10, 2012

- Whole genome sequencing at scale for the benefit of patients in the NHS
- Focusing initially on cancer, rare diseases and infectious diseases, we will efficiently sequence 100,000 whole genomes at diagnostic quality
- The Department of Health has committed up to £100m to this initiative that will be used:
 - to develop the necessary skills to support delivering the best patient outcomes
 - to support the linking of data and treatment outcomes which is essential for optimal patient care and future public and private research

UK Genomic Medicine Workforce Investment

- National oversight
- Deliver a pipeline of specialists in clinical bioinformatics knowledge and skills
- Building and developing the specialist genetics and genomics profession
- Develop skills for non-specialist healthcare professionals
- Investment in learning tools (e.g., e-learning and a competency framework)

Genome Canada and Canadian Institutes of Health Research

- Genome Canada: To harness the transformative power of genomics to deliver benefits to Canadians
- “Genomics and Personalized Health” RFA
 - to support projects that will demonstrate how genomics-based research can contribute to a more evidence-based approach to health and improving the cost-effectiveness of the health-care system.
- \$67.5 million available
 - \$40 million through Genome Canada
 - \$22.5 million through CIHR
 - \$5 million from the Cancer Stem Cell Consortium
- At least 50% of funding through co-funding
- 17 projects funded in 2013

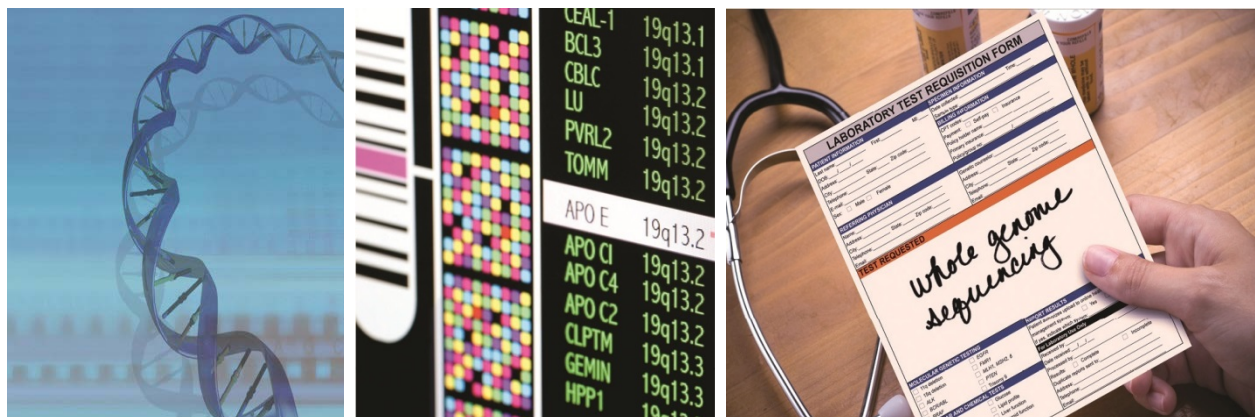


Canadian Institutes
of Health Research

Genome Canada – 2013 Project Examples

- **Biomarkers for Pediatric Glioblastoma**
 - Using mutations to guide therapy and measure outcomes
- **Personalized Genomics for Prenatal Aneuploidy Screening Using Maternal Blood**
 - Comparing effectiveness of different technologies for NIPT
- **Personalized Treatment of Lymphoid Cancer**
 - Sequencing to guide therapy, clinical CEA outcomes
- **Clinical Implementation and Outcomes Evaluation of Blood Biomarkers for COPD Management**
 - Use of biomarkers to diagnose COPD exacerbation and guide therapy
- **Viral and Human Genetic Predictors of Response to HIV Therapies**
 - Real time surveillance and monitoring drug resistance

Roundtable on Translating Genomic-Based Research for Health



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Roundtable on Translating Genomic-Based Research for Health

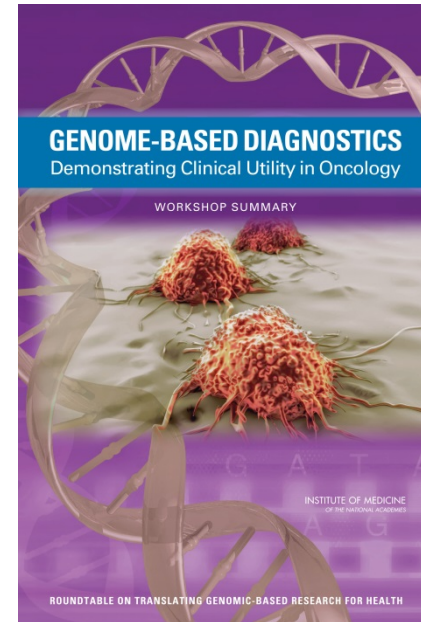
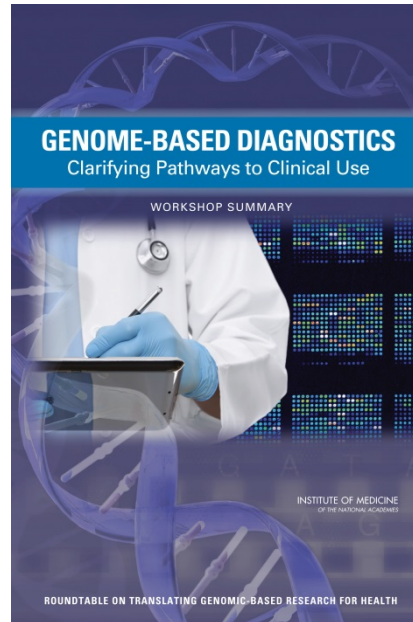
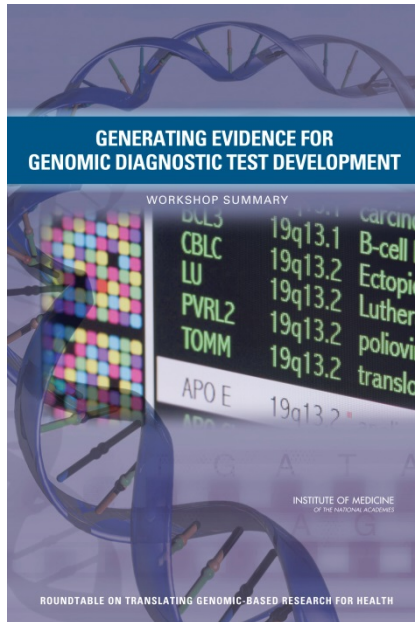
- Neutral convener for interested parties from government, academia, industry, and other stakeholders
- Explores and defines strategies for improving health through the translation of genomics and genetics research findings into medicine, public health, education, and policy
- Focal areas (2007-2012): Diagnostics, Drug Development, Public Health

Roundtable Membership

- **Wylie Burke (Co-Chair), UW**
- **Sharon Terry (Co-Chair), Genetic Alliance**
- **Naomi Aronson, BC-BSA**
- **Euan Ashley, AHA**
- **Paul R. Billings, Life Technologies**
- **Bruce Blumberg, Kaiser Permanente**
- **Pamela Bradley, FDA**
- **Philip J. Brooks, ORDR**
- **Ann Cashion, NINR**
- **C. Thomas Caskey, BCM**
- **Michael J. Dougherty, ASHG**
- **Victor Dzau, Duke University**
- **W. Gregory Feero, JAMA**
- **Andrew N. Freedman, NCI**
- **Geoffrey Ginsburg, Duke University**
- **Richard Hodes, NIA**
- **Mohamed Khan, AMA**
- **Muin Khoury, CDC**
- **Gabriela Lavezzari, PhRMA**
- **Thomas Lehner, NIMH**
- **Debra Leonard, CAP**
- **Kathryn McLaughlin, HRSA**
- **Kelly McVeary, Northrup Grumman**
- **Robert L. Nussbaum, UCSF**
- **Michelle Ann Penny, Eli Lilly**
- **Aidan Power, Pfizer Inc.**
- **Ronald Przygodzki, VA**
- **Allen D. Roses, Duke University**
- **Joan A. Scott, NCHPEG**
- **David Veenstra, UW**
- **Michael S. Watson, ACMG**
- **Daniel Wattendorf, Air Force**
- **Catherine A. Wicklund, NSGC**
- **Janet K. Williams, AAN**

Focal Area: Molecular Diagnostics

- Evidence generation
- Coverage and reimbursement
- Regulatory policies



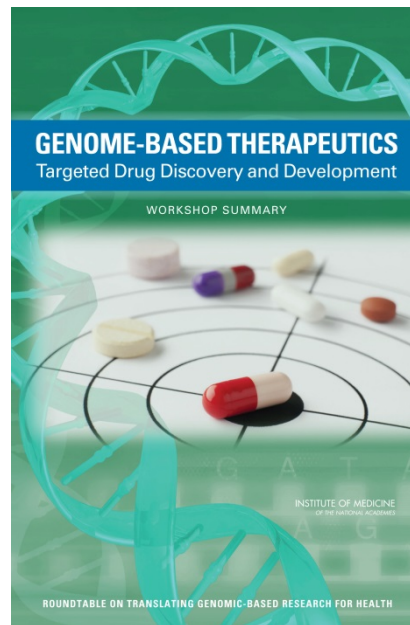
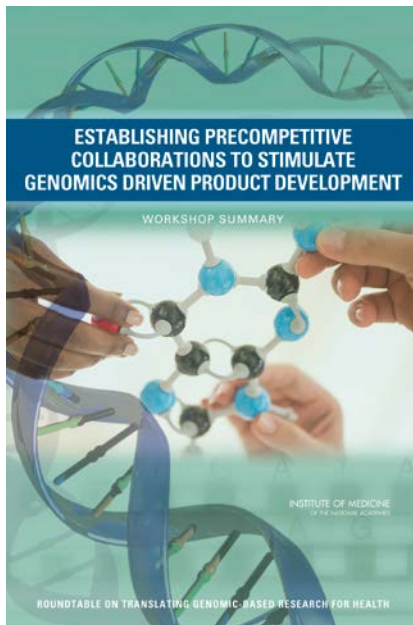
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Focal Area: Drug Discovery and Development

- Precompetitive collaborations/public-private partnerships
- Genome-based strategies for targeted therapeutic development
- Data sharing and biospecimens
- Drug Repurposing (June 24, 2013)



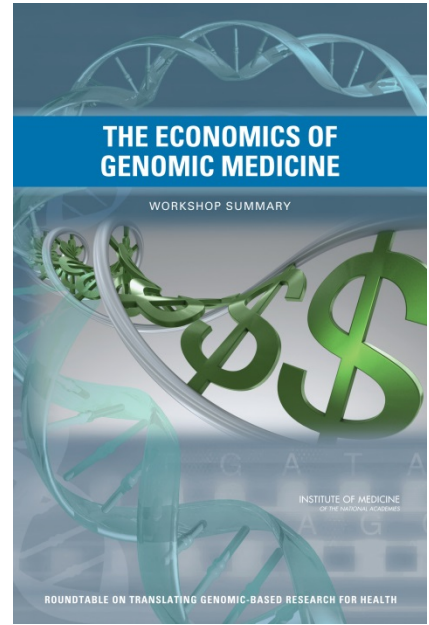
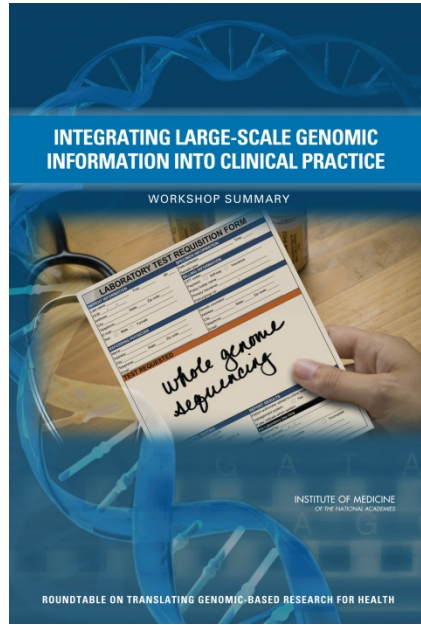
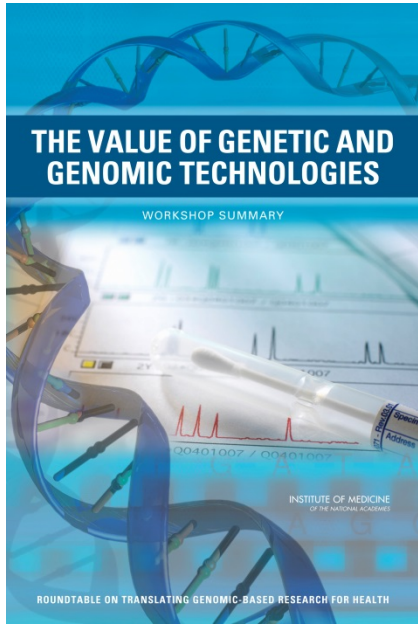
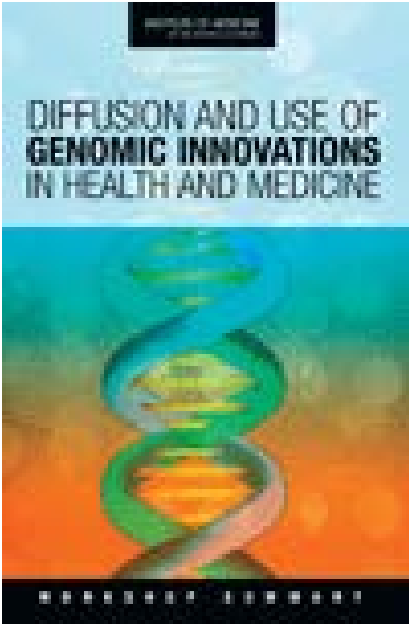
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Focal Area: Genomic Medicine and Public Health

- Implementation
- Genome technologies
- Health IT
- Health economics



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IOM Impact?

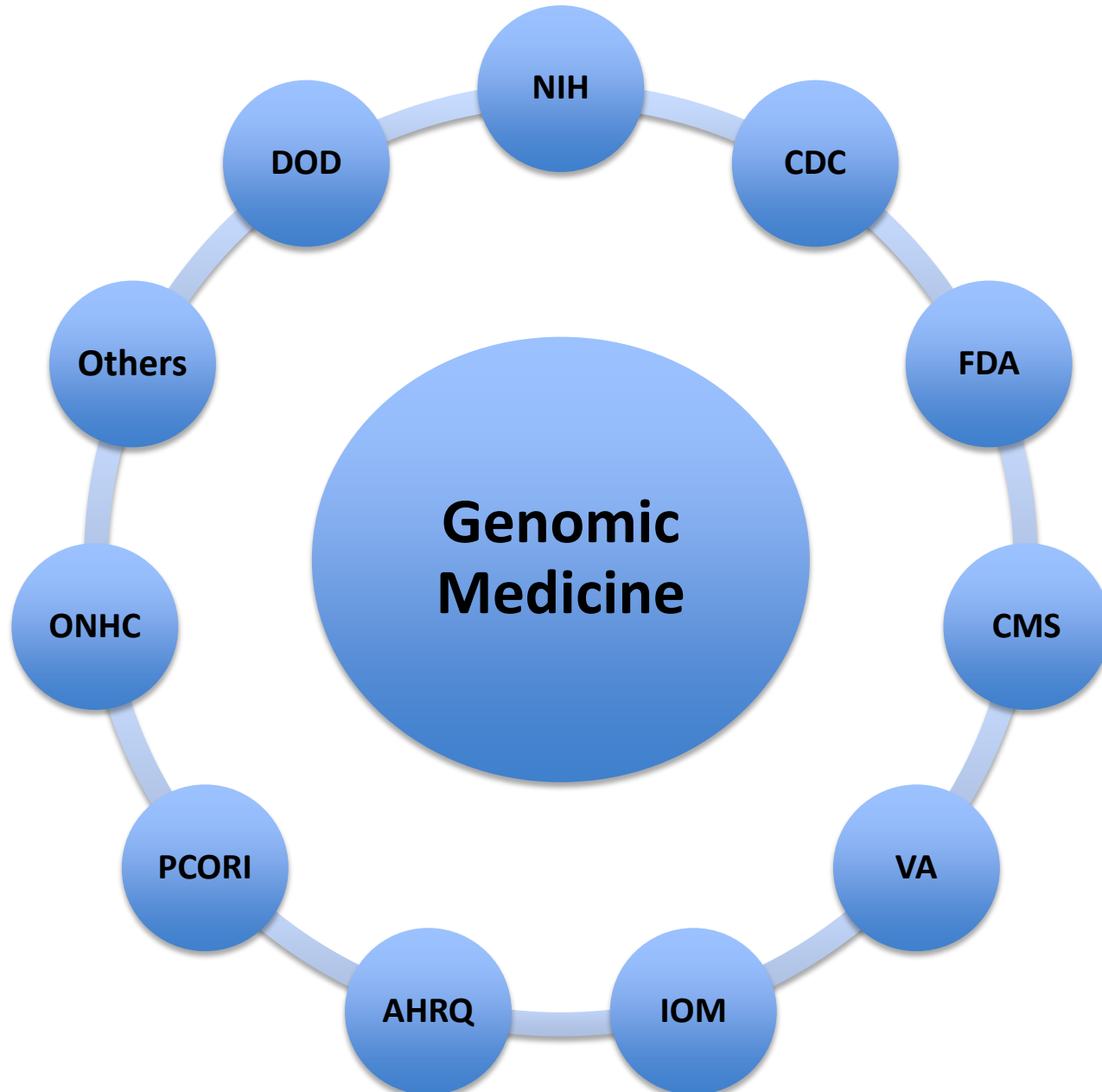
- Enabler of informed dialogue
- Fostering Collaboration and Partnership
 - Clinical Trial Comparator Arm Partnership
- Policy Impact
 - Data sharing and database development
- A Resource
 - Workshop proceedings
 - Whitepapers
 - Commentaries



IOM Focus Areas Advanced in 2013

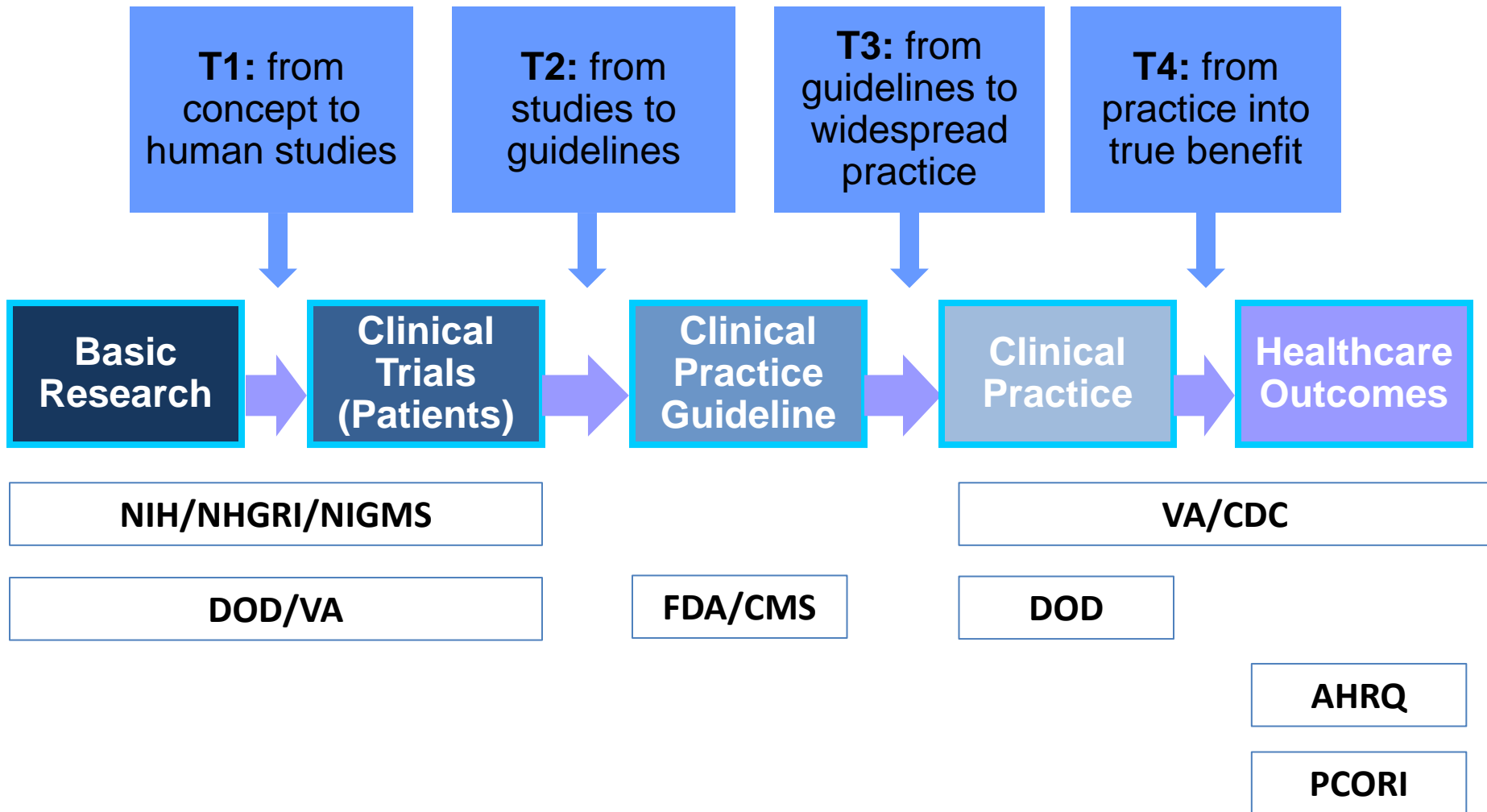
- Conflict of interest and medical innovation
- Education, engagement, and cultural change
- Developing a knowledgeable health care system for precision medicine
- Processes for evidence evaluation

The Genomic Medicine Federal Ecosystem



Translating Genomics to Health

Roadmap of Federal Agencies Along the Translational Pathway



GM5: Aspirations

- Understand the key issues in genomic medicine implementation facing each agency
- Is there consensus on the potential value of a US federal strategy
- How to go about developing such a strategy and gaining support for it
 - Key priorities
 - Major obstacles/challenges
 - Next steps?