#### Vision for the Cohort and the Precision Medicine Initiative

Francis S. Collins, M.D., Ph.D. Director, National Institutes of Health <u>Precision Medicine Initiative: Building a Large U.S. Research Cohort</u>

February 11, 2015

# President Obama's State of the Union Address: January 20, 2015



## Secretary Burwell Speaks Candidly at NIH: January 28, 2015





"And that's why we're here today. Because something called precision medicine ... gives us one of the greatest opportunities for new medical breakthroughs that we have ever seen."

> President Barack Obama January 30, 2015

#### The NEW ENGLAND JOURNAL of MEDICINE

January 30, 2015

## Perspective

#### A New Initiative on Precision Medicine

#### Francis S. Collins, M.D., Ph.D., and Harold Varmus, M.D.

Consight, I'm launching a new Precision Medicine Initiative to bring us closer to curing diseases like cancer and diabetes — and to give all of us access to the personalized information we need to keep ourselves and our families healthier."

- President Barack Obama, State of the Union Address, January 20, 2015

The proposed initiative has two main components: a nearterm focus on cancers and a longer-term aim to generate knowledge applicable to the whole range of health and disease. Both components are now within our reach because of advances in basic research, including molecular biology, genomics, and bioinformatics. Furthermore, the initiative

#### www.nih.gov/precisionmedicine

### **Precision Medicine**

#### **Concept is not new**

- Consider prescription eyeglasses, blood transfusions...
- Prospects for broader application raised by recent advances in basic research, technology development, genomics, proteomics, metabolomics, EMRs, Big Data, mHealth, etc.
- Reinforced by 2011 National Research Council report

#### What is needed now

- Development of rigorous research program to provide scientific evidence needed to turn concept into reality
- Recruitment of the best and brightest from multiple disciplines to join the team



## Precision Medicine Initiative: The Time is Right

- Advances in Scientific Knowledge
- Advances in Technology and Computing
- Americans' Growing Desire to Be Partners in Research
- Availability of Existing Research Cohorts



## Precision Medicine Initiative: The Time Is Right

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## The case for a US prospective cohort study of genes and environment

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National Human Genome Research Institute, National Institutes of Health, Building 31, Room 4B09, MSC 2152, 31 Center Drive, Bethesda, Maryland 20892-2152, USA (e-mail: fc23a@nih.gov)

Information from the Human Genome Project will be vital for defining the genetic and environmental factors that contribute to health and disease. Well-designed case-control studies of people with and without a particular disease are essential for this, but rigorous and unbiased conclusions about the causes of diseases and their population-wide impact will require a representative population to be monitored over time (a prospective cohort study). The time is right for the United States to consider such a project.



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#### **Precision Medicine Initiative**

Vision: Build a broad research program to encourage creative approaches to precision medicine, test them rigorously, and, ultimately, use them to build the evidence base needed to guide clinical practice.

- Near Term: apply the tenets of precision medicine to a major health threat – cancer
- Longer Term: generate the knowledge base necessary to move precision medicine into virtually all areas of health and disease

## Precision Medicine Initiative Proposed FY16 Support

Agency	\$ Million
National Institutes of Health <ul> <li>Cancer</li> <li>Cohort</li> </ul>	\$200 \$70 \$130
Food and Drug Administration	\$10
Office of the National Coordinator for Health Information Technology	\$5
TOTAL	\$215

#### **Precision Medicine Initiative: Near Term**

Apply tenets of precision medicine to cancer

- Identify new cancer subtypes, therapeutic targets
- Test precision therapies, with private sector partners
  - Wide spectrum of adult and pediatric cancers
  - Early stage to advanced disease
- Expand understanding of therapeutic response
  - Drug resistance
  - Combination therapy
  - Predicting and monitoring tumor recurrence



#### Precision Medicine Initiative: Longer Term

Generate knowledge base needed to move precision medicine into the whole range of health and disease

To reach this goal, the Initiative will support research to:

- Create new approaches for detecting, measuring, analyzing a wide array of biomedical variables: molecular, genomic, cellular, clinical, behavioral, physiological, and environmental
- Test these approaches in small, pilot studies
- Utilize the most promising approaches in greater numbers of people over longer periods of time to collect data of great value to both researchers and participants



### **Precision Medicine Initiative**

#### National Research Cohort

- >1 million U.S. volunteers
- Numerous existing cohorts (many funded by NIH)
- New volunteers
- Participants will be centrally involved in design and implementation of the cohort
- They will be able to share genomic data, lifestyle information, biological samples – all linked to their electronic health records



#### **Precision Medicine Initiative**

#### The National Research Cohort will:

- Provide scientists with a ready platform for:
  - Observational studies of drugs and devices
  - Tests of wearable sensors for monitoring health
  - More rigorous interventional studies
- Forge new model for scientific research that emphasizes engaged participants and open, responsible data sharing with privacy protections





### **Patient Partnerships**



#### EHRs



### Technologies



### Genomics



#### Data Science

## National Research Cohort: What Early Success Might Look Like

- A real test of pharmacogenomics—right drug at the right dose for the right patient
- New therapeutic targets by identifying loss-of-function mutations protective against common diseases
  - PCSK9 for cardiovascular disease
  - SLC30A8 for type 2 diabetes
- Resilience finding individuals who should be ill but aren't
- New ways to evaluate mHealth technologies for prevention/management of chronic diseases











## Precision Medicine: What Success Might Look Like

50-year-old woman with type 2 diabetes visits her doctor

#### Now

- Though woman's glucose control has been suboptimal, doctor renews her prescription for drug often used for type 2 diabetes
- Continues to monitor blood glucose with fingersticks and glucometer, despite dissatisfaction with these methods



## Precision Medicine: What Success Might Look Like

50-year-old woman with type 2 diabetes visits her doctor

#### Future: + 2 years

- Volunteers for new national research network
  - Sample of her DNA, along with her health information, sent to researchers for sequencing/analysis
  - Can view her health/research data via smartphone
- Agrees to researchers' request to track her glucose levels via tiny implantable chip that sends wireless signals to her watch, researchers' computers
  - Using these data, she changes diet, medicine dose schedule



## Other Diseases: What Success Might Look Like

50-year-old woman with type 2 diabetes visits her doctor

#### Future: + 5 years

- Receives word from her doctor about a new drug based upon improved molecular understanding of type 2 diabetes
- When she enters drug's name into her smartphone's Rx app, her genomic data show she'll metabolize the drug slowly
  - Her doctor alters the dose accordingly



### Other Diseases: What Success Might Look Like

50-year-old woman with type 2 diabetes visits her doctor

#### Future: + 10 years

- Celebrates her 60th birthday and reflects with her family about how proud she is to be part of cohort study
- Her glucose levels remain well controlled; she's suffered no diabetes-related complications
- Her children decide to volunteer for cohort study



## Building a Large U.S. Cohort for Precision Medicine Research

- NIH Workshop, February 11-12, 2015
- Representatives from a wide variety of fields
- Major areas of focus:
  - Cohort identification and participant recruitment
  - Participant engagement, data privacy, and novel ways of returning information to participants
  - Data collection, including mobile technologies
  - Informatics and electronic health records



### The Road Ahead

- Advisory Committee to the Director Working Group
  - Co-chairs Rick Lifton and Kathy Hudson
- Timeline
  - Planning in FY15
  - Begin building in FY16
- Need to expand the four workgroups
- Specific outreach meetings needed with:
  - Participants
  - Leaders of current cohorts
  - mHealth technology developers
- Coordinate with the White House and other agencies





#### www.nih.gov/precisionmedicine



Precision Medicine Initiative

What are the near-term goals? What are the longer-term goals? How is it different? Who will participate? NIH Workshop



The Precision Medicine Initiative: Infographic View larger (M (PDF - 163KB)

#### Precision Medicine Initiative

Far too many diseases do not have a proven means of prevention or effective treatments. We must gain better insights into the biology of these diseases to make a difference for the millions of Americans who suffer from them. Precision medicine is an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person. While significant advances in precision medicine have been made for select cancers, the practice is not currently in use for most diseases. Many efforts are underway to help make precision medicine the norm rather than the exception. To accelerate the pace, President Obama has now unveiled the Precision Medicine Initiative — a bold new enterprise to revolutionize medicine and generate the scientific evidence needed to move the concept of precision medicine into every day clinical practice.



Email Updates To sign up for updates please enter your e-mail address.



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#### **Related Links**

NEJM Perspective: A New Initiative on Precision Medicine

White House Precision Medicine Web Page

White House Fact Sheet: President Obama's Precision Medicine Initiative

Precision Medicine Initiative and Cancer Research

Storify: The Precision Medicine Initiative

## I not only use all the brains I have, but all I can borrow.

- Woodrow Wilson



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