Human Genome Project: 1990-2003
A vision for the future of genomics research
A blueprint for the genomic era.

Perspective
Charting a course for genomic medicine from base pairs to bedside.

Nature 2003
Nature 2011

Base Pairs to Bedside

2003 2011

Helix to Health
An emerging medical discipline that involves using genomic information about an individual as part of their clinical care (e.g., for diagnostic or therapeutic decision-making) and the other implications of that clinical use.
Green et al. 2011

Understanding the Structure of Genomes
Understanding the Biology of Genomes
Understanding the Biology of Disease
Advancing the Science of Medicine
Improving the Effectiveness of Healthcare

1990-2003 Human Genome Project

2004-2010 Basic Genomics Discovery Genomic Medicine

2011-2020

Beyond 2020
A Quarter Century of Genomics

Human Genome Sequenced for First Time by the Human Genome Project

Cost of Sequencing a Human Genome Reduced Nearly ~1 Million-Fold
Sequencing a Human Genome

Human Genome Project
(1st Sequence)

~$1B
~6-8 years

Today

~$2-4K
~1-3 days
A Quarter Century of Genomics

Human Genome Sequenced for First Time by the Human Genome Project

Cost of Sequencing a Human Genome Reduced Nearly ~1 Million-Fold

Tens of Thousands of Human Genomes Sequenced
Your Genome: By the Numbers

- ~6B nucleotides
- ~3-5M single-nucleotide variants
  - ~150K not in databases
  - ~60 not in either parent
A Quarter Century of Genomics

- Human Genome Sequenced for First Time by the Human Genome Project
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- Tens of Thousands of Human Genomes Sequenced
- Profound Advances in Understanding How the Human Genome Functions
A Quarter Century of Genomics

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- Significant Advances in Unraveling the Genomic Bases of Human Disease
Genomic Architecture of Genetic Diseases

Rare, Simple, Monogenic, Mendelian…

Common, Complex, Multigenic, Non-Mendelian…

Manolio et al., J Clin Invest (2008)
A Quarter Century of Genomics

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- Tens of Thousands of Human Genomes Sequenced
- Profound Advances in Understanding How the Human Genome Functions
- Significant Advances in Unraveling the Genomic Bases of Human Disease
- Vivid Examples of Genomic Medicine in Action Now Emerging
Routine Clinical Diagnostic Tools

Radiographic Imaging

Cancer Genome Sequencing
‘Hot Areas’ in Genomic Medicine

Cancer Genomics

Pharmacogenomics
Pharmacogenomics
“Children born with disorders not readily explained by standard tests can sometimes be diagnosed through genome sequencing and analysis.”

‘Hot Areas’ in Genomic Medicine

1. Cancer Genomics
2. Pharmacogenomics
3. Ultra-Rare Genetic Disease Diagnostics
4. Genomic Medicine ‘Test Drive’ Programs
‘Hot Areas’ in Genomic Medicine

- Cancer Genomics
- Pharmacogenomics
- Ultra-Rare Genetic Disease Diagnostics
- Genomic Medicine ‘Test Drive’ Programs
- Prenatal & Newborn Genomic Analysis
Noninvasive Prenatal Genome Sequencing

In 2025, Everyone Will Get DNA Mapped At Birth

Alice Park | @aliceparkny | June 30, 2014

Scientists have scoured trends in research grants, patents and more to come up with these 10 innovations that will be reality in 10 years (or so they think).

Everybody likes to blue-sky it when it comes to technology. Driverless cars! Fat-burning pills! Telepathic butlers! But the folks at Thomson Reuters Intellectual Property & Science do it for a living—and they do it with data.

Time (2014)
Genome Sequencing of Acutely Sick Newborns

The genomes of ill newborns can be sequenced in less than 24 hours to give clinicians a rapid diagnosis.

GENOMICS

Fast sequencing saves newborns

Rapid analysis of infant genomes is aiding diagnosis and treatment of inexplicably ill babies.

Nature (2014)
‘Hot Areas’ in Genomic Medicine

- Cancer Genomics
- Pharmacogenomics
- Ultra-Rare Genetic Disease Diagnostics
- Genomic Medicine ‘Test Drive’ Programs
- Prenatal & Newborn Genomic Analysis
- Clinical Genomics Information Systems
Generating a Human Genome Sequence is (Almost) Trivial
Clinical Genomics Information Systems
The Relevance of Genomics

Biomedical Researchers

Healthcare Professionals

Patients (and Friends & Relatives of Patients)
Genomics and Society
Breaking News
I. The Story
II. The Vision
III. The Plan(ning)
I. The Story
II. The Vision
III. The Plan(ning)
President Obama: Long-Standing Interest in Genomics

Senator Obama, 2006
Precision Medicine

A broader context for ‘individualizing’ medical care to advance human health
Precision Medicine

- **Today:** most medical care based on expected response of the average patient

- **Tomorrow:** medical care based on individual genomic, environmental, and lifestyle differences that enable more precise ways to prevent and treat disease

How do we get from today to tomorrow?
Precision Medicine

- Concept is not entirely new:
  - Prescription Eyeglasses
  - Blood Transfusions

- What is needed now:
  - Rigorous research program
  - Multidisciplinary approach
  - Audacious scale and scope

How best to ‘jump start’ precision medicine?
“...the budget I send this Congress on Monday will include a new Precision Medicine Initiative that brings America closer to curing diseases like cancer and diabetes, and gives all of us access, potentially, to the personalized information that we need to keep ourselves and our families healthier.”

President Barack Obama
January 30, 2015
Obama seeks $215 million for personalized medicine effort

Obama to Unveil Research Initiative to Develop Tailored Medical Treatments

Obama Announces $215 Million Precision-Medicine Genetic Plan

Obama's $215 Million DNA Sequencing Project Is A Great Idea

A Path for Precision Medicine

Obama Seeks Millions for 'Precision Medicine'
White House fleshes out Obama’s $215 million plan for precision medicine

Obama Enumerates Precision Medicine Initiative
The President requests $215 million to launch his push for personalized clinical care.

Obama to seek $215 million for precision-medicine plan
Details emerge as White House prepares to release budget request to Congress.

U.S. to Develop DNA Study of One Million People
An Obama initiative seeks to channel a torrent of gene information into treatments for cancer, other diseases.
I. The Story
II. The Vision
III. The Plan(ning)
A New Initiative on Precision Medicine

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

“Tonight, 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 near-term 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
Precision Medicine Initiative: The Vision

- **NEAR TERM**: Cancer as a Model of Precision Medicine
  - Cancer as leading edge of precision medicine, yet more to learn
  - Ramp up current efforts to include more cancer types

- **LONGER TERM**: Expanding the Model to Other Diseases
  - Create national research cohort of >1 million volunteers
  - Generate knowledge base for precision medicine

- **POLICY CHANGES**: Remove Barriers to Clinical Implementation
  - Update federal rules protecting research participants
  - Advance FDA oversight of precision medicine products
## Precision Medicine Initiative: Proposed Fiscal Year 2016 Funding

<table>
<thead>
<tr>
<th>Agency</th>
<th>$ Million</th>
</tr>
</thead>
<tbody>
<tr>
<td>National Institutes of Health</td>
<td>$200</td>
</tr>
<tr>
<td>• Cancer</td>
<td>$70</td>
</tr>
<tr>
<td>• Cohort</td>
<td>$130</td>
</tr>
<tr>
<td>Food and Drug Administration</td>
<td>$10</td>
</tr>
<tr>
<td>Office of the National Coordinator for Health Information Technology</td>
<td>$5</td>
</tr>
<tr>
<td>TOTAL</td>
<td>$215</td>
</tr>
</tbody>
</table>
I. The Story
II. The Vision
III. The Plan(ning)
NEAR TERM: Cancer as a Model of Precision Medicine

Ramp up current efforts to include more cancer types.

LONGER TERM: Expanding the Model to Other Diseases

Create national research cohort of >1 million volunteers.

Generate knowledge base for precision medicine.

POLICY CHANGES: Remove Barriers to Clinical Implementation

Update federal rules protecting research participants.

Advance FDA oversight of precision medicine products.

Precision Medicine Initiative: The Vision

NEJM (February 17, 2015)

Cutting the Gordian Helix — Regulating Genomic Testing in the Era of Precision Medicine

Eric S. Lander, Ph.D.

POLICY CHANGES: Remove Barriers to Clinical Implementation

Update federal rules protecting research participants.

Advance FDA oversight of precision medicine products.
National Research Cohort

- >1 million U.S. volunteers
  - Numerous existing cohorts (many funded by NIH)
  - New volunteers

- Participants to share genomic data, lifestyle information, biological samples – all linked to their EHRs

- Forge new model for ‘doing science’ that emphasizes:
  - Engaged participants
  - Open, responsible data sharing
  - Strong privacy protections

- Convert healthcare system into a ‘learning system’
The case for a US prospective cohort study of genes and environment

Francis S. Collins

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.

Precision Medicine Initiative: Next Steps

- **Working Group of Advisory Committee to the Director**
  
  - Intense planning for next ~4-6 months
  - Interim report in September

- **Additional meetings/workshops**

- **Implementation**
  
  - Trans-NIH implementation model
  
  - Fall 2015: Initial funding opportunities announced
  
  - Fiscal Year 2016: Funding begins
Emphasis on Transparency
www.nih.gov/precisionmedicine
PRECISION MEDICINE INITIATIVE

Events

The following are upcoming and past events held by the NIH to gather input from participant, scientific, and other stakeholder groups as it plans the development of the President’s Precision Medicine Initiative and the vision for building the national participant group. Please sign up for updates or check back frequently for additional meeting information.

All public workshops will be video-cast live.

UPCOMING EVENTS

- Public workshop on unique scientific opportunities for the national research cohort
  Tuesday, April 28, 1:00-5:30 p.m. ET and Wednesday, April 29, 2015, 8:30 a.m.-12:30 p.m. ET
  National Institutes of Health, Bethesda, Md.
  Registration details

- Public Workshop on Obesity and Electronic Health Records
  Thursday, May 28 - Friday, May 29, 2015, Time to be Determined
  Vanderbilt University, Nashville, Tenn.

- Public workshop on participants and community engagement
  Wednesday, July 1 - Thursday, July 2, 2015, Time to be Determined
  National Institutes of Health, Bethesda, Md.

- Public workshop on mHealth
  Monday, July 27 - Tuesday, July 28, 2015
  Intel Corp., Santa Clara, Calif.

PAST EVENTS

2015

- Public Workshop on Building a Precision Medicine Research Cohort
  Wednesday, February 11-Thursday, February 12, 2015
  National Institutes of Health, Porter Neuroscience Building, Bethesda, Md.

www.nih.gov/precisionmedicine/events.htm
‘Precision medicine’ takes aim at tumors, one by one
You’re already carrying a powerful medical research tool.

Hundreds of millions of people around the world have an iPhone in their pocket. Each one is equipped with powerful processors and advanced sensors that can track movement, take measurements, and record information — functions that are perfect for medical studies. The sheer number of them being used across the globe opens up new possibilities for researchers. With ResearchKit, researchers can easily create apps that take advantage of iPhone features to gather new types of data on a scale never available before.

Medical research is hard. It’s complex, it’s crucial, and it’s how we’re constantly improving the health of people around the world, and ourselves. To help, we’ve created ResearchKit to make it easy for researchers and medical institutions to create apps that gather data from their patients.

ResearchKit is already being used today. Several of the world’s leading medical institutions are already using ResearchKit to gain further insight into some of our most serious diseases. Learn more about their stories below.

You can also download these apps from the App Store.

www.apple.com/researchkit
Déjà Vu, All Over Again?

Human Genome Project
Circa Spring 1990

Precision Medicine Initiative
Circa Spring 2015