



National Human
Genome Research
Institute



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Health



U.S. Department
of Health and
Human Services

Precision Medicine

A broader context for
'individualizing' medical care to
advance human health

Teri Manolio, M.D., Ph.D.
Cross-Cohort Collaboration Meeting
March 7, 2015

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





“And that’s why 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



THE PRECISION MEDICINE INITIATIVE



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.

“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

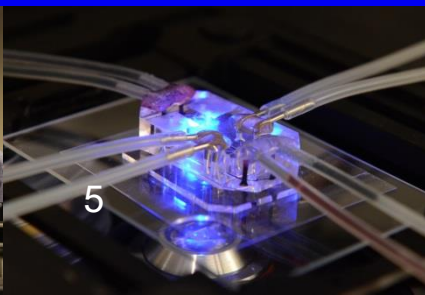
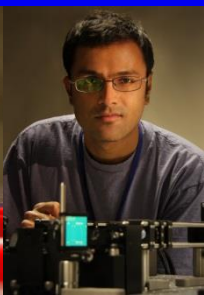
Precision Medicine

Concept is not new

- Consider prescription eyeglasses, blood transfusions...
- Prospects for broader application raised by recent advances in basic research, technology development, genomics and other -omics, EMRs, Big Data, mHealth

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

	Ten Years Ago	Now – 2014 (most recent data)
Cost of sequencing a human genome	\$22,000,000	\$1000 - \$5000
Time to sequence a human genome	2 years	<1 day
Number of smart phones in the United States	1 M (<2%)	160 M (58%)
EMR adoption, (% providers)	20-30%	>90%
Computing power	n	n x 16



Precision Medicine Initiative: The Vision

- Goal: Build a broad research program to encourage creative approaches to precision medicine, test them rigorously, use them to build the evidence base needed for clinical practice.
- Near Term: Cancer as a Model of Precision Medicine
Ramp up current efforts to include more cancer types
- Longer Term: Expand the Model to Other Diseases
Create national research cohort of >1M volunteers
- Policy Changes: Remove Barriers to Clinical Implementation
Privacy, regulation



Precision Medicine Initiative: Proposed Fiscal Year 2016 Funding

Agency	\$ Millions
National Institutes of Health	200
Food and Drug Administration	10
Office of the National Coordinator for Health Information Technology	5
Total	215



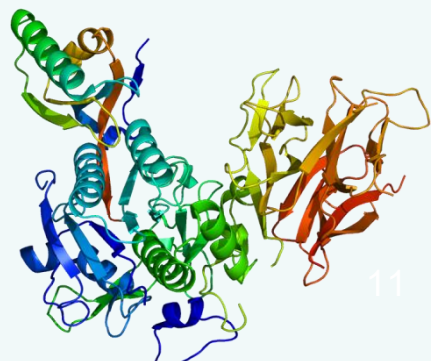
National Research Cohort



- **>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**
- **Provide scientists with a ready platform for myriad new studies to propel understanding of health and disease**
- **Forge new model for ‘doing science’ that emphasizes engaged participants and open, responsible data sharing with strong privacy protections**

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 LoF mutations protective against common diseases
- Resilience – finding individuals who should be ill but aren't
- New ways to evaluate mHealth technologies for prevention/management of chronic diseases



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



- NIH workshop held February 11-12, 2015
- Representatives from a wide variety of fields
- Major areas of discussion:
 - 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

Creation of a National Cohort

- What is the problem the cohort is trying to solve?
- Can we develop a set of compelling use cases, yet avoid being too prescriptive?
- What's the right blend of bringing existing cohorts together and starting new ones?
- What's unique about this initiative?
- Why is 1 million the right number? Need more?
- How does the cohort support discovery and also allow testing of interventions?
- How critical is it that point-of-care health care delivery is included in cohort structure?
- How will health disparities be addressed?

Creation of a National Cohort (continued)

- Are children included?
- Sustainability-- value grows over time
- Learn from prior successful and unsuccessful cohort projects
- Link with other international cohort projects
- Define the basic phenotype, lab, and genotype data to be sought for all participants
- Consider more than one tier:
 - Fully engaged, fully participating, with EHRs, laboratory data,...
 - EHRs and permission for recontact only – “Cohort Lite”

Next Steps

- Stand up Working Group of ACD
 - Name members (February 2015)
 - Collect further information based on output from this meeting
 - Begin outlining the plan
 - Interim Report to ACD in September to inform NIH decisions in FY16
- Consultation and Information Collection
 - Cohort leaders
 - mHealth folks
 - Potential participants



Precision Medicine Initiative

- [What are the near-term goals?](#)
- [What are the longer-term goals?](#)
- [How is it different?](#)
- [Who will participate?](#)
- [NIH Workshop](#)

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.

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](#)

Possible Use Cases (more needed)

- Identification and testing of biomarkers and predictive strategies for many common diseases
- Unbiased quantitative determination of risk (environmental exposures, penetrance, GxE)
- Powerful test bed for mHealth applications to health maintenance and chronic disease mgmt
- Rigorous test of PGx
- “The Human Knockout Project” – determining phenotype of LoF mutations
- Test bed for improving utility of EHRs for all
- Test ability of health care delivery systems to respond quickly to evidence – learning system

Participants as Partners

- Partners aren't just "at the table," but planning the meal (and bringing the food) – avoid the pt subcommittee model, aim for joint membership
- How do we identify those who can fairly represent the participant voice for this initiative?
- What research data are returned to participants?
- Define specific consequences of violating prohibition against re-identification-- GA4GH
- Can anyone join or must you have certain characteristics (like an interoperable EHR)?
- Emphasize opportunity to contribute to a larger national purpose, not just one's own care