

DEPARTMENT OF HEALTH AND HUMAN SERVICES  
NATIONAL INSTITUTES OF HEALTH

Precision Medicine Initiative

Witness appearing before the  
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Good afternoon, Chairman Alexander, Ranking Member Murray, and distinguished Members of the Committee. I am Francis S. Collins, M.D., Ph.D., and I am the Director of the National Institutes of Health (NIH).

It is an honor to appear before you today, alongside my dedicated colleagues, to discuss how we, as a Nation, can advance the health of the American public by accelerating progress toward a new era of precision medicine.

As the Nation's premier biomedical research agency, NIH's mission is to seek fundamental knowledge about the nature and behavior of living systems, and to apply that knowledge to enhance human health, lengthen life, and reduce illness and disability. I can report to you today that NIH leadership, employees, and grantees continue to believe passionately in that mission.

In January of this year, the President announced a new Precision Medicine Initiative—a bold, new research effort to revolutionize how we diagnose and treat disease, including a \$215 million investment in the President's Fiscal Year (FY) 2016 Budget. We believe that the time is right for this ambitious initiative, and the NIH and our partners, the U.S. Food and Drug Administration (FDA) and the Office of the National Coordinator for Health Information Technology (ONC), will work hard to achieve this vision.

Historically, physicians have had to make most recommendations about disease prevention and treatment based on the expected response of the average patient. This one-size-fits-all approach works for some patients and some conditions, but not others. Precision medicine is an innovative approach that takes into account individual differences in patients' genes, environments, and lifestyles. This concept is not new; blood typing, for example, has been used to guide blood transfusions for more than a century. Prescription eyeglasses are tailored

specifically to the patient's individual needs. Moreover, the identification of the *BRCA1* and *BRCA2* genes has made it possible to provide options for women at high risk for breast and ovarian cancers. And, the gene implicated in cystic fibrosis has led to widespread availability of screening and targeted therapeutics.

The prospect of applying this concept broadly has been dramatically improved by the development of powerful and affordable methods for characterizing personal biological attributes (such as genomics and metabolomics), the widespread adoption of electronic health records, the recent revolution in mobile health technologies, and the emergence of computational tools for analyzing large biomedical data sets. These advances will help make possible the dream of personalizing a wide range of health applications.

With this in mind, we are excited to take a lead in the two key components of the President's Precision Medicine Initiative that will be managed by NIH: a near-term goal that will focus on cancer and a longer-term aim to generate knowledge applicable to the whole range of health and disease. Both components are within reach, due in large part to scientific breakthroughs in basic research. Furthermore, the initiative will tap into converging trends in connectivity, through social media and mobile devices, and Americans' growing desire to be active partners in medical research in a way that protects their privacy.

Oncology is the clear choice for enhancing the near-term impact of precision medicine. Cancers are common diseases and are among the leading causes of death nationally and worldwide, and their prevalence is increasing as the population ages. They are especially feared because of their lethality, their symptoms, and the often toxic therapies used to treat them. Cancer research has been leading the way in precision medicine for many years. Thanks to advances in DNA sequencing and efforts such as The Cancer Genome Atlas project, we now

have a better understanding of the molecular changes that drive many cancers and we can define the driver mutations in individual tumors and use this information to design the ideal therapy for each patient. Genomic information has already helped shape the development of some cancer treatments. For example, the drug, imatinib (Gleevec), was designed to inhibit an altered enzyme produced by a fused version of two genes found in chronic myelogenous leukemia.

While we've made significant strides in recent years to learn the molecular signatures of many cancers, much more remains to be done. The National Cancer Institute will accelerate the design and testing of effective, tailored treatments for cancer by expanding genetically based clinical cancer trials, exploring fundamental aspects of cancer biology, and establishing a national "cancer knowledge network" that will generate and share new knowledge to fuel scientific discovery and guide treatment decisions. Furthermore, we aim to understand the development of resistance to targeted therapy, apply non-invasive methods to track patients' responses to treatment such as liquid biopsies, and explore the efficacy of new drug combinations targeted to specific tumor mutations.

As a longer term goal of this initiative, NIH will launch a national research cohort of one million or more volunteers who will play an active role in how their genetic, environmental, and medical information is used for the prevention of illness and management of a wide array of chronic diseases. This component will pioneer a new model for doing research; one in which people who participate are true partners. Not subjects, not patients—partners. The goal will be to expand the benefits of precision medicine into myriad aspects of health and health care. Participants will voluntarily share clinical data from electronic health records, results of imaging and laboratory tests, lifestyle data and environmental exposure recordings tracked through real time mobile health devices, and genomic information – all with appropriate privacy protections.

Participants will be at the center of the project design, and they will have access to their own health data, as well as research using their data, to help inform their own health decisions. As volunteers, each individual will participate because they choose to be a partner in this bold research effort. Through this dynamic community, researchers will be able to advance the information derived from this cohort into new knowledge, approaches, and treatments.

Researchers from many organizations will, with proper protection of patient information, have access to the cohort's data so that the world's brightest, scientific and clinical minds can contribute insights.

In order to help inform the vision for building the national research cohort of one million or more volunteers, a Precision Medicine Initiative Working Group was recently created. This group of experts in precision medicine and large clinical research studies is seeking public input from the diverse stakeholder community interested in the development of this initiative, including the patient community, and will articulate the vision for advancing participant engagement. They will help define what can be learned from a study of this scale and scope, what issues will need to be addressed as part of the study design, and what success would look like in the near and longer term. With the guidance from this team of experts, we will move ever closer to realizing the goals of this ambitious research program.

A project of this magnitude will lay the foundation for a myriad of new prevention strategies and novel therapeutics. Although the initiative will likely yield its greatest benefits years down the road, there will be successes in the relatively near future as well, especially in the areas of cancer and pharmacogenomics—how to provide the right drug at the right dose to the right person at the right time. Moving forward, this pioneering research initiative will require the involvement of many different sectors of science and society, including biologists, physicians,

technology developers, data scientists, and especially the American people. Given related efforts in a few other countries, we will aim to forge collaborations on a global scale.

With sufficient resources and a strong, sustained commitment of time, energy, and ingenuity from the scientific, medical, and participant communities, precision medicine's full potential can be realized to give everyone the best chance at good health. There's no better time than now to embark on this ambitious new enterprise to revolutionize medicine and generate the scientific evidence necessary to move this individualized approach into everyday clinical practice.

With your support, the future of medicine can be very bright. This concludes my testimony, and I look forward to answering your questions.