

Obama's Precision Medicine Initiative

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ON JANUARY 30, 2015, I had the great honor of attending the President's announcement about the Precision Medicine Initiative in the East Room of the White House. Along with many hard-working, visionary luminaries in clinical science, informatics, advocacy, and policy, I listened with great enthusiasm to the fabulous announcement. It was clear from the introduction, given by Elana Simon, that the focus for this initiative will be people.

Elana is a college freshman at Harvard University. So are about 2000 other young people. It was she who introduced President Obama on this special day because Elana is a "citizen scientist." When she was 12 years old, she was diagnosed with fibrolamellar hepatocellular carcinoma, a rare liver cancer. When she was 15 years old, she took on this disease that she herself had beat. As a computer-literate adolescent, Elana thought she could put those skills to work, and as she outsurvived so many friends affected by this rare disease, she decided to do a genomic analysis of her cancer. Realizing that an obvious suspect, like an oncogene, was responsible, it would have been discovered, Elana went after the whole genome and RNA sequencing rather than just exome analysis. She consulted with several scientists and institutions—they were generous with their time and resources. This is a rare cancer, so samples were scarce and some institutions were not willing to share. So she fired up a plea using social media, found other patients through Facebook, and created a YouTube video asking patients to liberate their samples from the hospitals where they had their surgeries.*

Elana and other patients with fibrolamellar found a single deletion in the DNA of one copy of chromosome 19 in every affected individual. She published a paper in *Science* (Honeyman et al., 2014). With her dad, Sanford Simon, a scientist at Rockefeller University, she has set up two clinical trials and created a blood test for the cancer.

This is all remarkable, and meeting Elana and her family was one of the highlights of my experience at the White House. However, Elana's experience represents an expression of the bigger message for the day: the intersection of participant engagement and precision medicine. In the beginning of her remarks, Elana specifically thanked the community of patients who had made this work possible. She said, "Patients contributed in different ways, including fund-raising, collecting patient data, donating tissue and by

working in the lab." President Obama clearly stated in his description of the Initiative that people would be *participants* in this new initiative. He acclaimed the patient rights' advocates that we present as critical to the process.

The National Institutes of Health (NIH) describes a central component of the effort this way (emphasis mine):

Creation of a voluntary national research cohort: NIH, in collaboration with other agencies and stakeholders, will launch a national, *patient-powered* research cohort of one million or more Americans who volunteer to participate in research. *Participants will be involved in the design of the Initiative* and will have the opportunity to contribute diverse sources of data—including medical records; profiles of the patient's genes, metabolites (chemical makeup), and microorganisms in and on the body; environmental and lifestyle data; *patient-generated information*; and personal device and sensor data. Privacy will be rigorously protected. This ambitious project will leverage existing research and clinical networks and build on innovative research models that enable patients to be active participants and partners. The cohort will be broadly accessible to qualified researchers and will have the potential to inspire scientists from multiple disciplines to join the effort and apply their creative thinking to generate new insights. The ONC [Office of the National Coordinator for Health Information Technology] will develop interoperability standards and requirements to ensure secure data exchange with patients' consent, to empower patients and clinicians and advance individual, community, and population health (NIH, 2015).

In a small luncheon following the event for research participants and advocates, Francis Collins, director of the NIH, spoke with great enthusiasm of the need to gain better insights into the biology of all diseases to make a difference for the millions of Americans who suffer from them. Dr. Collins paved the way for this day with his visionary leadership in sequencing the human genome and in leading the NIH. He was clear that we, the people, must lead this initiative.

NIH Deputy Director for Science, Outreach and Policy Kathy Hudson "The Initiative will set the foundation for new ways of engaging research participants, sharing health data and information, and employing technology advances to mine the information for comprehensive results. This is not lip service, this is real commitment."

What does this have to do with genetic testing and molecular biomarkers? Obviously these sciences, as they mature, are critical to realizing the promise of this Initiative. The challenges intrinsic to the science—analytic and clinical validity

*(https://www.youtube.com/watch?v=Y5lkp_uK9Ww).

and clinical utility—are key. Finding ways to accelerate evidence development and understanding the issues inherent in implementation are essential to advancing precision medicine. Policy issues, including regulation, are also key aspects of what must be elucidated and solved. The President himself stated: “We’re going to work with the FDA [Food and Drug Administration] to develop new approaches for evaluating next-generation genetic tests. The way we approve a new gene-sequencing technology is going to be different than the way we approve a new pacemaker or prosthetic device. And we need to make sure that our approach reflects the difference in technology” (Obama 2015).

The science has come of age. Now it remains for us to motivate the public to truly participate and to provide incentives for researchers and clinicians to collaborate to build a new culture of openness that allows the existing networks in the nation to weave together the large cohorts that will be necessary. There exists a fabulous foundation in NIH’s Clinical and Translational Science Awards, the Patient-Centered Outcomes Research Institute’s PCORnet, and FDA’s Sentinel, among many other networks in various integrated health services organizations. Add to this the people—the participants both formally organized and those in loose affinity groups—and let the revolution begin!

References

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