# Precision Health for All: NIH’s Precision Medicine Initiative Cohort Program

<https://www.youtube.com/watch?v=CjPPcTnme9w>

## Title Slide

Precision Health for All

NIH’s Precision Medicine Initiative® Cohort Program

Eric Dishman

Director, PMI Cohort Program

National Institutes of Health

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## Scene Change

Mr. Dishman speaking at the front of a conference room. He is holding a remote control for a projector. Behind him is a lectern with “National Institutes of Health” inscribed on it.

## Mr. Dishman:

I want to go on record in saying Eric [Green, Director, National Human Genome Research Institute, National Institutes of Health] specifically and Terry and Carolyn Hunter have been fantastic at both trying to get me at least a little less stupid about what NHGRI does, as well as just helping me settle as we move across the country from Portland to the area. So that’s which grocery stores to go to, which traffic to avoid, all of those are very important codes for how to live here successfully. So I call this precision health for all, and what I’m going to do—Eric, tell me how long we have, because I could go for days or weeks or months. I was going to be a Dickens scholar at one point, paid by the word, so it’s like I’ve never seen a word I can’t use.

## Scene change

A PowerPoint slide appears on the screen. Its heading reads: Know Enough To Be Dangerous…And Appreciative

Below this, in the left column, is a box with the heading Genomics and Society. The box contains a short article that discusses psychosocial and ethical issues in genomics research, psychosocial and ethics in genomics medicine, legal and policy issues, and broader societal issues. Below this box is a sentence that reads: Understand how to situate this new knowledge in our culture!

In the right column below the slide’s heading is a logo that reads, emerge network, Electronic Medical Records and Genomics.

Below this logo is a sentence that reads, Learn from your pioneering work on EMRs and biorepositories!

Below this is a logo for ClinGen, Clinical Genome Resource. Below this logo is a sentence that reads, Continue to drive data sharing, curation, & trust in “meaningful use” of variant data!

## Mr. Dishman continues:

So at this point, I would say I’ve read your strategic plan. Certainly the part of the strategic plan around genomics and society is particularly near and dear to my heart as a social scientist and as somebody who has spent a whole lot of my career focused on the legal, policy, and social implications of capabilities. Back in the early days of telehealth, I built very successful telehealth equipment and even workflows with clinical teams when I worked for Paul Allen, the co-founder of Microsoft, all those years ago.

## Scene change

The screen splits, with the previously described slide on the left of the screen, and a camera view of Mr. Dishman speaking in the lower right corner.

## Mr. Dishman continues:

And technically it worked, but the payment model, the entire sort of reimbursement scheme, all of the legal and social implications and the development of workflow were not ready, and in many ways we’ve taken 25 years to catch up with the technology and start to incorporate those. So I am very sensitive to those and the need to work on those early on, when you’re trying to do bold new innovation. I know enough of that emerged to be dangerous and I’m excited what we can already learn from the work that you’re doing on EMRs and biorepositories and certainly on ClinGen as well in terms of—you know, I don’t mean it in a negative way, but how do we make meaningful use of variant data and clinical practice?

## Scene change

The previously described slide again takes up the entire screen.

## Mr. Dishman continues:

Part of the discussion that you were just having and not only do I think a lot of the experts and a lot of the people that are frankly in our awardee community are part of, we’re already part of the awardee communities at NHGRI. So you’ve helped fund both training wheels and sort of moving into the 10-speed and then we’ll eventually get to 27-speed bicycle racing on this as we go forward in time.

## Scene change

A new PowerPoint slide appears. Heading: Appreciative of Your Work More Than You Know.

In the upper left corner is a photograph of Eric Dishman as a teenager, lying on a couch with a puppy. Caption: First week of very imprecise chemo, summer 1989, Chapel Hill at age 19.

## Mr. Dishman continues:

I’m very appreciative of your work, as Eric mentioned. I won’t go through the whole story here, but the long story short is, this is me the first week of a very imprecise chemotherapy regimen, the first of about 60 over the next 23 years that I went through. That’s me in the summer of 1989, Chapel Hill. My wife was wise enough to get a dog, or a puppy, to help me survive that first round of cisplatin and other nasty things and all the pre-nausea–drug days. So I was a pretty small guy at that point in my life, at age 19, and got a lot smaller really quickly. And I did manage to somehow—and I have donated my genetic material to serve two different resilience and survivor studies, because you look at my clinical history and say, “He shouldn’t be alive, just based on the drugs that we actually put him on.” And it was a whole genome sequence, as I was in my role as an executive at Intel years ago, four or five years ago, going around to different genome companies that needed our highest in computing to be able to even make their research or their business model work. And one of these had seen me speak at a nephrology conference years before and said, “Well, why don’t we do a whole genome sequence and we’ll help you understand our technology that tries to take that and start to visualize that data.” I didn’t think much of it. I mean, I did. We arranged to have tissue and blood and all of that done. I signed forms that said they could share it with my clinical team.

## Scene change

Mr. Dishman speaking at the front of the conference room.

Banner at bottom of screen: NIH NHGRI logo. Eric Dishman, Precision Medicine Initiative Cohort Program, National Institutes of Health

## Mr. Dishman continues:

At that point, it took three months of processing on Intel’s highest-end equipment and the top sequencers that were out there to compare me to the “not me.” And then, I didn’t know, but my clinical team took another four months trying to get help from startups, from clinical research, to actually the first time they’d ever done it, makes sense. And they basically came to a lucky well-educated guess. They were very precise when they told me that 92 percent of everything they’d ever put me on was destined never to have worked. I now understand that they couldn’t actually know that, but it was a good guess. But they got enough data and understanding to say, “It looks more like the mechanisms that cause pancreatic cancer are what are causing your cancer; we’re going to put you on a pancreatic cancer drug.” And they did, and I became cancer-free really quickly. Full kidney failure. Intel employee donated a kidney to me because my old ones were failed, and suddenly I’m healthier now at age 48 than I was at age 28 or even age 20. So I came out of that. My wife will tell you, in the ICU that—I can’t remember it, but she took notes because we knew that I wouldn’t remember at all—she said I’ve got to figure out how to make this kind of stuff available to everybody else.

## Scene change

Split screen, with the previous slide at left, and a camera view of Mr. Dishman speaking in the lower right corner.

## Mr. Dishman continues:

And that’s been my mission when I came back to Intel and ever since then, so this fits in.

## Scene change

Content is added to the previous slide. Below the first photo appears a photo of a stack of spiral notebooks with the caption, Have kept a journal since 3rd grade.

To the right of the photos appears the following text:

WORLD FELL APART: Sept 11, 2001, Portland

Exhausted. Quick. Much more later. Like I need to write this to remember, but planes destroyed WTC in NYC and Pentagon attacks. Saw horrors wish I could un-see. Wonder if more will hit tonight.

Ash and I can’t sleep—talking mostly about fear and, in a way, glad this whole ride has meant we never brought kids into this insane world. Am not afraid of dying in a terror attack (though I do worry bombings are now in our daily lives as in many parts of world). Cancer will certainly save me from an explosion—there’s one benefit!

## Mr. Dishman continues:

I’ve kept a journal since third grade, and yesterday I was actually looking back at what I wrote on 9/11, 15 years ago, and it was kind of interesting.

## Scene change

The slide now takes up the entire screen.

## Mr. Dishman continues:

You can start to see some of the mindset that I wrote in here. “I saw horrors that I wish I could unsee. I wonder if more will hit tonight,” meaning attacks. And then I said, “Ash and I can’t sleep, we’re talking mostly about fear, and in a way, I’m glad this whole ride, this whole cancer ride, has meant we never brought kids into this insane world. I’m not afraid of dying in a terror attack, though I do worry bombings are now in our daily lives as in many parts of the world. Cancer will certainly save me from an explosion, there’s one benefit.

I could never have imagined, each year they would say, ‘Well, you’ve got about 9 to 14 months to live.’ And after about 10 years, I said ‘Stop doing that.’” And I now recognize that what was happening was sort of two things: a complete lack of data and understanding to know how to come up with a definitive diagnosis with me that stuck, but also, it’s pretty clear I was morphing, my tumors were morphing in response to the various things that were happening. So it’s true that I didn’t probably have one diagnosis that made sense during that period of time.

## Scene change

The next PowerPoint slide appears on screen.

Heading: Good, Consistent Advice from Many Leaders in Genomics…

Left column:

One thing Rehm would definitely not recommend? Spending all of the Precision Medicine Initiative budget—President Obama proposed 215 million dollars to start—on sequencing.

“I would argue you need to spend more time focusing on the effective collection of phenotypes to then correlate that [genomic] data to,” she says, advocating for smaller pilot projects to successfully link genotypes and phenotypes before sequencing the whole cohort.

The best way to design infrastructure, she believes, is to define projects where you are seeking answers to specific questions, and use the questions and expected answers to drive the building of infrastructure.

Right column:

A photograph of a woman standing in front of a display of the human genome. Below this is a diagram with the ClinGen logo illustrating how patients, clinicians, laboratories, and researchers share genetic and health data to answer ClinGen’s critical questions:

* Is this gene associated with a disease? (clinical validity)
* Is this variant causative? (pathogenicity)
* Is this information actionable? (clinical unity)

All of which lead to building a genomic knowledge base (ClinVar & other resources) and improved patient care through genomic medicine.

At the bottom of the slide is the following credit: From Bio-IT World, “ClinGen and Lessons for the Precision Medicine Initiative,” by Allison Proffitt, June 10, 2015.

## Mr. Dishman continues:

So, as we think about that, I’ve been talking to lots of folks within NHGRI and people that you fund, even back when I was on the working group and we were hearing from people around the country. So there’s been some good consistent advice from many leaders in genomics, which I thought was summed up well in the *Bio-IT World* piece that Allison Proffitt wrote, and it says, “One thing Rehm would definitely not recommend? Spending all of the Precision Medicine Initiative budget—President Obama proposed $215 million to start—on sequencing. ‘I would argue you need to spend more time focusing on the effective collection of phenotypes’”—something that we’re exploring and trying to execute. And towards the bottom, and we should “use the questions” that we come up with and “expected answers to drive the building of the infrastructure as we go.” That theme of learning and iterating as we go is one that I certainly bring from industry and it’s one that I’m going to describe to you here in terms of the mindset and the processes that we’re setting up to be able to go for the long run of building out this program.

## Scene change

A new slide appears, which reads, “A review and status update of PMI CP.”

Immediately another new slide appears.

Heading: The Precision Medicine Initiative® Cohort Program

* One million or more volunteers, reflecting the broad diversity of the U.S.
* Opportunities for volunteers to provide data on an ongoing basis
* Data shared freely and rapidly to inform a variety of research studies

At the bottom of the slide appears the PMI logo, comprising silhouettes of a diverse group of people of various genders, ages, professions, and abilities. A silhouette of a woman stands in front of the others and is highlighted.

## Mr. Dishman continues:

So I’m going to give you a quick review. Eric shared with me previous slides, so I think most of you know the basics. I’m going to run through this part quickly, but then I’ll dive down a little bit more deeply on the sort of status of things right now.

So I believe you know that the Precision Medicine Initiative Cohort Program is part of the broader Precision Medicine Initiative. Our piece is to, you know, 1 million or more volunteers reflecting the broad diversity of the U.S., opportunities for volunteers to provide data on an ongoing basis, and data shared freely and rapidly to inform a variety of research studies. That’s sort of it, in a nutshell.

## Scene change

The next slide appears. Heading: Mission: Accelerate Science & Breakthroughs that Drive Towards Precision Health for All!

Below this is a circular flow chart in which Questions, Problems, & Hypotheses lead to Capturing, Secure, Clean, & Share Data; which leads to Unleashing Science & Diverse Scientists; which leads to Translate Into Action, Practice, & Meaning; which in turn leads back to Questions, Problems, & Hypotheses.

## Mr. Dishman continues:

The mission—and this is not a formal mission—but as I came, I arrived on a Monday and they said, “Oh, you need to do some White House prep and briefings because we’re about to announce the first 33 awards”—it’s really eight huge awards, but to 33 organizations—“and you need to lead a three-day workshop starting Wednesday because they’re all coming to town.” So I said, “OK, all right, I can do this. I know where the bathroom is, that’s a start. And how to park, and then we’ll do this.” And I wrote on that first day when we all came together to present, for the first time, the initial awardees. I said, really, this mission is to accelerate the science and breakthroughs that drive precision health for all, and using the broader word “health” because medicine is a key part of it but it’s not the only part of it.

And if you think about sort of all the different—you can go back and read all the different models of how science works or how inquiry works. But you know there’s different versions of it, but it all comes down to three or four stages of questions, problems, hypotheses. How do we capture data? And are kinds of data secure, and clean it and share it because it’s a very complicated thing to do. How do we unleash science and diverse scientists on top of that data, and how do we then translate that into action, practice, and meaning? As you’ve just been discussing, the challenges of getting people to accept anything that’s non-human data as they’re informed. And if you think about that knowledge turn, a turn that comes from economics that used to be the way they would evaluate the potential growth of a company, uh country, was based on its knowledge turns, its ability to ask questions and have its workforce walk through the cycle—that takes a certain period of time.

## Scene change

The circle and arrows at the center of the flow chart shrink.

## Mr. Dishman continues:

And when we succeed, we’re trying to shrink the period of time it takes to move through those and let lots more people actually move through those cycles by building the baseline infrastructure of both a million people who are going to trust us, to engage with us, and provide different kinds of data over time as well as different kinds of data types.

## Scene change

The next slide appears. Heading: A Transformational Approach to Diversity

Reflecting the country’s rich diversity to produce meaningful health outcomes for historically underrepresented communities.

Next to this are several photographs of people of various ages, races, genders, and ethnicities, including a man in a wheelchair, a group of teens, two children and a senior man blowing out candles on a cake, a mother holding a baby, a man of mixed ethnicity, and an African American woman. Superimposed over these photographs are the labels people, health status, data types, and geography.

## Mr. Dishman continues:

So how do we accelerate that? We certainly are trying to make this a very signature piece of the platform or of the philosophy, it’s a very transformational approach to diversity. I call it often quadruple diversity. So the diversity of people and the American melting pot is going to be able to generate a wide range of cultures and ethnicities, and making sure that we oversample and over-recruit the understudied with regards to a wide range of people, a wide range of health status. It’s not a disease-specific cohort program. Many of the people will be well at start and will understand the unfolding of their various health aspects over time. A diversity of geography, and in that we mean meteorological diversity, because people’s experiences in different parts of the country are very different—different times of year; rural, urban, and all points in between. And you’ll see as we’re making more and more awards, we’re working on building out networks of health provider organizations that can help us do that, as well as a diversity of data types. It’ll start with things like surveys and EHR data pulling in, but we will be doing things with wearables, both the existing ones that people already have and eventually pieces of technology that we might want to put out into the cohort program.

Of course, the challenge is everything times a million gets very expensive, so anticipating the cost curves and everything from –omics to mHealth are some of the challenges that we’ve got to figure out what’s the right time to go do these things?

## Scene change

The next slide appears. Heading: A Transformational Approach to Participation.

Participants in the PMI Cohort Program will be true partners—not patients, not subjects—in the research process.

Involved in every step of program development

* What data we collect
* What lab analyses we do
* What research is conducted
* How data gets returned

In the lower right corner of the slide is a photograph of several people in a huddle, with their hands stacked on top of each other in the center of the huddle.

## Mr. Dishman continues:

And then also a transformational approach to participation. We’re not just changing the word “subject” to “participant” and being done with it. It doesn’t also mean that we know how to do all of this yet, but participants involved in every aspect of it, from governance to an invention of questions of what we’re going to be doing. I’ve just come from and will return to some of the reviews of our current round of health provider organizations, where many of them are already experimenting with, you know, what are the questions that our participants want to know and what’s the kind of science that they want done out of these particular kinds of studies.

## Scene change

The next slide appears. Heading: A Transformational Approach to Data Access

* Data sharing will be swift to both researchers and participants
* Data collection will start small and will grow over time
* Privacy and security will adhere to the highest standards
* Will invest to level the playing field so diverse researchers can play

In the lower right corner of the slide is a diagram illustrating that PMI-CP data will be shared between PMI and colleges and universities, industry, citizen scientists, and community colleges and high schools.

## Mr. Dishman continues:

And then a transformational approach to data access. You know one of my goals is to make sure that we don’t just use taxpayer money to let the Tier 1’s who already have this infrastructure might even be able to do a million-person cohort on their own and just advance them. What is it going to take for us to enable citizens, scientists, industry, community colleges, and high schools to meaningfully participate in using the data and generating science on top of that. I came in on the sort of tail end of the discussion many of you are having about education and your challenges in that regard. It was something I spent a lot of time on in my Intel career of how do we anticipate having a workforce both to do the science and deliver the precision medicine so that technology and science don’t get there but it can’t scale because we just don’t have a workforce to do that. And I think that’s part of our investments and part of our thinking here as well.

## Scene change

The next slide appears. Heading: Two Methods of Engagement

In the center of the slide is a diagram. At the center of the diagram is a list that reads, participant-provided info, em Health data, consent, EHR data, baseline exam, and biospecimens. An arrow points from the top of this list to a cloud containing zeroes and ones, representing the cloud. Another arrow points from the bottom of this list to a line of test tubes.

To the left of this list are five stick figures labeled direct volunteers, and a drawing of a doctor wearing a stethoscope. A double-headed arrow points between these two illustrations. An arrow points from the direct volunteer stick figures to participant-provided info, em Health data, consent, and EHR data in the list. Another arrow points from the doctor illustration to baseline exam and biospecimens in the list.

To the right of the list are five stick figures labeled HPO volunteers and an icon of a building with a medical cross. A double-headed arrow points between these two illustrations. An arrow points from the HPO volunteer stick figures to participant-provided info, em Health data, and consent in the list. Another arrow points from the building with the medical cross to baseline data and biospecimens in the list.

Below this illustration are several photos of people of various genders, ages, races, and ethnicities.

## Mr. Dishman continues:

Two primary methods of engagement—you’ve probably heard this before—is through what we call the HPO path, so health provider organizations—as well as DV, direct volunteer. So we are working on developing the capacity to get the quality biosamples and physical evaluations that we actually need from anybody that calls a 1-800 number or pops onto the website or approaches through their church or through their health provider organization. That’s what we’ve got to be able to scale out and do over time with a kind of wide range of data going into that. So the DV path and the HPO path.

## Scene change

The next slide appears. Heading: Right Now: Building Interdisciplinary Platform Team

* Act as one team with diverse participation
* To deliver 1 million engaged volunteers & rich, diverse data over long period of time
* 33 awardee partners so far: 11 Working Groups/Departments … Steering & Exec Committees
* Biobank, Data and Research Support Center, Health Care Provider Organizations, and Participant Technologies Center
* Hire core NIH leadership team
* Deputy, CTO, CMO, Comms Chief, User Experience Chief …
* Develop transition plans with multiple government agencies to maintain & grow mutual support
* Build robust community partners network
* Use known industry methods: user-centered design, platform development process

In the lower right corner of the slide is an illustration comprising a number of hexagons, fitting together like puzzle pieces. Several are labeled with the names of government agencies and other entities, including community partners; biobank; industry; participant technologies center; U.S. Digital Service; En Eye H team; HPOs: Vee A medical centers; eye see partners; White House; research community; HHS, HRSA, Oh en see; HPOs: community health centers; data and research support center, Department of Veterans Affairs; HPOs: regional medical centers; communications support; and providers.

## Mr. Dishman continues:

So what’s going on? I just want to give you sort of a snapshot of what it looks and feels like to be part of this startup team. One of the challenges is to get everybody to act as one interdisciplinary platform team. When I said the word “platform”—and we basically are a platform company stitched together from a mix of technology and academic grants as well as staff that we’re hiring into the NIH—more than three quarters of the room and they didn’t even really know what I meant by the word “platform.” It’s one of the first things I said at the first work group meetings. I hardly knew Francis Collins or anybody, and I happened to be sitting next to him and I whispered to him and said, “You do realize this is kind of NIH basically trying to act like a platform company, right?” And I guess he thought about it for a while because somehow I ended up here. Be careful what you say in those opening working group meetings. You never know where you’re going to end up.

So getting those 33 initial awardees—as well as the more than 14 or 15 government agencies who are part of the PMI umbrella—to act as one team, so we can deliver this million-person engaged volunteers and these diverse data sets, is no small feat.

## Scene change

Split screen, with the previous slide at left, and a camera view of Mr. Dishman speaking in the lower right corner.

## Mr. Dishman continues:

And it’s the most important thing that I do out of the gate, because if we don’t build the culture, we don’t build the processes, then we will not be sustained for 10, 20, 50, 100 years for what we want to do here. Thirty-three awardee partners so far, really divided up into 11 working groups. I can’t do it today because it’s too complex and I can’t make PowerPoint do it, but I will eventually come in and share with you our departments, and these departments will look like what it would be like if a company was actually going out and building this infrastructure, and it will be made up of people from NIH, from corporate entities, from academic research centers as we go through.

So 11 working groups, the Steering and Executive Committees, are actually set up and actually moving forward. And you can see some of the working groups actually listed there. I’m still working to build the core team. I will probably send it out to you soon some additional senior leadership positions that we actually need. There’s sort of about seven or eight people and then another about 20 that are holding all of this up right now, because part of the challenge is they’re so busy working on the working groups or the protocols, and they’re the same people that need to be writing the position descriptions and doing the searches as we go out here. So a lot happening, hiring that core NIH leadership team.

Part of what we’re also doing, as you can imagine—this being the President’s Precision Medicine Initiative—all of those government agencies and partnerships—and I have to tell you, as somebody new coming in to be a Fed—this has been an unprecedented collaboration that I’ve ever witnessed or seen in terms of the different parts of the Health and Human Services family being brought together by the Secretary to work towards this common effort, on everything—like what challenges are we going to have with HIPAA going forward, to what are the policy changes that we’re going to need in place. We want to make sure that that support continues regardless of what happens in the election. So we’re working on transition plans and making sure that we have federal employees as the mesh network keeping this alive regardless of what happens politically above us. This takes time and energy to do right, and it’s a key thing that we’re working on right now.

And then we were building a robust community partners network. There’s a top 50 list of community partners that we’re working our way through who are national but also have local chapters to help us with everything from recruiting to educating providers, educating community members who will be on the front lines to help us hold this together, as well as a workshop next week with Health and Human Services on all the different community partners they’ve used coming together to say, “OK, what can we do to work together towards this effort?”

And then also we’re using known industry methods. I am teaching people and we are instituting and we are hiring competencies so that we do user-centered design and platform development process. And I’ll talk a little bit about that in a minute, because I’m assuming this is the first discussion that we’ll have for probably years to come. So I want to get you right into the mindset out of the gate so that it doesn’t become alien to you as I give you these updates and these discussions going forward.

## Scene change

Split screen, with a new slide at left, and a camera view of Mr. Dishman speaking in the lower right corner. The slide reads as follows:

Heading: Right Now: Testing & Piloting Pieces of the Program

* Rolling out personas built upon ethnographic work in homes/focus groups
* Building diverse 5,000-person community of beta testers
* Testing consent language & 5 survey modules and defining next 12 modules
* Iterating website, smartphone, 1-800#, & data center infrastructure
* Getting feedback on content for educating the public and participants
* Testing name, brand, look and feel
* Experimenting with recruitment methods
* Drafting full protocol for V1 launch
* Cybersecurity testing

In the top right corner of the slide is a box that reads,

Not for me Jasmine, 34

“I worry about the government knowing more about me than I do about myself. Do I want people to have access to my data? Even if it means I could be healthier? Even if it means they could catch my cancer sooner? I’m not sure.”

The box contains the photo of a woman between two smaller photos of two men.

## Mr. Dishman continues:

The other piece is—thanks to the great work that NIH had already done—the working sessions and listening sessions, some of the pilot grants awards that were actually funded at Vanderbilt and elsewhere, as well as what we did were these implementation papers where we said, “OK, well, we can anticipate these are the kinds of problems everybody’s going to have as soon as you bring all these awardees together.” Here’s what’s going on right now. We’re actually rolling out, and it will soon be fully public—there’s an image of one of them. We’ve developed based on focus groups and ethnographic studies of people in their home with a wide range of what the kinds of participants that we expect to come in here, what we call the ecology model. And it’s basically a segmentation model that cuts across ethnicity and race and age to help us identify these are going to be the challenges, the “not for me” persona segment cuts across lots of difference. What would we need to do to convince the not-for-me’s that this is something that they can trust and what are their concerns. So there’s about nine or 10 segments that you’ll hear about, and for years we will come to talk about our different success of trying to pull them in.

We have a 5,000-person community of beta testers already, and we’re actually recruiting additional to increase the diversity of that 5,000 initial test bed. We’re testing consent language in the first five survey modules, with great progress on the next 12. We’re iterating a website, the smartphone application that people will be able to come through, a 1-800 number, and all of the data center infrastructure going through security and stress testing. If the president and the national media stand up and say, “We want millions of people,” then we have to have a way of catching that.

Incidentally, we expect to have three tiers. Those who are just interested, click here. This exists on our website now. There’s a lot more content that’s going to fill that website very soon. Those who may want to participate in the surveys and could do it for years to come, we could potentially take millions and millions of people for that effort. And then those who will have full biosample and physical evaluation as we go forward in time. So we want to be able to set up for people to have different levels of engagement. And if you have millions and millions of people that sign up to be sort of members and fill out surveys and consent, it helps you figure out, “OK, who from those do we want to invite to move to the third tier?” so that you can help achieve some of the diversity challenges that you have.

We’re getting feedback on content and the name, brand, and look and feel. We’ll announce the name of the whole program soon; it won’t be called the PMI Cohort Program. It doesn’t mean very much—the word “cohort”—to real people when you go out and actually talk to them, and it doesn’t sound or feel very good to a lot of them.

We’re experimenting with different recruitment methods. Amongst our recipients, have different strategies of pulling in from buses and vans that pull up into parking lots, to church-based, to other online tools. And we’re drafting a full protocol for the V1—version one—launch. It’s coming along nicely in terms of the physical evaluation, the survey data, and the biospecimen. And then also doing cybersecurity testing. No better way to lose the trust of the country than coming out of the gate. And so certainly partnering with the different parts of government and industry to make sure that we’re using the best methods.

So a lot is happening and all these pieces are coming together.

## Scene change

Split screen, with a new slide at left, and a camera view of Mr. Dishman speaking in the lower right corner. The slide reads, What I mean by “platform approach.”

## Mr. Dishman continues:

I’ll close with the section here that just says, “What I mean by platform approach.”

## Scene change

Split screen, with a new slide at left, and a camera view of Mr. Dishman speaking in the lower right corner. The slide reads as follows:

Heading: Platform Dev Lifecycle: Innovation to Implementation

At the center of the slide is a diagram consisting of a cone and several labels. Six labels point to the center of the cone: human challenges, scientific discoveries, researcher needs, research questions, new technologies, and crazy ideas.

The cone is divided into four sections. The first section is labeled exploration R and D, V3. The second section is labeled platform definition, V2. The third section is labeled advanced development, V1. The final section is labeled production. Outside the narrow end of the cone is a label that reads launch.

Below the diagram is the following text:

* Repeatable, phase-gate process
* Develop multiple versions at once
* Each release may have new:
* Features for participants
* Features for researchers
* Scientific focus areas/measures
* Data capture capabilities

## Mr. Dishman continues:

So this is a classic diagram from innovation literature.

## Scene change

The current slide now fills the entire screen.

## Mr. Dishman continues:

Almost every company in Silicon Valley, or every company that does innovation around the country, has some version of what’s called the Innovation Funnel. And there’s usually three or four different phases or steps to it. Now out here at the edge of the funnel, it’s like you’re trying to understand human challenges, scientific discoveries that are happening, that may change the game. What are the researcher’s needs? What are the research questions? What are the new technologies? What are the crazy ideas? And that funnel is always open and will have a part of our function that’s always sort of trying to keep that funnel open. But then you come through all of that and say, “OK, well, with all of these kinds of inputs, we’re in the exploration and R&D mode of, what are the measures that we’re going to do, what are the questions that we’re going to ask, what are the recruitment methodologies that we’re going to use?” At some point, you’ve got to stop in time and say, “We’re going to find this platform.” And by “platform,” I mean everything from what the communications and media plan is and who it’s targeting of those particular persona segments, to the technology platforms that are in place both to take participant input as well as researchers’. Using that, you move that into advanced development. And the reason it gets smaller is you’re starting to throw and focus, you’re starting to throw features and capabilities that you thought were ready but aren’t ready for primetime out of the boat. And then eventually you move into production and launch. And this is a repeatable process. It’s a phase-gate process where you say, “Look, this particular program, we’re not moving from here to here until it meets these phases. We’re not even going to spend the money on the rest of the pieces.”

Eventually we will have three teams. We’re not there right now. We’re kind of right here with V1 of the launch, where all those pieces I just described a moment ago are in advanced development and we’re moving towards production. But since this process wasn’t used, we have to go back and do some platform definition because each awardee and NIH all had slightly different assumptions about what they thought the users might do, and you will not launch a product or a platform until you’re really clear: this is the definition, this is the minimum, and so forth.

So this is kind of where we are. We will ultimately have teams. The Operational Team that’s moving from advanced development to production will always be the largest. It’s the most difficult. Somebody back in here, in Platform Definition, is a smaller team. And then further back in R&D is an even smaller team, so your resources are usually almost in inverse proportion to the way the funnel works.

So each release and the mindset that you should be in is, “This is not a one-time we’re taking data one time.” This is every 12 to 18 months, we’re releasing a new release of the PMI Cohort Program platform. And in any one of those releases, somewhere between two and four of these things may be true. We have new features for participants about ways that they can engage with one another or provide data to us. We have new features for the researchers—and we’re not going to have a lot of features for researchers out of the gate, but we have a plan to build those out over time. By the way, we will borrow heavily and reuse things from eMERGE or whatever programs. We very much believe in borrow and license and use what’s already out there and build only when we absolutely have to. Scientific focus areas and measures will change over periods of time even though it’s a general population study. And then the data capture capabilities. “Oh, we’re now ready to take in data from anybody’s wearable. Oh, we’re now ready actually to have a set of neurological games that we want people to play and can actually take that kind of data in as well.”

## Scene change

A new slide appears on screen. Heading: Defining Our Target “Landing Zones.”

* Must have clear definitions of the “product” to build reliable schedules
* Cannot solve for “features” and “schedule” and “resources” at same time
* Landing zones allow for tradeoffs of time and money

Below this text is a table of four columns and eight rows. The four column headers read Departments/Work Streams, Minimum, Goal, and Stretch Goal. The rows under the column heading Departments/Work Streams read, User experience: Participants, Researchers, Providers, and Community Partners; Research questions/protocols; Technologies/touchpoints; Security & privacy; Communications/messaging; Outreach/partnerships; and Policy & legal. The other cells in the table are empty.

## Mr. Dishman continues:

So just a little bit more on that. You’re going to hear me over time use the word “landing zones.” This is an industry term that just means the targets for what you’re trying to do for a launch. So part of what I’m trying to get the teams right now to do—and a little bit of this is retroactive—is define for all the swim lanes or all of the departments of what we’ve got to deliver, from knowing what the user experience is, down to the policy and legal things that we need to have in place before that particular launch. What’s the minimum that we would need to have in place to launch, very crisply defined? What’s the goal, and then what’s the stretch goal? You cannot solve for features of a platform, schedule, and resources all at the same time. And when you define things in this way, it lets you make some tradeoffs of saying, “Well, we could be ready earlier or spend less money if we just went ahead and launched minimum.” Nope. It’s really important, we’ve got the money, and we can actually expand out in time to do stretch goal.

## Scene change

A new slide appears on screen. Heading: Sample Framework for Research Questions & Requirements.

Initially, the slide comprises eight vertical bars labeled Infectious diseases, neuro/cognitive, mental health and addiction, heart and lung, movement musculoskeletal, metabolic conditions, cancer, and chronic pain. A box in the lower right corner of the slide reads, examples of scientific bodies of knowledge.

## Mr. Dishman continues:

And then lastly, what I want to talk about here is—because this is going to come into the fore very quickly now. This is a framework I made up. This is not the final framework. I’m just using this as a conceptual framework for you. If you think about bunches of areas of scientific medical knowledge—infectious diseases, neurocognitive, heart and lung, metabolic conditions, cancer, chronic pain. And we have a scientific working group actually working on this right now about what’s the right framework.

## Scene change

Six horizontal bars appear on the slide, overlaying the eight vertical bars. The horizontal bars are labeled Prevention & wellness, Caregiving & compliance, Reducing disparities/improving access, Genomics, em Health, and Environment & exposure.

## Mr. Dishman continues:

Over time, as we do releases, there’s a bunch of horizontal capabilities that you’re going to apply across this. Prevention and wellness is something that you would want to consider in a lot of these buckets. What do we know, reducing disparities and improving access, what research, what science do we know within these buckets, caregiving, genomics, the mHealth capabilities from smartphones and wearables, you know, environmental exposure. So there’s a bunch of things that are horizontal capabilities.

## Scene change

A new slide appears on screen. Heading: Versions in Time: Different Requirements/Res. Questions.

Initially, the slide contains the same eight vertical bars from the previous slide, labeled Infectious diseases, neuro/cognitive, mental health and addiction, heart and lung, movement musculoskeletal, metabolic conditions, cancer, and chronic pain.

## Mr. Dishman continues:

Here’s a way to think about the versions over time. I mentioned V1, V2, V3.

## Scene change

At the bottom of each vertical bar on the slide, a box appears labeled Vee 1. The Vee 1 boxes are of various heights.

## Mr. Dishman continues:

If you take these areas of thought, our V1 launch may only get—in terms of what these different areas need—this much progress. And the way that we’re going to be doing this is basically going through a bunch of these areas with thoughts and defining requirements and looking for capabilities that can cut across many—three, four, or five—of these particular areas. So V1 might get us this far.

## Scene change

On each vertical bar, above the Vee 1 boxes, appears another box labeled vee 2. The vee 2 boxes also are of various heights.

## Mr. Dishman continues:

V2 might—we might have called this the heart and lung release even though there were capabilities that were about more than heart and lung, but we’re ready to add some capabilities that are going to be a game changer for heart and lung.

## Scene change

On each vertical bar, above the Vee 2 boxes, appears another box labeled Vee 3. The Vee 3 boxes also are of various heights.

## Mr. Dishman continues:

Or V3 might really boost up where we’ve been on neuro and cognitive. I’m getting you into this mindset, because as we engage with you and as you engage with other bodies of thought and other institutes, trying to understand what’s ready when, and we want to get the public and the researchers excited about these different releases over time.

## Scene change

A new slide appears on screen. Heading: Ultimately Want to Focus on Where Human Need Is Greatest.

The slide comprises a number of boxes of various sizes and shapes that fit together in a mosaic, like a jigsaw puzzle. The boxes are arranged in three color groupings. Each box is labeled with a medical condition. The blue group, which takes up just over half of the slide, includes the following labeled boxes: Eye H Dee, stroke, H tee en h dee, cee em pee, are h dee, aort an, and other cardio; Lung cancer, liver cancer, stomach cancer, colorectal cancer, breast cancer, oth neopla, cervix cancer, brain cancer, lymphoma, esophagus cancer, ovary cancer, lip oral cancer, kidney cancer, leukemia, and pancreas cancer; depression, anxiety, drugs, schizophrenia, A ess dee, alcohol, bipolar, conduct, eye dee, and other mental; congenital, skin, sense, and oral; diabetes, see kay dee, hemog, urinary, gyne, and endocrine; see oh pee dee, asthma, eye el dee, pneumocon, and other resp; back and neck, osteoarthritis, rheumatoid arthritis, and other em ess kay; med head, migraine, epilepsy, Alzheimer, e mess, and other neuro; pee you dee, ileus, pancreatitis, gastritis, and eye bee dee; and cirrhosis alc, cirrhosis hep bee, cirrhosis Hep cee, and other cirrhosis.

The red and green groups take up the rest of the slide. The red group includes the following labeled boxes: El are eye, diarrhea, meningitis, intestinal infection, measles, encephalitis, whooping, and you are eye; en en preterm, en en encephalitis, en en sepsis, and other en en; HIV, tee bee, malaria, leish, eff bee trema, el eff, and schisto; and iron, pee ee em, STD, hep, other mat, and other infection.

The green group, which is the smallest group, includes boxes labeled falls, mech, fire, drown, poison, med treat, eff body, animal, and other unintentional; road injury, self harm, violence, and war.

Below these boxes is the following source citation: Credit: Causes of DALYs globally, 2013. Dr. Christopher Murray, Institute for Health Metrics and Evaluation, University of Washington <http://vizhub.healthdata.org/gbd-compare>

## Mr. Dishman continues:

We could look at a framework around, you know, ultimately focusing where the human need is, a dailies approach, I don’t know what this framework will ultimately be, but you want to make sure that you’re getting requirements from it.

## Scene change

A new slide appears on screen. Heading: Next Up: Research Questions Workshops.

* Premise to Adopt for the Workshops: Imagine PMI CP is only to accelerate your body of thought and research in a particular domain!
* Three Big Questions:
1. Near Term: what are low-hanging fruit questions/measures for which the scale of PMI CP could help accelerate knowledge & breakthroughs?
2. Mid-Term: what kinds of questions might this CP answer where additional work selecting among measures/instruments is needed?
3. Long Term: what kinds of questions are ripe for CP of this size but for which we need fundamental science & tech to develop the instruments?
* Kinds Of Attendees:
* Research experts, co-funders, participant/advocacy orgs, providers, etc.
* What We’ll Do:
* Factor analysis of common needs across multiple knowledge domains = our “versions”
* Partner with domain stakeholders to grow funding for unique areas = sub-studies

## Mr. Dishman continues:

So what’s next—in terms of the spirit of this—is what I’m calling research question workshops. And we started—we did this back in the working groups but they were very high level. And what we need is to convene areas around these bodies of thought and then basically the premise of these workshops says this. Imagine the entire PMI Cohort Program of a million people is focused on just accelerating scientific knowledge and breakthroughs in your particular domain. Near term, what are the low-hanging fruit questions and measures for which the scale of something as large as this would be useful? Medium term, what kinds of questions might this platform—the Cohort Program—answer where additional work is needed to select amongst measures to answer those kinds of questions? And long term, what kinds of questions are ripe for the Cohort Program of this size but which need fundamental science and technology development to know the instruments to go answer those?

The kinds of attendees that we will want to come to these workshops are research experts in that particular field, co-funders. I don’t have the money to pay for all the research on this. We have the money to build the foundation. If we’re not building something that’s advancing NHGRI’s agenda and the various agendas of funders around those domains, then we will fail. We will have built a “field of dreams” problem where we build and nobody comes. So we want other co-funders, participant advocacy groups, and providers. And then what we’ll do—this is just classic product development. You look across for common capabilities across those needs and put it into those releases. And then negotiation that I will get into over periods of time is, “Hey, is that really something that needs a million people?” If the thing is unique only to your body of knowledge, then we’re going to have to work together to get a co-investment to be able to carry that forward because that just doesn’t apply to enough other domains. Does it require a sub-study, or does it require all million people?

## Scene change

A new slide appears on screen, which reads, Closing thoughts…

Immediately another new slide appears on screen. Heading: Key Opportunities for En H Are Gee Eye Collaboration

* Defining our genomics roadmap and plan in detail
* Research question workshops—genomic strategy on each domain
* Find ways to drive more ethical, legal, social implications research as we go/grow
* Explore what CLINGEN becomes over time as PMI CP capacity kicks in

At the bottom of the slide appears the PMI logo, comprising silhouettes of a diverse group of people of various genders, ages, professions, and abilities. A silhouette of a woman stands in front of the others and is highlighted.

## Mr. Dishman continues:

So that’s the mindset. And what I would just close is—again, as I said at the beginning—I know enough to be dangerous about all the scope of NHGRI, and I kind of know a little bit about Caesar in here, you were just talking about Caesar. But I don’t really know what it is and I’d be in trouble and have to use a lifeline to Carolynn or to Eric or somebody, but defining our genomics roadmap and plan in detail, it’s not done. And the reason we haven’t asked your input yet is because we are trying to get the basic infrastructure up and running, and one of the pieces of advice that I shared with the quote from *Bio-IT World* at the beginning—Eric gave it to me again last week—is get the basics up and running. The longer you can wait to do the genome piece, you’ll get more bang for your buck, and it’s a complicated issue, given the kinds of return of results issues that we’ve got. So we’ve got a great sort of 60 percent draft based on all the public comments and all the input previous. But over the next few weeks, as I get more senior people involved, we’ll start a process and build a work group around helping us build our actual genomics plans and the timelines for that.

Research question workshops—as we think about the genomic strategies in each of those domains, find ways to drive more ethical, legal, and social implications research as we go and as we grow. I appreciated the LC memo. I saw it more recently, I think I actually saw it back in May when I was on the work group. Those kinds of issues I worry more about than actual technology than cost because I’ve been burned so many times of having everything ready except for the, “Oh, it has to get sutured into the fabric of society.” Yeah that, that’s a little bit of a challenge. And then I think we also should explore what ClinGen becomes over time as our capacity kicks in. I think understanding what the roadmap and where ClinGen is going, that’s something that we’ve supported and I would expect us to continue to support. Over time I think that’ll be important for us.

## Scene change

A new slide appears on screen: Heading: From Luck for the Few to Precision Health for All

Below this appears a photo of Mr. Dishman during his chemotherapy at age 19, lying on a couch with a puppy. An arrow labeled luck and pluck points to two more current photos of Mr. Dishman, one of him in a hospital gown, sitting in a chair in a hospital room following his transplant, connected to medical equipment, and the other of him wearing a medical face mask, standing with his wife in his home.

## Mr. Dishman continues:

So the last slide, if you think about my own experience—this was the transplant experience four years ago that was never supposed to happen, that’s Tracy from Intel, who donated to me having never met me before. And it was luck and pluck that got me there, and my personalized health and my precision health experience took 23 years to get there. That’s not a very scalable model for a global population.

## Scene change

On the same slide now appears the same series of photographs below the first series. The label in the arrow has changed to read, routine care.

## Mr. Dishman continues:

So how do we make that happen? Routine care and infinitely more quickly than we could otherwise.

## Scene change

At the bottom of the slide appears the following: Look forward to working with you make this dream a reality!

## Mr. Dishman continues:

And I look forward to making this dream possible with NHGRI and your extended communities. Happy to take your questions.

Audience applause

## Scene change

Camera view of Mr. Dishman at the lectern. He points to someone in the audience.

## Mr. Dishman:

Sure.

## Dr. Robert Nussbaum

(Chief Medical Officer, Invitae, Inc.)

I had two, one quick and one maybe a little more extended. The quick one is the vertical bars you had of the different disease areas. Were those just examples, or is that meant to be comprehensive?

## Scene change

Camera view of Dr. Nussbaum sitting at a table.

## Mr. Dishman:

No, those were Eric’s examples, and we were actually—there’s a meeting coming up with the IC directors…

## Scene change

Camera view of Mr. Dishman responding to the question.

## Mr. Dishman continues:

… and a scientific working group that’s helping us figure out what those would be.

## Scene change

Banner appears at bottom of screen: NIH NHGRI logo. Eric Dishman, Precision Medicine Initiative Cohort Program, National Institutes of Health

## Mr. Dishman continues:

I don’t want them to all be organ, body parts, or diseases. I mean—there’s a certain level—and I think the way that we’ll have to set it up and talk about it with the scientific community might be slightly different. So you can imagine us doing a breathing campaign that creates for all of our participants the number of conditions and the number of institutes that that would cross over would be enormous.

## Scene change

Camera view of audience members in the conference room sitting around a table, looking at Mr. Dishman speaking at the lectern. The projector screen with the last slide appears to Mr. Dishsman’s right.

## Mr. Dishman continues:

So it’s not like a real one-to-one mapping with all the institutes, but that’s partly why we need the framework in place.

## Scene change

Camera view of Dr. Nussbaum.

## Dr. Nussbaum:

Okay, I was just going to say, I think you might garner a lot of interest if you had a nutrition and diet bar. The other comment I wanted to ask, I just joined a genetic startup company about a year ago. So a lot of what you’re saying is resonating with my learning experience over the last year, and I was wondering—while the government is wonderful for doing certain things in terms of stability and competence of the staff, and there are great things the government can do—but there are also some incredibly irritating and difficult around …

Mr. Dishman laughing.

## Mr. Dishman:

I’m learning that more and more each day—

## Scene change

Mr. Dishman laughing, then nodding and listening to the question.

## Dr. Nussbaum:

… particularly around software and around contracting. There’s not a wonderful track record of software development and maintenance in many of the agencies in the government. I’m wondering whether—do you occupy a somewhat special or privileged position in the government workings that would allow you to …

## Mr. Dishman

If I’ve been given a magic ring yet, I haven’t been told that.

## Scene change

Camera view of audience members in the conference room, with Mr. Dishman at the lectern.

## Mr. Dishman continues:

My assumption is we’ll use the right partner to do the right thing. I mean, we’re already looking at—there’s a lot of companies large and small that are saying, “How can we donate capacity and capability to you?”

## Scene change

Mr. Dishman standing beside the lectern.

## Mr. Dishman continues:

There is one aspect of the program, a particular kind of authority called Other Transactional Authority, which does—and if we didn’t have it, we could not have attracted some of the companies and others that we’ve already brought into it. Because a lot of these folks have never …

## Scene change

Dr. Nussbaum sitting at a table in front of a laptop computer, with his arms folded.

## Mr. Dishman continues:

… even this particular round of health provider organizations that we’re doing.

## Scene change

Mr. Dishman standing beside the lectern.

## Mr. Dishman continues:

The benefit of the Other Transactional Award Authority—there are several—but one of it is it really allows in kinds of players who wouldn’t normally participate, integrated delivery networks and others who are like, “I’ve never applied for an NIH program in my life.” And it also allows us flexibility on the other side of the award to say, “Oh, wow, we didn’t think this part through as we’ve iterated and learned as we’ve gone, we need to add an additional capability onto that.” So it’s a legitimate government capability that’s been used in other places, and NIH has started using it for Common Fund and others more recently.

## Scene change

Camera view of audience members in the conference room, with Mr. Dishman at the lectern. To his right, Dr. Eric D. Green, Director of the National Human Genome Research Institute, National Institutes of Health, is speaking.

## Dr. Green:

Not everybody has that authority. There’s no NHGRI dollars associated with OT Authorities—Other Transactional Authority.

## Scene change

Dr. Green sitting at a table, speaking.

## Dr. Green continues:

But the way the PMI money was sort of flowed through into NIH, it was put into a place that has that authority. So the program does have OT Authority. Yes.

## Scene change

Camera view of audience members in the conference room, with Mr. Dishman at the lectern.

## Mr. Dishman:

Yeah. But I very much believe in use the right tool for the job.

## Scene change

Camera view of Mr. Dishman speaking beside the lectern.

## Mr. Dishman continues:

And if we have to make software—and even with our DRC, which stands for the Data and Research Support Center—the award is to Vanderbilt and Verily—Google Verily—and Broad, who have had experience doing those tools. In the case of the PTC, which is the Participant Technology Center, so focusing on that direct volunteer capacity, they have a partnership with Walgreens to help with the national capacity as well as a software company called Vibrent Health. And in many cases, we’re using off-the-shelf commercial software that’s reskinned or added to and going through a FISMA security process. And that’s important to do but it’s much easier when you’re doing it with fundamental Google layer components that are in thousands and thousands of secure commercial products.

## Scene change

Camera view of the conference room and audience, from the front of the room looking to the back.

## Dr. David Walt

(Professor, Tufts University)

I’m interested in the biospecimens and biobanking. It seems that this is going to be a tremendous resource over time, and technology changes over time.

## Scene change

Camera view of Dr. Walt speaking.

## Dr. Walt continues:

And so what you can measure today is not necessarily what you can measure 10 years from now or 25 years from now, so what’s being done with respect to the kinds of samples that are being collected and the ability to keep enough of those available so that they become even more valuable in the future?

## Scene change

Camera view of Mr. Dishman speaking beside the lectern.

## Mr. Dishman:

Yeah. Now, this is one of these things where it’s like, “Wow, if I really felt stupid,” it was in the meetings where Mayo won the award for our biobanking, and Steve Thibodeau there is just fantastic. And they in the working group and in the very near-term—the protocol’s not completely done—but in the very near-term it will be blood and urine, and they’re going through all the tradeoff analyses of how much do we want to hold, how much do we want to put out for awards, what’s the range of kinds of things that we want to do? But we’re also looking at the pros and cons of storage around daily drinking water brought in with the participants from different sites, hair, fingernail clippings, so there’s a wide—that tradeoff list is being done. And the other thing is we’re trying to anticipate what kind of metadata and other kinds of things do we need to capture anticipating when we run out of those capabilities. And, you know, the blood draws three or four years from now and in three or four years thereafter are sort of set in stone. Partially is that innovation process I described is making sure that we’ve got people in the biobanking world that are looking out at the edge of that funnel of what’s starting to move through diffusion of innovation and then wrap that into our 5-year strategic plans. That’s the best answer I can give you.

## Scene change

Camera view of Dr. Walt speaking.

## Dr. Walt:

And I would recommend that, you know, microbiome stool samples.

## Mr. Dishman:

Yeah. All of those things are considered. And I think we’ll get to over time.

## Scene change

Camera view of audience members in the conference room, with Mr. Dishman at the lectern.

## Dr. Dan Roden

(Senior Vice President for Personalized Medicine, Vanderbilt University Medical Center)

That was a really inspirational talk. I just want to ask a question about managing expectations.

## Mr. Dishman:

Hmm.

## Dr. Roden continues:

So there’s a lot of money going into this. There are going to be people within the NIH and in the outside community who say, “Well, you know, that’s just taking stuff away from me.”

## Mr. Dishman:

Uh huh.

## Scene change

Camera view of Dr. Roden speaking.

## Dr. Roden continues:

And so the question is one of early wins, do you have a sense of how you’re going to manage that? And I will say that Senator Alexander visited us last week …

## Mr. Dishman:

Mmm hmm.

## Dr. Roden continues:

… the week before, and as far as he’s concerned, this is the genomics project.

Mr. Dishman laughing.

## Dr. Roden continues:

And that’s sort of—you know, Eric, as far as Senator Alexander, who is one of the biggest advocates in the Senate for this …

## Mr. Dishman:

That’s right.

## Dr. Roden continues:

… it’s all about human genomes. So there’s a whole bunch of expectations that you have to manage. Nobody’s going to expect an early win in the next two months. But what are your thoughts about that?

## Scene change

Camera view of Mr. Dishman speaking beside the lectern.

## Mr. Dishman:

Yeah. There’s a manager I had at Intel and he had an equation: S equals R minus E—success equals results minus expectations. And so Collins’s Law is with me full-on. I mean, to be honest with you, I think some of the expectations were that major science would start being done by the end of this year, and people had, “Hey, we’ll have a half million people by the end of the year,” and I came in and said, “No, no, no, no, no, no, you will hurt us if we try to drive towards crazy expectations like that and we’ll never be able to get off the ground or we’ll do something that’s a flash in the pan but doesn’t have the infrastructure or the process for the long haul and could potentially violate our trust.”

The scientific working group—and it’s part of the protocol that we’re considering. I mean, the early wins are going to be just understanding characterization of survey data linked to geodata. The clinical, even the EHR data—we are certainly—the health provider organizations who have won the awards, have demonstrated, and we’ve done pilots with them to show that they can get the EHR data to us relatively quickly, though they’re still slogging with each one to make sure that there’s interoperability there. Even for being able to get the EHR data for those coming into the direct volunteer path, I mean, that’s just an enormous challenge. Right?

## Scene change

Camera view of Dr. Roden.

## Mr. Dishman continues:

They come in from—they may not even have one. And if they do, it could probably—they can be completely uninsured, have no access to health care, and they’re going to be invited into our study.

## Scene change

Camera view of Mr. Dishman speaking beside the lectern.

## Mr. Dishman continues:

So trying to figure out that over time is going to be part of the key. Part of the expectations management has been to try to get everybody to understand the principles, and there are a lot of people that think this was a genomic study. I think in some ways, candidly, it’s partly why Francis and others said, “Look, there’s the practical cost issues,” and it’s not even so much the cost of sequencing but the cost of wrapping the human beings that helped make this a responsible giving back of information. And so, because of that expectation that precision medicine equals genomics, it’s another reason why we’ve actually not just started out of the gate doing that. I think we will pilot things early next year at some of the sites where we can just start to learn what it’s like if you get whole exome or whole genome, and are systems ready for that. But in terms of scaling those out, there’s a set of criteria and phase gates that we’re trying to do to make sure that we wouldn’t actually do this until all of these things were in place.

And some of—we have not communicated a lot about the program yet; as you can imagine, a program this visible has lots of masters. So there’s a whole set of communication materials and branding and educational material for different audiences, including the provider community, that we’re building the capacity to go do. But I’m just waiting for the right timing. It’s just back to, I don’t want to—if we’re going to go out and talk to providers, I want to make sure that we’ve got the content and the understanding of the kinds of providers that we’re going to do and the support organization’s in place.

## Scene change

Camera view of Dr. Roden.

## Mr. Dishman continues:

We will, I believe, pilot elements. I’m trying to get off of the notion of one big all-singing, all-dancing launch.

## Scene change

Camera view of Mr. Dishman speaking beside the lectern.

## Mr. Dishman continues:

We will, I believe, pilot elements. I’m trying to get off of the notion of one big all-singing, all-dancing launch. If you’re going to start in regions and then scale out, you’re going to start with some particular health provider organizations who are more ready and scale out, and you’re going to start with four or five different approaches to an enrollment of especially diverse populations and then save the other 12 to 15 until you start to learn from those. So this is partly why—and the other thing that we’re going to be doing is, and we’re setting up the infrastructure to do it—we will be talking to the cohort, to the participants, in a very regular way, saying “Hey, we think we’re going to do this about then.” But telling them, “You know what, we think there’s going to be a two-month delay and here’s why.” I mean that level of transparency I think is just going to be required out of the gate in terms of expectations management.

## Dr. Green:

I see many hands. Jeff and then Mark, then Gail, and then Sharon.

## Scene change

Camera view of the conference table, participants, and Mr. Dishman at the head of the room by the lectern.

## Dr. Jeffrey Botkin

(University of Utah)

Thank you for this presentation. I’m a little uncertain still about the role of children in the PMI.

## Mr. Dishman:

Mmm hmm.

## Dr. Botkin continues:

And in particular, given the potential influence of prenatal …

## Scene change

Camera view of the conference room and audience, from the front of the room looking to the back.

## Dr. Botkin continues:

… influences and environment on future health—are you going to be recruiting pregnant women and collecting samples relevant to pregnancies as part of the PMI, either now or down the road?

## Mr. Dishman:

Yeah, we will.

## Scene change

Camera view of Mr. Dishman speaking beside the lectern.

## Mr. Dishman continues:

And particularly the children piece is something that we’re not going to start out of the gate with, nor will we do those with cognitive impairment or incarcerated populations and so forth. But we have a roadmap and a plan for tackling all of those things, with work groups that are going to be working on those. The variability of state law in some of these particular areas is going to be particularly challenging for us, so an analysis of all the state laws that add another layer to it. But we will take children, we will take pregnant women, and we will get to the point where we figure out how to take those with different developmental disabilities. Some of the players that we have on the network, or in the HPO network, have experience with that. So it’s one of these things where it’s like we want to learn from—and you need the trusted brands of those and relationships of those local organizations.

## Scene change

Camera view of Dr. Botkin seated at a table with a laptop computer.

## Mr. Dishman continues:

And in some places they’re doing this quite well and how do we extrapolate and teach the rest of the HPOs to do that.

## Scene change

Camera view of Mr. Dishman speaking beside the lectern.

## Mr. Dishman continues:

But all of that is in the plan to work on. And we just—it’s one of these things where it’s like, focus on a minimum definition, get out the door, learn and iterate as we go.

## Dr. Mark Johnston

(Professor and Chair, Department of Biochemistry and Molecular Genetics, University of Colorado School of Medicine)

The initiative was announced, I don’t know, seven months ago …

## Scene change

Camera view of the conference room and audience, from the front of the room looking to the back.

## Dr. Johnston continues:

… and you’ve already awarded 8 grants to more than 30 organizations. It seems remarkably fast. How did you pull that off? What was the process?

## Scene change

Camera view of Mr. Dishman speaking beside the lectern.

## Mr. Dishman:

Partly, the air cover of the president, the secretary, and the director of NIH is both a blessing and a curse, and the blessing part of it was you were able to pull multiple departments—and so many parts of NIH stepped up to help. I’m based in the director’s office, it doesn’t have a lot of the infrastructure that something like NHGRI does or—Gary was here earlier in terms of heart and lung. So basically leveraging their expertise and their capabilities to get it out of the door. The other transactional award will be the fastest that we’ve done. We started—for this round of health provider organizations, it went out—it’s going to be a two-month period from end to end. In fact, I have like four minutes and I need to run because I got to get to a review in about 10 minutes away from here.

Mr. Dishman laughs.

## Mr. Dishman continues:

But that helps us a lot.

## Dr. Green:

The OT makes a big difference on some of these fast turnaround things.

## Scene change

Camera view of the conference room and audience, from the front of the room looking to the back.

## Dr. Johnston:

And do they have the usual peer review, the usual levels of your review?

## Scene change

Camera view of Mr. Dishman speaking beside the lectern.

## Mr. Dishman:

It’s slightly different, but the spirit of what traditional peer review is, is in there, in terms of the way that it’s done.

## Dr. Green:

Gail?

## Dr. Gail Henderson

(Professor, Department of Social Medicine, and Director, Center for Genomics and Society, University of North Carolina School of Medicine)

Well, first, I’m from Chapel Hill. I work at UNC.

## Mr. Dishman interjects:

Go Heels!

## Scene change:

Camera view of the conference table, participants, and Mr. Dishman at the head of the room by the lectern.

## Dr. Henderson:

Yeah.

## Scene change:

Camera view of Mr. Dishman speaking beside the lectern.

## Mr. Dishman interjects:

There are so many Duke people around, and I love you, but come on, I need my Tar Heels.

## Dr. Henderson:

Yeah, I know.

## Scene change

Camera view of Dr. Henderson sitting at a table with a laptop computer.

## Dr. Henderson continues:

So my question, it probably should wait till later, but it does have to do with the very difficult and contentious debates around how you define race and ethnicity in the context of ancestry populations, forgetting about the social factors that are clearly implicated in race and ethnicity and culture. And it sounded to me like what you were saying was something that was really quite interesting, that you’ve got proposals that are doing some different things and you’re going to see how it works.

## Mr. Dishman:

Uh huh.

## Dr. Henderson continues:

And then you’re going to look at it and go forward. Is that your plan about this really thorny topic?

## Scene change

Camera view of Mr. Dishman speaking beside the lectern.

## Mr. Dishman:

That’s part of it. And each of the awardees had to show us a plan of what their catchment area was, what diversity was on a lot of different axes, and what their strategies were. And they’ve signed up to say, “Hey, we will deliver certain numbers of these people over this period of time.” But I think it’s one of these things where you’re going to want the collective mindset of, “Hey, this is working really well here, here, and here. Should we make that a national thing that’s available across all of the folks that are there?” And there’s really thorny IRB issues with this as well. And we have one central IRB for everybody, which is I think the way NIH is going for these larger projects, but we’re on the cutting edge of figuring out what that means …

Mr. Dishman laughs.

## Scene change

Camera view of Dr. Henderson nodding.

## Mr. Dishman continues:

… getting a whole lot of locals to be OK with that.

## Dr. Green:

Sharon?

## Scene change

Camera view of the conference table, participants, and Mr. Dishman at the head of the room by the lectern.

## Dr. Sharon Plon

(Professor, Baylor College of Medicine)

So I’ll be really quick. Congratulations. I would just say, apropos of this question, how did you do it so fast? I think a lot of the kind of industry terms—like, I was sitting here taking notes—and approaches are things that many of us in academics could really benefit from, and many of us running consortia.

## Scene change

Camera view of Dr. Plon.

## Dr. Plon continues:

And it would be really nice to see how some of the work going into this would then translate into other aspects of NIH funding and planning and the iteration phase and things like that.

## Scene change

Camera view of Mr. Dishman speaking beside the lectern.

## Mr. Dishman:

I think I’ve already taken elements of sort of user experience and product development classes that I’ve taught for years and I finally just said last week to Francis, “Francis, we’ve got to find more people that know this because I cannot go teach this a thousand times.” But at some point when I can breathe, I would love to teach some of these basic infrastructure to both government and academic folks, because it works. And as somebody who has been a not-for-profit, academic, large for-profit, and now government, so there are some methods in here that are very powerful and really tested for a lot of different things.

## Dr. Green:

Jonathan?

## Scene change

Camera view of the conference table, participants, and Mr. Dishman at the head of the room by the lectern.

## Dr. Jonathan Pritchard

(Professor, Departments of Genetics and Biology, Stanford University)

Can you say something briefly about data release, how that’s going to work?

## Mr. Dishman:

It’s going to work very well. See, that was very brief.

Laughter from audience.

## Mr. Dishman continues:

It’s like on Star Trek when they talk about the Heisenberg compensators.

## Dr. Pritchard:

Slightly more about that.

## Scene change

Camera view of Mr. Dishman speaking beside the lectern.

## Mr. Dishman:

It will be a—it’s a risk-based model and certainly non-risky data that’s de-identified to the degree, you know, when calculating the risks of different levels, much of the data will be released to anybody that wants to do a query on top of it and then it’s basically just a risk-based model of credentialing for access to increasingly risky data as we go. But it’s partly—that’s a certain aspect of the answer. But as I mentioned, there are programs that we want to create and that we have the seeds of for. It’s like, how do citizen scientists use this data and what’s the education and training that needs to be in place to teach them how to use it, not just open up a website and say, “You can have it.” And the same thing is true for community college as well. So it’s—full disclosure—it’s like Vanderbilt and their colleagues own developing those data and research support tools, and they’re well down that path and many of those same people are involved in eMERGE and others.

## Scene change

Camera view of Dr. Pritchard, seated at a table, listening to Mr. Dishman’s response.

## Mr. Dishman continues:

So they have experience doing this as well as BD2K projects.

## Scene change

Camera view of Mr. Dishman speaking beside the lectern.

## Mr. Dishman continues:

But we’re not—we haven’t spent a lot of time on it yet, because it’s going to be a while before we start to have enough meaningful data. That being said, on the survey data and there’s other kinds of things, we expect to try to release that very quickly and let people mine it and characterize their own geography or their own community right out of the gate as soon as we can.

## Dr. Green:

Before we [unintelligible] is there anybody on the phone? We have several council members on the phone.

## Scene change

Camera view of the conference table, participants, and Mr. Dishman at the head of the room by the lectern.

## Dr. Green continues:

Did anybody want to get a quick question in? No? No queries to Rudy.

## Scene change

Camera view of Dr. Green seated at a table.

## Dr. Green continues:

OK, respectful of your time and your need to get to this review, I think we’re going to end there. Thank you so much.

## Scene change

Camera view of Mr. Dishman speaking beside the lectern.

## Mr. Dishman:

Sounds good. I hope you’ll have me back. I can’t wait to learn more from you. Thanks so much.

## Dr. Green:

Yeah, yeah, yeah, absolutely.

## Scene change

Camera view of Dr. Green.

## Dr. Green continues:

Rudy, are we going to take a break now?

Inaudible response

## Scene change

Camera view of the conference room and audience, from the front of the room looking to the back.

## Dr. Rudy Pozzatti

(Deputy Director, Division of Extramural Operations, Scientific Review Officer and Co-Chief, Review Branch, National Human Genome Research Institute, National Institutes of Health)

How about 20 ’til? Run upstairs and get your caffeine, everyone.

Mr. Dishman walks into the frame and shakes Dr. Green’s hand.

## Closing Slide

Logo of the Department of Health and Human Services

Logo of the En Eye H National Human Genome Research Institute

78th Meeting

National Advisory Council for Human Genome Research

Open Session

Monday, September 12, 2016