

# WHITE PAPER



## **ACCELERATING PRECISION MEDICINE**

Potential and Key Barriers Related to Policy & Technology

Presented by





## ACCELERATING PRECISION MEDICINE

### Potential and Key Barriers Related to Policy & Technology

The fully realized vision of precision medicine includes the capability to rapidly diagnose, sequence and develop a personalized treatment plan based on combined analysis of genotype, phenotype, environmental, clinical and behavioral data. Precision medicine can be advanced through the “All of Us” research initiative of the Precision Medicine Initiative, the most ambitious accumulation of human genomic, personal health and clinical information in history; but accelerating precision medicine to achieve this vision will require new capabilities and speed in personal health, clinical and genomic data storage transfer and analysis, as well as in interoperability and security. Further, precision medicine will demand new private sector, legislative and regulatory approaches for review and approval of genomic sequencing technologies and other software and devices needed to advance knowledge of disease factors that demonstrate variability among individuals.

The *Accelerating Precision Medicine* program, sponsored by Intel and hosted at the Personal Connected Health Alliance’s 2016 Connected Health Conference, consisted of a main stage panel and two workshops focused on the technology and policy barriers to accelerating precision medicine. Panelists were invited based on their specific expertise, and participation was open to the public. Main stage panelists were selected to represent perspectives from policy, clinical care, patient advocacy and technology. Insights from these sessions, collected and summarized below by HIMSS Analytics, will be provided to the White House Office of Science and Technology Policy and organizations affiliated with the Precision Medicine Initiative.

## BACKGROUND DATA TOOLS & POLICY AS DOCUMENTED BARRIERS TO PRECISION MEDICINE

In an April 2016 publication, “Information Needs in the Precision Medicine Era: How Genetics Home Reference Can Help,” published in the *Interactive Journal of Medical Internet Research*,<sup>i</sup> authors discuss the need for “new tools for researchers and clinicians to store, manage and analyze large amounts of data...as a key factor in the implementation and success of precision medicine.” The announcement of the Precision Medicine Initiative by the Obama Administration in January 2015, which includes a volunteer research cohort of one million people to explore the intersection of lifestyle, environment, genetics and individual variability on disease and prevention, brings urgency to the need for these new tools.

Policy challenges of accelerating precision medicine are also recognized. For example, authors of “Integrating precision cancer medicine into healthcare — policy, practice, and research challenges,” an opinion published in *Genome Medicine* in October 2016,<sup>ii</sup> concluded, “Without necessarily creating a complete new set of legal and ethical issues, the advent of PM [precision medicine] has made the border between research and healthcare increasingly porous. This uncertainty challenges policymakers to find new policy tools and solutions to protect traditional principles and norms such as informed consent, return of results, privacy and confidentiality, and benefit sharing. Given that research is an increasingly international endeavor, whereas healthcare is still defined at the national or regional level, these questions will need to be answered at different geographical layers, while promoting normative coherence and integration.”

At the same time, Research and Markets’ report, “Precision Medicine Market Analysis & Trend — Therapeutics, Application — Forecast to 2025”<sup>iii</sup> finds that the global market for precision medicine, including applications in companion diagnostics, targeted therapeutics, biomarkers, big data analytics, pharmacogenomics, next-generation sequencing, molecular diagnostics and other applications, will grow at an compound annual growth rate (CAGR) of about 11.12% over the next decade, reaching approximately \$112.62 billion by 2025. According to the Personalized Medicine Coalition, from 2014 through 2016 nearly one in four drugs approved by the FDA was a personalized medicine, which the organization defined broadly as ‘those therapeutic products for which the label includes reference to specific biological markers, identified by diagnostic tools, that help guide decisions and/or procedures for their use individual patients.’<sup>iv</sup> As of 2015, according to a separate study sponsored by the Personalized Medicine Coalition and conducted at Tufts Center for the Study of Drug Development, 42% of all medicines and 73% of cancer medicines in development are potential personalized medicines.<sup>v</sup>

Recent research conducted by HIMSS Analytics showed limited evidence of precision medicine programs across the U.S. healthcare market, but found that many precision medicine programs are in place at larger, research-based organizations such as academic medical centers (35 percent), multi-hospital health systems (25 percent), or organizations with over 500 beds (41 percent).<sup>vi</sup>

## PRECISION MEDICINE PROGRAM AT CONNECTED HEALTH CONFERENCE

Recognizing the current state of precision medicine, and pressures to accelerate the discovery of technology and policy solutions to meet its potential, Intel embarked on the *Accelerating Precision Medicine* program to coalesce expert and public opinions about technology and policy barriers. The program consisted of a main stage panel, “Accelerating Precision Medicine,” and two workshops: “Part I: Defining Policy Changes Needed for Accelerating Precision Medicine” and “Part II: Overcoming Technology Barriers to Precision Medicine”. This publication, developed by HIMSS Analytics and the Personal Connected Health Alliance, is the output from that program.

## CORE QUESTIONS

Panel participants were asked to identify the top two or three barriers to achieving precision medicine. Workshop participants took part in focused sessions on technology or policy, while the main stage panel covered technology, policy and other issues of concern to panelists. Comments pertaining to barriers and concerns are consolidated with input from workshop participants in the ‘Key Program Insights’ section of this document. Direct quotes are included from the main stage panel, which was recorded, while workshop participants’ contributions were captured in notes highlighting conversation threads and are incorporated as summary material.

# PANEL PARTICIPANTS

## PANEL PARTICIPANTS

## MAIN STAGE PANEL ACCELERATING PRECISION MEDICINE

**Moderator: Susan Dentzer**, President and CEO, Network for Excellence in Healthcare Innovation (NEHI)

**Kevin Johnson**, MD, MS, Senior Vice President of Health Information Technology, Vanderbilt University Medical Center

**Michael McManus**, PhD, Senior Health & Life Sciences Solution Architect, Intel

**Bray Patrick-Lake**, Director of Stakeholder Engagement, Duke Clinical and Translational Science Institute; Co-Chair, NIH Advisory Committee to the Director Working Group on the Precision Medicine Initiative

**Greg Simon**, JD, Director, Biden Cancer Initiative at Biden Foundation; former Executive Director, White House Cancer Task Force

## WORKSHOP DEFINING POLICY CHANGES NEEDED FOR ACCELERATING PRECISION MEDICINE

**Facilitator: Alice Borrelli**, MPA, Director Global Health and Workforce Policy, Intel

**Aaron Black**, Director of Informatics, Inova Translational Medicine Institute

**Samantha Burch**, MA, Senior Director, Congressional Affairs, HIMSS Government Relations

**Jeff Coughlin**, MA, Senior Director, Federal Affairs, HIMSS Government Relations

**Shannon Curtis**, JD, Assistant Director, Federal Affairs, American Medical Association

**Greg Downing**, DO, PhD, Executive Director of Innovation, US Department of Health & Human Services

**Greg Eley**, PhD, Chief Technology Officer, Inova Translational Medicine Institute

**Rob Havasy**, MS, Senior Director, Connected Health, HIMSS



**Sean Khozin**, MD, MPH, Senior Medical Officer, Office of Hematology and Oncology Products, FDA

**Christopher Khoury**, MS, MBA, Vice President, Environmental Intelligence and Strategic Analytics, American Medical Association

**Tom Leary**, MA, Vice President, HIMSS Government Relations

**Edgar MacBean**, MBA, Senior Manager, Market Development, Population Sequencing Illumina

**Michael McManus**, PhD, Sr. Health & Life Sciences Solution Architect, Intel

**Michael Strubin**, Manager, Personal Connected Health Alliance

**Scott Thiel**, MBA, Director, Navigant Consulting

**Afton Wagner**, PhD, Manager, Federal Affairs, HIMSS Government Relations

## WORKSHOP

### OVERCOMING TECHNOLOGY BARRIERS TO PRECISION MEDICINE

**Facilitator: Rick Cnossen**, PhD, MBA, Vice President, Enterprise and Government R&D Philips Government Solutions Group

**Aaron Black**, Director of Informatics, Inova Translational Medicine Institute

**Doug Bogia**, PhD, Platform Solution Architect, Intel

**Greg Eley**, PhD, Chief Technology Officer, Inova Translational Medicine Institute

**Julian Goldman**, MD, Med Director, BME/Dir Interoperability, Partners Healthcare System, Inc.

**Joseph Kvedar**, MD, Vice President, Partners HealthCare, Connected Health

**Michael McManus**, PhD, Sr. Health & Life Sciences Solution Architect, Intel

**Andrew Omidvar**, PhD

**David White**, MBA, Market Research Analyst, Philips Healthcare

# KEY PROGRAM INSIGHTS



## KEY PROGRAM INSIGHTS

This section contains consolidated insights from the main stage panel and the workshops. Direct quotes from main stage panelists are included where comments were particularly insightful or representative of participant feedback.

### THEMES: BARRIERS AND CONCERNS

**Integrating Precision Medicine into Clinical Care**, for example, educating providers to make appropriate decisions about when to use precision medicine approaches and counsel patients based on genomic or other biomarker information.

Converting data into knowledge that will drive improved outcomes was a common theme among participants. Clinicians, patients and healthcare organizations will need an end goal, a definition of success, that will help promote the adoption and impact of precision medicine. Even when genomic data is available, there are few who know how to identify the clinically meaningful data necessary to develop actionable insights or knowledge. Only when healthcare providers are able to associate validated, clinically relevant biomarkers with patient data will precision medicine be effective.

### QUOTES:

#### Greg Simon

“...How many people are really going to need precision medicine? Is it going to help them as compared to what we would think of as traditional medicine if people know more about their family history? We don’t often even need their genomics sequenced, but very few people actually go to the trouble of really knowing their family history. So, we don’t need to make everything hard. As they say, if you hear hoof beats, think horses — not zebras. Precision medicine will often be a zebra and we need to watch out for that....And then, when we get down to should we be paying for sequencing for the average patient, the question would be: Have you really looked at the most important data about that patient, which is their family history?”

Simon went on to give a personal example related to family history:

“So my genetic test from Navigenics, many years ago, showed I had no risk for glaucoma. Unfortunately, I already had glaucoma, which I got at the driver’s license exam because I have monovision from LASIK surgery. So, I can’t read with both eyes. This is my reading eye. This is my distance eye. They said, ‘You have to go to an eye doctor and certify that.’ Went to the eye doctor. The eye doctor said, ‘You have glaucoma.’... We’ve got to improve the funnel that captures people with problems we can deal with, because

when I got diagnosed with glaucoma, I thought, geez! I asked my mom. I said, ‘You know, your dad used to sit right against the television and scream obscenities at the Saturday afternoon wrestlers. Did he have glaucoma?’ ‘Yeah, he did.’...Well, let’s keep the simple stuff working and then we’ll work our way up to the expensive stuff.”

### Kevin Johnson

“There are data that suggest that it’s going to help in certain cases. There are data that suggest that what we thought was going to help last year probably isn’t going to help now that we know a little bit more. But two use cases that I think are very pervasive and I find sort of challenging myself: One very simple one: Homeless patients should not receive a medication that requires refrigeration. That’s precision medicine and that’s doable today.

“...The obstacles that keep me up at night start with physician and nurse and professional education. We still don’t teach — most of us who are physicians or nurses or dentists — about the role of precision medicine in healthcare....There was a child, for example, whose DNA parents sequenced, and they found a few things about the child that were interesting. One such finding was that the child had a risk for supralvalvular aortic stenosis, one of the different types of heart disease one can have and now the questioning really begins...If all we know is that this child is at risk, what do we do differently? The parents are asking everyone: ‘What do I do? I know my child’s at risk.’ No one knows what to do. We’ve never learned this. We’ve never really confronted this. For the next 10 years, a major obstacle is going to be to learn how to manage the information that we may glean from this data.”

**Health Equity & Patient Participation** was raised by several participants. Issues identified relate to equal access to precision medicine; consumers’ perceived value of precision medicine and its relationship to technology access; potential disparities that could arise if collection of sequencing data is dependent on having a premium insurance plan; and recognition of the fact that the data set of sequenced genomes currently skews heavily to a Caucasian population of European ancestry and must become diversified for new knowledge in precision medicine to be applicable to the general population.

Patient willingness to provide personal data will also be a challenge. They will need to be properly incentivized to participate in any precision medicine endeavors and allowed access to their genomic data. How will that data be stored, accessed and protected, and will their genomic data need to be re-sequenced eventually? These are all barriers that will need to be addressed from the patient perspective.

## QUOTES:

### Greg Simon

"...How does this affect the 10 to 15 minutes [a] doctor spends with you now, if you have to go in with your SNP analysis, and how do we distribute the benefits of precision medicine to parts of our country where they can't even get a mammogram or a lung X-ray in their community without driving for hours? We cannot continue to develop the high end without bringing everybody along, and we're a long way from that.

"...We have to figure out when sequencing is going to help people, and in order to do that, you have to sequence a lot of people that it doesn't help. CMS really doesn't like paying for things that don't help, so there's this debate going on about how do you do enough sequencing to find out when it will change the course of treatment and make it less expensive in the long run and when you're just gathering information about sequencing rather than treating people."

### **Financial Considerations**, such as cost and reimbursement of precision medicine.

Currently reimbursement and insurance questions abound as precision medicine is somewhat uncharted territory from a financial perspective. Additionally, the incentives for precision medicine appear to be wrong: healthcare organizations are not financially incentivized to begin programs and clinicians/consumers have not been shown the value in genomic data. It remains to be seen whether precision medicine policy can be shaped to address the needs of all healthcare organizations and identify the appropriate investments across the market to make the creation and sustainability of programs worthwhile.

## QUOTES:

### Kevin Johnson

"We have to think of a way that we can improve the reimbursement system in a fee-for-value structure, which is where we're headed, that will actually support what we're trying to do with precision medicine."

### Michael McManus

"Patients can get a covered expense, a lab can get reimbursed for a test they run and ultimately that creates the revenue flow that drives all the downstream efforts like educating physicians to be more knowledgeable about genetic disease and to understand the genomics process a little more. They don't have to be experts, but they need to understand a bit more about the origin of genetics and inheritance and how that affects disease. Those are really the improvements I'd like to see done in the future. I would like to see all this reimbursement taken care of, so you don't have to think about it. And I'd like to see the insurance companies realize that running sequencing early gives them a huge downstream benefit of preventive care, and intervention, rather than reactive care. I'd like to see physicians graduating with deeper training in genetics and understanding its relationship to disease."

**Data Sharing, Interoperability & Standards**, including attitudes about the sharing of data in healthcare, lack of interoperability and data standardization.

A common barrier identified throughout the Connected Health Conference sessions revolved around standards for precision medicine via creation of policy as well as medical and technical parameters. Currently there are limited standards set forth from a policy standpoint, as the healthcare industry has only initiated precision medicine on a limited scale.

Direction for standards needs to come in the form of policy intended to drive adoption and innovation, however policies must also address medical/treatment and technical issues concurrently. Initial technical efforts should focus on the standardization of data across the healthcare continuum and throughout the current healthcare IT infrastructure. This could mean standardizing the way data is collected and entered, the type of data collected, how it is stored and for how long, and where it can be found. Current thoughts on storage and retrieval of precision medicine data are focused on Electronic Health Record (EHR) solutions, but it is acknowledged that these solutions face challenges with regard to storage capacity. The Global Alliance for Genomics in Health is working on a non-EHR focused effort for interoperable genomics data exchange, and has developed Genomics API to help providers and consumers work together on a global scale. The API has already been adopted by Intel to share data on variant annotation.

New medical standards related to patient data privacy and ownership are also needed, and patient fears around privacy and genomic data ownership could limit participation, hindering progress in the field. Patients may not want to participate because of other reasons, such as the impact an individual patient decision about sequencing their genome may have on family members. Additionally, medical professionals and most healthcare organizations are not incentivized to adopt precision medicine. Efforts need to be made to expand the level of clinician expertise in understanding the impact of genomics and incorporating their precision medicine knowledge and experience into patient care.

Building on the theme of standards is the barrier of interoperability.

The healthcare industry has seen an unprecedented wave of IT adoption over the last eight to nine years in a rush to digitize medical records, provide better care and reduce costs. However, as healthcare professionals and organizations implemented these solutions the lack of interoperability between solutions became apparent. Hospitals can share data within their own ecosystem, but that is not enough to facilitate precision. Capitalizing on the potential of precision medicine will require capability to leverage existing data to the fullest extent, to share data across different/multiple ecosystems and provide data in a uniform format. This will help existing precision medicine programs and organizations

with plans to implement precision medicine programs move forward collectively. Lack of existing interoperability across healthcare ecosystems contributes to the misaligned financial incentives for healthcare organizations to become more interoperable and eventually contribute to precision medicine programs.

## QUOTES:

### **Michael McManus**

“There are insurance reimbursement issues around genomics which when solved will start the flow of revenue. This revenue flow is what will drive the effort to better educate physicians about genomics.”

“I would like to see insurers realize sequencing a patient early in life provides a huge downstream benefit being able to offer preventive care for a latent disease versus a more reactive care after that disease has manifested.”

“Interoperability. How do we actually interoperate between silos of data and how do we reduce those silos so we have our clinical data and our genomics data all integrated into a single data store?”

“There are regulatory issues, including FDA oversight of CLIA that affect how data are processed and stored.”

“Restrictions on the movement of data across sovereign boundaries is a big issue. It reduces our ability to encompass the breadth of genetic diversity in our ongoing analysis of the human genomes.”

## QUOTES:

### **Greg Simon**

“Nothing is fully dealt with, yet. We have been pounding on the cancer community about the data sharing since Day One of the Moonshot. The fact that Vice President Biden had to have his son-in-law, who’s a doctor at Jefferson Hospital, fly a CD of the Vice President’s son’s MRI at Jefferson to MD Anderson to get it there in time for a second opinion is crazy.

“So, we have initiated a conversation with all the cancer center community that’s now being run out of NCI to come up with the principals under which they will share data, and we want to make it a condition of being designated as a cancer center – that people adhere to those principles and share that data.

Just to give you an example, there's a network called Orion in southeast U.S. Has 15 cancer centers in it. They have common data standards for describing cancer. That doesn't exist in the rest of the community. They have common pathology standards for getting the tissue, characterizing it and preserving it. That doesn't exist in the rest of the cancer community. They have a common way of characterizing the patient, so they have the information ready to go when they get news of a new trial. That doesn't exist in the rest of the community.

"So, we ask the obvious question: If you can do it, why can't everybody do it? Let's expand that attitude and even those standards. Let's quit arguing about it because if this were finance, the world economy would have crashed a long time ago if I couldn't use my credit card anywhere but in the ZIP code of my bank, and that's where we are with medical data.

"So, we are aggressively pursuing this and, by the way, aggressively pursuing the electronic health record industry who got \$35 billion of your money to create proprietary, by design, non-interoperable, non-patient friendly systems. And we'll be having a meeting before we leave office with that cohort to demand that they come on the team to provide patients free, immediate access to the complete medical record in a way they can use it."

### **Storage and Management of Data, including policy and regulation**

The volume of data that has been collected as a result of the digitization of healthcare is already overwhelming clinicians and healthcare organizations. While some of this data is contained in EHR systems, clinicians still have to sift through massive amounts of data to piece together a complete longitudinal patient record. As precision medicine is implemented, it will greatly impact not only the volume of data but the velocity of data accumulation. The volume and velocity of data aggregation will need to be addressed through a substantial data management program which could include metrics to address data sharing parameters, analysis guidelines and best practices and the incorporation of machine learning. Additionally, future precision medicine initiatives could also incorporate an individual patient's social determinants of health, which will offer contextual data on top of the data already collected.

Patient willingness to provide personal data will also be a challenge. They will need to be properly incentivized to participate in any precision medicine endeavors and allowed access to their genomic data. How will that data be stored, accessed and protected, and will their genomic data need to be re-sequenced eventually? These are all barriers that will need to be addressed from the patient perspective.



## QUOTES:

**MICHAEL MCMANUS**

“We also have storage problems. For example, a whole genome, depending on what coverage you run it at--can be close to a terabyte in size. Do we have to keep all that data? Can we get rid of some of that data and store the essential data that doesn't require a huge storage load? Imagine if one hospital sequenced a thousand patients and kept all the data, that's a terabyte times a thousand. It's a lot of data just for a thousand patients. How do we manage it?

Then, if you take a step back, we have regulatory bodies like the FDA and CLIA, who are in this tussle about who's going to control what happens, say, inside of a CLIA lab. Are they going to be self-governed or are they going to be under the FDA's jurisdiction?

“The FDA is scrutinizing machine learning tools for feature detection in pathology images and variant detection tools in genomics because they want it to be safe. If you make a decision with these tools, downstream care will be based on these decisions.

This tussle has to end. It's been going on for over 25 years, and somehow, we have to resolve this so that we can get all the things in place — so the machine-learning and variant detection algorithms can be used and you know what data you have to store. If these issues all get settled, then that's a huge barrier that's out of the way.

“It's an ephemeral issue. If sequencing is cheap enough, if it was not that expensive and we could just take those bits of data that are important and throw away the rest and just re-sequence the same way you take another x-ray, then the issue would go away. But right now, in the current state, we keep these data because even though the technology is somewhat stable, it is about to change dramatically. So, in the early days of sequencing, people were keeping files that were even larger and then they realized that they didn't have to. And part of it is regulatory issues in the clinical world, part of it is just fear of losing something that they might want to go back and look at again. Your genome doesn't change, but the devices that measure what your genome contains do change, and that's an argument for re-sequencing later, but only if you can get the price down.”

# VISION & PRIORITIES

## VISION & PRIORITIES

Main stage panelists were asked to discuss their vision for precision medicine 10 years from now and to share their personal ‘wish list’ for accelerating its attainment. Their key comments are supplied below, and perhaps may provide the beginnings of a roadmap.

### 10-YEAR VISION & PRIORITIES SUMMARY THEMES & QUOTES

#### **Ability to Make Good Decisions with a Holistic View of Patients**

##### **Greg Simon**

“We will have a better understanding of where we need and do not need precision medicine. For example, with diabetes or other conditions where simpler approaches will suffice; cancers that can be identified and treated with chemotherapy alone...We will have made progress in rationalizing the science — figuring out where and when precision medicine is really necessary.”

##### **Bray Patrick-Lake**

“What I am looking for is the holistic picture of bringing the person and all the pieces into one picture of health and also integrating one system.”

##### **Kevin Johnson**

“Another one — of course, we talk about a lot — is pharmacotherapy that’s being tailored to some specific set of biomarkers in the patient, whether it would be genomic or other social and behavioral determinants or even potentially family history. I think those are where we should expect...in 10 years, I’m hoping that will be common place. Another one is precision diagnostics. The best example I give of that — I think we heard from Greg just a second ago — is disease sub-phenotypes. We all know that all asthma is not the same, all sickle cell disease is not the same. All of a lot of the diseases — hypertension, cancer — are not the same. I think the challenge now is to understand that. I believe that once we start to make more breakthroughs in that space, as we’ve started to do with certain diseases, we will then start to be able to come up with very important therapeutic changes to improve the outcomes of those patients. The third area that I think about a lot is precision screening. The best example I can give you of that is, for example, patients who are LGBT have increased suicide risk, yet we have to know that they are LGBT and know that data to be able to ask that question. Patients who have glaucoma, who have it at an early onset, now may be able to be diagnosed before they have acute losses in their vision because we

understand some of the genetics around glaucoma. Most of us who treat those patients as primary care providers may not know that yet, but it's in the literature and it's out there.

"... I think in 10 years the infrastructure will be in place to do a number of these things. Health information exchange will be available. I think wearables and ingestibles, which are actually also on the horizon, will become available, but I do think that there are some significant obstacles."

## **Hope for Reimbursement, Early Sequencing, Medical Training & Data Sharing**

### **Michael McManus**

"I would like to see all this reimbursement taken care of, so you don't have to think about it. And I'd like to see the insurance realize that running sequencing early gives them a huge downstream benefit of preventive care, intervention, rather than reactive care. I'd like to see physicians getting deeper training in genetics and understanding its relationship to disease...I'd really love to see medical records be able to be standardized across many institutions and be able to deal with genomic information and not just a few variants that you find but the larger information somehow accessible for people who want to use that information either for population health or for other endeavors."

## **Larger Volume of Data & Different Data Sources in Use, but Unpredictable Arc**

### **Kevin Johnson**

"The iPhone...it did not exist ten years ago. That type of black swan event could catapult us into a future that none of us could ever chart. The present is pregnant with the future, therefore what is present right now that is not widespread is what will be around in ten years...We will be drawing on different data sources. The most obvious routine data we are starting to use is quantifiable self-data and internet of things data. We will use and exploit those differently than we do now, but will also be collecting much more data and in different forms."

## HOW TO ACCELERATE THE VISION: THE WISH LIST

### Greg Simon

“The critical thing is to have artificial intelligence applied to the big data of patient outcomes first, rather than hunting for SNPs and alleles, etc. Let us get big data around patient outcomes for predictive purposes and longitudinal retrospective analysis of how did this get this way and what determined the outcome....I want to keep the patient at the center of it, patient outcomes, put that data at the center of it in a way that can be analyzed with computed intelligence and then allow the patient to have their record so that third parties can help them make sense of it.”

### Kevin Johnson

“...I would give patients the data and allow other businesses to develop methods that they can take some of what’s in the literature and help patients to understand their own data. I think that’s the fastest because that’s the group who has the highest level of incentive. I would pay 99 cents for someone to tell me something really smart about myself.”

### Michael McManus

“I would really like to do a few things. I’d like to make sure we sequence babies when they’re born. There are 143 million new births a year, all over the world. Let’s just sequence them, wherever we can. I’d like to be able to sequence people in remote areas in Africa. There are handheld sequencers now. Oxford Nanopore just announced they did a whole genome sequence on their MinION handheld sequencer, so it is possible to sequence in remote areas. Of course, storing the data is another issue.

“I’d like to solve the interoperability issues and some of the sovereign boundary issues, which we haven’t talked about. For example, the State of Texas will not share medical information outside of Texas...so, we have systems in place that enable one institution to benefit from the knowledge of another institution’s data without the data crossing state boundaries. I think that interoperability is a necessary evil in the era of silos. I’d love to see much more standardized medical records. That would be a huge development. I’ve worked at six startups in my career and I don’t know where my medical records are.”

### Bray Patrick-Lake

“I would like one record that can be shared for care and research, and that speaks to creating value. We have to let people know how we’re using their data, what we’re learning and how we’re using the technology to actually improve their health if we are going to keep their buy-in and keep people sharing and keep people interested in improving their own health.”

# CONCLUSIONS & PRIORITIES FOR ADVANCEMENT



## CONCLUSIONS & PRIORITIES FOR ADVANCEMENT

The Accelerating Precision Medicine program began with addressing some of the barriers and concerns related to precision medicine. One of the major barriers is not having the right data tools available. As panelists pointed out, there are big data tools available for healthcare. The integration of genotype, phenotype, environmental and behavioral data together, key to precision medicine, are not usually included in these tools. Policy issues are the second major barrier to precision medicine. While the previous White House helped launch the Precision Medicine Initiative and the Cancer Moonshot, comprehensive policies are yet to be developed for long term support of these initiatives.

The next major challenge, integrating precision medicine into clinical care, has seen some success, but not all providers are convinced of the value. Many oncologists see the value as subtypes of cancer are identified through genomic profiles. But for many other conditions, the value of precision medicine has yet to be proven sufficiently.

Health equity and patient participation were also discussed. While initial results show enthusiasm among white, well-insured patients, those who lack adequate means of payment and those who may be suspicious of how their data will be used or managed may not participate. Also, the general challenges of getting millions of patients to donate their data to the initiative may be the greatest challenge due to lack of awareness and access.

Financial considerations, such as cost and reimbursement of precision medicine, create barriers to routine use of these technologies in a broad clinical setting. The cost of genetic studies, while falling quickly, still represents a significant cost for the patient.

Data sharing, interoperability and standards, including attitudes about the sharing of data in healthcare, continue to frustrate efforts to expand precision medicine. Sharing data, particularly of cancer patients who are likely to seek a second opinion, is a major hurdle, especially for genomic data.

Storage and management of data is a technical challenge in precision medicine. While more data is available, storage of large files for millions of patients pushes the limits of what health systems can provide, pushing them to consider cloud storage. However, policy and regulation are yet to clearly address the use of the cloud and storage of large data files, like complete gene sequencing, and procedural practice has not been established by providers.

In spite of this extensive list of challenges, program participants expressed hope for accelerating precision medicine and were clear in expressing their priorities. First, the ability to make better clinical decisions based on a more holistic view of the patient. Second, there is hope for reimbursement particularly in oncology and rare diseases. Early sequencing, such as at birth, is under experimentation and could lead to significant benefits in the future. Establishing precision medicine as a routine part of medical training is also seen as critical. Participants also voiced the need to apply artificial intelligence to interpret the large volumes of data necessary to facilitate personalized clinical decisions. Finally, educating patients to understand their own data, and creating a 'one record system' to engage people in improving their own health, were expressed as priorities to accelerate precision medicine.

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