



The Integration of Personalized Medicine into Healthcare: Advancing the Promise to More Patients

By Elissa Quinn, Associate Director, Precision Medicine, Blueprint Medicines

ONE OF THE MOST important opportunities facing the medical community today is the integration of personalized medicine into healthcare. A broad range of stakeholders across the healthcare ecosystem must ensure that the innovative, life-saving tools we have brought to the

forefront address the urgent needs of the patients we serve. During a virtual Precision Medicine Leaders' Summit event this past May, I was the featured speaker among a panel of experts* spanning precision diagnostics, targeted therapies and healthcare delivery to discuss the current state

of personalized medicine adoption and a recently published study, "A Quantitative Framework for Measuring Personalized Medicine Integration into U.S. Healthcare Delivery Organizations."¹

Panelists agreed that personalized medicine strategies should become the standard of care ▶

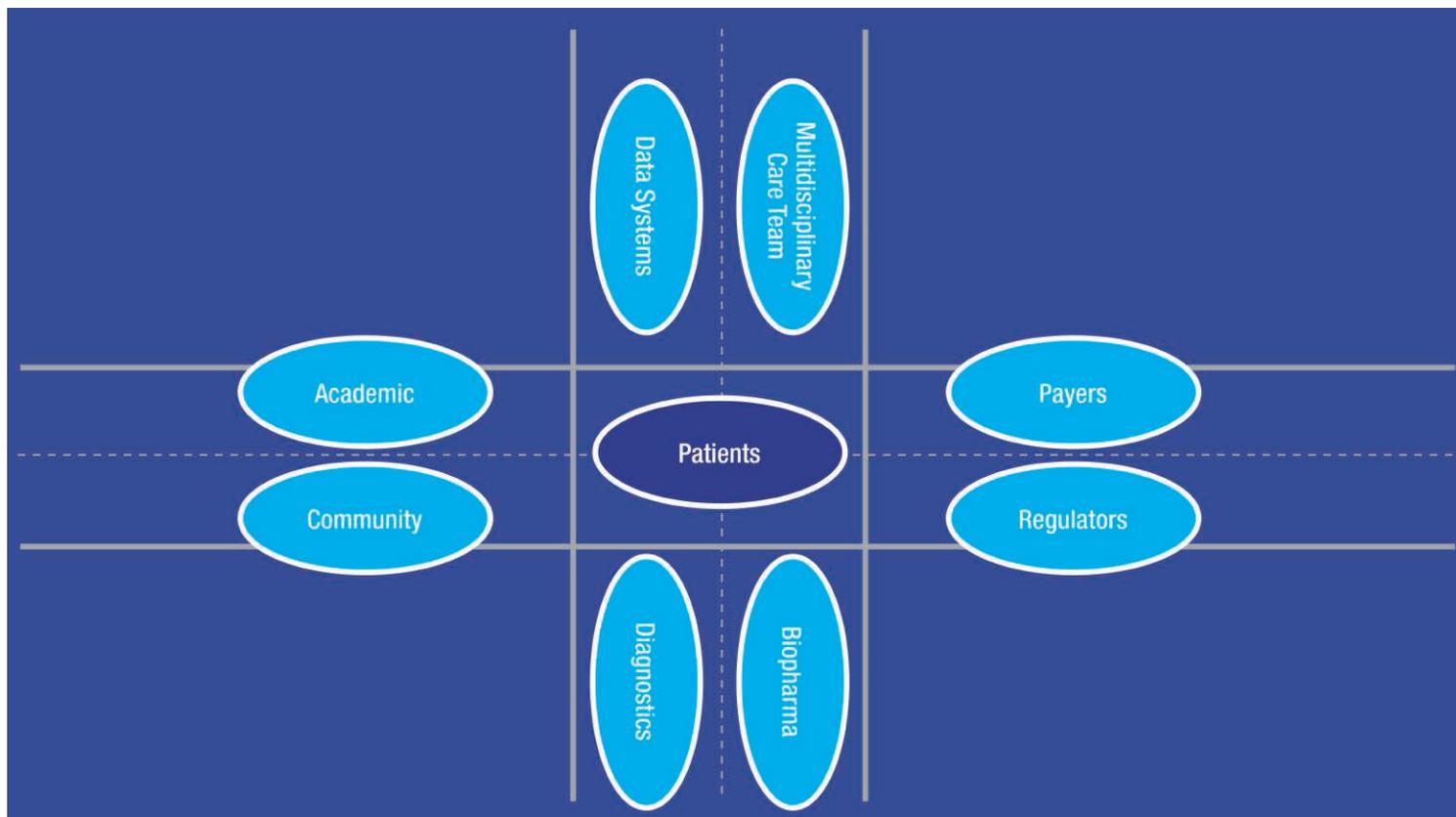


Figure 1: Intersection of Personalized Medicine Stakeholders

in many diseases; however, the challenges for integration and patient access are very real. In spite of dramatic progress including advances in test methodology, real-time clinical guideline updates and more supportive government and payer policies, personalized medicine falls short of reaching all the patients it is designed to help. Panelists highlighted that enhanced education across clinical stakeholders is critical, but insufficient by itself. Without healthcare system leaders establishing systematic standards and accountability and investing in resources to overcome operational barriers, personalized medicine data will not be acted upon across a broad range of patients.

Characterizing the Scope of Personalized Medicine Approaches

To lay the groundwork for the panel discussion, several of us shared our views on how personalized medicine should be defined. In opening remarks, I highlighted that it is a complex matrix of contributing factors and data points about a person including genetics, lifestyle and environmental influences that are intentionally used to inform a diagnosis, tailor treatment and impact patient outcomes. Daryl Pritchard, Senior Vice President for Science Policy at the Personalized Medicine

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Coalition, described personalized medicine as encompassing a “whole picture” of patient data that supports clinical decision making for that patient; on the other hand, precision medicine can refer more specifically to the science of developing medicines tailored to a person’s clinical, multi-omic, and molecular profiles, as well as the molecular biology approaches involved in the mapping of these measures.

In a 2015 paper published in *Science Translational Medicine*,² Hawgood *et al.* stated that biomedical research, health and health care are at an inflection point, poised for precision medicine and therefore moving us to integrate unstructured data into a comprehensive knowledge network (see image reproduced from Hawgood *et al.*). To me, this concept of a comprehensive knowledge network reflects the need for interdisciplinary care

coordination across diverse stakeholders, which is critical to realize the full promise of precision medicine and impact personalized care for patients. These stakeholders consist of clinicians and healthcare leaders on the internal practice side, as well as external industry organizations involved in advancing diagnostics, pharmaceuticals, data solutions and clinical decision support systems. Viewed holistically, personalized medicine is an intersection of mission-critical stakeholders, harboring different areas of expertise, goals, incentives and working norms (Figure 1). Most importantly, at the center of that intersection is the patient. We are all working to reach the ultimate objective of personalized care for the individual, so we may advance patient outcomes. Ultimately, we are looking to achieve this objective at scale, so that as many patients as possible can benefit from personalized medicine.

Advancements Driving the Need for Personalized Medicine Integration

To understand the myriad factors influencing personalized medicine, it was important for our panelists to assess where we have been, where we are currently, and where we have the opportunity to go. The previously cited Hawgood *et al.* paper was published in 2015, when the reality of personalized

medicine was very different than it is today. For example, in oncology, emerging biomarkers were predominately seen in colorectal cancer, lung cancer and breast cancer, rather than the broad range of relevant tumor types today. Analyses of these biomarkers were principally done by single analyte testing ordered on a “two ply” requisition form, and results were faxed or mailed back to the ordering physician, typically the pathologist. Only a handful of laboratories were offering multi-analyte testing, including next-generation sequencing (NGS), a high-throughput technology that determines the sequence of DNA or RNA nucleotides and is used to study genetic variation associated with diseases. Even fewer companies had dedicated field-based teams speaking with the medical community about the relevance of NGS, where it fits into clinical care, and how to leverage this tool to inform a diagnosis and tailor treatment.

Numerous targeted therapies have now been approved across disease areas, including 19 in 2020, representing 39% of all new molecular entity approvals by the FDA.³ The Center for Biologics has approved eight cell-based or gene therapies, including two already in 2021.⁴ NGS has moved from being offered by only a handful of laboratories to being broadly available, and test results are now transmitted electronically to both the ordering and the treating clinician and are stored digitally. Targeted treatment options have expanded significantly, including drugs that have been approved or agents that are being developed and for which patients may be eligible for clinical trial enrollment.

Calibration Model to Measure Integration

With personalized medicine applicable to a rapidly expanding set of patients, the Personalized Medicine Coalition recently commissioned a Health Advances study to quantitatively assess clinical adoption across healthcare systems in the United States and inform strategies that can be used across institutional programs. The study, “A Quantitative Framework for Measuring Personalized Medicine Integration into US Healthcare Delivery Organizations,” seeks to offer unique perspectives by reaching beyond academic institutions to survey a broad range of community hospital systems, geographies, therapeutic areas and adoption levels. In addition, the research extends beyond traditionally used metrics, such as testing usage, to include a broad, multi-factorial definition of personalized medicine adoption and integration.

Gary Gustavsen, Partner and Managing Director Precision Medicine Health Advances, explained that the adoption model incorporated



Figure 2: Eight Dimensions of Personalized Medicine Adoption¹

“By leveraging a consistent tool for interrogation of each healthcare system’s unique personalized medicine adoption “fingerprint” (or profile), the study is designed to highlight specific integration challenges and potential solutions that may be insightful for the healthcare system and other industry stakeholders.”

eight dimensions of personalized medicine (Figure 2), including:

1. Collection of genomic data
2. Other “omics” data (e.g., metabolomics, proteomics, epigenomics)
3. Non-laboratory data including clinical outcomes, economic, and social determinants of health;
4. Testing guidance and data accessibility
5. Utilization of data
6. Data sharing
7. Internal personalized medicine leadership
8. Funding of personalized medicine initiatives

By leveraging a consistent tool for interrogation of each healthcare system’s unique personalized medicine adoption “fingerprint” (or profile), the study is designed to highlight specific integration challenges and potential solutions that may be insightful for the healthcare system and other industry stakeholders. The model calibrates

how each individual institution is faring on the personalized medicine adoption curve. An ultimate goal is to encourage institutions to audit their own organizations and characterize their level of adoption, and more importantly, work to build on their current scoring and track progress over time.

Gustafson stated, “The beauty of this would be if people can stand behind their institution score and say ‘hey, we’re a level four precision medicine center,’ and for that to actually mean something.” He went on to say: “As you move up the adoption curve, we’ve shown there is a really tight correlation between a number of important outcome measures, be it economic or clinical, that really resonate with the C-suite, and then [it’s important to] have them try to push adoption forward.”

The publication of the initial study was based on a survey of a representative sample of 153 healthcare providers and systems. The analysis shows that 83 percent of the institutions studied scored a two or higher on the five-point scale used to examine their integration efforts. The analysis also showed that 22 percent of the institutions studied scored a four or a five on the personalized medicine integration scale. These findings suggest that a baseline set of integrated personalized medicine approaches are being applied across institutions, yet there remains an important opportunity to expand on these initial efforts across many healthcare providers.

Cross-Stakeholder Perspectives from the Panel: Driving Integration within Healthcare Systems

During our panel discussion at the Precision Medicines Leaders’ Summit, consistent themes emerged about how to accelerate personalized

medicine strategies for the benefit of patients. The following is a review and summary of four themes:

1. Securing Leadership Commitment and Tailoring Initiatives to Healthcare System Objectives

At the healthcare system level, the importance of leadership buy-in and tying to corporate healthcare objectives were highlighted throughout the panel discussion. Damon Hostin, Lead, Healthcare Systems, Market Access of Illumina discussed mapping the benefits of personalized medicine with the goals of the individual healthcare organization, so external stakeholders can tailor their conversations with administrators, staff and providers to the “currency” of how healthcare systems make investment decisions. Positioning personalized medicine in line with other innovations that enable cost avoidance, physician and patient satisfaction, and differentiation within the healthcare system’s regional footprint is important to convey the associated value and potential for improved outcomes. He added: “Knowing a patient deeper enables better decisions and that is a strategic investment.”

Catherine Hajek, Associate Professor of Medicine at University of South Dakota Sanford

School of Medicine and Medical Director, Sanford Imagenetics, added how her institution, an integrated healthcare system based in South Dakota, is incorporating personalized medicine into patient care. Hajek spoke of embedded genetic counselors throughout various clinics,

“Even with leadership endorsement and stepwise personalized medicine tools in place, adoption will lag in the absence of a system-wide belief in the purpose of these programs.”

preemptive screening, in-house testing, results processing, clinical decision support, and in-person consultation – many of the critical ingredients of an effective personalized medicine program. But how did they gain leadership support for this comprehensive initiative? Similar to Hostin, Hajek explained that it started by aligning this overarching program with the goals of the healthcare system – ultimately to improve patient outcomes and decrease costs. These objectives also aligned with increased patient satisfaction

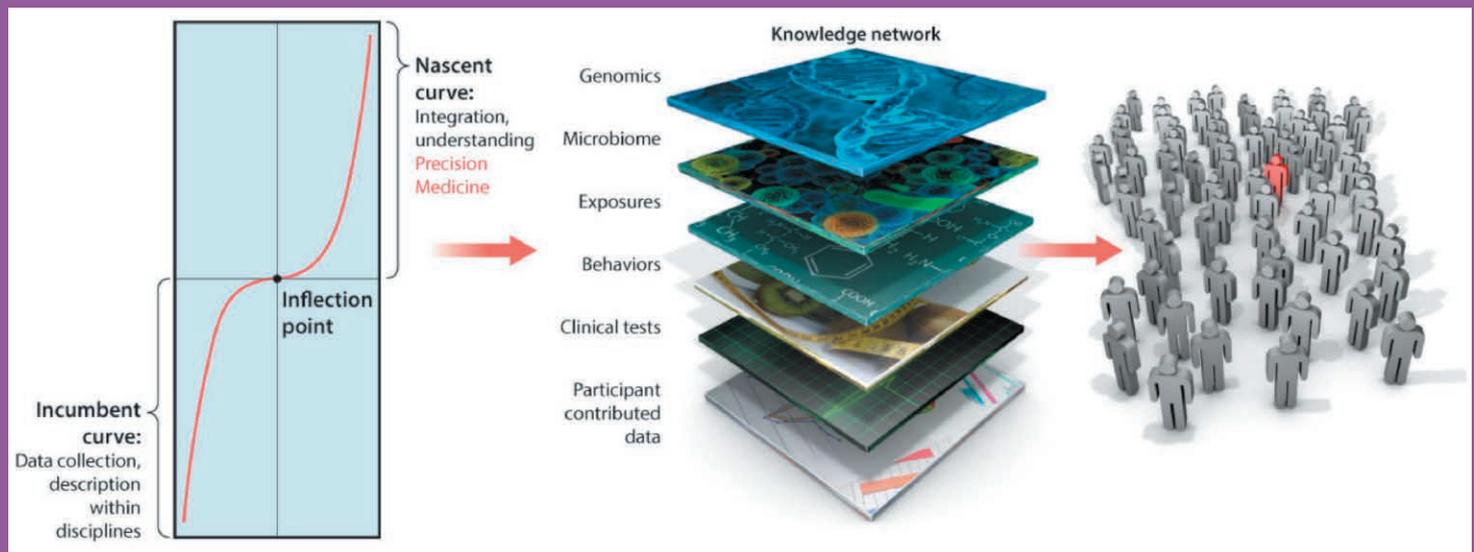
and differentiation from other healthcare systems. The approach helped Sanford overcome the initial barrier of convincing the healthcare system, providers, and patients that an integrated personalized medicine program would be beneficial for all involved.

2. Empowering Internal Stakeholders with Education and Driving Standardization & Accountability

Even with leadership endorsement and stepwise personalized medicine tools in place, adoption will lag in the absence of a system-wide belief in the purpose of these programs. Education is key: information must be applicable, timely, scientifically vetted, and importantly, coupled with relevant and ongoing training that is right sized for each audience.

Hajek described how Sanford Health delivers formalized educational content (including information geared to non-geneticist professionals) as required training in the form of 20-minute electronic modules. Providers, staff, and even administrators complete these modules quarterly on an ongoing basis over a couple of years. In addition, panelists highlighted how clinical decision support tools should be considered part of this larger educational program, because they can provide on-demand, guideline-supported >

A View From 2015: Poised for Precision Medicine



Surpassing single-layer healthcare. An inflection point marks an opportunity or moment of dramatic change between the first, or incumbent, curve, marking steady progress, and a second, or nascent, curve, indicating transformation and accelerated progress. In biomedical research, health, and healthcare, *we are at an inflection point, poised for precision medicine*. Whereas Google Maps links layers of transportation, land use, and other data, precision medicine aims to integrate and apply data from biomedical research, clinical practice, social/behavioral studies, and participant contributed observations toward better diagnosis, treatment, and preventative strategies.

Image and excerpted caption from Hawgood et al. Image credit: C. BICKEL/SCIENCE TRANSLATIONAL MEDICINE

information at the point of care. At Sanford Health, formalized education is bolstered by these embedded and automated decision support tools, as well as access to a team of genetic counselors and geneticists. Hajek believes these “safety nets” have allowed for improved uptake from a practice perspective.

In terms of accountability, once leadership voices establish a precedent for how personalized medicine will be practiced in the system, subsequent communication plans should be grounded in clinical evidence and guidelines, supported by resources and infrastructure, and reinforced with training that is required and repeated. Education is the thread that weaves together the individual fibers of personalized medicine – from applications for diagnosis and treatment to overall care practices – for an entire hospital, network or region. With standardization and the expectation that what is taught will be applied, measured, and improved upon, the implementation of personalized medicine will continue to strengthen and become a reality for all patients.

3. Bridging Differences Between Academic and Community Settings

Because institutions have varying patient populations and system-wide priorities, one implementation strategy does not fit all. At a broad level, there are often differences between the provider and patient experience in academic medical centers compared to community settings (e.g., rural vs urban). Experts have discussed the challenges and opportunities across these settings in roundtable forums countless times and inevitably, we tend to come up short on tangible solutions. In addition, a recent publication concluded that “rapid advances in molecular diagnostics, the advent of targeted therapies and the introduction of precision medicine have amplified differences between community and academic oncology practices.”^{5,6}

To overcome these challenges, a growing number of alliances are being forged between academic and community centers, leveraging their complementary differences for mutual benefit. For example, the greater diversity in economic, social, racial, and ethnic patient populations in community care enables more inclusive representation in clinical trials and, subsequently, strengthens drug development. In addition, the increased degrees of specialization, research and focus on training typical of academic medical centers are important; extending those benefits to patients traditionally seen in the community setting will be critical to advance equitable access to personalized medicine.

4. Collaborating Across Industry and Healthcare Practice Stakeholders

Beong Yoon, Executive Director of Oncology Global Medical Affairs at Amgen, highlighted educational as well as operational barriers to integrating personalized medicine into patient care, and called on industry stakeholders to partner closely with healthcare systems to help address these challenges. For example, he highlighted that in academic centers, all of the key clinical stakeholders are integrated into one healthcare system, whereas in diverse and dispersed community settings, that type of coordination is much more difficult. A critical question is how to address operational barriers in these settings, such as ensuring tissue availability for tumor biopsies to support personalized cancer care.

While various approaches leverage the power of data to advance personalized care, real-world adoption is limited in the absence of effective knowledge sharing. The panelists highlighted these challenges in the context of precision medicine, where a key goal is to deliver the right drug to the right patient based on testing results. As explained by Luca Quagliata, Vice President, Global Head of Medical Affairs at Thermo Fisher Scientific, without an understanding of what test results mean and the critical significance for treatment decisions, a physician will not order a test. Without testing, there is no way of stratifying the patient. Quagliata went on to say: “Biomarkers are only good if they are effective; effective means getting the right drug to the patient.”

Accelerating Progress

We collectively agreed that precision medicine has taken significant steps into healthcare systems in the 20 years since the introduction of the Human Genome Project. Today, we must reassess where barriers exist that continue to prevent personalized care from reaching patients. The panel raised and addressed relevant factors that have enabled us to form a comprehensive knowledge network, which will help us to bring personalized medicine to the patients that we serve. Have we created sufficient momentum to continue to challenge ourselves to re-evaluate how we are strengthening that network and solving problems? What biases and historical norms are we relying on to inform our approach to building solutions? As a personalized medicine ecosystem, we should now focus on inspiring healthcare leaders’ involvement and commitment, further investing in infrastructure, driving “right sized for the right audience” education, and partnering across stakeholders to advance toward our shared aim: ubiquitous and equitable personalized care for all who may benefit. 



Elissa Quinn

Elissa works in partnership with leaders across academia, professional societies, advocacy, and national labs to improve patient access to personalized medicine. With more than 20 years of industry experience in cancer and rare disease, Elissa’s unique understanding of the interplay between diagnostics and therapeutics has established her as a trusted ally within the precision medicine ecosystem. In her role at Blueprint Medicines, Elissa leads programs addressing barriers to adoption and integration of genomically guided care.

Acknowledgement

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- Elissa Quinn, Associate Director, Precision Medicine, Blueprint Medicines, Featured Speaker;
- Daryl Pritchard, PhD, Senior Vice President, Personalized Medicine Coalition, Moderator;
- Gary Gustavsen, MS, Partner and Managing Director, Precision Medicine, Health Advances;
- Catherine Hajek, MD, Associate Professor of Medicine, USD Sanford School of Medicine and Medical Director, Sanford Imagenetics;
- Damon Hostin, MS, Lead, Health Systems, Market Access, Illumina;
- Luca Quagliata, PhD, Vice President and Global Head of Medical Affairs, Clinical NGS and Oncology Division, Thermo Fisher Scientific;
- Byeong Yoon, PhD, Executive Director, Oncology Global Medical Affairs, Amgen

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