Illumina CIO Carissa Rollins; CEO Jacob Thaysen, PhD; and Head of Region—Americas Nicole Berry kick off the summit.

Illumina helps pioneering health systems deliver precision medicine

At a private summit, Illumina convened a group of health system leaders to discuss strategies for implementing genomics into clinical care

THIS MAY, 30 LEADERS representing 12 genomics-forward US health systems gathered at Illumina's San Diego headquarters for a two-day summit to share their lessons learned, challenges, and opportunities associated with integrating genomics into care delivery. The agenda included outlining strategies for supporting the Quadruple Aim (that is, enhancing patient experience, improving population health, reducing costs, and improving the work life of health care providers) along with priorities in quality, equity, and value-based care.

"It will take all of us working together to make genomics part of the standard of care," said Illumina CEO Jacob Thaysen, who during his first seven months at the company has traveled the globe to meet and spend time with customers. "We are here today to talk about our shared, greater purpose."

The United States has a unique and fragmented setup for care delivery, compared to countries with single-payer or national health systems. The task ahead, in the US, is implementing genomics into the clinical workflow while staying current with evolving clinical guidelines. In contrast with single-payer structures, it is important for American health systems to work with payers to show that conducting genomic testing earlier and more broadly will improve patient outcomes and provide value for everyone. Thaysen hopes that very soon all patients who could benefit from guideline-based tests will hear about and have access to appropriate and necessary genomic profiling."

Physicians need engagement and support

The summit attendees represented health systems that are implementing genomic technologies in different clinical care workflows, such as population screening, pharmacogenomics, oncology, cardiovascular disease, rare genetic disease, and even research into polygenic risk assessments.

The consensus among speakers was that primary care is an ideal setting in which to initiate testing because many people will follow through if their doctor says it's a good idea. Testing, if triggered by primary care, might have the broadest impact. Primary care physicians, however, are busy and would need any new processes to be simple. Many of the leaders at the summit had

experienced hesitation from physicians, and they emphasized that any genetic testing program would require provisions for educating physicians and for embedding decision support into routine care.

During the summit, "Make it easy" was a recurrent theme. One system found that doctors were more likely to adopt genomics if they could give away free screening tests. The system started with 1000 free tests and soon increased to 10,000. Another speaker suggested a way to keep it simple that came up during their COVID-19 screening: bundle testing with other routine or regular care—a patient could give their history and provide a sample at the same time they are in the clinic for a mammogram, for example. Yet another strategy is to assign staff who are not physicians to collect the samples and information, or to provide information that doesn't require a specialist.

Some genetic testing belongs with specialists, as there are specific guidelines for suspected genetic disease across clinical areas—but this narrow view is being challenged by emerging genomic capabilities driving whole-person care. The genomic data may be more helpful if the patient is sequenced comprehensively and early to assess potential disease risk or to avert adverse outcomes. Indeed, one clinician cited a cardiovascular study participant who, through testing, discovered they had leukemia. The patient was grateful for the early detection and the care that followed.

Recruiting clinical champions

Many at the summit agreed that the key to successful implementation was identifying clinical and administrative champions within their system. These individuals help educate peers and increase buy-in and adoption from physicians.

For example, in preparation for a study at a health system in the midwestern US, one such clinical champion contacted doctors and specialists to explain what kinds of results would come back and to ask who would be interested in seeing what—dividing up the types of information. After several rounds of communication, a growing number of doctors began to feel comfortable. He found that recruiting over time helped educate others and placate the protestations (such as "many conclusions drawn from genetic information have yet to be proven," "the results vary too much," "they're too complicated," and "testing isn't always covered by insurance").

Additionally, this clinical champion advised his peers not to wait for physicians to get on board before making things happen. Ideally, the process would be semi-automated. Recruiting, educating, reminding patients, gathering info, collecting samples, and interpreting and delivering results should have some degree of protocol and electronic assistance. Finally, he gave those in attendance a great tip: Don't position genetic testing as a research study, but rather as a quality initiative that will support the care delivered at their institution and ensure equitable access for all.

More testing, higher patient engagement, greater value

One health system found that patients who participated in genetic testing logged "hundreds" more encounters and engaged in more follow-up screenings or risk-reducing activities than those who didn't. This is important in terms of intervening earlier, before disease progression leads to unfavorable outcomes and poor quality of life. The leaders recommended gathering as much information as possible, as early as possible, to prevent diseases from developing and becoming painful, cumbersome, and costly for all.

"Health systems, payers, and patients spend a disproportionate amount of resources on chronic and poly-chronic care. The economic burden of cardiovascular disease is enormous," said Damon Hostin, Illumina's Health System Market Access lead. "Thankfully, cardiovascular care is moving in the direction of oncology, where risk assessment, prognostics, and specific diagnoses are based on genetic biomarkers, leading to more informed treatments. In this area, both the medical community and patients are beginning to reap the benefit of a more comprehensive approach."

In one health system's implementation, the patients were more enthusiastic about the testing than the physicians were. It was clear that any genetic test needs to provide value for everyone involved: patient, family, caregiver, and organization.

On reporting, synthesizing, and educating

Another hot topic of the summit was data management and reporting.

On the clinical informatics side, there's the challenge of integrating data from multiple sources, including reports from external laboratories or providers. Is it useful to integrate a PDF report into an electronic medical record (EMR)? When will discrete fields in an EMR be standard for physician EMR instances per system? How

should a pathologist disseminate results to various specialists? What is the best way to reanalyze existing data against new questions or updated publications and findings? And how can we best provide physicians with decision support?

Doctors, in addition to patients and families, need education. It's no small task. Illumina's medical affairs department, patient advocacy team, market access group, and the company at large are committed to educating health care professionals, patients, and families. That, in fact, was the standout recurring theme of the inaugural summit: Educate, educate, educate.

Today and beyond

Some of the speakers provided arguments for working through these challenges today. One cited cardiovascular complications as the second-leading cause of death in cancer patients, behind cancer itself. From this perspective, not conducting cardiovascular and pharmacogenomic testing is a drug safety and liability concern.

Another speaker posited that sequencing patients would encourage them to stay loyal to a single health system over many years, and stay committed to maintaining their diet, fitness, and health.

Overall, the summit's two-day dialogue was candid and productive—and one to carry forward.

"This is our first health systems summit," said Nicole Berry, Illumina's head of region for the Americas. "It's critical that we learn from each other on how to implement genomics and accelerate access." She shared that she had already fielded requests from health care leaders in other countries to host similar gatherings. Dozens of leaders worldwide are asking similar questions, finding innovative solutions, and looking to collaborate." ◆