



Precision Medicine Institute cofounders Gregor Stoddard, Alisa Gaskell, and Scott Demarest. Photo courtesy of Children's Hospital Colorado

Children's Hospital Colorado delivers precision medicine to kids

How an entire institution successfully integrated genomics into everyday care

OVER THE LAST 20 YEARS, precision medicine has become the standard of care, but for many hospitals, the complexity of the process has made it challenging to integrate or manage efficiently and at scale. To be successful, health care organizations need to think about how to educate physicians and staff system-wide, how to manage large datasets, and how to deliver results in a consistent way. The clinical workflow must make sense for patients, physicians, and hospitals.

Children's Hospital Colorado (Children's Colorado) was once in that position, balancing resources and refining processes in order to fully integrate precision medicine.

"We wanted to approach it from a holistic point of view and not that one specialty would be precision medicine and everything else would be just medicine," says Alisa Gaskell, PhD, scientific director of the Precision Medicine Institute at Children's Colorado. "We stood up three pillars—diagnostics, therapeutics, and education—to create a cohesive, institutional-level vision."

Integrating genomic diagnostics into day-to-day care

Children's Colorado formally took on the challenge in

July 2023, when it established the Precision Medicine Institute,¹ cofounded by Gaskell; Scott Demarest, MD, medical director; and Gregor Stoddard, administrative director. The institute delivers whole-genome sequencing data to clinical settings, conducts research, and generally leverages genomics to improve care.

"We create systems that help clinical groups maximize their ability to implement genomic testing," Demarest says. "We want them to feel confident they are delivering accurate diagnoses, based on the testing, and can confidently act on those diagnoses."

The precision medicine team realized early on that the hospital's electronic health record (EHR) system would be the best conduit to ensure that genetic information is easily accessed, clinically relevant, and fully responsive to the most current genomic research.

The first step in institutionalizing genomics is to ensure that the results are stored as discrete entries within the EHR. At Children's Colorado, the clinical genomics laboratory accomplished this in 2020. Having made genomic results searchable, the precision medicine team was well positioned to bring that data closer to

1. childrenscolorado.org/doctors-and-departments/departments/precision-medicine

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clinical practice. Now, if a provider is making decisions that could benefit from genomic data, the system scans for relevant results. If it finds them, it shares those insights. If not, it recommends a genomic test.

“Integrating that information in a more sophisticated way has been key to ensuring patients and physicians have the right information at the right time to make the right decisions,” Demarest says.

One early success has been in pharmacogenomics, which measures how genetics influence a patient’s responses to specific drugs. Timely information can help physicians make the most appropriate choices.

“It prompts them, saying their drug or dosing choice may be influenced by a pharmacogenomic result, and here are the guidelines to make that adjustment,” Demarest says. “We’re building it into the workflow to make it as easy as possible for physicians to implement.”

Genomic knowledge is constantly growing, and the system is designed to keep up. In the case of pharmacogenomics, new research may show that a certain gene variation makes a medication dangerous for some people. A patient’s sequencing data may be two years old, but the EHR now synthesizes the existing data with the new research. If a physician tries to order that medication, the prescription will be flagged.

“If we build something that’s only attuned to today’s level of knowledge, it’s going to age very quickly,” Gaskell says. “If a provider is choosing a blood medication, today there may be four different drugs and two genotypes that influence the decision. A year from now, there may be six drugs and 10 genotypes. Because we’re capturing whole-genome data that’s dynamically linked to our EHR, we can make real-time adjustments by just pushing a button.”

This dynamic connectivity between the EHR and large genomic data extends beyond pharmacogenomics. Considering that their hospital is dedicated entirely to children, the precision medicine team has a grand vision of delivering usable and dynamic information across the system of care.

“While pharmacogenomics is a great example of how genomics can impact a health care system, the same approach can deliver life-changing information in the rare disease and cancer spaces, leveraging system builds to support access to testing and actionability,” Gaskell says.

The feedback loop

One of the Precision Medicine Institute’s most important missions is education—increasing each provider’s genomic intelligence quotient.

To keep that knowledge flowing, the Precision Medicine Institute has created a clinical liaison committee, which includes 20 clinical areas and meets monthly to discuss the best ways to implement precision medicine.

“When we’re developing pharmacogenomics and other programs, it helps us understand whether we’re getting the details right,” Demarest says. “That helps us get the big picture to make sure we’re delivering programs that benefit providers and improve care.”

illumina’s role from the start

illumina has been partnering with the Children’s Colorado genomics team for around seven years, long before the Precision Medicine Institute was established.

“illumina and our teams have been committed to seeing our hospital take on clinical genomics,” Gaskell says. “We’ve leveraged our team’s know-how with illumina’s products to get to that excellent clinical implementation.”

The Precision Medicine Institute has integrated a suite of illumina products to manage their data pipeline. Clarity LIMS tracks samples and helps manage the genomics workflows. illumina Connected Analytics helps analyze and query large datasets. DRAGEN provides data analysis, and Emedgene automates interpretation and research.

In turn, the Children’s Colorado team developed a custom application programming interface to coordinate the hardware and software. The Precision Medicine Institute has three illumina sequencers—two NovaSeq 6000 Systems and one NovaSeq X Plus System—and the data pipeline has been fully integrated. This is particularly useful in complex cases, such as rare diseases, in which patients and their parents are all sequenced.

This has been a collaborative project from the start. illumina shared the applications’ underlying code so Children’s Colorado could knit them together and scale them for its incredibly high volume—around 450 datasets a month.

“We developed an operational dashboard to integrate all the data,” Gaskell says. “That means I can see where a sample is with just one click. If there’s a question about family samples, it takes me less than a minute to answer it. That’s really the goal: take complex data and make it easy to understand and actionable. Putting everything together, we give our researchers and clinicians better tools to succeed.” ♦