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Health Care

Children's Hospital Colorado unveils rare DNA lab that can read the human genome

The technology takes a "team of teams" to run effectively, the hospital said.



Alisa Gaskell, chief genomics officer at Children's Hospital Colorado, works in the hospital's whole genomic sequencing lab.

CHILDREN'S HOSPITAL COLORADO



By Analisa Romano - Senior Reporter, Denver Business Journal Mar 24, 2025 **Updated** Mar 24, 2025 9:12am MDT



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Children's Hospital Colorado has launched an onsite lab with the capacity to read a patient's entire genetic code, joining a small percentage of U.S. hospitals that employ the technology in-house.

The hospital's whole-genome sequencing lab was launched in September and announced publicly on March 11. Since then, the lab has run sequencing for about 3,500 people, helping clinicians to diagnose diseases and contribute to a more complete understanding of patients' health, said Alisa Gaskell, chief genomics officer at Children's.

Children's began using genome technology with the formation of its Precision Medicine Institute several years ago. That technology looked at specific regions of a patient's genome that most helped with diagnosing rare diseases and identifying effective treatments. However, it analyzed just 2% to 3% of the 3.3 billion base DNA sequences that make up a person's unique genetic code, Gaskell said.

"We knew that if we're really going to be at the forefront of making sure that we understand every patient that comes through the door, what's driving their disease on that molecular level, we had to get to a point where we needed to look at 3.3 billion bases," she said.

Many labs have the capacity to sequence specific regions of the genome to answer specific medical questions, such as those looking at cancer, but very few can look at a person's entire code, Gaskell said.

Children's has long planned strategically to invest in the capital and operational costs needed to run such a comprehensive lab, she said. Gaskell did not have an estimate for the cost of the lab, but she said it takes about 40 people to run and interpret the data.



Scientists at work in Children's Hospital Colorado's whole genome sequencing lab. CHILDREN'S HOSPITAL COLORADO

In addition to genomic scientists who work on moving specimens through the lab, Children's has research and development scientists, technology development specialists, and data analysts who ensure its quality, she said. Once the test is run, a separate team of scientists and molecular geneticists step in to analyze and interpret the results.

The data helps clinicians in departments across the hospital, from oncology to cardiology and neurology, Gaskell said.

Children's uses technology from Illumina, a biotech company headquartered in San Diego, to run the lab. The company is a frontrunner in genomic sequencing with technology and excels at serving the health care sector, Gaskell said.

"They really understood that to build this in a health care system, they have to be a partner," Gaskell said. "They cannot just bring a tool."

Getting health insurance companies to cover utilization of the lab hasn't yet "landed at perfect," Gaskell said.

"If you're in a really emergency situation in the NICU and you have a high suspicion of genetic underpinning, you cannot wait 30 days to get authorization for testing," she said.

The road forward involves more discussion, Gaskell said.

"You can see how there has to be dialog on both sides to enable that," she said.

"But also as an internal hospital, we need to build those inroads to ensure that we are providing the testing at the right time with the appropriate kind of approvals."

In addition to allowing patient samples to be collected with swabs on the cheek instead of blood collection, the in-house lab allows Children's to get genomic results back much more quickly, and the results integrate with patients' electronic health records to build toward more comprehensive treatment in the future, the hospital said.

One success story was a patient who had been in the Childrens' hospital system for 14 years with symptoms of very frail bones and frequent fractures.

[&]quot;There's a whole litany of genes that can cause that," Gaskell said.

After running the patient's DNA through the whole genome lab, Gaskell said her team identified a gene that makes the patient insensitive to pain. The issue wasn't fail bones at all, she said.

"That is the power of genomics," Gaskell said. "You can figure out what's really happening rather than what you're seeing from the outside."



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