

Pioneer Spotlight

Children's Hospital Colorado

"Precision medicine approaches contribute to a balanced, learning health care system, where continuous improvements are spurred on by individualized breakthrough discoveries, resulting in efficient care that is integrated, coordinated, and nimble," says Alisa Gaskell, Ph.D., the Scientific Director of Precision Medicine Program and Scientific Director of Precision Diagnostics Laboratory at [Children's Hospital Colorado](#).

Alisa Gaskell, Ph.D.

Co-chair and Scientific Director of Precision Medicine at Children's Hospital Colorado



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We at Illumina agree with Dr. Gaskell's assessment—we believe genomics-powered precision health is the next big step towards improving patient care, delivering better healthcare outcomes, and opening new avenues for healthcare discoveries and insights.

As the clinical, operational, and financial dynamics of healthcare continue to evolve, health systems will need to make innovative yet strategic investments to support this transformation. To see how these investments can pay off in a clinical diagnostic setting, we reached out to Dr. Gaskell to discuss how Children's Colorado developed strategies aimed at transforming laboratory operations to support a comprehensive precision genomics program.

Transforming Infrastructure

Cancer and rare disease are two key areas of clinical need for institutions that serve pediatric patients like Children's Colorado. 50 percent of primary childhood tumors harbor a potentially targetable genetic event, and pediatric cancer patients show somatic mutations in known cancer genes less frequently than adult cancers,¹ with a pattern towards more complex genetic alterations. As a result, there is often a great deal of overlap in the diagnostic journeys for pediatric oncology and rare disease, and genomics plays a key role.



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"We came to realize how many commonalities there were in genomic testing needs across the institution," says Dr. Gaskell. "For us, it just felt more natural to build one infrastructure rather than many separate ones." In 2018, she and her colleagues set out to evaluate various operational and structural models to update clinical testing programs at Children's Colorado.

Their design goal was to have a scalable and expandable testing infrastructure that could support the needs of patients and providers at Children’s Colorado while enabling the growth required to keep up with precision medicine advancements and evidence-based changes to clinical practice, which continues evolving rapidly. “We knew the structure should reflect the specific vision, goals, and needs of our healthcare system—and that customization should be true for any precision health initiative,” says Dr. Gaskell.

Streamlining Chemistry and Data

Dr. Gaskell and her team began by re-evaluating existing laboratory operations and requesting feedback on genomic testing processes from the entire institution. They questioned the need to provide a large menu of testing options regardless of test utilization, and explored opportunities to consolidate testing on a common chemistry to reduce testing complexity.

“We started to ask ourselves: Can a single chemistry serve most of our clinical needs across oncology and rare disease spectrum of testing?” says Dr. Gaskell. As a result, the precision diagnostics laboratory moved to run the majority of our genomic testing on a whole-exome backbone. This approach allows scientists to interrogate all genes in the genome regardless of the clinical question, but only report on the genes that are immediately actionable for clinical use. As a result, we no longer need to play catch-up with ever-evolving disease gene lists. Furthermore, the data outputs are not dependent on service line or test ordered, providing seamless interoperability across the entire institution, eliminating the need to harmonize data after the fact, and reducing operational burden.

The team identified 6 key elements of a successful precision genomics testing infrastructure:

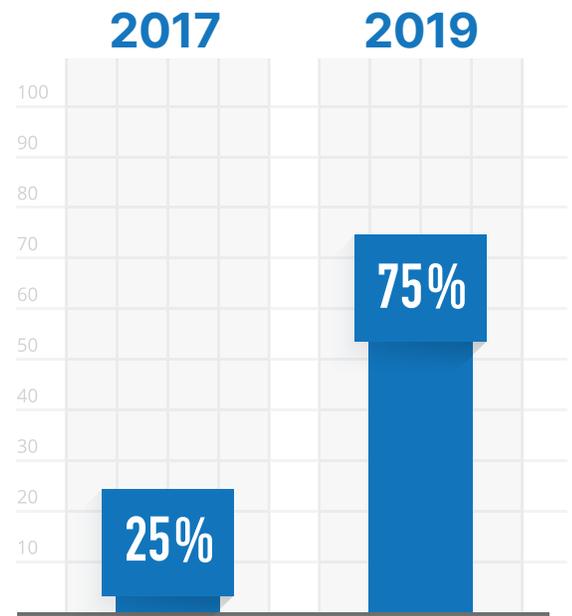
- 1 Single ordering access point
- 2 Nimble lab practices
- 3 Tailored bioinformatics solutions
- 4 Data architecture that supports [FAIR](#) (findable, accessible, interoperable, and reusable) data principles
- 5 Integrated and transparent clinical interpretation processes
- 6 Discreet data ingestion into the EMR, with pathways to record and support subsequent clinical decisions

Clinical, Operational, and Financial Benefits

Back in 2017, only approximately 25 percent of total oncology volume at Children’s Colorado was directed to genomics testing; by 2019, 70-75 percent of oncology tests were run on the genomics platform to enable precision health practices. By 2020, they expanded further to bring on large-scale genomic testing to streamline and consolidate testing processes.

“The value proposition of genomic testing is very clear in the clinical realm, but there is also immense value operationally and financially,” says Dr. Gaskell. The precision diagnostics laboratory at Children’s Colorado reduced the number of tests that needed to be staffed from 20 to 10, improving not only the throughput of the tests but also the turnaround times and the efficiency of their staff. Although the up-front cost to internalize genomic testing seems steep, Dr. Gaskell says it has brought significant savings. “When we look at the costs of sending out all those tests previously, we see that we’ve actually carved out over 50 percent cost savings with our internalized solution, and the data is fully interoperable with our EMR,” she says.

Today, Children’s Hospital Colorado is evaluating moving to whole-genome sequencing as the de facto backbone for genomic testing to serve most clinical needs. Their progress thus far demonstrated that a strategic investment in internalized testing for the whole institution is not only better for the clinical outcomes, but also provides value-added opportunities towards operation and financial outcomes in the long run.



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¹ Gröbner, S. N. et al. The landscape of genomic alterations across childhood cancers. Nature 555, 321–327 (2018).