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Roshi Subedi, Lannie Edwards, and Tinus Creeper of Territory Pathology. Photo courtesy of NT Health

Operating a speedy, high-tech NGS lab in remote Australia

Illumina bioinformatics solutions prove essential for a Northern Territory genomics program

DARWIN IS A MICROBIOLOGISTS'S DREAM. Located in the far reaches of Australia's Northern Territory, the region is hot, humid, and host to a wide range of pathogens. From a public health standpoint, that makes the area a little precarious—and someone has to keep track of all those microbes.

The Genomics Department at Territory Pathology, based in Royal Darwin Hospital, is playing a crucial role in monitoring the infectious diseases coming into the Northern Territory. In addition to local pathogens, Darwin has a busy international airport and cruise ship terminal, which are entry points for an assortment of microbes especially respiratory viruses.

To complicate matters, the Northern Territory is vast—larger than continental France, Spain, and the UK combined. Territory Pathology has regional labs in Alice Springs (930 miles or 1500 kilometers away, with an arid climate) and other remote locations in Katherine, Gove, Tennant Creek, and Palmerston, but all genomics tests are conducted in Darwin.

The Department of Genomics began its services by offering SARS-CoV-2 whole-genome sequencing (WGS) for epidemiological surveillance in the Northern Territory. It then focused on developing WGS services for organisms of public health importance. Validation is currently underway for *Salmonella*, *Shigella*, and invasive Group A *Streptococcus* (iGAS), and with a plan for tuberculosis. Cloud-based analysis solutions have enabled this small, remote laboratory to expand its capabilities.

During the test-trace-isolate period of the COVID-19 pandemic, using genomics for tracking and tracing the source of cases was important, and having local sequencing capacity was key to providing results in an actionable timeframe. The Darwin lab started with an Illumina iSeq 100 System but quickly outgrew it. As the pandemic escalated, the group bought a MiSeq System and a NextSeq 2000 System, as well as a liquid handler to automate their processes. These platforms are now pivotal in developing a broad range of WGS services, spanning microbial analyses and oncology panels.

At the time, the laboratory did not have a bioinformatician, so the team leaned on cloud-based analysis solutions such as BaseSpace Sequence Hub (BSSH) and the DRAGEN COVID Lineage pipeline to perform lineage analysis of the virus.

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Cloud-based bioinformatics and automation for overnight analysis

Illumina specialists helped the lab staff increase their efficiency by providing hands-on training, both with wetlab and *in silico* operations.

In addition to using BaseSpace Sequence Hub for bioinformatics, the laboratory has now employed Illumina Connected Analytics (ICA)¹ to scale their cloud bioinformatics operations. The transition to ICA enables the lab to analyze runs overnight; its flexibility allows them to use a number of open-source pipelines to help identify different pathogens. For instance, the ShigaPass² *in silico* tool can be integrated with the Illumina pipelines in ICA for Shigella serotype predictions.

Expanding into cancer genomics

After establishing next-generation sequencing to monitor pathogens, Territory Pathology became interested in extending those capabilities into oncology. Currently, the tumor samples it analyzes travel as far as Melbourne for testing, nearly 3200 kilometers (2000 miles) away. The geographic distance and the shipping process prolong the turnaround times for these tests, which may result in delays to treatment.

The lab has been one of the first in Australia to adopt Illumina's cloud computing. As the team moves into oncology, they have been working with Illumina experts to optimize Illumina Connected Insights³ for performing downstream variant interpretation of heme samples. The team did some initial pre-validation work with a myeloid panel to screen for blood-based cancers such as acute myeloid leukemia, and now is shifting toward replacing this panel with a new pan-heme panel. They intend to expand their capacity for solid tumors after completing validation and offering the service for hematological malignancies based on requirements from the National Association of Testing Authorities, Australia and National Pathology Accreditation Advisory Council. By providing this service locally, the team can reduce the turnaround time for hematological malignancy panel testing from several weeks to just a few days, ultimately improving health outcomes for Territorians.

2. pmc.ncbi.nlm.nih.gov/articles/PMC10132075

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^{1.} illumina.com/products/by-type/informatics-products/connected-analytics.html

^{3.} illumina.com/products/by-type/informatics-products/connected-insights.html