Computational biomedicine at the forefront of epigenomics

The advent of new genetics technologies has exploded in the last decade. Since the unravelling of the human genome project in 2003, and with recent discoveries in the field of epigenetics, we are starting to understand not only how and when genes are expressed, but which post-translational modifications are important in disease, and how we can manipulate them therapeutically. Professor Christopher E Mason has developed new technologies to catalog genetic and epigenetic changes caused by everything from cancers to novel viruses, and these tools have proven invaluable during the COVID-19 pandemic.

Since the first discoveries in the field of epigenetics in the 1950s, it has been understood that all the inheritable changes occurring within a cell cannot be explained simply by the DNA sequence itself. We now know that both environmental and lifestyle changes can influence whether genes are expressed or silenced, and these factors can have potentially damaging effects on our health, such as making us more prone to developing certain diseases. Professor Christopher E Mason is an expert in computational and systems genomics, having worked in the field for over 15 years. He is one of the founding directors of the MetaSUB International Consortium. Through his research, he has developed a wide knowledge of the epigenome and epitranscriptome – DNA- and RNA-level modifications, respectively – to gather vast amounts of data that will allow a better understanding of how some of these seemingly unrelated pathways interact in disease progression. Dr Mason explained: "I oversee a combination of molecular and computational biology research programs using 'wet work' at the lab bench and the design of new algorithms and machine learning to process and make sense of the data that is generated and discover its biomedical meaning."

"Prior to the COVID-19 pandemic, we mostly processed cancer and microbiome samples, the latter as part of a project called MetaSUB, which is a large consortium tracking microbial changes in urban environments. Initially, we were doing everything by hand, but our rapidly growing workload meant that we urgently needed a robust platform that could reliably and continuously process the large numbers of samples we had coming in. Crucially, it needed to be something we could easily fit into our lab and allow us to hit the ground running. I heard about the DreamPrep™ NAP workstation through a number of our collaborators, and Tecan has a good reputation in the field, so I contacted the company to see what we could do."

"We were quickly able to set up and run a high throughput platform using the DreamPrep NAP workstation to extract and process genetic material collected from either clinical samples, or from the environment – sewage, public benches, city subways, etc. I am very much an omnivore of genetics, so when I'm looking at viruses, I like to find them all, which is why we use a shotgun sequencing approach. This is the real fun part of science – pure discovery – and in the environmental samples we've been collecting, we've seen anywhere from 30 to 60 percent new species, annotating them and tracking their evolution as we went. This very broad, flexible approach meant that, when the pandemic hit, we were already very well set up to provide a rapid platform to track SARS-CoV-2 genetic material from various sample types, including both patients' samples and swabs from hospital surfaces, etc."

The lab still had to adapt its workflow to accommodate the unexpected increase in throughput due to testing of COVID-19 patient samples. Dr Mason expanded: "Since the beginning of the pandemic, we have been involved in numerous projects, both for getting new testing methods onto the market and for tracking the emergence of new strains as a result of viral evolution, to help understand the implications for disease development, treatment and vaccination. This means that our workflow has been pretty much 5am to 10pm every day. We really felt that this was a time where we could be most useful to the wider society, and so we worked as much as we could!"

"The DreamPrep NAP has fitted seamlessly into our workflow, saving us time, energy and costs. Without it, I don't think we would have been able to process anywhere near as many COVID-19 samples as we did, especially with the need to reduce the number of staff we had in the lab at any one time. It has been a true workhorse, and very reliable, with the preprogrammed protocols working well. The support from Tecan has also been very responsive. Any time we've had an issue or questions, we've had someone on the phone really quickly, or coming to check it out, which is essential in this kind of situation," Dr Mason concluded.

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To find out more about Tecan's DreamPrep NAP workstation, go to www.tecan.com/ dreamprep-nap-workstation

For general information on nucleic acid purification visit www.tecan.com/nap

To learn more about the MetaSUB consortium, visit www.metasub.org

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