

Bringing Bioinformatics Pipeline In-House Reduces Costs and Decreases Turnaround Time

Phosphorus uses the DRAGEN™ Bio-IT Platform to perform genomics data analysis onsite and at an accessible price point.

Introduction

Alexander Bisignano is a molecular biologist and a serial entrepreneur, founding several biotechnology companies over the past 10 years. He is now cofounder and Chief Executive Officer of Phosphorus, a US-based direct-to-consumer genomics company enabling people to uncover their own unique genetic profiles. With a current complement of 24 employees, Phosphorus is strategically focused on improving human health with genomics.

In 2017, the Phosphorus team re-evaluated the company's bioinformatics infrastructure amidst expanding sample throughput requirements and alignment on strategic goals. They wanted to bring bioinformatics analysis in-house cost-effectively. After reviewing their options, the Phosphorus team adopted the Dynamic Read Analysis for Genomics (DRAGEN) Bio-IT Platform as the bioinformatics engine for the company's targeted sequencing assays.

iCommunity spoke with Mr. Bisignano to discuss the decision-making processes behind the evolution of the company's technology and bioinformatics platform, and the practical outcomes of those decisions.

Q: What was your motivation for founding Phosphorus?

Alexander Bisignano (AB): We started Phosphorus because we saw that there was immense opportunity to expand the scope and impact of genomics. We felt that by bringing more cost-effective and accurate technology to market, we could increase people's access to genetic testing and work toward increasing its utilization. We now offer a portfolio of genomic assays, all using next-generation sequencing (NGS).

Q: What is the significance of the Phosphorus company name?

AB: Phosphorus is a special and interesting element. It was discovered by the Greeks and means "bringer of light." When you think about carbon as the backbone of organic life, phosphorus is the energy. It's a central part of adenosine triphosphate (ATP), which is how energy moves throughout life.

Phosphorus is a group of scientists that are combining our energy to enlighten the world with genetic information. For us, the symbolism is apt.

Q: What are the principal elements of the Phosphorus technology platform?

AB: We use MiniSeq™ and NextSeq™ Systems for NGS and our own target enrichment capture panels. Our bioinformatics is now

based on the DRAGEN Germline Pipeline implemented on the DRAGEN Platform.

Q: How did you choose the marker sets for your sequencing assays?

AB: Our assay markers were chosen based on well-understood and well-documented gene-phenotype relationships. We took time to curate markers that are impactful and highly penetrant. We mined the literature and really narrowed the panels down to a core set of genes.

We've developed six targeted sequencing assays, each with a different marker set. For example, our PhosphorusOne panel includes 375 genes. Illumina NGS systems enable us to look at every coding and significant noncoding part of all gene transcripts to find genes related to cancer, heart disease, infertility, neurodegenerative disease, and more.

Q: What bioinformatics solution did you use initially?

AB: Initially, we were using a bioinformatics solution from another vendor that was deploying their version of GATK. At the time, we were just beginning to run these types of analysis pipelines. Having a third party performing the analysis made the most sense, as opposed to the complexity of building a bioinformatics team on our own. As our assay volume grew, we realized that the cost of outsourcing would continue to climb. We started looking at other bioinformatics options on the market.



Alexander Bisignano is cofounder and CEO of Phosphorus headquartered in New York City.

Q: What prompted you to choose the DRAGEN Platform?

AB: As we learned more about the DRAGEN Platform, we saw that its analysis speed was fantastic. We were also very excited that the DRAGEN Platform was significantly more cost-effective at our scale of sequencing. It also gave us the option to grow into other applications on our roadmap, such as whole exome, which was an important factor behind our decision.

Q: What was your experience transitioning to the DRAGEN Platform?

AB: We have a local DRAGEN server that was easy to implement. We were up and running quickly and found that it isn't difficult for us to support. Even though we are mostly a cloud-based stack, the local DRAGEN server integrates easily into our NGS workflow.

"As we were performing concordance studies...we ended up with accuracy above where we originally started. We are seeing better sensitivity and specificity in multiple areas with the DRAGEN Platform. It is faster, more cost-effective, and more accurate. That's a nice trifecta."

Q: How has the DRAGEN Platform performed?

AB: When we began implementing the DRAGEN Platform, we had enough data to perform validation experiments ourselves and test it. We compared basic variant calling and found some subtle differences between the DRAGEN pipelines and the other pipelines we were using. We also looked at more complex indels, homodimer repeats, and short tandem repeats (STRs).

As we were performing concordance studies and looking at different mutation types, we obtained good support from the DRAGEN team that helped make our validation process straightforward. We ended up with accuracy above where we originally started.

We are seeing better sensitivity and specificity in multiple areas with the DRAGEN Platform. It is faster, more cost-effective, and more accurate. That's a nice trifecta. It provides better access to testing through lower cost, high-quality analysis.

Q: What has your adoption of the DRAGEN Platform meant for Phosphorus and your customers?

AB: We are trying to make genomics more available, so price is an important feature. We are continuing to drive down our cost of goods wherever we can. With the DRAGEN Platform, we have been able to reduce our data analysis costs by 25-30% on a per-sample basis, including the cost of the physical server over a two-year timeline.

We are able to deliver higher quality assays than our competitors. The DRAGEN Platform is contributing to the work we are doing to gain market share.

"With the DRAGEN Platform, we have been able to reduce our data analysis costs by 25-30% on a per-sample basis, including the cost of the physical server over a two-year timeline."

Q: How does the DRAGEN Platform relate to your strategic vision?

AB: We are hoping to grow across several different segments. We are a genomics technology company with a laboratory in Secaucus, New Jersey and are actively growing that laboratory with referrals and laboratory-to-laboratory contacts. We are also a technology-enabled services company that helps to build and manage other genomics laboratories.

We are seeing great success in many of these areas and are hoping to see continued growth in all those segments over the next few years. Illumina NGS is the technology that is driving our genetic assays and DRAGEN Platform is enabling data analysis between the instrument and the report. The DRAGEN Platform is definitely a significant piece of how we will reach our goal.

Learn more about the products and systems mentioned in this article:

DRAGEN Bio-IT Platform, www.illumina.com/products/by-type/informatics-products/dragen-bio-it-platform.html

MiniSeq System, www.illumina.com/systems/sequencing-platforms/miniseq.html

NextSeq 500 System, www.illumina.com/systems/sequencing-platforms/nextseq.html