Illumina teammates Samuel Bunga, Daniel Diaz, Jelan Waly, and Nathan Castro-Pacheco pose with the MiSeq i100. Photo by Illumina

How an Illumina bioinformatics engineer contributes to the company mission

For Daniel Diaz, it's not just a job: He's motivated by the belief that one day we'll have a better understanding of all diseases

IN HIS FIRST JOB OUT OF COLLEGE, Daniel Diaz worked on firmware testing for a hard drive manufacturer. Next, he moved to an investment fund, where he met some great mentors. But it was still early in his career, and he knew he could afford to change direction. He felt a pull to do something that he connected with, at a company whose mission that resonated with him.

Diaz first found Illumina in a book. He was a software engineer living in Orange County, California, just an hour's drive north of the company's global headquarters, and he'd picked up a copy of *The Gene* by Siddhartha Mukherjee. "I remember reading in the final chapter that Illumina was the biggest genome sequencing company at the time," he says, "and that it was based in San Diego!"

He joined Global Information Services at Illumina in September 2019, and later transferred to the software engineering organization in R&D. As a staff bioinformatics engineer, he works on Illumina DRAGEN (Dynamic Read Analysis for GENomics),[1] a secondary analysis tool that gives researchers the ability to call genomic variants with award-winning accuracy[2] and speed.[3]

In his current role, Diaz's specialty is software testing, making sure that DRAGEN passes specific performance metrics. He tests that it calls variants correctly, and that DRAGEN apps perform well on the MiSeq i100. The 3 billion base pairs of every sequenced whole genome amounts to approximately 80 gigabytes, so it's quite a feat to identify the variants that might be causing a disease—and to do it with speed.

"It's really fast at doing certain computations that are important for genomic analysis," Diaz says. "Workflows that could take hours and hours can be completed in an hour, and that means a lot to our customers." The results speak for themselves: the better the variant caller, the better our understanding of the genome will be.

Diaz's knowledge of infrastructure and his cloud experience are useful for his current team, and in his time at Illumina, he's been able to expand his skill set beyond software and learn more of the biology side. He says, "It is unbelievable the combination of electrical engineering, computer engineering, software engineering, bioinformatics—all these disparate disciplines come

1. illumina.com/products/by-type/informatics-products/dragen-secondary-analysis.html
2. precision.fda.gov/challenges/10
3. bio-itworld.com/news/2017/10/23/children-s-hospital-of-philadelphia-edico-set-world-record-for-secondary-analysis-speed

together in DRAGEN to allow customers to do genomic analysis faster."

A personal investment

Diaz has Type 1 diabetes. This complex autoimmune disease is multifactorial—it has a combination of genetic and environmental causes.

Early during his tenure at Illumina, he underwent genetic testing. With a simple blood draw, he had his whole genome sequenced. He was surprised to learn that he did not have a variant in any of the genes known to be associated with susceptibility to Type 1 diabetes mellitus.

"It's possible I have a variant or a set of variants that are not well understood, or that gave me a predisposition that isn't well understood, or I just developed it through some other mechanism," he says. "But what that tells me is there's so much more to learn about Type 1 diabetes and what causes it. I actually think that's kind of cool, because it just means there's so much more to do. That's indicative of all of genomics in general—we're still at the very beginning."

He found the experience motivating. "I want to be part of that future that better understands diseases, including Type 1 diabetes. And I believe I can contribute to that by being here, through my work with DRAGEN."

Diabetes at work

Diaz says that diabetes is largely an invisible disease; you can't tell he has it just by looking at him. But because he's insulin-dependent, he must continuously wear a glucose monitor and take insulin manually.

He says there are times when he might have to miss a meeting because of a doctor's appointment: "My manager and my team have always been super understanding and supportive. I can't speak more highly of them. I'm even part of a clinical trial at University of California, San Diego for a new type of insulin, and my manager and my director completely support me taking a day off so I can do the overnight study there. It's a really good feeling when you know you have their support."

Currently his team is focusing on new applications for the MiSeq i100 System that will increase the number of use cases—and biological insights—available to customers. And that excites him. "I work at Illumina fully knowing and believing that we're actually helping people," he says. "And we're making an impact on people that are on diagnostic odysseys, people that have rare diseases, people that are trying to understand more about disease. I think that's incredible. The last 20 years of technology and innovation have all been driven by computers and software. In my opinion, the next 10 to 20 years are all going to be about biology."

Science, technology, and a dash of culture

Diaz also feels supported in other aspects of his life at Illumina. This month, Illumina and its employee resource group iLatinX will celebrate Latino and Hispanic Heritage Month with an opening ceremony in the campus amphitheater that will include mariachi music and paletas.

Last year, a mariachi band also played on campus. Diaz took a video and sent it to his parents, who were impressed. "I loved the experience," he says. "I thought it was really cool, and it just helps you feel seen, really. As a person who works at this really great company that's already incredibly diverse and you're represented as part of that diversity—honestly, it just feels good." ◆