Fabric Genomics Looks to Sew Up Clinical Genome Interpretation Market

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Premium

CHICAGO (GenomeWeb) – When Fabric Genomics changed its name from Omicia in March 2017, the idea was to alter the public perception that Omicia's strength was in the laboratory market rather than in clinical genome interpretation.

"Many people thought that we were a research company, for translational research, and not a clinical company. When our revenue was 99 percent coming from clinical customers, we decided to rebrand," President and CEO Martin Reese explained.

"We wanted to make a statement to all our customers and say, 'We are now a clinical reporting company, and not a research company anymore,'" Reese said. "We thought that a rebranding would signal to our customers that we can solve the problems of hospital systems."

This transformation dates to the end of 2013, when the company closed a $6.8 million Series A investment round. That money, along with a partnership with Laboratory Corporation of America consummated around the same time, helped what is now called Fabric Genomics launch its clinical application.

Reese started the Oakland, California-based company in 2009 with three others, including Illumina cofounder John Stuelpnagel, current Natera Chief Medical Officer Paul Billings, and bioinformatics entrepreneur Edward Kiruluta. Reese relinquished the CEO role for a time, serving as CSO and president, but took the top spot again in June 2017 after Matt Tindall stepped down.

"In the early days, we basically built a software platform for analyzing genomes," Reese said. Omicia actually worked with the first 10 genomes ever to be clinically analyzed.

"Because we did it so early, the first thing we learned is, we cannot do this manually anymore. Genomics was at that time hand curation, hand [authoring] of software scripts. Doing a full genome, we immediately needed automation. We needed to reduce the time of genome interpretation," Reese recalled.

A collaboration with the University of Utah's ARUP Laboratories got the ball rolling on the lab side.

In 2009, Omicia received a $590,000 phase II Small Business Innovation Research grant from the National Human Genome Research Institute to develop a computational system to predict novel genetic disease associations, in collaboration with Mark Yandell's group at Utah.

That followed a $400,000 grant Yandell received a year earlier to developed a "gene interference system" to predict candidate genes and mutations that may play a role in a disease and to develop software for the automatic annotation and targeted analysis of whole-genome sequence variation data.

That software became Omicia's VAAST, for Variant Annotation, Analysis, and Selection Tool, which allows individual individual scientists to pull relevant results from whole-genome variant files for specific diseases, such as breast cancer.

Fabric began publishing its pipeline as early as 2013 with ARUP, but needed something to interpret the genomes with. Thus, the clinical platform was born.

A $23 million Series B round in 2016 included participation from new investor UPMC Enterprises, an affiliate of the University of Pittsburgh Medical Center. Reese called the subsequent partnership with UPMC a "pivotal moment" for Fabric Genomics.

"They wanted to bring sequencing in house, but guess what they were missing? The software," he said.

UPMC initially asked Fabric Genomics to integrate the analysis software with its Cerner electronic health record. "We were genomic guys. We did PDF reports. We just had our engineering team work on integration into Cerner," Reese said. Now, the parts are fully connected.

Longtime partner LabCorp has its own clinical information system, which Fabric also is connected with. Fabric has integrated with one Epic Systems customer, but health systems with Epic typically have their own integration teams. All Fabric Genomics has to do now is connect to the EHR's application programming interfaces.

This year, Fabric signed on Intermountain Healthcare, the large, tightly integrated health system based in Salt Lake City. In June, the company announced 10 additional customers, including its first in Africa and the Middle East, namely the Centre for Proteomic and Genomic Research in South Africa and King Fahad Medical City in Saudi Arabia.

While Intermountain has a long history in the field, Many of Fabric Genomics' recent customers are hospital systems just getting into genomics.

"The hospitals, they don't want to do send-outs anymore. They don't want to exclusively send out the genomic data. They want to bring it in house," Reese said. Just like with UPMC, they can buy sequencing machines, but these organizations need interpretation software.

"Our goal is to bring this now to every single hospital and every single testing lab around the world, not only at these top institutions," Reese said. While there is plenty of competition in the interpretation market, Fabric Genomics has compiled a database of 50,000 genomes from public and private sources to train its algorithms in a form of machine learning.

One piece of its technology is the Phenotype Driven Variant Ontological Re-ranking tool, or Phevor, which came along in 2013.

"You can enter a phenotype of a patient and you combine it with the genome data, and that allows you to do better diagnosis, all automatically," Reese said. "What a typical scientist does is they find genes, and then they try to link the genes to the phenotype from the patient."

Reese previously said that Phevor offers a more automated means of selecting the variants that likely play the most pertinent roles in a particular patient's phenotype from lists of candidates that existing software such as the Variant Annotation, Analysis, Search Tool (VAAST) or Annotate Variation (Annovar) identify. It's an alternative to relying on disease experts to make the connections between phenotypes and genotypes

based on their prior knowledge of the disease or by manually searching for information from the literature, he said.

"We looked at that and we said, 'Hey, we can do that better automatically,'" Reese explained this week. We have a very nice tool called Phevor that analyzes phenotype data with [Human Phenotype Ontology] terms and then integrates that in a statistical framework with the gene burden test, and then you get the best possible ranked list in an automatic fashion," Reese explained.

It took a while for such tools to catch on, but Reese said that the market has taken to automation over the last five years. "It's a great shift in the market for us," he said. Fabric Genomics can process an entire genome from the computational side in less than 10 minutes, according to Reese, in line with others in the industry.

All along, work has continued on one of the company's largest efforts, its contract with Genomics England. In 2015, the then-Omicia inked a deal to become one of three interpretation service providers — along with Congenica and WuXi NextCode — for the UK's 100,000 Genomes Project.

Just last week, Illumina CEO Francis deSouza said that Genomics England has now finished sequencing 70,000 genomes and is on track to finish the remainder by the end of the year. While the 100K Genomes Project is winding down, deSouza noted that it will really be just the beginning of the UK's foray into population sequencing, since the country plans to transition into diagnostic whole-genome sequencing within the National Health Service for rare diseases and some cancers.

Last November, the NHS said that it planned to start offering diagnostic whole-genome sequencing routinely in October of 2018. At the time, Anna Schuh, director of molecular diagnostics in the University of Oxford's department of oncology, said that this will go along with a "complete restructuring of the NHS genetics labs." Rather than each individual NHS lab having a WGS protocol, sequencing will be centralized and performed at the same Genomics England facility used in the 100,000 Genomes Project. Meantime, other types of genetic diagnostics will be phased out, Schuh said.

Reese sees a major opportunity for Fabric Genomics here. "We're very excited because hopefully we can extend [our work] into the NHS," he said. "Every genome for rare diseases will flow through our pipeline."

Reese has his gaze fixed on other national genomics programs because he believes that Fabric Genomics' cloud-based platform offers the scale that massive sequencing and interpretation efforts require.

"You can imagine now in the world how many country projects are coming. It's just insane. That's another big focus for our products," Reese said.

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