Sequencing.com Hopes to Shepherd Growing Market of DIY Genomics Tools

Sep 27, 2016 | Molika Ashford

NEW YORK (GenomeWeb) – Sequencing.com, a new company hoping to play a unique role in the future of personal genomics, announced its global launch last week.

The last few years have seen the emergence and propagation of companies that help users in possession of their own genetic data to access and understand what this information means for their health, skirting the rules and regulations surrounding the marketing of genomic tests directly to consumers.

For example, genetic analysis company 23andMe had to stop communicating information about customers' health gleaned from its microarray-based analysis of their DNA after the US Food and Drug Administration cited it for offering a genetic test without the proper clearance. Since then, the company has launched individual analyses through the FDA, but still cannot perform the comprehensive analysis it launched with.

However, customers can currently take their raw genetic data from 23andMe or other companies and link it to their biology or learn about its implications for their current and future health, using services offered by other companies.

Anticipating a future in which use of these tools continues to grow, Sequencing.com aims to be a one-stop shop that will collate and organize available analysis engines or apps in a single space, and facilitate access to them by consumers who have a range of different types of genomic data at hand.

The company's platform has been live online since the beginning of the year, in what Brandon Colby, Sequencing.com's founder and CEO said was a beta period, in which it tweaked and optimized its system.

Now the company is hoping to get the word out about what it can offer users and app developers.

In essence, Colby told GenomeWeb, Sequencing.com wants to be both a safe space for genomic data, and a modern-world agora, or marketplace for tools to understand that data and put it to use.

"Our perspective on the data being generated by all these genetic testing companies is that it is owned by the individual. It is a person's asset, that they have, and that we store to make it useful for them," Colby said.

"One of the problems I have seen with the industry is that it's very fragmented in terms of the tools and different ways that exist to analyze and interpret genetic data," he added. "You might have data from 23andMe or be an early adopter of whole-genome sequencing, but you don't know what to do with it."

According to Colby, Sequencing.com has a two-part structure. First, the company offers free, unlimited, and HIPPAA-compliant storage of a user's genetic or genomic data.

"We have researchers with terabytes of FASTQ and BAM files. It's always free. We don't see storage as something that should be monetized," he said.

The second side is providing a marketplace that brings together what the company hopes will be all the best available tools or apps for analyzing and interrogating genomic data.

The site started with 25 internally developed apps, and now has 27, Colby said. And with its official launch this month, it is now anticipating adding its first externally developed apps in the next few weeks, with ongoing discussions with numerous developers.

In light of the company's focus on personal ownership and safety of users' data, it has a process for evaluating and vetting potential third-party apps. "We want customers to feel confident that there is a certain floor an app can't pass below," Colby said.

This process included the company's own bioinformatics team looking at the structure and claims being made by a potential new app, to make sure "nothing audacious is being claimed, and everything is based on published literature."

"This means no 'you have this gene, and you're going to live to 1,000 years, but you have to take our vitamins,'" Colby said.

Sequencing.com can host third-party apps on its own platform or, in cases where other companies have put a lot of time and effort into creating their own product and user interface, they can take advantage of Sequencing.com's app market API, which creates a required link with an outside platform.

The company's own apps currently range from those geared toward users with professional genomic experience, like bioinformaticians, to simpler consumer-oriented apps that address potentially actionable health information.

On one side, Colby said, the site's main bioinformatics app, called EVE (abbreviated from the word 'everything') allows users to create a pipeline for variant calling, alignment, and interpretation, without needing to do their own coding or be LINUX masters.

Since most of the tools that are a part of this app are open source, the app is free, Colby said.

On the other side of the spectrum, the current slate of apps in the Sequencing.com marketplace also includes an app called "Wellness & Longevity," which analyzes user data for links to genetic risk of a range of "preventable diseases," including cancers, heart disease, diabetes, multiple sclerosis, blood clot or bleeding disorders, as well as pharmacogenomic determinants of drug response.

"Knowing your risk means having the chance to lower it," the app description reads.

Another app in the online agora, "Healthcare Pro" is geared more toward generating a report for use by a medical professional.

"In addition to the genetic report, the Healthcare Pro app results also include detailed information about the specific genes and genetic variants analyzed as well as the medical references that link the variant to a specific disease, medication reaction, or trait. This information is easy to browse and search with some content linking out to additional information.

According to Colby, even though the end result of obtaining a traditional genomic test may look strikingly similar to what a user can do through a combination of genomic testing and app-directed analysis, by separating the actual testing of human samples from the study of the resulting data in the context of scientific and medical literature, the process remains outside the purview of the FDA.

"For all the associations that are made, the actual studies are included, so that there is a transparency in where the information is coming from. We make that very clear and obvious," Colby said.

"Apps do provide analysis of different genetic variants associated with diseases or phenotypes that could cause harm, but it's the choice of the person with the data whether they want to learn that information," he added.

A disclaimer at the bottom of Sequencing.com apps reads "the genetic analysis and statements that appear in this app have not been evaluated by the United States Food and Drug Administration. The Sequencing.com website and all software applications … are not intended to diagnose, treat, cure, or prevent any disease."

Although it doesn't do any actual genomic testing, Sequencing.com does have a preferred provider network. Companies included in the network automatically makes the genomic data they generate available through Sequencing.com if a customer chooses to contract with them.

Since the company doesn't make money from storing users' data, and it doesn't conduct or charge for any actual sequencing or other DNA analyses, there are only two aspects of its business that are revenue generating, although Colby said that the company is completely confident that its model will work, regardless of how much user data it eventually has to contend with for free.

The first source of revenue is actual app usership. Some of the apps, like the bioinformatics tool EVE, are free, while others currently on the site cost up to $10 per use.

Third-party app developers will work with Sequencing.com to price their own apps at a level that they think is right, Colby said.

Aside from that, the main revenue generator for the company is another type of API, a real-time personalization API that allows the integration of genetic and phenotypic data from users of Sequencing.com into mobile or everyday-use types of apps.

For example, an app could take advantage of whether a person is likely, based on their genome, to have blue eyes or be sun sensitive. "They can make calls against this and then personalize the app experience based on what they get back," Colby said.

At first, using the API is free, he explained, but as usership grows, developers have to sign up for a subscription. "That's really where we see our growth," he said. "That's what we see generating revenues for us and allowing us to provide our free services."

App users have to approve the connection for their data to be a part of this personalization API, Colby said. Unlike other companies which retain ownership of user data, and make money by selling or licensing that data to pharmaceutical or other biotech companies, a user of Sequencing.com can decide not to use any apps, and still freely store their data — or even delete their data from the system entirely.

Sequencing.com is not alone in addressing what many have argued will be a growing market for personal or consumer genomics.

For example, Illumina and a number of private equity partners have been working on bringing out a new company, Helix, to provide both affordable sequencing and database services for consumer samples

obtained through third-party partners, with access to a marketplace of on-demand apps to gain insights into the genomic data.

Other companies, like newcomer Genomics Personalized Health — which also works with Sequencing.com — aim to facilitate, or help customers navigate their options for DNA testing and analysis.

According to Colby, Sequencing.com's free storage is already attracting users, and data, rapidly. He also stressed that developers are attracted to the company's data agnosticism, which gives them an easy way to attract more users, and users with a variety of different formats of data.

For example, he said, "a lot of third-party apps were designed for 23andMe data and they are seeing good adoption for this, but they want to expand out to [users with data from] another company like Ancestry or they are looking at exomes."

"It can be a quagmire dealing with different formats of data, but when they join Sequencing.com they receive full compatibility."