OnQ Blog

# The Bio Boom: A Cottage Industry Around Cheap Genomics

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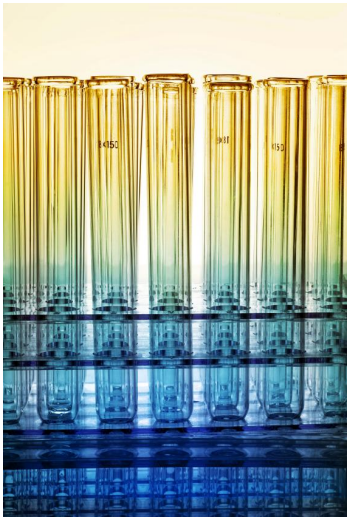
Can genome mapping change the nature of healthcare?

In June 2000, President Bill Clinton announced the Human Genome Project, an international effort to map all 20,000 to



25,000 genes in the human DNA. The project, which took 13 years and $3 billion to complete, inspired scientists and entrepreneurs to explore the possibility of mapping every person's DNA (genome) sequence.

This was a far-out idea in the early 2000s. Mapping a genome means decoding all three billion letters in someone's DNA, a process that took months and cost six figures to complete at the time. But the idea was nonetheless compelling.

Your genomic data can tell you your disease risk factors, how you might respond to certain drugs, and even what traits you might pass down to your children. But could that information empower or burden you? And what would you do with it?

After years of hard work and rapid technological improvements, the reality of direct-to-consumer genome testing is finally a reality. Genome sequencing can be done in days instead of months. Consumers can pay as little as $99 for a limited view of their data and around

$9,500 for their full genome.

As the technology accelerates and prices come down, Dr. Robert Green, genetics professor at Harvard Medical School, says genomic testing will soon go mass market. And from there, anything can happen.

"A lot of people compare the genomic testing revolution to the rise of the personal computer," he says. "There have been a number of ways we've used the personal computer, all of which have gone beyond anything that could have been imagined when Steve Jobs was creating the first Macintosh."

## The Genomic Holy Grail

For now, scientists are focused on one lofty goal: achieving the $1,000 full sequence. And in the meantime, "budget conscious consumers have options of doing smaller tests, just not a whole genome sequence," says Catherine Afarian, spokesperson for 23andMe, a genetic testing company based in Mountain View, California.

23andMe offers one of the most popular direct-to-consumer options. For $99, a consumer can order a testing kit, spit into a tube and send it back to the company.

From there, 23andMe will send the sample to a lab and come back with a *genotype* report. Genotypes differ from *genomic sequences* in that the data provides patients access to around 1 million genomic "markers." A full sequence includes 3 billion letters, giving consumers a more complete view. But 23andMe's less expensive offering produces more than 240 personalized genetic reports on how you will respond to medication, your risk for specific diseases and physical traits (such as do you blush when intoxicated, have wet/dry ear wax, etc.), Afarian says. Couples can learn the probability of traits in their offspring when both parties get tested and use the company's Inheritance Calculator.

"The tests will also tell you your carrier status of things like Cystic Fibrosis—things you want to know before starting a family," she says.

In early 2012, the company released an Android app titled 23andMe Mobile where consumers who've already done the DNA test can view results. (About 180,000 have taken the test so far.) The app includes a deep dive into the history of each disease a user is susceptible to and links to original research publications related to genomic

results. As Afarian explains, the app has tabs for doctors and information on disease-specific support groups.

## iGenome

For those who want to sequence their entire genome, Illumina is a company that can help. Based in San Diego, its TruSight Individual Genome Sequencing service helps consumers interested in preventative care. The catch: it is only accessible with a doctor's prescription, since it requires a blood test. In addition to preventative care, the test is also useful for physicians looking for specific diagnosis information. Preventative care tests cost $9,500. Medical tests cost $7,500 as part of a subsidy program.

In April, Illumina came out with MyGenome, an iPad app that allows users to interpret their entire genome in a graphical fashion. Dr. Jordan Stockton, director of product marketing, computational biology at Illumina, explains that one doesn't need to have their genome sequenced to use the app.

"The primary purpose of MyGenome is to make people aware of the direction healthcare is going," Stockton says. "It is

meant to start discussions as to what a healthcare system empowered by genomic medicine could look like."

Also in San Diego is Portable Genomics, an early stage software company slated to release a digital product in the next few years that will allow consumers to analyze genomic data. The product, says founder Dr. Patrick Merel, a molecular diagnostic expert, will offer a graphical interface that will direct patients to nearby specialists and provide medical testing reminders. The biggest draw will be the cost: $100.

"I believe that this will be possible by 2015," says Merel.

While he waits for technology to improve and testing prices to go down, Merel is busy working on GeneGroove, a free app (available on iTunes) that takes genomic data (from a 23andMe test) and translates it into music to "get patients comfortable using genomic data" he says.

## Privacy Concerns

Over the years, attention has been paid to a host of privacy concerns around genetic testing, from discriminatory insurance practices to the public sharing of

information. In October, the President's Bioethics Commission released a report addressing the uneasiness around the topic. The report contained recommendations urging federal and state governments to develop processes to protect the privacy of persons interested in retrieving genomic data.

But while the public shows concern for privacy around issue of genome sequencing, experts in the industry, including Merel, don't seem bothered. To him, genomic data will one day be considered similar to having banking information on one's personal computing device.

Green doesn't see privacy as an urgent issue either but does concede that with the rise of genetic data out there, the opportunity increases for it to land in the hands of someone who shouldn't be looking.

"But most of the time you will realize that there isn't stuff in there that you would feel extremely protective about," Green says. "Mostly, it is some health markers, traits and ancestry issues."

Nonetheless, genetic markers are

important clues to your health—even if you never get a disease for which you have a genetic marker. The path ahead for genome sequencing lies in the availability of the technology and the entrepreneurs who will find ways to leverage and optimize the code within.

*This article is commissioned by Qualcomm Incorporated. The views expressed are the author's own.*

Healthcare

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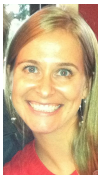

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