Has FDA approval of 23andMe tests put the personal genetics revolution back on track?

“23AndMe Will Decode Your DNA for $1,000. Welcome to the Age of Genomics” ran headine in Wired magazine in November 2007. 23andMe, a Google backed Silicon Valley startup was pioneering the personal genetics revolution by launching its send-me-your-spit-in-a-tube genetic test to consumers. By 2012, the product which had once been selected by Time Magazine as the Invention of the Year reduced its price to $99, a tenth of the original cost and increased the number of health traits it reported on from 14 to close to 250. 23andMe said it was “democratizing” personal genetics, making it possible for everyone to learn about their own genetic makeup.

Soon after however, questions started arising about the validity of the tests and how the results could be interpreted. Not all consumers understood what the genetic risk assessments given in their report meant and some reports suggested that consumers had even taken to YouTube to ask questions like “It’s all so vague; what does [higher risk] even mean?” and “I know [genetic testing] is still in its infancy, but how do I know any of this legitimate?” Several experts questioned the validity of the risk assessments based primarily on genetics alone. Amy Sturm, a genetic counselor at The Ohio State University Wexner Medical Center said that the results provide “a very incomplete view” of risk for a given disease.

Amidst all these concerns, two years ago, the Food and Drug Administration ordered that 23andMe stop marketing and selling its direct to consumer tests which gave a genetic risk assessment for more than 200 health conditions as they were not vetted and validated by the FDA. This ignited a furious debate as many thought that the FDA ruling would hurt the progress medical advances fueled by personal genetics. Noted science writer David Dobbs argued that this was “this is a bone-headed move hostile to public health” and geneticist Razib Khan said the FDA ruling wouldn’t mean much in the long run writing “The data is coming.The institutions designed to protect the public from fraud need to think more about empowerment rather than engaging in *fiat* paternalism.”

This week however, 23andMe announced that it had successfully worked with the FDA to get some of its tests approved and was bringing back the product back on to the market. In its most recent iteration of the direct-to-consumer version however, 23andMe offers a fewer number of tests, all approved by the FDA, and reports the results differently. It also cost twice as much ($199) as it did before.

What do experts think about the new test? And what does this development mean for personal genetics?

‘Carrier Status’ Reports and ‘Wellness’ Reports

The analyses that most concerned the FDA were those that were relevant to the individual’s health such as providing information about the genetic risk of developing a certain disease. And as expected, these were the ones subject to the most significant changes.

Several experts commented on this development to the Genetic Expert News Service, noting the primary difference that 23andMe was no longer offering the tests that most concerned them before. Cecile

Janssens, a Professor of Epidimiology at Emory University said “The company is not testing the diseases that raised most concerns in the past. It does not test predisposition to common diseases such as heart attack, asthma and hip fractures, for which lifestyle factors are often more important; it does not test high risk variants such as BRCA1 and BRCA2 for breast and ovarian cancer and APOE for Alzheimer; and it does not include pharmacogenetic testing.”

Dietrich Stephan, a Professor of Human Genetics at the University of Pittsburgh Medical Center said “[23andMe] no longer are able to communicate medical risk information on common chronic diseases directly to consumers, which is an important distinction with respect to their previous offering.”

Indeed, more than half of the approximately 60 reports that each test result contains are those that reveal ‘Carrier Status’ –whether a person carries a genetic variant that has been connected with a specific condition like Cystic Fibrosis or Sickle Cell Anemia. These carrier status reports are primarily intended to show what genetic predispositions a person has inherited and could pass on to their children. Speaking to the carrier tests, Cecile Janssens said, “the company has substantially improved the information that they provide to customers. For the predisposition tests, they accurately say that DNA is only one of the factors that matter, and that the information should mainly be considered to learn ‘how DNA relates to these traits’.”

However, some questions still remain, according to experts. Brian Zikmund-Fisher at the University of Michigan said that while carrier testing been available to prospective parents through their physician’s offices for many years, “people may not know how to process the information [about carrier mutations] without help. In the 23andMe case, there is a risk that someone will decide to get tested because they are interested in one disease but end up getting potentially disturbing information about another one, one that they perhaps were less prepared to consider.”

This thought was echoed by Dietrich Stephan who said “while FDA has approved carrier tests there still isn’t infrastructure to support the consumption of that information by consumers, and that’s where I have concerns.”

Privacy concerns

Stephan also touched upon privacy and ownership of data, topics that have created a fair amount of controversy when it comes to personal genetic information. “[Consumers’] genetic data and their self-reported survey data are being pushed to internal pharmaceutical development, and may be shared with external pharmaceutical partners outside of 23andMe. It’s legal and above board, but important for consumers to be aware of.”

The questions of who owns the data and what rules govern sharing are still debated. 23andMe asStephan pointed out asks permission from its customers (and gets it more often than not) to use theirinformation anonymously for research purposes. In January this year 23andMe announced that wassharing genetic data from over 650,000 individuals who had given consent with pharmaceutical giantsPfizer and Genetech in order to find new drugs for diseases like Alzheimer's. The deals were said to bethe first in a total of 10 such agreements that 23andMe planned to execute in 2015.

In the meantime, it is also working with the FDA to expand its current offering of tests reports Jullia Belluz of Vox.

> *According to a 23andMe spokesperson, the company is working on getting FDA clearance for other reports but he wouldn't comment on the specifics while discussions are ongoing. This means 23andMe could eventually start offering interpretations on a person's genetic responses to certain medicines, for example, or their risk of developing serious diseases like Alzheimer's.*

23andMe has now established a path forward for other companies to follow in the arena. While many concerns do remain and are being addressed as we speak, it's evident that the personal genetics revolution is back to pushing the boundaries. And it's up to the regulators, genetic counselors, physicians, consumers and everyone else to catch up to it.

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