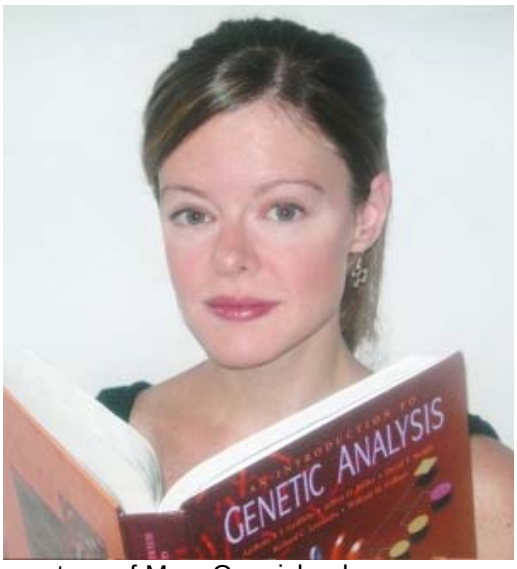
Newsweek

DNA Dilemma: Day Four

Should Genetic Tests Be on the Market?

by Mary Carmichael (/authors/mary-carmichael.html) **August 05, 2010**



courtesy of Mary Carmichael

My five days to decide whether to take a consumer genetic test are almost up, and it's time to examine the issue that inspired me to embark on this project: how the tests should be regulated in the wake of the recent dramatic congressional hearing. Should I be allowed to buy a genetic test without a doctor's permission? Will heavy regulation prevent me from getting one in the future? How is the FDA approaching these issues? I sought the opinions of a genomics lawyer, the executive vice president of the American Society of Human Genetics, and a consultant who follows the industry. I also spoke with two of the highest-ranking FDA officials now charged with regulating the tests. The outside experts provided good context for understanding some of the puzzling things the policymakers had to say. Let's hear from everyone in order:

THE TEST MAY NOT BE HERE TOMORROW
Dan Vorhaus
Attorney, Robinson Bradshaw & Hinson

"DTC [direct to consumer] genetic testing has faced down the specter of heightened regulation before, and over the long term I am confident it will continue to do so. Nevertheless, in the short term it is possible that DTC genetic testing will be subjected to a substantially more restrictive regulatory framework. Will DTC continue unchanged while regulators and companies engage in protracted negotiations? Will oversight weed out the 'snake oil salesmen' and permit legitimate companies to flourish? Or will it drive all genetic testing (temporarily) out of the hands of consumers?

I cannot advise you to take the test or not, but I can say that if you want to proceed there is no time like the present, for there is no guarantee that the option will still be on the table tomorrow."

Read an in-depth version at The Genomics Law Report (http://www.genomicslawreport.com/index.php/2010/08/05/the-past-present-and-future-of-dtc-genetic-testing-regulation/) .

Mary's take: I'm not opposed to regulation. I want to see "snake oil salesmen" put out of business. But Dan's parting comment troubled me, because it suggested I'd better hurry up and decide. If I choose not to take the test,

will I be comfortable with that being a (semi)final decision if the tests go off the market?

(/tag/dna-dilemma.html)

- Should I Take a Genetic Test? (/2010/08/02/dna-dilemma-one-writer-s-week-long-quest-to-determine-if-she-should-take-an-at-home-genetic-test.html)
- What Do Genetic Tests Show? (/2010/08/03/dna-dilemma-day-two-what-can-i-learn-from-at-home-dna-tests.html)
- How Reliable Are At-Home DNA Tests? (http://www.newsweek.com/2010/08/04/dna-dilemma-day-three-how-reliable-are-at-home-dna-tests.html)
- Should Genetic Tests Be On the Market? (/2010/08/05/dna-dilemma-day-four-should-genetic-tests-be-on-the-market.html)
- FAQs (/2010/08/02/dna-dilemma-the-faqs.html)

WHO ARE WE PROTECTING?
Joann Boughman
Executive VP, American Society of Human Genetics

"In our statement on DTC as well as all genetic testing, our first principle is transparency. There are also already two levels of regulation that are not being enforced in genetics the way they are in some other areas. The first is simply that every lab that turns back results needs to be CLIA [Clinical Laboratory Improvement Amendments]-certified. (Most are.) Certification from the College of American Pathologists also sends 'unknown' samples to labs to be tested. These systems are set up to find the few bad actors. That's what regulation is supposed to do. It's not supposed to overwhelm good laboratories.

[The] concept of protecting the public has so many levels. Who are you protecting the public from? Bad actors? Bad tests? Bad physicians? Good physicians who don't know everything in the world? Are we protecting people from themselves? If you want to get a test out of simple curiosity and you want to pay for it, that is an individual's choice. I love people who are curious. As a scientist, I also want people to have good information, interpreted correctly."

Mary's take: Boughman had reasonable suggestions for weeding out "bad guys" while keeping good actors in, and there were already existing systems to put her proposals in place. But what struck me most was her first comment on a theme that has threaded throughout this week's discussions: transparency. I agreed that it would be crucial to make regulation work.

DON'T STIFLE INNOVATION
Jim Prutow
Director at PRTM, a management consultancy with a health-care specialty

"Some of the companies in the GAO report were making clearly misleading statements. That was concerning. But I'm also worried we're going to see a pendulum swing toward much stronger regulatory authority over DTC genomics, and we don't want to stifle innovation.

It's hard for the FDA to justify why they haven't done anything in this market in the last few years. They're in catch-up mode, and they're going to want to be seen as strict. The challenge for them will be balancing the need to act quickly and the need to have a pathway for allowing these tests into the marketplace. Also, how is the FDA going to approve a panel with a hundred different tests? Is it going to go test by test? That's a big burden for both the agency and the companies.

Based on the letters that the FDA has sent, so far, it's not 'cease and desist, don't try to sell any more of this product.' But they could effectively shut down this market for 18 months while they approve or don't approve various panels. And a few of the more concerning tests—those that truly are medically diagnostic information—they might have to take those off."

Mary's take: Prutow's warnings gave me perspective. I was agonizing over whether a genetic test was right for me—imagine the people at the FDA, making that decision on a macro level. The agency would strike the right balance, I thought, only if its thinking on all the issues—the validity of the science, the risk to consumers, the need to balance benefits of regulation with its costs—was clear.

It was time for my interview with Alberto Gutierrez and Elizabeth Mansfield, respectively the FDA's director of the Office of In Vitro Diagnostics in the Center for Devices and Radiological Health and its director for Personalized Medicine. I hoped they could bring some much-needed clarity to the debate. Alas, much of the interview left me more confused about potential regulation than I was before. (Excerpts are presented below, but you can read the full interview (/blogs/the-human-condition/2010/08/05/dna-dilemma-the-full-interview-with-the-fda-on-dtc-genetic-tests.html) here.)

On the day of the interview, I was troubled by a rumor that one of our project's contributors, Daniel MacArthur, had reported earlier that morning (http://scienceblogs.com/geneticfuture/2010/08/will_the_fda_kill_direct-to-co.php) : the FDA might cut off the testing firms' supply of Illumina DNA chips, the technology used to identify gene variants. This would be a subtle but effective way of keeping DTC genetic tests off the market. Without chips, the companies can't test genes. The only long-term alternative to going out of business is to buy other chips, probably abroad, or set up shop in factories in, say, Singapore, which could lead to lower-quality testing (http://www.genomicslawreport.com/index.php/2009/08/26/the-wild-wild-east-of-dtc-genomics-and-the-need-for-meaningful-self-regulation/) . Personally, the rumor was another impetus for me to hurry and decide what I wanted—because once there are no chips left in the supply chain, there's no test for me to take or decide not to take.

I asked if it was true that the agency had "requested" that Illumina stop selling chips to consumer genetics companies. The officials' response made me dizzy. "We have not specifically requested that [Illumina] stop selling [chips] to anybody," said Mansfield. When I asked if that meant the company could continue to supply chips to DTC test makers, she said the FDA was "working with them on that."

Since the chips were marketed for "research use only," I asked if selling those chips to commercial companies violated the FDA's rules. "If they continue to label them that way, yes," she said, "that would be something that we would most likely take a little further action on. At the moment they're working with us, and we will see what they can bring to us in a reasonable amount of time without completely blowing up their business or the market or anything."

I asked one more time if the chips would remain on the market for now. I just wanted a yes or no. But after a pause, Gutierrez said, "I think Illumina needs to figure out how they're going to move forward."

A few hours later, 23andMe issued a formal statement (http://scienceblogs.com/geneticfuture/2010/08/update_statement_from_23andme.php) about the rumor, saying that "23andMe is engaged in an ongoing process with the FDA. 23andMe understands that Illumina is also engaged in such a process. 23andMe has no reason to believe there will be an interruption in the supply of Illumina chips at this time." When asked to respond, the FDA e-mailed me with, "We have not asked Illumina to stop, but if they continue to make these chips available, we could explore other regulatory action." I still couldn't see much difference.

I asked Gutierrez and Mansfield for another clarification during the interview. What, exactly, was the problem with DTC genetic tests: the data they provide or the interpretation of it? For instance, would a company need FDA approval just to give people raw data about their own genomes? Mansfield said as long as companies "don't make any medical claims about that data, then they're free to provide information as far as we're concerned." I asked what constituted a "medical claim." Gutierrez said there was an official definition. At my request, he sent the two relevant policies to me—the FDA has a new and laudable transparency task force (http://www.fda.gov/aboutfda/whatwedo/fdatransparencytaskforce/) devoted to putting out information in a user-friendly format—but he told me they "wouldn't be that easy for people to follow." They were, in fact (http://www.accessdata.fda.gov/scripts/cdrh/cfdocs/cfcfr/CFRSearch.cfm) , impenetrable (http://www.fda.gov/RegulatoryInformation/Legislation/FederalFoodDrugandCosmeticActFDCAct/FDCActChaptersIandIIShortTitleandDefinitions/ucm086297.htm) .

There was a final point I wanted to nail down. Had the FDA thought about the risk Prutow brought up—that overly strong regulation might stifle an innovative (if rough-edged) industry? Gutierrez said that "we don't actually get to make a decision that's based on the economic issues for [a] new device." He confirmed that: "No. No. Our review does not, no, we don't take into account cost." Mansfield agreed: "And, Mary, that cuts agency-wide. That is not considered in any of our reviews." Gutierrez also told me it was "a more complicated question than you actually think."

It *is* a complicated question. There's a policy on the FDA's Web site (http://www.fda.gov/AboutFDA/ReportsManualsForms/Reports/EconomicAnalyses/default.htm) that reads: "The Economics Staff in the Office of Planning conducts economic analyses of all important proposed and final regulations issued by the Food and Drug Administration. Each economic analysis includes an assessment of the costs, benefits, and cost-effectiveness of the action." Gutierrez said the policy didn't apply to DTC genetic tests because what the agency was considering was "neither a rule nor a

regulation." Apparently, applying an existing standard to new devices doesn't count as "proposed and final regulation," so no need to worry about whether it might be economically damaging. I'm still not sure I understand why the policy applies in some cases but not others, or why Mansfield told me that none of the FDA's reviews considers cost when some actually do.

After I hung up, I thought about what I want from government against the backdrop of what I've learned this week. DTC testing companies are often called out for explaining risk badly, for lacking transparency, and for making vague and potentially misleading statements. Meanwhile, the companies' critics are accused of paternalism (http://scienceblogs.com/geneticfuture/paternalism/) , of telling people that some topics aren't that easy to follow, of decreeing that the issues are, well, more complicated questions than you actually think.

If the FDA is going to rope in varying and vocal people and define standards for the industry, maybe it should set a better example on those points itself.

Tomorrow: To Test or Not to Test? I Make My Decision

Follow Mary on Twitter (http://twitter.com/mary_carmichael) as she makes her decision, or add your opinion using the #DNADilemma hashtag.

DNA Dilemma: The Complete Series

Monday: Is Now the Time to Test? (http://www.newsweek.com/2010/08/02/dna-dilemma-one-writer-s-week-long-quest-to-determine-if-she-should-take-an-at-home-genetic-test.html)
Tuesday: What Can I Learn From This Test? (http://www.newsweek.com/2010/08/03/dna-dilemma-day-two-what-can-i-learn-from-at-home-dna-tests.html)
Wednesday: How Meaningful Are the Results? (http://www.newsweek.com/2010/08/04/dna-dilemma-day-three-how-reliable-are-at-home-dna-tests.html)
Thursday: Should These Tests Be on the Market? (http://www.newsweek.com/2010/08/05/dna-dilemma-day-four-should-genetic-tests-be-on-the-market.html)
Friday: My Genome, Myself: I Make My Decision
Recommended Reading: Further Articles, Essays, and Web Sites
DNA Dilemma FAQs: You're Doing What, Exactly? (http://www.newsweek.com/2010/08/02/dna-dilemma-the-faqs.html)