Clearing a Path for DTC Oversight

by Dan Vorhaus

In a few hours, the FDA will kick off a two-day public meeting to consider the future of clinical direct-to-consumer (DTC) genetic tests. Few corners of the personal genomics landscape have generated as much attention from regulators, consumers and, especially, the media as DTC genetic testing. Thus, when the meeting was first announced last month, we applauded the FDA's attempt to examine DTC's unique set of issues separate from other larger and ongoing regulatory conversations, including whether and how to regulate the far more numerous category of laboratory developed tests (LDTs).

So just what should we expect from the next two-days? 2010 saw a flurry of DTC-related regulatory and legislative activity but, ultimately, little in the way of new oversight or concrete guidance. Both regulators (including the FDA) and industry appear to have responded in 2011 with a more measured approach, and this week's meeting is an opportunity to thoroughly examine the state of DTC genetic testing and develop a clear, sensible strategy for future oversight of the industry.

Over at *Genetic Future*, Daniel MacArthur has already weighed in, adopting a tone of cautious optimism in advance of the DTC meeting. Meanwhile, with just a few hours left until the meeting kicks off, here are three key points I'll be emphasizing in my own talk tomorrow morning (slides):

1. What does "DTC" mean? Direct-to-consumer, direct access, over-the-counter, home use...the terminology is exhausting and rarely used consistently. A key goal for the next two days will be to establish, as clearly as possible, precisely what "direct-to-consumer genetic testing" means, as well as which particular varieties of DTC genetic testing the FDA is interested in overseeing. For instance, in the Executive Summary provided in advance of the meeting (pdf), the FDA indicated that it is:

...focused specifically on issues regarding clinical genetic tests that are marketed directly to consumers (DTC clinical genetic tests), where a consumer can order tests and receive test results without the involvement of a clinician.

That's helpful, because it identifies specific types of DTC genetic testing (e.g., clinical tests marketed to, and ordered by, consumers, without physician involvement) that concern the FDA and, by extension, other types of DTC genetic testing that, for now, appear to be in the clear (e.g., non-clinical tests). Still greater specificity is possible, however, and will be critical to developing clear guidance.

2. Planning for the DTC of tomorrow. To develop an appropriate system of oversight for DTC genetic testing, we must consider the DTC genetic testing industry not only as it exists today *but also* as it is nearly certain to exist tomorrow. With the rapidly declining cost of whole-genome sequencing (WGS) it is only a matter of time before we transition from SNP-based DTC genetic testing to WGS-based DTC genomic interpretation. This transition won't happen in 2011, but it *will* happen, and it will happen soon enough that any DTC oversight developed today must be designed with WGS firmly in mind.

DTC's inevitable shift from comparatively limited genotyping to whole-genome sequencing, just as it did in clinical and research genetics, will carry with it numerous important changes. First, it will obliterate the increasingly tenuous clinical/non-clinical distinction at the level of the DTC test, with DTC providers offering both clinical and non-clinical data (along with copious amounts of data of uncertain significance) as an unavoidable feature of every whole-genome sequence.

Second, it will accelerate a trend already in place today: the separation of testing (data generation) from interpretation (data analysis). Two and a half years ago, DTC spit parties graced the Fashion & Style section of *The New York Times*. Two and a half years from now, odds are that consumers at the forefront of DTC will have long ago mailed in their last "spit kit" and, with whole-genome sequence in (virtual) hand, will have a bevy of software-based DTC interpretative services from which to choose.

3. Coordination and Leadership. Finally, while an important part of the personal genomics landscape, clinical DTC genetic testing is still only *part* of that landscape. Close coordination with other federal agencies (e.g., CMS and FTC), as well as with states and international bodies, will be necessary to ensure not only meaningful oversight of clinical DTC genetic testing, but also the development of a comprehensive, coherent system of oversight for personal genomics and personalized medicine. An important part of the "path to personalized medicine" (outlined by FDA Commissioner Hamburg and NIH Director Collins last summer) will be close coordination among agencies and other forms of oversight, not mere legislative or regulatory Band-Aids.

It will also require leadership. For the second decade of human genomics to be as groundbreaking as the first, we need strong and even visionary leaders at the federal level who are focused on addressing the policy, as well as legal and social challenges that threaten to serve as barriers to scientific and medical progress. Those are not shoes the FDA will be able to fill on its own, but the FDA should be encouraged to participate fully in that process.

I will likely have more to say as the meeting progresses (you can follow along on Twitter at #FDADTC) and ultimately concludes. In the meantime, feel free to share your thoughts about what you would like to see from Washington over the next two days in the comments below.