Direct-to-Consumer Testing 2.0: Emerging Models of Direct-to-Consumer Genetic Testing

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CME Activity

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Learning Objectives: On completion of this article, you should be able to (1) evaluate some ethical challenges in the direct consumer access to genetic testing; (2) summarize ongoing changes to the regulation of direct-to-consumer genetic testing in the United States; and (3) predict possible consequences of expanded access to direct-to-consumer genetic testing of clinical practice.

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Abstract

Direct-to-consumer (DTC) genetic testing emerged in the early 2000s as a means of allowing consumers to access information on their genetics without the involvement of a physician. Although early models of DTC were popular with consumers, they were controversial in medical and regulatory circles. In this article, we trace the history of DTC genetic testing, discuss its regulatory implications, and describe the emergence of a new hybrid model we call DTC 2.0.

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A Spit Party is kind of like a Tupperware Party, only the plastic containers are smaller and they're not for leftovers.
23andMe[1]

In December of 2007, an early direct-to-consumer (DTC) genetic testing company celebrated its launch with a Spit Party in which attendees danced, drank, and submitted DNA samples for sequencing. Within weeks, partygoers would have access to a comprehensive report, including their genetic preference for vegetables, whether their tongue curled, and their risk of developing breast cancer. In Silicon Valley, at the height of the dotcom boom, 23andMe was at the vanguard of a wave of interest in personal genomics. Competitors deCODE and Navigenics

also offered genome panels, whereas companies such as Ancestry.com offered to interpret people's DNA and trace their ethnic ancestry.

As the name suggests, DTC genetic testing companies offer genetic tests independent of a physician. Some tests include nonmedical "infotainment" such as ear lobe attachment or the propensity to flush when drinking alcohol. In the past, however, these tests were bundled with risk factors for complex diseases such as type 2 diabetes and osteoporosis or monogenic disease such as *BRCA1* and *BRCA2* for breast and ovarian cancers.[2] Although these genetic analyses had been technically possible for years, the cost of testing was financially prohibitive. In 2007, the cost of a DTC panel hovered around $1000.[3-5] Three years later it dropped to between $300 and $400. By 2012, it dropped to $99 and 23andMe announced their goal of collecting 1 million users.[6]

The users of DTC test products reported that viewing personal genetic risks made them think more carefully about diet and exercise.[7] Online tools allowed users to track the contents of their genome and compare it with that of others. Some products allowed users to conduct a "family search" of the database to determine whether other users may be relatives. "Our DNA is a fascinating aspect of who we are, and we feel strongly that anyone who wants their genetic data should be able to get access to it," the authors of the 23andMe blog posted.[8] Some scientific sources agreed. The journal *Science* named human genetic variation the "breakthrough of the year" and highlighted 23andMe in its coverage. "The best outcomes [of DTC genetics]," wrote the editorial board of *Nature Genetics*, "would be to convert patients into active investigators and navigators of their own health, to make genetics the foundation of medical education [,] and to expand the scope of genetic counseling as a profession."[9]

Fast forward 5 years to 2012, when most DTC companies offering medical information in the United States had gone out of business (although DNA-based ancestry testing remained commercially available). Only one of the early pioneers in this sector remained, and the US Food and Drug Administration (FDA) had temporarily barred it from selling medical information panels. Nevertheless, there was a movement toward a new form of consumer-initiated genetic testing, what we call DTC 2.0.

Fast forward again to 2017, when the FDA has authorized the first DTC test as an approved medical device. Energized by this regulatory development, other companies are actively working to obtain similar approvals. In this article, we review the history of DTC genetic testing products in the United States from an ethical and regulatory perspective. Although the status of these products remains in flux, we will attempt to characterize DTC 2.0 and its potential implications. Direct-to-consumer 2.0 represents a new model of disseminating, using, and interacting with genetic health data, a model that has the potential to be either transformative or disruptive, depending on how key ethical and regulatory challenges are addressed.

RISE AND FALL OF DTC 1.0

The initial rise of the DTC model of genetic testing was, at least partially, a reaction to traditional health care models of providing genetic testing. The medical model is characterized by a dependence on expert knowledge and the structural elements of the health care system. A medical professional, operating within a fiduciary patient-provider relationship, orders clinically indicated genetic testing and licensed, board-certified medical genetic providers interpret and deliver results. The health care system is the mediator of genetic information, responsible for its quality, creation, interpretation, delivery, protection, and implications. In particular, the medical model is committed to protecting the privacy of health information, including genetic information. Patients retain the ability to control which organizations have access to their data. Like all medical care, the model is designed to maximize patient benefit and promote informed clinical decision making while minimizing associated risks.

This medical model has its drawbacks. Private sector actors frequently complain that it innovates too slowly because of regulation and professional resistance to new practices. Some have argued that resistance to new medical decision-making technologies stems from the desire of medical actors to preserve professional autonomy.[10] There is also a lack

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