

The Ethics of Direct-to-consumer Testing

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Introduction to DTC:

With the advent of personal genomic companies like “23andMe” and “everygenome.com,” it has become more commonplace to have individual genomes sequenced for as little as \$300. With this powerful genomic information at hand, consumers (now more than ever) worry what will happen as a result of this information; will we *truly* feel more empowered?

A key component of this debate over personal genomics is the growth of direct-to-consumer (DTC) testing, which is currently a three billion dollar industry offering over 1100 various tests to detect risk and diagnosis of everything from depression to breast cancer. DTC testing is distinct from genetic testing; genetic testing is the analysis of DNA, RNA, protein, or metabolites to diagnose a heritable human disease, predict the course of the disease, or guide treatment decisions. These tests are usually administered by a certified physician, and include both pre-test and post-test counseling. Common examples of this test include the karyotype testing for Down’s syndrome for embryos. DTC testing, however, are tests that are directly sold, delivered, and used by the consumer without any intervention by the healthcare provider. Many of these tests are available to purchase via the Internet, and are heavily marketed via the use of pop-up ads, etc. An example of this test includes the test for the BRCA1 and BRCA2 genes, both which are implicated in breast cancer. While proponents argue that these tests increase patient self-empowerment, many opponents argue that patients are taking tests of poor quality, and lack the adequate support before and after taking the test. Due to this and other reasons, the Food and Drug Administration (FDA) and the Federal Trade Commission (FTC) should further regulate the DTC industry due to the risk posed to the consumer (both practical and ethical), as well as public health concerns (Hogarth et al. 2008).

DTC Harm to Consumer:

DTC tests pose much harm to consumers. First, many DTC tests are of dubious quality, and many are not even clinically validated. A study of over 127 tests showed that only three percent of the tests were supported for the general public as screening tools; further, even if the tests were targeting a specific testing group, only fifteen percent of the tests had been supported to test within a specific community. For 23% of the tests, there was insufficient evidence as to the validity of the tests, and there was no guidance for the other half of the tests (Lovett et al. 2012). Further, recent studies on CYP450 (test that helps guide selection of antidepressant) has shown that there is a lack of support for the clinical validity of this test (Udesky 2010). These results, in conjunction with previous observational studies, shows that only very few of these tests are qualified to be used as possible screening tools in patients.

Second, there is no system of pre-test or post-test counseling. With genetic tests, there is a physician that guides the patient about what the test means, what their results means, etc. However, since physicians are removed from the picture in the DTC business model, there is lack of patient counseling. For example, over a survey of 127 DTC tests, 95% lack any sort of counseling for the patient (Lovett et al. 2012). This can be very traumatic for a patient who has received a positive test result on the BRCA1 DTC, and doesn't understand that breast cancer is a multifactorial disease with multiple factors playing a role in its causation. Also, even if the test shows up with a positive result for a mutation within the BRCA gene, it could be a mutation of uncertain significance. This means that the mutation could be benign or carcinogenic; currently, there isn't enough data to determine which case patients fall into. However, most patients are unaware of this, and can thus lead to unnecessary worries. Therefore, it can lead to more harm than good for the patient (Udesky 2010).

Third, due to the misleading nature of DTC tests, patients often assume that if they test positive for a gene that plays a role in a disease, that they will develop that disease. For example, many DTC's claim to "screen" for depression, but they are only

checking for one gene involved in causing depression. Depression is a multifactorial disease with genetics and environment playing a key role in causing the disease. With the patient misled about their ability to get the disease (and confusing this with their probability of getting the disease), this can lead to distress for the patient, which is the exact opposite of what the patient expected from the test (Udesky 2010).

Fourth, the labs that perform the actual sequencing (and thus serve as the intermediate between the DTC testing company and the consumer) are of dubious quality. Under the Clinical Laboratory Improvement Amendments (CLIA), labs that perform sequencing are required to be certified, but there is no widespread regulation of this rule. Further, when consumers purchase the medical test, they are not told whether the lab that takes their private genetic information is CLIA-certified or not (certification is done by the Center for Medicare and Medicaid Services). Additionally, none of the tests performed by the lab are certified, meaning that there is no proper protocol (and lack of regulation of proper protocol) within these labs, leading to poor quality of test results (Udesky 2010).

Finally, many DTC-testing companies use misleading advertisements to get consumers to buy their product. For example, MYRIAD, a DTC-testing company, ran an advertisement campaign encouraging women to take the BRACAnalysis test if they felt they were predisposed to breast and ovarian cancer. In Denver, one of the cities in which the campaign ran, there was a 300% increase in calls for women interested in BRACAnalysis, yet 30% decrease in referral for high-risk women for breast and ovarian cancer. Further studies showed that the ads didn't accurately portray the test's ability to predict cancer or to reach out to their physician to discuss whether they should have this test ("Direct to). Another example is the advertisement for the Reward Deficiency Syndrome (RDS) gene test; the DTC company that sold this test ran the following ad campaign:

“Are you compulsive? Have you ever wondered why you crave certain things and/or act in an irrational manner? Would you like to know if you have the genetic predisposition to abuse drugs and alcohol? Are you concerned about your children’s future? Does your child have the genetic trait that leads to disruptive and addictive personalities? DNA testing can help you understand and manage a child’s behavior before it gets out of control. Imagen will test a panel of dopaminergic related Reward Deficiency Syndrome (RDS) genes. This will allow you to know if there is a genetic predisposition towards any of the associated addictions. The Reward product line is then available to treat the genetic predisposition towards RDS.” (Berg et al. 2007)

This ad uses a lot of complicated terminology to make it sound like it predicts whether a patient becomes a drug/alcohol abuser, when in reality it checks for one condition for a multifactor disease. Further, it provides no evidence for the clinical validity of the test, or encourages patients to talk to their doctors about the test before purchasing one.

DTC Ethics and Public Health Concerns:

There are several ethical and public health concerns with DTC testing. First, DTC promotes a mindset of “genetic determinism,” the belief that one’s fate is determined solely by their genetic makeup. Advertisements promote this idea that testing positive for one gene means that you will get this disease, or have a predisposition for a certain disease. However, there is more to genetics than just the genome; with the rise of epigenetics (study of heritable change in mechanisms other than the DNA sequence), we are beginning to realize the effect of other factors in phenotypic expressions of genes (Berg et al. 2007).

Second, this emphasis on genetic determinism undermines public health initiatives by diverting scarce health resources away from research into environmental and social risks associated with this disease, and into genetic screening and interventions.

This prevents patients and physicians from obtaining a clearer picture about the disease. Further, the prominence of using genetics to predict disease outcome unnecessarily increases the amount by which genetic variation is turned into a medical syndrome (i.e. unnecessarily pathologized). Coming up with these excess tests might not help, and may end up overwhelming the patient; thus the test ends up doing the opposite of what it is supposed to do (Samuel et al. 2010).

Finally, access to this genetic information might lead to genetic discrimination. If the information is not properly secured, insurance companies and employers can discriminate by knowing genetic information. Even though there are legal mechanisms to prevent this from happening (i.e. Genetic Information Nondiscrimination Act or GINA), these prevent the employers/insurance companies from discriminating based “solely” on genetic predisposition. However, employers/companies can claim to discriminate against someone based on genetic predisposition and other factors, and can thus get around GINA via this loophole (Samuel et al. 2010).

Counterarguments:

Proponents of DTC often argue that increased access to genetic information is good for the patient because it empowers them to take charge of their health. However, if the information itself is of dubious quality (as shown above), then there is no actual “empowerment” of the patient. For example, if the patient worries about an incorrect diagnosis, or gets a false positive, then there is no use for that patient to have that piece of information (Berg et al. 2007). Further, there is no clear delineation in the DTC test advertisement for who may/may not benefit from the tests, which leaves the consumer in a quagmire of excess information without any way to filter who needs to take this test. This might discourage the consumer for taking charge of their health since they feel that there’s just too much ground to cover, which is the opposite for what the test wants people to feel (Lovett et al. 2012).

Another major argument used by proponents is that once patients know their genetic information, they have greater control over who gets access to it, thus increasing patient privacy over their genetic information. However, most DTC companies don't even disclose their privacy policy, which makes it hard to understand what the company will do with the consumer's genetic information. Also, since DTC companies outsource their sequencing to external labs, there is no guarantee that these labs have the adequate security system to properly protect people's genetic information. Additionally, the DTC industry is not subject to the Health Insurance Portability and Accountability Act (HIPAA), which means that they are not required to protect patient's private information. Finally, if the patient decides to discuss their test results with the doctor (which they inevitably will if they get a positive test result) then that information becomes part of their medical record, and employers/insurance companies have access to it (Hudson et al. 2007).

Reform:

There are many ways by which the DTC industry can be reformed. First, there needs to be more FDA regulation of the industry. The FDA should implement laws that require DTC companies to limit DTC tests as public health tools with organized target audiences and protocols for performing the test. Further, they should require DTC companies to disclose risks, scientific evidence, sensitivity and specificity of the tests, predictive value of tests, CLIA certification status of the labs, and privacy policy. This level of transparency will ensure that even if the test is of dubious quality, the consumer can make an informed choice. The FDA should also require the DTC companies to offer (or at least encourage) pre-test and post-test counseling, so the patient makes an informed decision about using the test and their results. Also, they should educate physicians about DTC tests, since only about 44% physicians (on average) are aware of DTC's. If physicians don't know about DTC's, then they can't give their patients adequate advice

whether they should take the tests (Hogarth et al. 2008). Further, the FDA should require the CMS not only to regulate the labs that provide these tests, but also the tests themselves. As shown previously, many of these tests are of dubious quality, and there isn't enough scientific evidence to validate their necessity. Thus, some sort of rating would be helpful to consumers in making their decision to purchase this test (Udesky 2010).

Finally, there needs to be increased FTC regulation of the industry. The FTC is responsible for protecting consumers and competition. As shown previously, most advertisements run by DTC companies mislead the consumers about test validity, quality, and security. Thus, the FTC should intervene and ensure that the advertisements run by the DTC companies (both on and off the internet) should not be misleading. If the tests lack scientific validity, then this should be very explicit in the advertisement and especially on the website through which these companies sell their product (Hogarth et al. 2008).

Conclusion:

In conclusion, the DTC industry poses a risk to the consumer due to its unethical nature as well as its tendency to undermine public health initiatives. However, the solution to this problem isn't to eliminate the DTC industry, but to increase FDA and FTC regulation to ensure that consumers can make informed choice to purchase DTC tests. This can greatly impact the greater overall efficiency of the healthcare industry via patients making informed decisions to predict, diagnose, and understand a possible inherited disease, and thus make better treatment decisions.

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