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The Ethical and Social Consequences of Direct-to-Consumer Genetic Testing

Abstract

Genetic testing has been an effective way for individuals to attain information regarding their predisposition to particular conditions. This growing industry has recently incorporated a new aspect that allows consumers to receive genetic test results without the inclusion of a medical professional. In this process, termed Direct-to-Consumer (DTC) genetic testing, an individual sends a sample of saliva or blood to a company and receives genetic test results for multiple conditions and diseases; sometimes, recommendations for the consumer are provided along with the results. Advocates for the process deem that it creates autonomy and allows individuals direct access to medical information. However, many opponents propose that this novel form of genetic testing forgoes all aspects of traditional medicine including autonomy, beneficence, maleficence, and justice. This paper addresses the concerns about these principles and the ethical and social implications of DTC genetic testing. Research indicates that the defects within the testing process show that the principles of medicine need to be further imposed in the industry.

Josh was in his forties and worked for a large, private medical research company. When his employer decided to start a study on the effect of genetic predisposition testing on behavior, he willingly chose to participate because he saw “no harm in it.” When he submitted his sample to the genetic testing company, Josh was slightly worried about possibly having diabetes mellitus. Surprisingly, when Josh received his results several weeks later, they did not show a link to diabetes but rather a predisposition to Alzheimer’s disease. The consequences of these test results impacted Josh’s life more than he could have ever imagined (Messner 5).

Genetic testing is a growing phenomenon in the United States and, at a smaller rate, around the world. These genetic tests depend on the power of the internet, the possibility of large amounts of profit, and the growing desire for consumers who want self-created healthcare. With this in mind, various companies are beginning to offer genetic testing results directly to consumers. In this process, a consumer writes a check, ranging from about \$1,000 to \$2,500, and sends his or her saliva through the mail. Then, several weeks later, the consumer receives the

results for a multitude of genetic conditions (Pray 1). The procedure ultimately forgoes an interaction between a patient and physician or genetic counselor, but instead directly targets consumers. DTC testing is neither definite nor accurate and may provide individuals with false information that may lead to unnecessary treatments (Hogarth, Javitt, and Melzer 161). In most cases, individuals do not receive accurate interpretation of their results from companies, so the consequences are severely detrimental (Hawkins and Ho 2). Ethicists question the impact that these results and the regulations within this consumer-driven industry may have on purchasers of the product.

Alice K. Hawkins, a Doctoral Candidate at the University of British Columbia, and Dr. Anita Ho, the director of Ethics Services at Providence Health Care, co-wrote the journal article “Genetic Counseling and the Ethical Issues around Direct to Consumer Genetic Testing.” This piece presents a theoretical argument regarding the ethics of DTC genetic testing called “The Ethic of Care.” Hawkins and Ho determine that DTC testing poses a detriment to consumers because it directly contradicts four basic principles within the field of medicine: autonomy, beneficence, maleficence, and justice (2). Their discussion questions the beneficial effects of DTC genetic testing, if there even are any advantages. Their theories lead to the question of what are the ethical and social implications of DTC genetic testing? It seems that the regulation of the laboratories that conduct these tests and the companies that manage the procedures is insufficient because there are many faults in the administrative system. Due to the defects within the current standards of DTC testing and its various dangers toward the wellbeing of consumers, it seems that the principles of the Ethic of Care need to be implemented into the system by imposing new laws.

Direct-to-consumer genetic testing has been gaining prominence over the past several years. Currently, DTC genetic testing is allowed without limitation in about half of the states in the United States, and all states can be subject to DTC testing inspection at a federal level (Genetics and Public Policy Center). In July 2006, the Government Accountability Office (GAO) issued a report that showed peculiar marketing practices by some DTC testing companies. Consequently, the Federal Trade Commission (FTC) issued a consumer alert advising “consumers to be skeptical about claims made by some DTC companies” (“Federal Trade Commission”). The DTC tests include tests for single-gene disorders, such as cystic fibrosis, and predisposition to multifactorial diseases, such as cardiovascular disease and depression. In addition to the results of the genetic tests, some companies make suggestions regarding lifestyle changes, such as modifications in diet or use of nutritional supplements (Hudson, Javitt, Burke, and Byers 635). Today, there are more than 1,100 genetic tests available, and many of those tests are being offered directly to consumers without any involvement of health care providers or counselors (Hudson and Javitt 59).

This paper focuses on DTC genetic testing that is strictly based on a consumer-company relationship, without the involvement of a healthcare professional as a liaison. Sometimes, “DTC” refers to tests advertised but not sold directly to consumers, but those tests will not be discussed in this paper. Additionally, while DTC testing also includes paternity and ancestral testing, this research paper strictly addresses the genetic tests that make health-related assertions or directly affect healthcare decision-making (Hudson, Javitt, Burke, and Byers 1). Also, for the purposes of this research paper, genetic test refers to an examination of human DNA, RNA and protein(s) in order to determine or predict an inheritable condition, suggest treatment or predict the occurrence or recurrence of a condition (Hogarth, Javitt, and Melzer 163).

One of the main principles within genetic medicine is minimizing harm—non-maleficence—and promoting the well-being—beneficence—of patients (Hawkins and Ho 2). Under the protocol in which DTC testing is currently performed, non-maleficence and beneficence are not used as main principles. Companies do not assure that consumers’ best interests are taken into account when conducting tests because they are only concerned about their own financial well-being. DTC testing actually has the adverse effect of enhancing harm and does not take into account the well-being of consumers.

As Hawkins and Ho note, DTC genetic testing “challenges [the care] ethic specifically because...[i]f individuals undergoing genetic testing are consumers (rather than patients or clients), a ‘buyers-beware’ attitude may ensue. As such, the importance of the therapeutic relationship is worryingly diminished” (Hawkins and Ho 3). Clearly, the relationship within a patient and a qualified professional is lost. DTC genetic testing does not create a patient-physician relationship, but rather a buyer-seller relationship. As a result, companies are not seeking the best interests of consumers as patients or healthcare recipients. The companies fail to care for consumers after they provide them with life-altering information.

Normally, in a professional setting, genetic tests would require the involvement of genetic counselors in order to oversee the physical and psychological welfare of the patient and to provide medical interpretation of the results for the patient. Without these normal guidelines within DTC testing, the consumer is left with the results of a test that they probably do not understand. A recent study in Public Health Genomics reported that “although members of the general public are likely to think that DTC genetic test results are easy to understand, they may not interpret them correctly” (“Interpreting Risk”). According to this study, it seems that the presence of a genetic counselor would allow an individual to accurately interpret results in order

to make the procedure more useful to the consumer. Appropriate interpretation of results by a qualified genetic counselor is crucial in order to minimize harm and maximize the potential benefits of the procedure (Hawkins and Ho 2). For example, Dr. Shannon Behrman, who has a Ph.D. in molecular biology and is a health communications fellow at the National Cancer Institute's Office of Cancer Genomics, noted the likelihood of an individual misinterpreting results: "If I look at the results for my melanoma risk, it shows I have a twofold elevated risk of 2.4 percent. For the average person, when they see these numbers, they may just think, 'that's a twofold risk!' But people receiving such a result should note that even though the risk may be double the average person's risk, it is still, at 2.4 percent, a fairly small risk" ("Interpreting Risk"). Clearly, the advice and knowledge from a medical professional greatly enhances the accurate analysis of genetic testing results.

The process of DTC genetic testing undermines the ideal of beneficence and non-maleficence because companies are not providing consumers with medical information about the various conditions in the DTC tests; therefore, the consumers do not have the proper knowledge to accurately interpret results and know how to adequately use the information afterwards. Often in DTC testing, the patient is given misleading reassurance based on falsely negative results. Essentially, the consumer is given false results that state that he or she does not have a genetic condition because the tests are inaccurate. On the other hand, a consumer might get unnecessarily anxious due to false positive results for a disease. DTC companies ultimately promote unnecessary frustration and false relief within consumers (Favaloro, Lippi, and Plebani 1225).

These false results are most prevalent in multigenic gene disorders for which there is the most risk of being misdiagnosed. In this case, a result of a genotype for a specific condition does

not always correlate with a matching phenotype, or physical trait. As a consequence, a patient will receive treatment for a condition he or she does not have; this can be very detrimental to the individual's health because treatment involves diet change or large amounts of medication. These dramatic changes in an individual's lifestyle often lead to health complications for a healthy person (Favaloro, Lippi, and Plebani 1225). Clearly, it is unfair to give inaccurate results to a patient for a condition that will potentially and dramatically impact the individual's life. This situation actually enhances the harm for the consumer.

For example, cytochrome P450 (*CYP450*) is a diverse group of genes that is tested using DTC genetic testing. A presence of this set of genes suggests an effective type of antidepressant an individual should utilize; this may be harmful to the patient if they take the results into his or her own hands. In a typical situation, a patient who is informed of his or her *CYP450* profile might decide to change his or her dose of antidepressant medication. Leslie Pray, an independent science writer and editor, notes that “[a]ntidepressants have been associated with suicidal thoughts and suicide attempts, among other adverse effects, and physicians are warned to carefully monitor their patients, particularly when adjusting dosage, which may prove difficult with increased access to DTC testing” (1). Obviously, this situation is detrimental to the consumer and will happen on a grander scale with the growing prominence of DTC testing.

Some may argue that DTC testing promotes beneficence and non-maleficence in that it allows a person to make better health and lifestyle decisions. However, this statement is quite questionable due to the current nature of transparency regarding DTC testing. A common concern relating to the DTC industry is “the fidelity of the marketing of DTC genetic tests” (Hawkins and Ho 3). Commonly, the advertising tactics for the testing greatly exaggerates the importance of the information. Timothy Caulfield, the Research Director of Health Law at the

University of Alberta, conducted research that indicates that many advertising techniques are “unfairly grandiose” and “from a health perspective... [the] genetic information is not very useful” (4). The DTC companies continuously promote themselves as a helpful service for consumers in order to dramatically change their lifestyles for their own good. Essentially, the companies are promoting harm because consumers do not necessarily need to know about the results of this test because it may cause unnecessary effects, such as anxiety and depression, and exacerbate the onset of a particular condition.

In the case of Claire, a journalist in her fifties, she experienced bouts of depression after receiving DTC genetic tests that indicated her predisposition to Alzheimer’s disease. She described her thoughts after receiving the results: “It’s like somebody is handing you a death sentence. It’s awful. I mean you just feel like you’re going to die. This is your end result... I’m actually a really happy person most of the time. But there’s this . . . depressive presence over me every day” (Messner 4). Initially, after reading several DTC advertisements, Claire thought that the genetic test results would be very helpful. Unfortunately, afterwards, Claire did not receive genetic counseling, and she was left with feelings of depression. This is an example of one of the many detrimental consequences that the advertisements and the results of DTC genetic testing can have on the psychological wellbeing of consumers.

Furthermore, the Federal Trade Commission (FTC) Act deems unlawful “‘unfair or deceptive acts or practices in or affecting commerce’ and directs the Commission to prevent such activities” (Hogarth, Javitt, and Melzer 174). Currently, DTC companies do not abide by this law because it is not strictly enforced; therefore, they are committing illegal acts.

For example, “some companies claim that at-home genetic tests can measure the risk of developing a particular disease, like heart disease, diabetes, cancer, or Alzheimer’s disease. But

the FDA [The Food and Drug Administration] and CDC [The Centers for Disease Control and Prevention] say they aren't aware of any valid studies that prove these tests give accurate results" ("Federal Trade Commission"). Many companies make promising claims about the benefits of their genetic tests despite the primitive research which forms the basis of most results. Many times, having a specific gene does not guarantee that an individual will develop a particular disease. Also, most conditions involve the interaction of multiple genes and even this does not definitely mean that an individual will or will not have a disease. In addition, many conditions are influenced by various environmental factors and physical alterations in chromosome structure that are not accounted for in the DTC genetic test results (Flaker and Mazza 8). Although many times companies claim to reveal pertinent information about an individual's predisposition to certain conditions, this statement cannot always be made because many conditions require extensive analysis of genes that DTC companies do not provide ("Federal Trade Commission").

Therefore, when DTC genetic testing companies, such as 23andMe, make claims on their websites about their services such as "[u]nlock the secrets of your own DNA' and 'shed new light on yourself," they are using false advertising techniques (Pray 1). The descriptions used on their websites do not indicate the implications that genetic test results may have on an individual. The "new light" that is shed on an individual is not necessarily accurate and may not have any significance. If the results are not definite, it is useless for an individual to know about them because the results are not accurately describing the consumer ("Federal Trade Commission"). In general, advertisements make claims about the effectiveness of their services, but do not emphasize the risks; there is not a fair balance of information delivered to consumers when comparing efficacy to risk. Moreover, the way that information is presented may compromise the

intended message because when large amounts of information are given this might result in confusion and/or misinterpretation. Charlotte E. Sibley, the director of the Global Healthcare Businesswomen's Association (HBA) and a member of the Board of Directors of American Pacific Corporation (AMPAC), notes that “consumers have reported that they do not have sufficient time to study information in certain types of broadcast advertising, and thus important risk information may not be fully communicated” (qtd. in Sheehan 161). Essentially, the advertisements do not allow consumers to retain all the information that is presented, so they cannot make informed decisions.

Additionally, DTC genetic testing companies make false claims about the accuracy of their tests. A class action lawsuit was filed in February 2006 against Acu-Gen Biolabs for its Baby Gender Mentor test. The Massachusetts company was issued a lawsuit, on behalf of women who used their service and were dissatisfied with the incorrect results about their fetuses. “The lawsuit...focused on the company’s failure to honor its ‘200% money back guarantee’ and its claim of ‘99.9% accuracy.’ In addition, the lawsuit claimed that the company provided incorrect medical advice to women about the health of their fetuses, which caused them emotional distress and led them to undergo unnecessary testing” (Hogarth, Javitt, and Melzer 176). Clearly, this company made inaccurate claims about their services and this is an issue of great importance within the DTC genetic testing industry. Due to the use of advertising techniques, genetic testing companies do not inform consumers of the inaccuracies of their business because ultimately, their intent is to sell a product. The genetic testing companies are involved in a service that makes profit from selling genetic tests, so that is their main priority.

Autonomy is the state of being independent and making decisions as an individual. Dr. Raanan Gillon, chairman of the Institute of Medical Ethics and a member of the British Medical

Association's Medical Ethics Committee and International Committee, states that this concept also involves the practice of respecting a person's "intrinsic right and capacity to think, make decisions and take actions based on their values and beliefs" (qtd. in Hawkins and Ho 3). The idea of respecting an individual's autonomy is another fundamental principle within genetic counseling.

Proponents of DTC testing argue that their practices encourage autonomy because they make genetic information more directly accessible to the public, therefore promoting individual decision-making. Although a patient may be making a decision by his or herself, the decision cannot be fully described as autonomous until the individual fully understands the process of DTC genetic testing. Respecting autonomy in genetic counseling involves providing an individual with the information and tools necessary to make a decision (Fraker and Mazza 34). This also includes helping the individual understand the various options and consequences from his or her decision, and providing a supportive environment for questions and concerns (Hawkins and Ho 4).

Unfortunately, autonomy is never sufficiently utilized in the process of DTC genetic testing because the information that companies supply cannot fully address the necessary issues. Also, the basis of the information relies on advertising tactics that focus on luring consumers to buy a product. For example, "[w]hile writing [her] article, [Messner] searched Google for 'Alzheimer's prevention.' A sponsored link for Navigenics appeared at the head of the page declaring: 'Get tested for Alzheimer's & learn how you can Prevent Alzheimer's'" (Messner 9). Currently, Navigenics is one of the main DTC genetic testing companies and this type of advertising is a reflection of other DTC companies. This advertisement clearly offers a form of hope for preventing Alzheimer's disease, but this information is very misleading. Upon further

examination of the company's website, it becomes clear that the company cannot actually deliver reliable Alzheimer's disease (AD) prevention strategies. These are the types of advertisements that could lure consumer such as Josh, from the introduction, to get genetic testing. Eventually, these tests can lead to consumers commenting about DTC testing with the same response as Josh, "I feel at this point in time that I've been harmed by this and not helped by this . . . I would not recommend this procedure or testing to anybody... it really doesn't help, one way or another, to know this information" (Messner 6).

Additionally, some proponents of DTC testing support their claim of autonomy by allowing customers to opt out of certain diseases in the standard package for the genetic tests. However, the same issues occur concerning a consumer's informed purchase. It is nearly impossible for a consumer to make an informed decision about which tests to "opt" out of without a clear understanding of the implications of the various tests (Messner 9). The necessary counseling and informed consent process involved in offering a genetic test can be complex. The process includes supplying medical information, as well as the exploration of psychosocial and familial issues. Furthermore, this process is extremely time-consuming and the fact that DTC testing companies provide testing for multiple genetic traits would increase the complexity of the situation. Ultimately, "the information provided by DTC testing companies at their websites is not comparable to the information provided by clinical physicians or researchers for informed consent" (Messner 7). Without a genetic counselor or other qualified professional involved, obtaining an autonomous informed decision about testing is not only difficult, but almost impossible.

Moreover, the absence of autonomy in DTC genetic testing directly contradicts the principles under the National Society of Genetic Counselors (NSGC), a group founded in 1979

that consists of genetic counselors and various healthcare professionals who seek to advocate genetic medicine. Under the NSGC's Code of Ethics, genetic counselors "[e]nable their clients to make informed decisions, free of coercion, by providing or illuminating the necessary facts, and clarifying the alternatives and anticipated consequences" (Hawkins and Ho 3). DTC genetic testing companies are currently unable to provide essential information about diseases and conditions, so they are undermining the principles of genetics, and the practice of medicine.

The final component of medicine raises awareness about justice on an individual basis as well as on a societal spectrum. This principle involves the promotion of equity within healthcare. Essentially, an individual should have access to care that is nondiscriminatory and just. Within society, this involves even distribution of healthcare and the burdens associated with it (Hawkins and Ho 5).

Supporters of DTC genetic testing argue that the availability of tests promotes justice because individuals have access to information that is not always supplied by the healthcare system (Hawkins and Ho 5). Also, there is medical value within this form of genetic testing because it creates an opportunity to identify individuals who need more screening. With this information, consumers can receive an earlier diagnosis of diseases, such as breast cancer, and receive aggressive or preventative treatment; this may decrease mortality rates. Sometimes, an early intervention may also decrease healthcare costs because an individual will not have to treat cancer that has greatly metastasized (Flaker and Mazza 23).

Despite the hope of the benefits of DTC genetic testing, these promises cannot be maintained. Testing is more likely to increase healthcare costs because of the limitations. Usually, companies only provide results for conditions and do not follow-up on the patients afterward. Therefore, once a consumer receives test results that indicates a predisposition to a

certain disorder, he or she then returns to the traditional healthcare provider in order to conduct more tests. Therefore, this creates more burdens on the healthcare system because if the results are inaccurate, then the individual is paying for tests that are unnecessary. This situation has “societal justice consequences (e.g. increasing waiting times, use of tax payer dollars) and individual (e.g. insurance premiums, deductibles) justice consequences” (Hawkins and Ho 3). Ultimately, although DTC companies earn profit from their business the burden always falls on the healthcare system and citizens because as a recent study revealed “78% of those considering using DTC genetic testing services indicated that they would ask their family physician for help in interpreting results” (Hawkins and Ho 4).

Although those who support DTC genetic testing would say that it is a service for anyone, this claim is not supported by the costs of the testing. Since the tests can exceed \$2,000, obviously it cannot be afforded by everyone. Unlike the traditional healthcare system, which provides financial support for those of lower socioeconomic status, DTC companies do not provide this. DTC genetic testing is a market that targets people who have means to use it; this makes the service inequitable and inaccessible to everyone (Flaker and Mazza 25). According to studies about DTC genetic testing, “the demographic characteristics of those who pursue DTC testing are higher socioeconomic status and Caucasian ancestry” (Hawkins and Ho 3). Clearly, this indicates a form of discrimination against those who cannot afford the service. Also, even if individuals were able to afford the product, it would discriminate against those who are not well-educated about the implications of the results. A 2006 survey conducted by the Kaiser Permanente Center for Health Research found the same correlation between awareness of DTC genetic testing and socioeconomic status as well as education. The survey showed that “among respondents with low, intermediate and high family incomes, the percentages aware of the tests

were 11, 13, and 16, respectively. More college graduates (19 percent) were aware of the testing than respondents with either limited or no college education (15 and 9 percent, respectively)” (Flaker and Mazza 16). Clearly, the awareness of the consequences of testing indicates that an individual has a higher form of income or education. It seems to be that DTC genetic testing, despite its claims, promotes injustice within society.

Clearly, DTC genetic testing undermines the four basic principles of medicine: autonomy, beneficence, maleficence, and justice. Since, these are not only violations of genetics, but also of medicine there are obvious changes that need to be imposed within the industry. Despite the purported promises and benefits, DTC genetic testing ultimately does not provide results to consumers in a way that minimizes harm. Individuals are not well informed about the process and its implications and as a result of their own curiosity, are left with dire consequences. DTC genetic testing companies exaggerate the promise of their product and do not accentuate the risks, therefore promoting maleficence on an individual and societal level. The “Ethic of Care” argument by Hawkins and Ho promotes the incorporation of autonomy, beneficence, maleficence, and justice within the system and is necessary. One can only hope that adjustments, such as only giving results that are definitely accurate or providing genetic counseling, can be implemented within the Direct-to-Consumer genetic testing industry; this would minimize harm and maximize the benefits of the process. Individuals should be more informed about the interpretation of results and should be knowledgeable about the accuracy of the results, so they do not make detrimental changes to their lifestyles. Without regulations, DTC genetic testing becomes a form of healthcare that serves more harm than good.

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