

## FOCUS: RESEARCH AND CLINICAL ETHICS

# Direct-to-Consumer Genetic Testing: A Comprehensive View

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Direct-to-consumer (DTC†) genetic testing refers to testing sold directly to consumers via the Internet, television, or other marketing venues without involving health care professionals. As the recent Supreme Court ruling eliminated the patentability of human genes, this rapidly evolving segment in the laboratory testing industry is starting to attract increasing scrutiny by government, scientists, consumers, and other interested parties. This article provides a panoramic view of the DTC genetic testing industry, including reasons for seeking DTC testing services, benefits and concerns associated with the industry, and potential development and prospects of this relatively new market under the current regulatory environment.

Direct-to-consumer (DTC) genetic testing refers to testing sold directly to consumers via the Internet, television, or other marketing venues without involving health care professionals. As a rapidly evolving segment in the laboratory testing industry, DTC genetic testing is starting to attract increasing scrutiny by government, scientists, and consumers alike. This paper aims to provide a panoramic view of the DTC genetic testing industry, including reasons for

seeking DTC testing services, benefits and concerns associated with the industry, and potential development and prospects of this relatively new market.

### WHY DO PEOPLE CHOOSE GENETIC TESTING?

According to global industry analysts, the global DTC genetic testing market is projected to reach the size of more than

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†Abbreviations: DTC, direct-to-consumer; DNA, deoxyribonucleic acid; GINA, Genetic Information Nondiscrimination Act; FDA, Food and Drug Administration, UCLA, University of California, Los Angeles.

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\$230 million by 2018 [1]. Consumers utilize DTC genetic services for various reasons. Even though some researchers group these reasons into many detailed subgroups, we can group them into three general categories.

### *Identity-Seeking*

This category includes testing for the purpose of identifying ancestry, paternity, and ethnicity. In general, these tests utilize mitochondrial DNA (maternal contribution), Y chromosome (paternal contribution), and markers on autosomes (ancestral information). Medical reasons are usually not involved. Some genetic companies provide extraordinary service in this area: for example, 23andMe not only gives you continental, regional, and subregional geographical information down to 0.1 percent of your genome, it even tells you what percentage of your genetic makeup comes from Neanderthal [2]. Philosophers are having an ongoing debate about the role that genetic testing plays in the modern search for identity in a pluralistic world [3], and the idea of defining “who I am” through genetic testing has a strong appeal to many [4]. Many companies promote this aspect of their service by including in their names first person pronouns or words that refer to origin: 23andMe, iGENEA, DNA Tribes, Family-Builder, etc.

### *Disease Risk-Testing that Complements Health Care*

This type of testing is more often ordered by physicians than patients and is a highly contested area in the context of DTC genetic testing due to lack of regulations. Testing is usually directed at specific genes and their corresponding diseases, such as *BRCA1* and *BRCA2*, which are responsible for some hereditary ovarian cancers [5]. The results of this kind of testing can potentially lead to important health decisions: Angelina Jolie, an actress and director, decided to have a double mastectomy after learning that she was a carrier of *BRCA1*. Statistical analysis of her results showed that she had an 87 percent risk of developing breast cancer and 50

percent risk for ovarian cancer [6]. As more companies are attempting to broaden the scope of their business using the latest advances in genetics, accountability of such tests is put in question as more scientists examine the soundness of the science that binds them [7]. A 2010 report by the Government Accountability Office expressed concerns over genetic testing companies that offer “expert” medical-related advice when they lack the legal capacity to do so [8].

### *Curiosity-Driven Testing/Searching for Better Lifestyle*

This is one of the most prevalent reasons for people seeking genetic testing. According to a survey conducted by Johns Hopkins University researchers on 1,046 gene testing customers, 94 percent did it out of curiosity, while 91 percent did it to learn about potential future diseases [9]. The diseases investigated by these tests are usually more common and, according to some researchers, provide limited medical value [10]. In her article, “Why 23andMe Genetic Testing Is A Waste Of Time And Money,” Elly Hart says that the results generated may be irrelevant [11]. While Hart is in her early 20s and of Asian descent, her report indicated she has a 8.9 percent risk of a heart attack, based on data that she is of European descent and between the ages of 40 and 79 because there was no option for other ethnicities [11]. The 50-plus traits and characteristics offered by the company even include earwax type and underarm odor [2], and some critics comment that the results only give users useless knowledge, such as “you should shower when you smell” or “it is not a good idea to touch heroin.” Research shows that the impacts on lifestyle improvements due to DTC genetic tests are often limited, resulting in no significant positive or negative changes [12].

## **BENEFITS OF DTC GENETIC TESTING**

There are many benefits associated with DTC genetic testing. Compared to traditional genetic testing as part of the health

care system, DTC genetic testing is known for its accessibility and affordability. Since consumers can have a testing kit mailed to them by ordering online, geographic locations no longer restrict one's ability to obtain the service. Prices range from less than \$100 [2] to more than \$1,000, depending on the type of test being ordered, and the industry has seen a rapid reduction in prices over the years. Thanks to the Genetic Information Nondiscrimination Act (GINA) signed into law in 2008, consumers need not worry about impacts on their health insurance policy and employment status as a result of pursuing genetic testing. Note, however, that this law is not perfect, as we will see in following discussion.

Some argue that the knowledge of potential diseases will lead individuals to make more healthy and preventive health decisions, thereby decreasing health care costs [13]. However, opinions on the subject have been divergent at best. Many physicians believe that DTC genetic testing can result in unnecessary health care costs when patients seek additional genetic counseling or visit their health care providers more often [14]. Currently, there is not enough empirical data to support either side, and ongoing researches still need more years of observation.

The most often used argument by genetic testing companies is empowerment. According to a study done by the American Marketing Association in 2008, more than 60 percent of genetic testing companies use empowerment as an emotional appeal to their customers [15]. Interpretation of the empowerment being discussed is two-fold: 1) the ability of consumers to make better informed health choices as a result of increased knowledge of their genetic risks and 2) the nebulous, ill-defined sense of enhanced power and being "in control." Even though a study shows that more than 80 percent of consumers derived some sort of satisfaction or empowerment from genetic tests [16], this benefit is highly subjective and debatable.

Lastly, DTC genetic testing raises the public's awareness of the topic of genetics

and serves as a tool for public education [17]. This role has been made more prominent in recent years, as the DTC genetic industry gradually attracts more attention being in a constant flux of change and evolution. Some DTC companies provide detailed patient education materials on many conditions, and as more consumers are exposed to the topic, more people will eventually gain the important knowledge of gene and the role it plays in our society.

## CONCERNS ABOUT DTC GENETIC TESTING

DTC genetic testing has attracted many criticisms and concerns, including its insufficient demonstration of clinical value, potential to reinforce inherited identity and prevent beneficial social integration, and aggravation of global health care inequity issues, just to name a few. For our discussion, we will explore some of the more quantifiable and defined issues raised in recent years.

### *Regulatory Loopholes*

As mentioned above, laws like GINA give consumers a certain degree of protection by restricting health insurers and employers from discriminating against certain populations based on genetic data. However, the law has multiple loopholes. It does not cover life insurance, long-term care insurance, or disability insurance [18]. Furthermore, according to an official government guidelines regarding GINA, health insurers are not prohibited from utilizing genetic results in determining insurance payments [19], which limits the scope of protection provided by GINA even more. Luckily, the market has yet to see significant impacts of these specific loopholes. In related research published in 2003, 636 women participated in a study to evaluate the impact of concerns about life insurance discrimination on use of *BRCA1/2* testing, and no respondent reported having life insurance denied or canceled [20]. While the regulatory implications remain a concern, exploitation of regulatory loopholes by DTC firms is unlikely to be-

come prevalent under tight public and government scrutiny.

On the other hand, genetic privacy may also become a big issue. Even though the majority of DTC genetic testing companies claim they will not share confidential genetic information with third parties without consumers' consent, these policies are usually self-imposed and not backed by laws. There are currently no laws that dictate what happens after a genetic testing company goes out of business, and very few states have laws that "broadly protect against unauthorized use of genetic information" [21]. Arguments for maintaining the status quo mostly come from researchers, who claim that the passing of such laws can hinder genetic research. The University of California recently submitted a formal letter against a proposed bill on increasing consent in the use of human-related bio samples, estimating substantial additional cost if such regulations were to be put in place [22].

The issue of privacy is further complicated by the nature of genetic testing. The same mechanism that has been employed by law enforcement for identification purposes is now being used to reveal the identities of "anonymous" research participants [23]. The identifiability of private data certainly raises legitimate concerns about balancing scientific needs with respect for individual privacy, and the privacy issue is likely to continuously present new challenges to the legislation as the technology evolves.

### *Emotional Cost of Learning Negative Results*

There was a legitimate concern that as patients learn about negative genetic testing results, they could potentially develop clinical depression, anxiety, and suicidal thoughts [25]. However, in a recent interview with Dr. Andrea Farkas Patenaude, psychology professor at Harvard Medical School, she pointed out that those individuals "who know their genetic test results indicate cancer risks are not likely to develop psychopathologies." This conclusion is supported by Dr. Lidewij Henneman at Vrije University, who claims that after extensive

study and psychoanalysis of genetic testing patients, "genetic risk information seems to be associated with little distress or anxiety" [26]. There are many hypotheses regarding the personal and societal factors that contribute to this result, but no definitive explanation has been provided.

### *The Science Behind DTC Testing*

According to Dr. Jeffrey E. Shuren, director of the Center for Devices and Radiological Health at the Food and Drug Administration (FDA), "None of the genetic tests now offered directly to consumers have undergone premarket review by the FDA ... to ensure results are accurate, reliable, and clinically meaningful" [24]. Although numerous associations between common genetic variants and diseases have been established, the quality of science behind these tests has been called into question at times [27]. This issue is highly correlated to the previous two topics discussed. Part of establishing a unified code of regulations requires building rigorous testing standards, and this can only be achieved through reliable and tested science.

## **THE FUTURE OF DTC GENETIC TESTING**

As a rapidly growing sector within the genetic industry, direct-to-consumer genetic testing is unlikely to go away in the short term. As pointed out by many professionals in the field, given the advancements in genetic testing techniques in recent years, more people should have access to the information stored in their genomes [28]. There are several possible directions in which DTC genetic testing might evolve, as we shall explore in the following discussion.

### *Become Part of the Existing Public Health Care System*

Even though DTC testing has gained substantial exposure to the public in recent years, it is still "very peripheral in the health care industry" [29]. Results offered by DTC testing companies are not connected to anyone's medical record, and the lack of physi-

cian involvement further aggravates the situation. In a physician survey conducted by the Washington State Department of Health, physicians serving a large percentage of Medicaid patients “were half as likely to have referred a patient to genetic counselors” [30], which signifies a low level of current integration between genetic testing and public health care.

One of the major hurdles DTC testing needs to overcome is unreasonable pricing. Our already fragile health care system is unlikely to be able to support DTC tests, unless they can be widely distributed at low cost. This goal became considerably more achievable with the recent Supreme Court’s ruling on the patentability of human genes [31]. The most conspicuous examples are *BRCA1* and *BRCA2*, which were previously owned by Myriad. Before the ruling, one-fifth of identified human genes had been patented [32], and some of them are linked to diseases such as Alzheimer’s disease, colon cancer, and others. The law of supply and demand dictates that companies like Myriad will no longer achieve high profit margins in the services they provide, and the subsequent lower prices will lower the financial pressure for Medicare, Medicaid, private insurance companies, and consumers. The soon-to-come cheap and wide distribution can be coupled with increased involvement of knowledgeable health care professionals, where testing results are interpreted in light of personal and family medical history, while potentially being included as a part of patient’s health care package.

#### *More Government Oversight While Remaining Independent*

Instead of being absorbed into the public health care system, it is highly possible that existing DTC testing companies will continue to exist independently while under increased government regulations. Since the emergence of private testing companies after the completion of the Human Genome Project, the tests that they offer have been subject to only minimal regulation [33]. As the FDA increases its scope of regulation by including all genetic tests as Class III medical

devices requiring pre-market reviews, effectiveness of these tests is likely to be more strictly moderated. For our government, the ultimate goal is to provide consumers with safe and reliable access to gene testing products and to address different social and medical issues raised by the DTC industry. This goal can be achieved more easily with more rigorous regulation than what our current system provides.

#### *Decrease Regulatory Agency Involvement and Promote Innovative Research and Development*

After the FDA reasserted itself in the regulatory environment in recent years, more and more scholars are warning about the undesirable effects that can come from an overly rigorous climate [34]. It is not yet clear how much more additional administrative expenses might be incurred from increased regulatory activities, but the amount can be substantial [22]. Also, it will be challenging for the FDA to achieve its stated goals without simultaneously stifling innovative research and development in the field [34]. Some other countries, such as Britain, have supported industry self-regulation [35], but in a mainly money-driven market such as the United States, this approach may be hard to realize.

## **CONCLUSION**

As a rapidly developing field, direct-to-consumer genetic testing is likely to see many new evolutions and changes in the near future. The field no doubt has tremendous potential, but the status quo is plagued with concerns and issues that urgently need to be resolved. Ultimately, direct-to-consumer genetic testing has to be centered around consumers, because without them the industry will be nonexistent. However, the data used in our discussion reveals that, unfortunately, the current genetic testing market can be both confusing and intrusive to customers, with misleading results and privacy issues overwhelming individual users.

The marketability and attractiveness of genetic testing is delivered by the reliability

and effectiveness of these tests, as these qualities will ultimately define the consumer experience. It is important to remember that testing results have familial implications as well and are therefore not restricted to individuals. Even though the firms still have many issues to work through, we hope that as the technology becomes more and more mature, the goal of assisting the health care system with DTC genetic testing will become more achievable and more people will be able to benefit from this affordable technology. Finding the delicate balance between ensuring appropriate government oversight and supporting innovative lab techniques can be a complex issue, yet at the same time it is a goal that can be accomplished through efforts and time.

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