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THE GROWTH OF THE DIRECT-TO-CONSUMER GENETIC TESTING INDUSTRY AND ITS POTENTIAL IMPACT ON THE FUTURE OF HEALTHCARE

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ABSTRACT

An emerging industry in the fields of technology and biology is direct-to-consumer genetic testing (DTCGT). This industry, encompassing companies such as 23andMe and AncestryDNA, provides genetic test results directly to consumers without a medical professional acting as an intermediary. As genetics research and DNA sequencing technology improves, these DTC genetic tests have become quicker and less expensive, leading to the rapid growth of this industry. This systematic review aims to uncover the public's perception of these services and the major motivations for pursuing DTCGT. Additionally, the potential uses of genetic data will be discussed, specifically in regards to its future utility in healthcare. Although there are various benefits of these DTCGT services, there also exists much criticism of the industry. Because there are so many concerns regarding DTCGT, several changes will need to be implemented in order for the DTCGT industry to continue to thrive. This systematic review incorporates research from the existing literature on this topic and presents both the positive and negative views on DTCGT. Using the information found within the existing research, predictions are made about the future of the DTCGT industry and the use of genetic data in healthcare.

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Chapter 1

Introduction

This chapter introduces the topic of direct-to-consumer genetic testing (DTCGT) and the motivation for selecting it as the focus of this systematic review. The history of the DTCGT industry and several concerns about DTCGT are also discussed. Also included in this chapter is a brief explanation about the biology behind DTCGT and the process consumers undergo when they pursue genetic testing. The research methodology is also explained followed by a list of the major research questions that will be answered in this systematic review.

Opening

Every human being is different due to a unique combination of 3 billion base pairs within his or her DNA. An individual's DNA has a major influence on his or her physical appearance, traits, and disease risk. With rapid advancements in DNA sequencing technology, it is now possible to fully sequence the human genome and discover the different traits and characteristics that it encodes. In the genetics field, a major trend within the past ten years is the rise in the availability and popularity of DTCGT services (Covolo et al., 2015; Skirton et al., 2013). These tests are available online and can be ordered directly by consumers, independent of physician referral or approval. Although the research behind the tests is conducted by medical professionals and researchers, consumers order the test without going through a medical

professional. Additionally, DTCGT companies do not provide consumers with help from medical professionals or genetic counselors in interpreting their results.

The emergence of these DTCGT companies has enabled individuals to learn about their own genomes and discover information related to their traits, health, and ancestry (Roberts et al., 2017; Skirton et al., 2013). While these services encompass an exciting new field of technology, there are many concerns about how the results are interpreted and how they may influence individuals' decision-making regarding their health (Covolo et al., 2015; Leighton et al., 2012; McBride et al., 2009). Additionally, there are ethical and security concerns due to the collection, storage, and analysis of such highly personal information (Borry et al., 2010; Skirton et al., 2012a). Existing research on this topic has focused on the public's attitude toward these DTCGT services and peoples' motivation for undergoing or refraining from testing (Gollust et al., 2012; Roberts et al., 2017; Vayena et al., 2014). Further research has also examined the potential use of these results, particularly focusing on how the results will influence health-related behaviors and decision-making (Kaufman et al., 2011; Nielsen et al., 2017). Other research concentrates on the negative aspects of DTCGT and how these services may cause more harm than good for consumers (Dohany et al., 2012; Leighton et al., 2012). The field of DTCGT is growing but the many current concerns regarding this industry may stunt its advancement. This systematic review aims to analyze the current research on this topic and propose a look into the future of DTCGT.

Motivation

Within just the past decade, the DTCGT industry has emerged and seen rapid growth. A report in January 2016 identified 246 companies that offer a DNA test to consumers online (Phillips, 2016). Several of these companies have already amassed databases with significant stores of consumer genetic data. Some of these companies include 23andMe[1], AncestryDNA[2], Gene by Gene[3], and Illumina's Helix Venture[4] (Phillips, 2016). Due to such high demand and intrigue of DTC genetic tests, Time named the genetic test offered by 23andMe its 2008 "invention of the year" (Kutz, 2010). As of 2018, the DTCGT industry is becoming widespread: advertisements for several DTCGT companies are regularly seen on television and online through various social media platforms. Since its founding in 2006, 23andMe alone has already amassed over 2 million customers. This increase from 800,000 in 2015 shows the recent surge in popularity of the DTCGT industry (Herper, 2017). The market leader in ancestry testing, AncestryDNA, has accumulated over 7 million customers since its founding in 2012 ("Company Facts," 2018). Although there exists much interest in this industry, medical professionals question the efficacy of these services and how they will impact the future of healthcare (Covolo et al., 2012). Understanding and analyzing these concerns is the major motivation for this systematic review. While the DTCGT industry continues to grow, what can be expected for the future of these services and their potential impact on the future of healthcare?

[1] https://www.23andme.com
[2] https://www.ancestry.com
[3] https://www.genebygene.com
[4] https://www.helix.com

History

One of the major sparks for the emergence of DTCGT services was the completion of the Human Genome Project in 2003 (Rehm, 2017). The Human Genome Project was a global initiative undertaken with the goal of sequencing the entire human genome. Researchers from institutions around the world collaborated to sequence the genomes of several humans who volunteered their DNA for research purposes. The researchers had the goal of uncovering the sequence of the 3 billion bases of human DNA with 99.99% accuracy ("1990: Launch of the Human Genome Project," 2013). There are four bases that are the basic building blocks of DNA. The order of these four bases within DNA codes for the amino acids that make up proteins within the human body ("Nucleotides and Bases," 2015). The goal of the Human Genome Project was to determine the order of these four bases within the volunteers' DNA. By 2003, the human genome sequence had been published and the researchers had discovered a total of approximately 20,000 genes in the human genome ("Timeline of the Human Genome").

In addition to successfully sequencing the human genome, the project resulted in vast improvements in DNA sequencing technology. The first billion bases of the human genome were sequenced in four years. With improvements in technology, the second billion bases were sequenced in just four months ("Bioinformatics: Introduction," 2013). In addition to the reduction in time, new technologies also allowed the DNA sequencing process to be less expensive. In 1990, at the beginning of the project, the average cost of DNA sequencing was 10 dollars per base. By the conclusion of the project in 2003, the cost had decreased to 10 cents per base ("Bioinformatics: Introduction," 2013). The completion of the Human Genome Project not only provided a human genome reference sequence, but also led to improved DNA sequencing

technology that was faster and less expensive. These factors opened the door for the growth of the DTCGT industry.

The first DTCGT company was founded in 2001 when Sciona, a UK nutrigenetics company, began to market to consumers via its website (Hogarth & Saukko, 2017). Before companies started to market to customers online, personal genetic tests were available only through a medical professional. However, this new method of genetic testing sold directly to the consumer saw growth as genetic testing was combined with the increasing use of the Internet as a medium for retail shopping. This new trend shifted genetic testing out of a medical setting and into the marketplace (Hogarth & Saukko, 2017).

Since 2001, there have been two identified phases of DTCGT. The first phase occurred before 2005 and consisted of companies like Sciona that marketed their tests for nutrigenetics. These tests mainly focused on identifying genetic variation and its involvement in nutrient metabolism. The main use of these test results was learning about dietary recommendations based on an individual's response to nutrients (Hogarth & Saukko, 2017).

The second phase of DTCGT rose in popularity after 2007. This second wave consisted of larger DTCGT companies such as 23andMe and Navigenics. Around 2007, these firms started offering polygenic risk tests that provided consumers with their risks for a multitude of common diseases including asthma and diabetes. The emergence of these tests stemmed from the improved DNA sequencing technology practice termed Genome-Wide Association Studies (GWAS). As GWAS technology improved, researchers were able to uncover more gene-disease associations. These known associations could then be searched for within an individual's genome to determine their risk for specific diseases (Hogarth & Saukko, 2017). The goal of uncovering health and disease risks persists as a major focus for DTCGT companies today. In

addition to health-related results, many current DTCGT firms specialize in offering further information to consumers such as ancestry, trait, and pharmacogenomic information. More controversial companies have even marketed tests that claim to determine if an individual is "compatible" with their partner or claim that they can predict a child's athletic talent (Topol, 2012).

While each DTC genetic test varies in price based on the company and results offered, a general trend is that DTCGT is becoming less expensive. For reference, a Health and Ancestry kit from 23andMe was priced at $999 in 2008 (Pollack, 2008). This test currently sells for $199 ("Our Services," 2018). Both the kit offered by AncestryDNA and the ancestry kit from 23andMe currently sell for $99 ("Ancestry," 2018; "Our Services," 2018). However, both of these companies regularly have sales that make the costs of the tests more reasonable for consumers.

Since many of these DTC genetic tests involve testing for health-related information without medical intervention, there have been major concerns about the lack of regulation of the tests (Topol, 2012). In its first several years, 23andMe provided customers with results that included their risk for 254 health conditions. In November 2013, the FDA forced 23andMe to withdraw all of its tests related to genetic health risk because the tests' efficacy had not been established (Herper, 2017). This ban presented the first roadblock to the continued growth of the DTCGT industry. However, in 2015, the FDA approved 23andMe to provide customers with risk information for 10 conditions including Parkinson's disease and late-onset Alzheimer's disease (Herper, 2017). More recently, in March 2018, 23andMe received the first-ever FDA authorization for providing cancer risk results to consumers. This authorization allows 23andMe to provide risk reports for *BRCA1*- and *BRCA2*-related genetic risk for breast, ovarian, and

prostate cancer ("23andMe Granted First FDA Authorization," 2018). These authorizations show how the FDA is focused on directing the DTCGT industry toward offering more regulated tests. This type of governmental regulation may help to assuage peoples' concerns about the efficacy of DTC genetic tests (Gniady, 2008). Conversely, many people argue that increased regulation will limit the public's ability to access their own genetic information (Gollust et al., 2017). This topic of regulating DTC genetic tests is controversial and will be discussed further in Chapters 4 and 5 of this review.

How Tests Work

In order to better understand the DTCGT industry, a brief explanation on the biology behind these genetic tests is needed. Within the human genome, there are four distinct base pairs that make up DNA. It is the unique combination of these four base pairs that codes for the proteins that make up the human body. It has been determined that human DNA is about 99.5% identical from person to person. The small differences within the human genome that make each individual unique are termed variants ("The Science Behind 23andMe," 2018). Many of these variants can be linked to an individual's health conditions, traits, and ancestry groups. The most common types of genetic variation among individuals are termed single nucleotide polymorphisms (SNPs). Each SNP is a single difference of a base pair. SNPs occur regularly throughout an individual's DNA, though most have no observable effect. However, some SNPs have an influence on an individual's disease risks, traits, and reactions to certain drugs (McGowan et al., 2012). Therefore, SNPs are the major focus of DTC genetic tests (Topol, 2012).

However, genetics is a very complex field and research is ongoing to uncover more associations between genes and susceptibility to disease (Aronson & Rehm, 2015). Genetics is such a difficult area to study because some conditions are caused by multiple SNPs in different genes, while different SNPs in a single gene may cause a range of diseases (Jameson & Long, 2015). Additionally, there are so many other non-genetic factors that can influence a person's health and traits. For example, an individual's environment and lifestyle can majorly impact his or her disease risk ("The Science Behind 23andMe," 2018). This complexity contributes to the concerns over the reliability of DTC genetic tests (Covolo et al., 2015).

The process of collecting and sequencing a consumer's DNA is standard across many DTCGT companies. The following explanation is the process that 23andMe customers undergo. A customer orders a test online from 23andMe and in return receives a kit containing instructions and a vial. The customer spits into the vial and sends it back to the 23andMe lab. Once in the lab, DNA is extracted from the cells in the saliva sample. The lab then processes the DNA using a genotyping chip. This genotyping chip reads hundreds to thousands of locations within the genome. These locations within the genome are known to vary between individuals and have been linked to certain conditions, traits, and ancestry groups. Once these locations are fully sequenced, the genetic data is analyzed using known genetic associations from scientific and medical research. 23andMe then generates a personalized report that displays the consumer's genetic test results ("The Science Behind 23andMe," 2018). Based on the company, results that could be presented include disease risk, appearance traits, ancestry, and pharmacogenomic information.

Research Methods

In order to capture sufficient information about the current status of DTCGT and project ideas about the future of this field, a systematic review of the existing literature was completed. I located existing studies and papers on this topic using the online search engines Google Scholar and PubMed. I also explored the websites of DTCGT companies to better understand how these services are marketed to consumers and what consumers experience when they view these websites. Additionally, I purchased a health and ancestry genetic test from 23andMe to better understand the experience of DTCGT as a consumer. These research methods allowed for a thorough understanding of the current status of the DTCGT industry and the aptitude to make predictions about the future of DTCGT.

Research Questions

The aim of this systematic review was to gain a thorough understanding of the DTCGT field in order to answer the following research questions. Each of the questions is addressed in a separate chapter of the systematic review.

1) What are consumers' main motivations for undergoing DTCGT?
2) What are the potential uses of DTCGT results?
3) What are the major criticisms of DTCGT?
4) How will DTCGT affect the future of healthcare?

Chapter 2

Consumer Motivations For DTCGT

Most of the existing literature regarding DTCGT focuses on consumers' perceptions of these services and their reasoning for undergoing or refraining from testing. This chapter outlines the major motivations for consumers to pursue testing, separated into medically-related and entertainment reasons. The major medical motivations include learning about disease risk, the opportunity to learn valuable information while family planning, and the opportunity to contribute to genetics research. The major entertainment motivations are learning about genetic traits and ancestry, as well as a general curiosity with DTCGT.

Learn About Disease Risk

Throughout the existing literature on DTCGT, one of the major motivations for consumers to undergo testing is to learn about their disease risk. Many DTCGT companies currently offer customers information about their health risks based on genetic variants that are known to be associated with a variety of conditions. Most DTCGT companies present disease risks to consumers as absolute risk[5,6,7]. This absolute risk prediction is derived from relative risk and average population risk. The relative risk is determined based on genetic risk markers found in an individual's DNA. The average population risk varies based on the population defined.

[5] https://permalinks.23andme.com/pdf/samplereport_geneticheath.pdf
[6] https://www.helix.com/shop/health/
[7] https://www.genebygene.com/pages/genetics

These values are multiplied together and presented to consumers as their absolute disease risk for a specific condition. Figure 1 below is a helpful visualization of how absolute disease risk is derived.

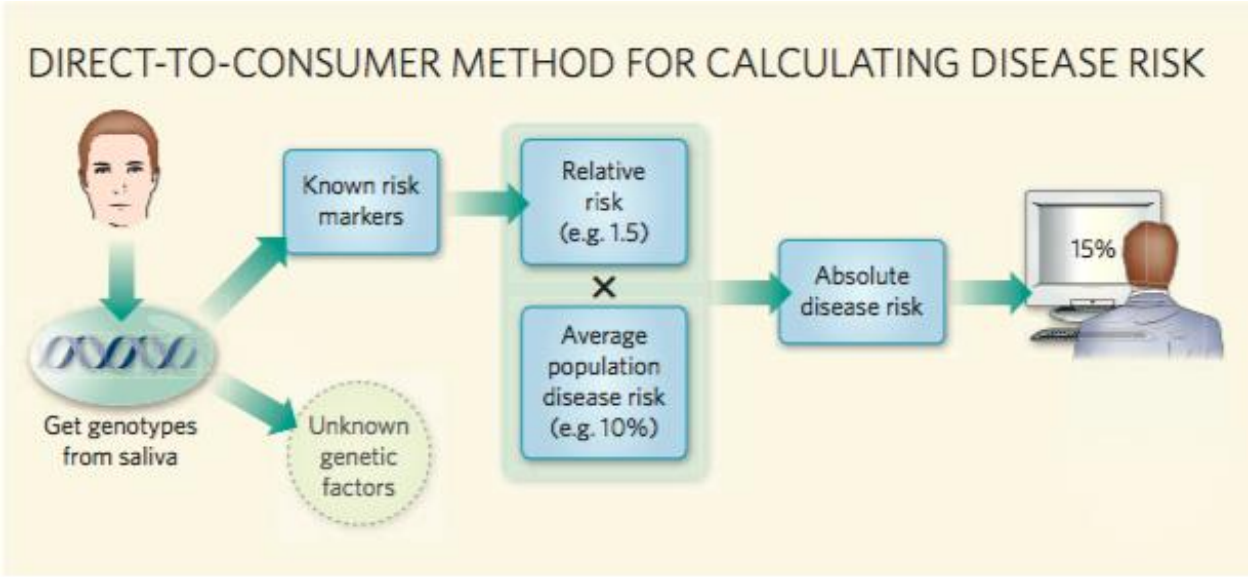

Figure 1. DTCGT method for calculating absolute disease risk. (Ng et al., pg. 724)

In a study of over 1,600 consumers from two leading DTCGT companies, 72% of participants indicated that they were very interested in learning about their disease risk, particularly for heart disease, breast cancer, and Alzheimer's disease (Roberts et al., 2017). Gollust et al. (2012) surveyed 369 individuals and found that many participants were motivated to learn about their disease risk in hopes of potentially improving their health. Vayena et al. (2014) conducted a similar study with a sample of university students and found that the second most rated motivation was an interest to find out disease risks. McGowan et al. (2010) investigated early users' reasons for utilizing DTCGT services. They found that the two most commonly cited reasons were to gain health-related information and to learn about individual genetic risk factors. In a qualitative study that examined the content of users' personal stories written about on online blogs and DTCGT company websites, one of the major themes identified

was a motivation to learn more about health. This included consumers' desire to learn about their risk for specific diseases, to potentially improve their quality of health and live longer, and to learn more about their medical history (Su et al., 2011).

Throughout the literature, a desire to learn health-related information ranks at or near the top of participants' motivation for undergoing DTCGT. Additionally, McBride et al. (2009) discovered a correlation between perceiving one's self as having health habits in need of changing and his or her desire to get tested. In addition to the desire to learn about disease risk for physical diseases, Wilde et al. (2010) conducted a study to examine participants' willingness to undergo testing for mental disorders. They found that the majority of participants were interested in having a genetic test for susceptibility to major depression. However, all participants who were interested in the predictive genetic test for depression risk reported that they would only do so through a trusted medical professional. This indicates that consumers would not want to learn their risks for psychological disorders through a DTCGT company. However, the general finding is that the opportunity to learn about disease risk is a major motivating factor for individuals considering DTCGT. Many participants are motivated by the prospect to then use their disease risk results to make lifestyle changes in hopes of improving their health.

Family Planning

Since genes are passed down from generation to generation, another major motivation for consumers to get tested is to learn about genetic risks when they are family planning (Rehm, 2017). Many diseases are heritable and can be tracked through a family. Therefore, DTCGT can

be a useful way to learn more about the genes responsible for disease within families. In a study of over 1,000 social networking users, of those who would consider DTCGT, 74% reported that they would use it gain knowledge about disease in their family (McGuire et al., 2009). Wilde et al. (2010) also found that participants believed a major benefit to testing was learning valuable information that could benefit their whole family.

In addition to learning about health information that could be useful for an individuals' family members, genetic test results can also be helpful for couples who want to have children. In a longitudinal qualitative study of 20 primary care patients who underwent DTCGT, many participants were motivated by their desire to help their families and the next generation (Wasson et al., 2013). The main way in which DTCGT results can be used in family planning is carrier status testing. Some diseases are genetically recessive, meaning that two copies of the disease variant are needed for an individual to have the disease. If an individual only has one copy of the variant, they are a carrier of the disease and do not show symptoms. However, if two carriers have a child, there is a greater probability of having an affected child. Therefore, learning the carrier status of both partners can provide valuable information for family planning. In her review article, Rehm (2017) provided a case study that demonstrates the utility of carrier status testing. In this example, a couple is considering having a child, so they undergo preconception genetic testing. It is found that they are both carriers for the *SMN1* gene. Because of this, there is a 25% risk that the couple has a child with spinal muscular atrophy. Due to this information, the couple decides to pursue preimplantation genetic diagnosis so they can select embryos that do not inherit both pathogenic variants. This example emphasizes the valuable utility of DTCGT results for family planning.

Opportunity to Contribute to Scientific Research

Another medically-related motivation for consumers to pursue DTCGT is the opportunity to contribute to scientific research (Vayena et al., 2014). When an individual undergoes DTCGT, he or she can opt for their genetic data to be used for scientific research ("Research," 2018). For example, 23andMe has utilized consumer genetic data to discover various associations between genes and diseases. Additionally, 23andMe consumers can answer questions about their health habits and lifestyle. These responses are then used by 23andMe researchers to make genetic discoveries. In their study, Vayena et al. (2014) discovered that the university students' major motivation to undergo testing was a desire to contribute their data to scientific research. In their study of early users of personal genomics companies, McGowan et al. (2010) cited that many participants viewed personal genome services with optimism for genomic research. Genetics is an exciting field and many individuals are motivated by the opportunity to play a part in helping researchers make genetic discoveries. Although contributing to genetics research does not directly benefit consumers, it could greatly advance our knowledge of genetics.

Learn about Genetic Traits

A more entertainment-related motivation for undergoing DTCGT is the opportunity to learn about genetic traits. Many DTCGT companies offer consumers information about genetic traits that are not associated with health or disease risk. Examples of trait information that are offered by 23andMe are eye color, skin pigmentation, the presence or absence of cheek dimples, and more random attributes such as asparagus odor detection. In addition, 23andMe offers

wellness reports that consist of information related to weight, muscle composition, sleep patterns, and propensity for caffeine consumption ("Reports," 2018). Although these attributes and characteristics are physically observable, consumers are still interested to learn how these traits and habits are linked to their genome. In the study of over 1,600 DTCGT consumers, 72% of participants indicated that they were very interested in trait information (Roberts et al., 2017). Vayena et al. (2014) also found that the second most rated motivation to pursue testing among the university student sample was an interest in learning about personal traits. While learning about trait information does not offer consumers information that could be used to make lifestyle changes, it is a commonly reported motivation for consumers. Learning trait information is more for entertainment purposes than being actually informative for consumers. Although learning trait information is not directly beneficial to consumers, it has been found to be a major reason for consumers to undergo testing.

Learn about Ancestry

Another major motivating factor related to family is the ability to learn about ancestry information. Many current DTC genetic tests offer insight into an individual's ancestry based on known genetic associations. The leader in genetic ancestry tests, AncestryDNA, can estimate an individual's origins to more than 150 ethnic regions around the world ("Ancestry," 2018). Many DTCGT consumers may not know their family history, so genetic testing provides an opportunity to learn more about their ancestors. Therefore, the opportunity to learn ancestral information through DTC genetic tests has become a major motivating factor for consumers. In a study of over 1,600 DTCGT consumers, Roberts et al. (2017) found that ancestry was the

greatest motivation for users, as 74% of participants indicated that they were very interested in learning about their ancestry. Su et al. (2011) conducted a qualitative study of users' experiences on online blogs and DTCGT company websites. They also found the common theme of genealogy amongst users' reasons for getting tested. Another motivation related to ancestry that was found in this study is the potential to learn about famous relatives. The opportunity to track one's heritage through his or her genes is an interesting entertainment-related aspect of DTCGT that motivates many consumers to pursue genetic testing.

Curiosity

Because DTCGT is a new and exciting field, many consumers are motivated simply by a curiosity or fascination with the process. In their review article on DTCGT, Covolo et al. (2015) found that within 37 papers, consumers interested in pursuing DTCGT had the main motivation of curiosity. Curiosity was also cited as the major reason for early adopters of personalized genomics to undergo DTCGT (Gollust et al., 2012). Wasson et al. (2013) also cited that that one of the major motivations amongst 20 primary care patients was curiosity. Su et al. (2011) found that curiosity was a major motivating factor written about on online blogs and DTCGT company websites. These researchers elaborated by citing that consumers were both curious and fascinated with the technology behind the tests as well as with the topic of genetics itself. It is a general pattern that when any new form of technology becomes available, many consumers are intrigued and want to learn more about it. This same pattern can be seen in the growth of the DTCGT market as increasingly more individuals are curious about the process of genetic testing and interested in learning more about their personal genetic information.

Chapter 3

Potential Uses of Genetic Data

This chapter summarizes the potential uses of DTCGT results and of genetic data in general. The potential uses of DTCGT results by consumers include the opportunity to make lifestyle changes to lessen health risks and the opportunity to share results with healthcare providers. Another valuable use of genetic data in the healthcare field is in pharmacogenomics, which enables more targeted treatment based on an individual's genomic information. The major use of consumer results by DTCGT companies is to conduct large-scale genetics research studies.

Make Lifestyle Changes

As stated in Chapter 1, one of the major motivating factors for consumers to pursue DTCGT is to learn health-related information. These health-related results can then be used as motivation for consumers to make lifestyle changes in hopes of improving their health. In her review article on this topic, Rehm (2017) stated that there is widespread recognition that an individual's access to his or her own genetic data allows for a more thorough understanding of his or her health and disease risks. With a more complete understanding of their risks, consumers may then be motivated to make lifestyle changes to reduce their risk. In a study of over 1,600 DTCGT consumers, after receiving their results, 59% of the respondents indicated that the results would influence the management of their health (Roberts, 2017). In another study of over

1,000 consumers, 43% of respondents sought additional information about at least one health condition covered in the genetic test results. Researchers of this same study found that one-third of the respondents indicated that they were being more careful about their diet after receiving their results (Kaufman et al., 2011). These statistics show that consumers are willing to act on the health-related results they receive. A similar discovery was found in another study of 1,000 DTCGT consumers. In this study by Nielsen et al. (2017), 30% of participants reported making a change to their diet and 26% of participants reported changing their exercise habits. These lifestyle changes were specifically motivated by consumers' genetic test results. Although these statistics only indicate modest changes in diet and exercise within the sample, they demonstrate that some consumers do act on the results they receive.

Although the studies above demonstrate that DTCGT results may motivate consumers to make positive lifestyle changes, other researchers found that DTCGT is limited in its power to affect change. In their study of over 1,000 university students, Vayena et al. (2014) found that of the respondents interested in DTCGT, many were not likely to be affected by or act upon the results. In another study of over 2,000 DTCGT consumers, Bloss et al. (2011) found no significant differences between baseline and follow-up in dietary fat intake or exercise behavior. Abdul-Husn et al. (2014) focused on genetic testing for lung cancer risk and found that genetic test results do not independently have a significant impact on consumers' willingness to change their behaviors, such as quitting smoking. The findings of these studies indicate that although individuals may purchase DTC genetic tests with the intention of making lifestyle changes based on their results, the majority of consumers do not actually change their behaviors.

However, a common theme throughout the literature is that consumers do not make changes in response to their results mainly because there is nothing on which to act (Wasson et al., 2013). Many individuals may receive results that are not concerning or they may learn that they have a predisposition for a condition that cannot be avoided. In these situations, a lifestyle change would not be necessary. Abdul-Husn et al. (2014) stated that genetic information may motivate individuals to engage in healthier behaviors only if the results pertain to conditions that they feel threatened by, or if there is something that can be done to reduce the disease risk. So although researchers of these studies found that DTCGT results are not regularly used to make behavioral changes, there may simply not be a reason to make changes. Even though the observed use of genetic test results to make behavioral changes among consumers is low, the opportunity to make health-related lifestyle changes still exists as a potential use of DTC genetic test results.

Share Results with a Healthcare Provider

Because genetics is such a complex topic, many DTCGT consumers may not have the background knowledge necessary to understand their results. Upon receipt of their results, many consumers will reach out to healthcare providers or genetic counselors for help interpreting their health risks. In one study of potential DTCGT consumers, 92% of participants intended to share their genetic test results with a physician (Gollust et al., 2012). McGuire et al. (2009) conducted a study involving social media users and found that 78% of those who would consider undergoing DTCGT would ask their physician for help interpreting their results. These

researchers also found that 61% of participants actually felt that physicians have a professional obligation to help interpret the results. However, many physicians do not currently have the knowledge or experience necessary to accurately interpret genetic results. This issue will be further discussed in Chapters 4 and 5.

Covolo et al. (2015) discussed that the significant benefits of DTCGT can be realized if consumers share their results with a medical professional. They stated that those who shared their test results were significantly less worried about being tested compared to non-sharers. Additionally, receiving help from a trained healthcare professional when interpreting genetic test results led to significantly higher consumer comprehension than just online delivery of the results. This review demonstrates that the utility of DTCGT can be augmented with the help of a healthcare professional or genetic counselor. Seeking assistance can help alleviate consumer misunderstanding and the healthcare professional can provide advice to the consumer on how to mitigate their health risks.

While there are apparent benefits to sharing genetic test results with a healthcare professional, the actual percentage of consumers who share their data is low. Covolo et al. (2015) found that the number of people who actually reported sharing test results with a physician was less than 30%. In one study of over 1,000 participants, only 28% discussed their results with at least one healthcare professional. Within this group, nearly half stated that sharing their results taught them something they did not already know that could be used to improve their health (Kaufman et al., 2011). This demonstrates the value of sharing genetic test results with a healthcare professional.

The decision to share genetic test results also varies based on the results themselves and the consumer's demographic information. For example, the higher a person's perceived risk of cancer, the more likely he or she is to share the results. Additionally, older participants were more likely to share their results with a healthcare provider (Kaufman et al., 2011). This may be explained by the generally lower genetics knowledge found in older generations. While many studies indicate that consumers plan to share their DTCGT with a healthcare professional, the actual percentage is relatively low. However, there appears to be major benefits to consumers who discuss their results with a healthcare provider. Therefore, as the DTCGT market continues to grow, there will be an increased need for healthcare providers and genetic counselors to help consumers interpret their genetic test results.

Pharmacogenomics

As more is discovered about the human genome, there has been a growth in the field of pharmacogenomics. Pharmacogenomics is the study of how the genome influences drug response (Abdul-Husn et al., 2011). In the past, drugs were prescribed to patients with little knowledge of how a patient would metabolize the drug. With increased genetic research, it has been discovered that an individual's genome can influence his or her drug response. For instance, a specific dosage for one patient may have minimal curative impact whereas the same dosage in another patient may be potentially dangerous (Aronson & Rehm, 2015). These differences in drug response due to different variants within patients' genomes makes treatment much more complex. Even when the cause of a disease is known and well-understood, a patient's specific

genetic variants can affect the efficacy of the treatment by changing how the drugs are metabolized or by increasing the possibility of harmful side effects (Aronson & Rehm, 2015).

As more research has been conducted on pharmacogenomics, there has been the growth in a new wave of disease treatment called personalized medicine. Personalized medicine aims to combine an individual's genetic data, along with known clinical, family, and demographic variables to more precisely guide a patient's medical treatment (Abdul-Husn et al., 2011). Instead of a "one-size-fits-all" approach to treatment, patient treatment will be personalized based on his or her genetic information. Personalized medicine will enable clinicians to quickly, efficiently, and accurately select the most appropriate and safest treatment plan for a patient (Aronson & Rehm, 2015). Esther Dyson, a strong proponent for personalized medicine and one of the first participants in the Personal Genome Project, stated the importance of personalized medicine: "You would no more take a drug without knowing the relevant data from your genome than you would get a blood transfusion without knowing your blood type" (Topol, 2012). This quote serves to demonstrate the importance of knowing an individual's genomic information when determining the most effective treatment plan. In a systematic review of the existing literature, Covolo et al. (2015) found that 70% of physicians felt that genetic tests would be helpful in managing patients, especially in regards to pharmacogenomics.

In addition to physicians' positive opinions about genetic testing for pharmacogenomics information, another study found that the general public has a high level of interest in receiving pharmacogenomics information (Abdul-Husn et al., 2014). This is suggestive of positive public attitudes toward personalized medicine. Although it is still in its early phases, personalized medicine is an exciting field that could greatly impact the future of healthcare. Because

pharmacogenomics is such a complex area, however, any genetic test that provides pharmacogenomics information would most likely need to be discussed with a healthcare professional.

Research

While the three subcategories above outline the potential uses of genetic test results for consumers, the major potential use of the results amongst the DTCGT companies themselves is the opportunity to conduct genetics research. As increasingly more consumers are undergoing DTCGT testing, these DTCGT companies benefit by being able to use consumers' genetic data to conduct scientific research. When consumers pursue DTCGT, they can allow for the companies to store their raw genetic data and use it for research ("Research," 2018). These large databases of participant genomes are then used to investigate patterns and find links between genes and diseases. In their review article on precision medicine, Aronson & Rehm (2015) noted that the line between clinical care and research is blurring. As increasingly more DTCGT consumers allow for their genomes to be used in research studies, they are allowing researchers to gain a better understanding of the role of genes in disease.

As more is understood about the genetic root of complex disorders, researchers will be better able to find cures and develop more effective treatments for patients. For example, researchers of one genetics research study examined the genetic data from more than 118,000 23andMe customers who consented to participate in research. With such a massive sample size, the researchers were able to identify new genomic regions associated with asthma, hay fever, and

eczema. They identified more than 130 different genes or gene regions associated with these allergic diseases (23andMe, 2017). Using this information, future research can be done to develop a more targeted approach to treating these conditions. Because DTCGT companies have amassed so much genetic data from consumers, exciting research like this can be conducted. The ability to conduct such large-scale genetic research studies is a major benefit of the DTCGT industry.

Chapter 4

Criticism of DTCGT

Despite the multiple benefits and potential uses of DTCGT outlined above, there are many concerns regarding the DTCGT industry. This chapter explains the major motivations for consumers not to pursue testing and the major criticisms of the DTCGT industry. The main motivations against testing include a fear of receiving negative results and the cost of testing. Major criticisms of the industry as a whole include the possibility for results to be psychologically distressing to consumers and the potential for results to be misinterpreted. Further concerns have been raised about the lack of genetics knowledge amongst healthcare providers, which limits DTCGT consumers' ability to get help understanding their results. Additionally, there have been major concerns about the clinical utility and the lack of regulation of these tests. Another concern is that some tests may be marketed misleadingly by claiming to provide information that is not scientifically proven. Another major concern is the privacy of consumer data and the fear of insurance or employment discrimination based on genetic test results. The final criticism that will be discussed is the lack of ethnic diversity within genetics research, which limits the amount and depth of results provided to consumers of certain ethnicities.

Consumers May be Fearful of their Results

While many individuals are interested in and motivated to undergoing DTCGT, many people are wary of these services and would refrain from testing. The major concern for many of

these individuals is the possibility of receiving negative health results. In a study of over 1,600 consumers of DTCGT, 62% of the consumers still considered the possibility of receiving unwanted information (Roberts et al., 2017). Although they decided to undergo testing, they were still fearful of receiving negative results. In another study of individuals who purchased a DTC genetic test, Bloss et al. (2010) found that approximately half still expressed concerns about the process. A major concern about DTCGT is that consumers will receive results that indicate an increased risk for an incurable disease. Since nothing can be done to change his or her risk, this type of result can be especially worrisome for an individual.

Privacy Concerns

Another fear involving DTCGT is the possibility of privacy issues (Skirton et al., 2012b). Genetic data is such highly personally information so many individuals are concerned about the security and privacy of their personal genetic data. Other perceived disadvantages of predictive testing are a fear of the risk of discrimination by insurance companies and employers, risk of fatalistic thinking, and an increased social stigma (Wilde et al., 2010). These fears were all cited as individuals' motivations for refraining from DTCGT.

Cost

Another motivational factor that could influence individuals not to pursue DTCGT is the cost of these services. Although the cost of genetic testing has decreased over the years ("Our Services," 2018; Pollack, 2008), many individuals may not have the economic means to purchase

a test. Others may not believe that the information provided by the DTCGT company warrants spending the money to purchase the test. While cost may be a barrier preventing someone from undergoing DTCGT, the major deterrent found in the literature is the fear of receiving worrisome results.

Psychological Impact of Results

Because DTC genetic tests can provide consumers with disease risks, a major concern is that consumers may experience anxiety or psychological distress upon receiving their results. The fear of receiving worrisome results is cited as one of the major deterrents to pursuing DTCGT (Skirton et al., 2012b). Nearly half of physicians (neurobiologists and psychologists) that were surveyed in one study believed that genetic testing could cause psychological harm to their patients (Covolo et al., 2015).

The following case study demonstrates the possibility for DTCGT to cause psychological harm to a consumer. This case study followed the experience of a woman who underwent DTCGT and discovered that she had a *BRCA* mutation, which indicates an increased risk of developing breast cancer. The DTCGT company did not provide any genetic counseling services, which led the woman to experience anxiety and distress. She also had minimal understanding of the implications of her results (Dohany et al., 2012). This case study demonstrates that significant psychological distress can occur for consumers who receive results indicating an increased risk for disease. However, the woman personally sought out genetic counseling to better help understand her risks and her possible treatment options. After genetic counseling, the

woman reported feeling fortunate that she learned about her *BRCA* mutation and that she could take steps to lessen her risk of developing breast cancer (Dohany et al., 2012).

While this case study demonstrates the potential for DTCGT results to cause distress, many other studies negate this finding. Researchers that examined 14 existing studies on DTCGT found that fewer than 30% of DTCGT customers reported a change in health anxiety even if they were mutation-positive. They also discovered that there was no significant difference between baseline and follow-up anxiety symptoms at both 3 months and 1 year after receiving the DTCGT results (Skirton et al., 2012a). Bloss et al. (2011) similarly found no significant difference in anxiety between baseline and follow-up for a sample of over 2,000 DTCGT consumers. Another systematic review indicated that only 2% of DTCGT consumers regretted being tested and only 1% reported that they experienced harm from their results (Skirton et al., 2012b). In a qualitative study by Wasson et al. (2013), half of the participants reported that testing had no significant impact on them. In fact, the most commonly reported emotional effect was being relieved or pleased with the results. Although there are concerns that the results of DTCGT will cause consumers anxiety or distress, most of the research on this subject indicates that DTCGT results have little impact on consumers' psychological state. For those consumers who do experience anxiety from their results, genetic counseling is a viable option to help consumers better understand their risks and help alleviate some of their worry.

Misinterpretation of Results

One of the most cited criticisms found throughout the existing literature on DTCGT is that consumers are likely to misinterpret their results. Since the essence of DTCGT is that results are available directly to the consumer, there are not genetic counselors or medical professionals provided to help consumers interpret their results (McBride et al., 2009). Many critics of DTCGT argue that the genetic variants tested for confer relatively little risk for common health conditions. They further believe that the general public has little knowledge about the complex and multi-factorial nature of these common diseases. Therefore, there are concerns that many consumers may misinterpret their results (McBride et al., 2009).

Additionally, critics cite that the absence or lack of quality genetic counseling and medical supervision will result in consumers having minimal help in interpreting their results (Su et al., 2011). Covolo et al. (2015) found that more than 80% of physicians expressed concerns about consumers' possible misinterpretation of their results and how this could lead to increased anxiety. Ormond et al. (2011) conducted a study focused on a small group of medical students enrolled in a human genetics course. They found that 70% of the students believed that DTC genetic tests needed interpretation. Additionally, after taking the course, 80% of the students indicated that DTCGT companies should provide genetic counseling to consumers.

In addition to physicians' concerns, the general public expresses concerns that they do not possess the knowledge necessary to understand their genetic test results. In one study, participants were recruited from a group of individuals who registered to attend an event on personalized medicine. Among this sample, 32% had misperceptions about personal genomic testing, such as believing that these tests provide diagnostic information (Gollust et al., 2012).

These individuals already had prior interest or knowledge in genetics, so the percentage of individuals who have misperceptions about genetic testing in the general public is likely to be even higher.

One of the greatest misconceptions about DTCGT is that the results are a medical diagnosis. McGuire et al. (2009) surveyed over 1,000 social networking users and found that 34% of the participants considered the results obtained from DTCGT to be a medical diagnosis. Since genetics is so complex, DTC genetic tests do not provide diagnostic results. Presently, many of the DTCGT company websites do inform consumers that the results do not provide diagnostic information. For example, 23andMe describes their Genetic Health Risk report as providing:

> Information about whether you carry genetic markers associated with risks for certain health conditions. They are not for diagnosis. Factors like lifestyle, environment and genetic markers not covered by this test can also play a role ("Reports," 2018).

While this statement is clearly visible on the website[8], consumers may still have the misperception that their DTCGT results provide diagnostic information. Additionally, without adequate counseling by knowledgeable medical providers or genetic counselors, consumers may fail to acknowledge this limitation of DTCGT (Sweeney & Legg, 2011). The fact that DTCGT results are not a diagnosis is a common misconception that may cause confusion and distress for consumers.

Leighton et al. (2012) specifically focused on comparing the perceptions of DTCGT between the general public and genetic counselors. The researchers of this study found a

[8] https://www.23andme.com/dna-health-ancestry/

significant difference between the way the general public and genetic counselors interpreted the meaning of DTCGT results. For example, the participants were shown sample results for a colon cancer test. As a whole, the general public believed that the results showed a greater risk for colon cancer than the genetic counselors did. Leighton et al. (2012) also found that the general public participants believed that the results would be significantly more helpful than the genetic counselors did. Another interesting finding in this study was that the majority of the general public respondents rated the results as easy to understand, but they often misinterpreted them. This misunderstanding may lead consumers to have unnecessary concern, false reassurance, or may cause them to make unnecessary changes in their health or screening behaviors.

Although the majority of prior research indicates that consumers of DTCGT may have difficulty interpreting their results, researchers of several studies have discovered high comprehension amongst DTCGT consumers. Findings of one study indicate that participants exhibited high overall comprehension of results, with an average score of 79.1% correct on comprehension questions. The higher level of comprehension was significantly associated with higher levels of numeracy, genetics knowledge, and education. It was also found that older age was associated with lower comprehension scores (Ostergren et al., 2015). McGowan et al. (2010) surveyed early users of personal genome services and found that nearly half of the participants believed that the results were not medically actionable. These participants understood that the results indicated disease risk, not a rigid disease diagnosis. However, the other half of these participants did not understand this limitation of DTCGT.

Overall, the pattern seen within the research on this topic is that the general public is not sufficiently equipped to accurately interpret DTCGT results. Additionally, the risk for

misinterpretation is greater for older individuals and those who are less educated. This possibility of misinterpreting DTCGT results has raised the concern of whether it is appropriate to offer genetic testing without medical supervision (Hogarth & Saukko, 2017). Moving forward, this concern will need to be addressed in order for the DTCGT industry to persist.

Lack of Genetics Knowledge Amongst Healthcare Providers

Due to the likelihood of consumer misunderstanding, consumers may reach out to healthcare providers with questions about their results. But because DTCGT is a relatively new field, many healthcare professionals have not been trained on how to handle patients who seek out their help in interpreting DTCGT results. In their systematic review on DTCGT, Skirton et al. (2013) found that there was a low level of awareness and experience of DTCGT in health professionals. If a DTCGT consumer is concerned with the results they receive, he or she may turn to a healthcare provider for help in interpretation. However, many primary care physicians are not currently equipped with the genetics knowledge or experience to help these patients. Abdul-Husn et al. (2014) found that since the availability of genetic information outside of the clinical genetics domain is so new, most practicing providers have no formal education in its use. This in turn limits their ability to help their patients interpret their DTC genetic test results. McGowan et al. (2010) found that early users of personal genome services believed that a lack of physician knowledge limits the clinical acceptance and clinical applicability of DTCGT results. The lack of genetics knowledge amongst healthcare providers limits the potential utility of these

results in a healthcare setting. In the future, medical professionals will need to have more formal education and training in genetics in order to help patients interpret their DTCGT results.

Lack of Clinical Utility

Another major concern regarding DTCGT is the lack of reliability and clinical utility of these tests. Many critics argue that DTCGT companies overstate the actual predictive power and utility of the results (Skirton et al., 2012a). Leighton et al. (2012) explained that SNPs used in current DTCGT are generally not causative of disease and may instead just confer a very small change in risk of developing the disease. Therefore, the presence of a SNP in a consumer's genome does not necessarily provide any useful information to the consumer. Furthermore, since many diseases are caused by a combination of multiple genes, the contribution of a single gene is often small. Each gene may indicate a small risk increase or decrease, and is thus clinically insignificant (Cho, 2009). Additionally, many of the genome-wide association studies that were done to determine associations between SNPs and disease outcomes were not considered clinically robust by medical standards of the time (Allyse et al., 2018). Since DTCGT exists outside of a medical setting, many critics are wary about the gene-disease associations that are made by these tests. Also, the disease risk is often presented in terms of a percent increase without providing information about the background risk that served as the basis for the risk estimate (Allyse et al., 2018). Without knowing the baseline risk, it is difficult to understand one's personal predicted risk.

Another argument that has been raised is that accurate predictive risk assessments for individuals cannot be made based on the results of large populations (Topol, 2012). These critics argue that association does not indicate causation. Even if one gene appears to be associated with a certain disease in a certain sample, there are so many other factors that could be involved. An individual may have a certain gene associated with disease, but this does not necessarily mean he or she will develop the disease.

Another concern regarding the clinical utility of DTCGT tests is the inconsistency of results amongst DTCGT companies. Several researchers have received different results for the same consumer when the DNA was sent to several different DTCGT companies (Topol, 2013; Ng et al., 2009; Abdul-Husn et al., 2014; Kutz, 2010). This lack of consistency amongst the DTCGT companies further suggests that the results are not clinically valid.

The uncertainty about the clinical utility of DTCGT has been noted by medical professionals. Covolo et al. (2015) found that 60% of primary care physicians had doubts about the clinical utility of DTCGT results. Researchers of the study on medical students in a human genetics class also found that the majority of students expressed concerns about the reliability and utility of results (Ormond et al., 2011). Since predicting disease risk is a major motivating factor for consumers to pursue DTCGT, it is concerning that there are such major doubts about the clinical utility of these services. Cho (2009) stated that it is far too soon for DTCGT services to promise clinically useful information. She believes that there is far to go to assure the validity of these DTC predictive tests and that more research has to be done to develop their clinical utility in the medical field.

Lack of Governmental Regulation

Because DTCGT exists outside the scope of the medical world, there is less governmental regulation for the DTCGT industry. Although the DTCGT industry is in the commercial marketplace, the product offered provides health-related information. Therefore, many critics argue that these tests should be governmentally regulated. The FDA's Alberto Gutierrez believes that "genetic tests are medical devices and must be regulated" (Topol, 2012). Among the concerns of these DTCGT critics are the lack of an adequate consent procedure and the possible inappropriate testing of minors (Su et al., 2011).

On the other side, proponents for DTCGT argue that regulation will limit the types of results that are available to consumers and may even completely restrict consumer access to this information (Borry et al., 2010). These individuals see genetic data as personal information that should be directly accessible by consumers. Among those who are in favor of reduced regulation of DTCGT are many consumers of DTCGT. In a study of over 900 DTCGT consumers, Gollust et al. (2017) found that 89.9% of participants believed that people have a right to access genetic information directly. Additionally, 68.3% of participants supported even wider availability of personal genome testing services. These statistics indicate that many DTCGT consumers support less regulated access to one's personal genetic data and DTC genetic tests that are more widely available to the general public without governmental interference.

Currently, there are several legislative acts that are relevant to genetic testing. These acts are the Health Information Portability and Accountability Act (HIPAA) and the Genetic Information Nondiscrimination Act (GINA) (Shoenbill et al., 2014). The HIPAA Privacy Rule considers individually identifiable genetic information to be health information. But since DTC

firms have direct interactions with consumers without going through physicians, HIPAA does not currently apply to DTCGT firms as a group (Laestadius, 2016). On the other hand, GINA has helped to alleviate some of the worries about employment or insurance misuse of genetic test results. However, there are clear limits to this law including that insurers can still request genetic information before covering procedures (Topol, 2012). Although these two laws exist, they do not directly apply to the DTCGT industry because these tests are available outside of a medical setting. This complicates the regulation of the DTCGT market and will be a point of contention in the future of this industry.

As discussed in Chapter 1, 23andMe initially offered disease risks for over 250 conditions. In 2013, the FDA intervened and banned 23andMe from providing this disease-related information to consumers. Many proponents of DTCGT believed that the FDA overstepped and should not have banned 23andMe from providing these results (Hogarth & Saukko, 2017). However, the FDA is continually authorizing 23andMe to provide risk information for more conditions (Hogarth & Saukko, 2017; "23andMe Granted First FDA Authorization," 2018). These authorizations show the potential future of regulation in the DTCGT industry. Regulation is important because it can help ensure that results are valid and easily understandable for consumers. However, regulation also limits the access consumers will have to their genetic data. Therefore, in the future, a balance between regulation and open access will need to be achieved in the DTCGT industry.

Misleading Marketing

In addition to the current lack of clinical utility and regulation of DTC genetic tests, a major criticism is that some DTCGT companies misleadingly market their products. While many DTC genetic tests offer disease risk and ancestry information that has been scientifically confirmed, other companies offer less scientific results. For example, there have been companies that market products claiming to be able to identify a child's athletic talent. Kutz (2010) identified two companies that marketed tests claiming to predict in which sports a child would excel based on DNA analysis. One expert in this field characterized this claim as "complete garbage" and indicated that there was no scientific evidence to back the claims made by these companies. Other genetic test companies market to consumers by claiming that their tests are able to identify with whom an individual is romantically compatible (Topol, 2012). Again, there is no current scientific evidence that confirms that compatibility can be inferred from one's genome. While these claims may entice consumers to purchase a DTC genetic test, the results are not scientifically proven. As a result, these companies marketing false claims damage the reputation of the DTCGT industry as a whole.

Privacy and Security

Another major concern regarding the DTCGT industry is the privacy of consumers and security of their genetic data. Genetic data is such highly personal information that can be used to individually identify a consumer. Because DTCGT results contain such highly personal information, many people are concerned about how this data is secured and how the privacy of

consumers is protected. Skirton et al. (2012a) found that the most commonly cited potential harm amongst DTCGT consumers was the risk that consumers' privacy and confidentiality would not be respected. More specifically, critics of DTCGT have concerns about the use of consumers' samples in research activities and the testing of minors (Borry et al., 2010). In one study of the public's perceptions of DTCGT for susceptibility to major depression, all participants were unanimously against accessing DTC predictive genetic testing over the Internet. Their major worries involved the security of their DNA sample and privacy of depression risk information (Wilde et al., 2010).

Although consumer genetic data is de-identified and anonymized when it is used for research, a genome-wide scan can still be used to identify a particular person. Therefore, it is important that DTCGT companies have practices in place that protect consumer privacy in any database that contains their genetic data (Topol, 2012).

Another related concern is if the consent process for consumers who purchase a DTC genetic test is adequate. Other concerns focus on the potential secondary use of consumer data for research by third parties without the consumer knowing (Middleton et al., 2017). Additionally, there is a major concern about the potential misuse of the data by the DTCGT companies themselves. For instance, what if these companies marketed the data to pharmaceutical companies? Also, what would happen to consumer genetic data if one of these companies were to go bankrupt (Topol, 2012)?

Because the DTCGT industry involves highly personal information, it is important that these companies take action to protect the security and privacy of their consumers. As the pervasiveness of hacking increases across the world, it is understandable that individuals are

concerned about the security of their genetic data. Therefore, DTCGT companies need to take the appropriate steps to protect and secure consumer genetic data. Rilack and Wernicke (2014) present a framework for securely storing genomic data on the cloud. The first step involves keeping the genetic data encrypted at all times, including during transfer, in storage, and during computation. In addition to this server-side encryption, all user data, file transfers, and platform services should communicate through encrypted SSL channels. The final suggestion is that computation environments be separated in an Amazon Web Services Virtual Private Cloud container to ensure a high level of isolation, security, and monitoring. Following these steps will help to ensure that consumer genetic data is stored and handled securely.

Insurance or Employment Discrimination

One of the major fears related to the privacy of DTCGT is that the results could be used discriminatorily by insurance companies or employers. In a survey of neurobiologists and psychologists, almost half of these physicians thought that genetic testing consumers could be exposed to possible insurance discrimination (Covolo et al., 2015). In their study of 20 DTCGT consumers, Wasson et al. (2013) found that a repeated concern was the confidentiality of results. More specifically, the patients in this study believed that their results may affect their health insurance or lead to denial of coverage. The researchers who focused on individuals' perceptions of DTCGT for major depression found that one of the major perceived disadvantages of predictive testing was the risk of discrimination by insurance companies and employers (Wilde et al., 2010). If a consumer's DTCGT results indicate an increased risk for disease, he or she may

be fearful of these results being used by insurance companies and employers. This possibility may even prevent potential DTCGT consumers from pursuing testing. The fear of insurance or employment discrimination emphasizes the need for DTCGT companies to protect the privacy of their consumers and ensure the security of consumer genetic data.

Lack of Ethnic Diversity

The final criticism of DTCGT deals more broadly with a criticism of genetics research as a whole. This criticism is the present lack of ethnic diversity within genetics research. Currently, the majority of genetics research focuses on people of European descent. Because the effect of genetic variants is dependent on the nature of the studied population, much more is known about genetic variants in people of European ancestry than in other populations (Abdul-Husn et al., 2014). Currently, the DTCGT tests available are not adequately sensitive to the influence of ethnic differences amongst consumers (Allyse et al., 2018). Not only is there more information related to ancestry for individuals of European descent, there is also more known about genetic disease risk. For example, a variant that is found to be associated with diabetes may indicate a different risk in someone of European descent than in someone of African descent. Similarly, differences in variants based on an individual's ethnicity may confer different drug responses. This means that certain drugs will be safer and more effective in some populations than in others (Popejoy & Fullerton, 2016).

Because the majority of genetics research has been conducted with genetic data from individuals of European descent, consumers of African and Asian ancestry are currently more

likely to receive ambiguous genetic test results. They are also more likely to receive results indicating that they have variants of unknown significance (Popejoy & Fullerton, 2016). Because of this, DTCGT consumers of European descent receive much more informative results than consumers of other ethnicities.

In 2006, the Government Accountability Office (GAO) investigated the role of ethnicity in DTCGT. They sent DNA samples of African American and Asian donors to four DTCGT companies. They found that none of the companies could provide these donors with complete test results. Additionally, none of the companies disclosed this limitation prior to the test purchase (Kutz, 2010).

Researchers of a more recent study in 2016 found that the percentage of individuals included in genome-wide association studies who are not of European descent has increased to nearly 20% (Popejoy & Fullerton, 2016). While this is an improvement, much of this rise is contributable to more studies being conducted in Asia on individuals of Asian ancestry. The proportion of individuals of African, Latin American, Hispanic, and indigenous descent has hardly shifted. These four groups of individuals represent less than 4% of samples analyzed (Popejoy & Fullerton, 2016).

The lack of genetics research conducted on these populations limits the amount of information that DTCGT companies can provide to these consumers. Therefore, it is recommended that fundamental changes be made within genetics research. For example, Popejoy and Fullerton (2016) suggested that funding agencies should develop financial incentives for studies that include diverse cohorts of participants. If steps like this are taken, more can be learned about the effects of genes in a multitude of ethnicities. By increasing the research

conducted in diverse populations, more informative results can be provided to DTCGT consumers of all ethnicities.

Chapter 5

Future of DTCGT

Despite the current criticism of the DTCGT industry, the DTCGT market continues to grow (Covolo et al., 2015; Herper, 2017). However, several changes can be implemented within the DTCGT industry to help assuage these concerns in the future. This chapter outlines the changes that I perceive as necessary in the future of the DTCGT industry. The first involves more background information and training being provided to consumers on how to interpret their results. Additionally, DTCGT companies should provide consumers with more information and resources about genetic counselors. Because genetic testing is becoming more widespread, healthcare professionals will also need more training and education on genetics to help patients interpret their results. To address concerns about the validity and reliability of genetic test results, regulations should be established to ensure that DTCGT companies are providing accurate and consistent results to consumers. There should also be greater regulation on the marketing of DTCGT so that consumers are fully aware of the limitations of these DTC genetic tests. Because genetic test results have the potential to majorly impact the future of healthcare, steps will also need to be taken to incorporate genetic data into electronic health records and databases. If these actions are taken to improve the DTCGT industry, I perceive that this market will continue to thrive and these tests will have the potential to greatly impact the future of healthcare.

Increased Information and Training Provided to Consumers

As discussed in Chapter 4, one of the major criticisms of the DTCGT industry is that consumers are prone to misinterpret their results. More specifically, comprehension of results varies by demographic characteristics and education levels (Ostergren et al., 2015). In a study by Carere et al. (2016), genetics knowledge and genetics self-efficacy of personal genome testing consumers was tested at baseline and 6 months after receiving results. The researchers found a slight increase in mean genetics knowledge, but a decrease in consumers' mean self-efficacy scores. The decrease in self-efficacy shows that consumers were less confident in their understanding of genetics following the receipt of their results.

Since genetics is such a complex field, it is probable that consumers become more confused after being tested. Therefore, it is important that DTCGT services provide adequate background information about their tests and training materials so that consumers can fully grasp the implications of their results. As a leader in this industry, 23andMe has established a satisfactory example of how results should be presented to consumers. Figure 2 shows the 23andMe results provided to a consumer for a late-onset Alzheimer's disease test. In addition to explaining the specific variant that was tested, 23andMe also explains the limitations of the test and provides scientific details that could help consumers better understand their results. Although the figure shows that the tested variant was not found within the genome, 23andMe explains that there are other factors and genetic variants not covered by the test that could confer an increased risk for late-onset Alzheimer's disease. All of this information provided to consumers can greatly improve their understanding of results.

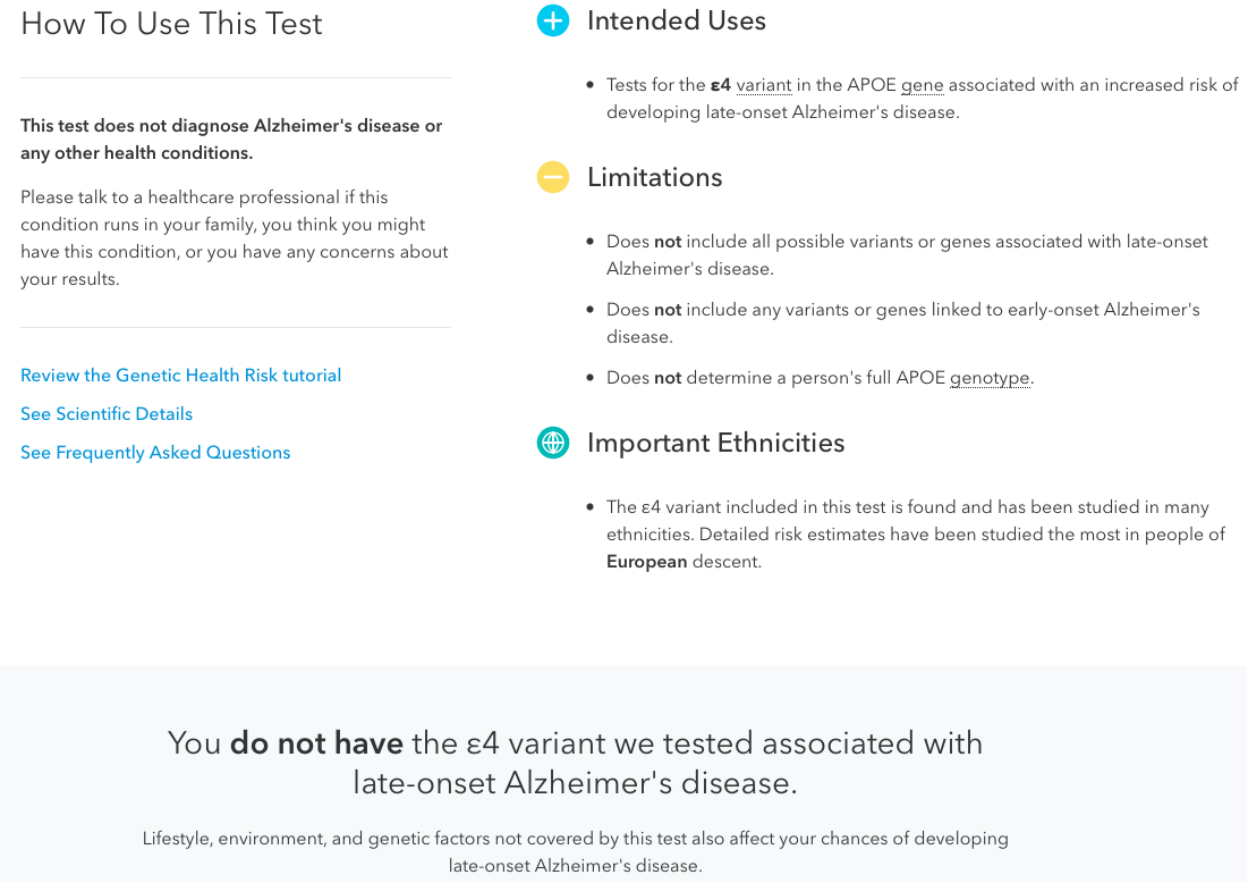

Figure 2. 23andMe consumer result for late-onset Alzheimer's disease.

23andMe does an exemplary job of presenting results to consumers. However, an improvement that has been suggested within the existing literature is the tailoring of results to individual consumers (Bloss et al., 2010). As discussed above, comprehension of genetics differs based on demographic characteristics and education. Therefore, it would be beneficial if DTCGT results were tailored to individual consumers, especially since these results are not delivered via a trained health care professional (Ostergren et al., 2015).

In addition to providing more background information and training on genetics, another improvement that could be made is the use of visualizations to display probability information. Shoenbill et al. (2014) stated that many individuals possess limited understanding of genetics and statistics. Since so much of genetics is probability and statistics, it would be beneficial if DTCGT

services provided visualizations to consumers to help them interpret their results. For example, the 23andMe test for cystic fibrosis shown in Figure 3 below indicates that the consumer has a 1 in 230 chance of carrying a variant not covered by the test. However, the visual provided displays one affected individual in a crowd of less than 230. It would be valuable if the visual accurately displayed the probability of 1 in 230.

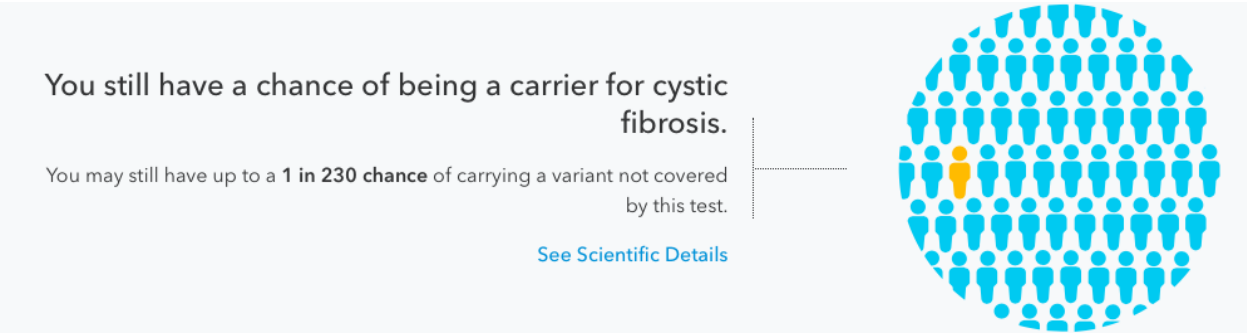

Figure 3. 23andMe consumer visualization for cystic fibrosis risk.

Using a figure to present results is both visually appealing and informative to consumers. Therefore, a recommendation to DTCGT companies is to provide accurate visuals to consumers to help them interpret their results. Overall, the general consensus in the existing literature is that DTCGT services should provide consumers with more tailored information and training to help in their understanding. Additionally, providing informative visuals can help consumers interpret their probability and risk results. These changes will help to alleviate the major concern of consumer misinterpretation of results.

Increased Information and Resources about Genetic Counselors

Although providing more information and visualizations to consumers may help improve their understanding, some may still need additional help interpreting their results. One option

that these consumers have is discussing their results with a trained genetic counselor. In their study of 20 DTCGT consumers, Wasson et al. (2013) found that the majority of participants indicated that the results were fairly easy to understand with the help of a genetic counselor. Within this group, only one participant stated that a genetic counselor was not needed to interpret the results. Additionally, nearly half indicated that they would not have understood their results without the assistance of a genetic counselor. This study demonstrates the benefits of discussing DTCGT results with a genetic counselor.

Dohany et al. (2012) reported that both pre- and post-test genetic counseling in combination with DTCGT can help to alleviate consumers' distress and empower them to utilize their results to make lifestyle changes. The American College of Medical Genetics has even advised that:

> A genetics expert such as a certified medical geneticist or genetic counselor should be available to help the consumer determine, for example, whether a genetic test should be performed and how to interpret test results in light of personal and family history" (Middleton et al., 2017).

While Middleton et al. (2017) do not advocate for mandatory genetic counseling to be provided by DTCGT companies, they believe that if the testing process could potentially cause psychological distress, then it is the responsibility of the company to provide support and resources to its consumers.

Despite the potential benefits of discussing results with a genetic counselor, the current state of the DTCGT industry involves very little information about counseling services. Covolo et al. (2015) examined 24 online companies and found that 75% recommended and arranged for counseling services. However, only one third of the companies directly provided counseling

services. Hennen et al. (2010) found that less than 39% of online companies provided genetic counseling and pre-test counseling was especially rare. Although the premise of DTCGT is that a test can be ordered without a medical professional acting as an intermediary, it would be very beneficial if these companies provided counseling resources to their consumers if necessary.

In addition to DTCGT providing more information to consumers about genetic counseling options, the field of genetic counseling itself will need to grow. In her article about evolving healthcare through personal genomics, Rehm (2017) stated that training programs must be expanded to produce more genetic counselors capable of working with patients and answering their complex questions. Because genetic counselors can greatly help DTCGT consumers interpret their results, there is an urgent need to train a whole workforce of genetic counselors to meet the demand for jobs with DTCGT companies (Middleton et al., 2017). Another researcher on this topic, O'Daniel (2010), stated that genetic counselors will play a critical role in facilitating the integration of genetic health risks into precision medicine. In the future, genetics will play a greater role in healthcare. Consequently, the field of genetic counseling will need to grow in order to help the general public understand the role of genetics on health and disease risk. As the genetic counselor workforce grows and DTCGT companies increasingly provide resources to their consumers, the understanding and utilization of DTCGT results can be augmented.

Increased Genetic Education and Training for Healthcare Providers

In addition to increased training for DTCGT consumers, healthcare professionals will also need increased levels of genetics education and training in order to help consumers interpret

their results. Although the DTCGT field exists outside the supervision of medical professionals, many consumers will approach their healthcare provider for help interpreting their results. Therefore, in the future, it will be important for healthcare professionals to be equipped with the genetics knowledge necessary to help their patients understand their genetic health risks. Because the rate of genetics research has been so rapid, there has been little time for health professionals to acquire the skills needed to accurately interpret genetic test results. Additionally, physicians are more commonly encountering the Internet-informed health consumer, such as consumers of DTC genetic tests (McBride et al., 2009). In the past, patients received their medical information only through a trained healthcare provider. Today, health-related information is readily available to individuals over the Internet. Therefore, it is important that physicians become better equipped to explain and answer patient questions.

As discussed in Chapter 3, sharing DTCGT results with healthcare professionals has been found to add perceived utility to the tests (Kaufman et al., 2011). Therefore, it is important that healthcare professionals possess adequate knowledge of genetics in order to help their patients. However, the current level of genetics knowledge amongst physicians is lacking. In a 2012 study, only 16% of primary care physicians declared themselves to be “very confident” in interpreting genetic test results (Covolo et al., 2015). In a survey of over 2,000 primary care physicians, the average respondent had a correct response rate of only 74% in interpreting a genetic diagnosis (Gniady, 2008). These studies indicate the current lack of physician understanding of genetics. This lack of understanding greatly limits the utility of DTCGT results and could lead to even more confusion for consumers. Furthermore, in a 2009 survey, only 10% of physicians thought that they had the necessary training and knowledge in genomics to use genetic testing to treat patients (Bloss et al., 2011). Since there is such great potential for using

genetic data to determine targeted treatment plans for patients, it is important that healthcare professionals have a better understanding of genetics. Therefore, efforts should be made to increase genomic literacy within physicians. This will allow for more informed decision-making and treatment of patients (O'Daniel, 2010).

Because healthcare professionals will have an important role in helping consumers interpret their results, it has been recommended that genetic education be improved in medical schools. There have also been recommendations to establish guidelines for genetic competency among primary care physicians (Shoenbill et al., 2014). As the field of genetics continues to grow and more is uncovered about the impact of genetics on health, it is important that healthcare professionals have adequate training and education on this subject. In regards to DTCGT, increased genetics training for healthcare professionals can greatly increase the utility of consumer results and help to alleviate consumer misunderstanding.

Increased Regulation to Ensure Validity and Consistency

As mentioned in Chapter 4, one of the major concerns of the DTCGT industry is the reliability and validity of the results. Since these DTCGT services provide consumers with personal health risks, it is important that the results are as accurate as possible. However, there are current concerns about the validity of DTC genetic tests because results differ between DTCGT companies. Abdul-Husn et al. (2014) found that for any given disease, there is little concordance between DTCGT companies in the variants that are studied, the effect sizes, the average population risk, and the algorithms that are used to calculate the disease risk. This has led to significant differences in disease risk predictions between the DTCGT. Topol (2012)

discussed that the results across companies could even be conflicting. The genotyping of the raw data is concordant, but there is no consensus on which studies and calculations are used to develop risk predictions. He explained that there are no current standards for the level of evidence that is needed to identify a genetic marker or how to determine the magnitude of its effect. Ng et al. (2009) compared the results provided from the DTCGT companies 23andMe and Navigenics. They found that the accuracy of the raw data was high as it agreed more than 99.7% of the time. However, they also found that only two-thirds of the relative risk predictions qualitatively agreed between the two companies. A similar study done by GAO involved sending consumer DNA to four DTCGT companies. The disease risk results often differed between the four companies, which indicates that identical DNA samples can yield contradictory results (Kutz, 2018).

Because of the lack of consistency in results amongst the various DTCGT companies, there have been recommendations to increase the regulation of this industry. Hogarth and Saukko (2017) advocate for the rigorous independent evaluation of DTCGT tests before they enter the market. More specifically, Allyse et al. (2018) suggest that the FDA should take a regulatory approach that seeks to find a balance between direct consumer access to their genetic data and stricter regulatory control over the more medically-related results.

While regulation is important to ensure the validity of DTCGT tests, it should not be a barrier to consumer access to results. A balance will need to be achieved in order for the DTCGT industry to continue to thrive. In a study of over 500 DTCGT consumers, the majority of participants (68.3%) believed that the test kits should be even more widely available to consumers, such as at outlets like drug stores. Additionally, only 27.8% believed that the government should enforce more regulation (Gollust et al., 2017). These statistics indicate the

public's desire to have direct access to genetic data without government interference. But without regulation, there is a great risk of DTCGT companies returning inaccurate or inconsistent results to consumers. Therefore, in the future, it will be important that a balance is struck so that consumers have access to genetic data that is as accurate as possible. Additionally, standards should be established within the DTCGT industry so that the results are consistent as possible between the different DTCGT companies. In her paper, Gniady (2008) advocates for regulation of DTCGT in order to protect the consumer without suppressing the medical revolution caused by genetic testing. She supports the regulation of genetic tests by the FDA as well as promotes the open market for selling tests directly to consumers. The regulation of the DTCGT industry is a controversial subject. But because DTCGT does provide medical information, it is in the best interest of the consumers that the tests are validated. As the tests become more regulated, consumers can be more confident that they are receiving accurate results. This will help to alleviate one of the major concerns regarding DTCGT.

Increased Regulation on the Marketing and Advertising of DTCGT

In addition to the need for increased regulation of the validity of DTC genetic tests, there is a great need to monitor how these tests are marketed to consumers. Covolo et al. (2015) explained how DTCGT is currently advertised despite the minimal and controversial nature of the supporting evidence used for these tests. They argued that more research needs to be done on these tests to determine if it is appropriate to market them to consumers at all. Webborn et al. (2015) also reported that there is concern within the scientific community that the current level of genetics knowledge is being misrepresented for commercial purposes. In other words, critics

are concerned that DTCGT companies are making inaccurate claims about the utility of DTCGT in order to entice more consumers to purchase these genetic tests. Currently, DTC genetic tests are being advertised using traditional persuasive strategies that are generally used for commercial products (Webborn et al., 2015). This involves emphasizing the benefits of the tests while downplaying the potential side effects. But since DTCGT involves providing vital health-related information to consumers, there are critics who advocate for improved guidelines for the advertisement of DTC genetic tests (Covolo et al., 2015).

Researchers studying the advertisement of DTCGT found that the majority of stories published on DTCGT websites focus on health-related benefits. However, most of the stories from non-DTCGT company websites emphasize consumer motivations related to curiosity and genealogy. This suggests that DTCGT companies are selectively using stories that promote health-related benefits of genetic testing (Su et al., 2011). However, as was discussed in Chapter 4, these DTC genetic tests do not provide diagnostic information. In fact, most DTCGT websites have disclaimers on their websites stating that their tests are not to be used as a diagnostic tool. Additionally, much of the existing literature indicates that the majority of consumers do not make health-related lifestyle changes based on their results. Therefore, the current marketing strategy of focusing solely on health-related benefits is misleading to consumers. Currently, these companies are overpromising the health-related utility of their tests in order to entice more consumers to buy them.

In addition to misleading marketing, Kutz (2018) found 10 examples of false marketing among DTCGT companies. Kutz uncovered claims made by four companies that a consumer's DNA could be used to create a personalized supplement to cure diseases. Also, two of these companies made claims that their supplements could "repair damaged DNA" or even cure

disease. Experts confirmed that there is no scientific basis for these claims. The findings of this study highlight the current level of deceptive marketing within the DTCGT industry. These companies try to entice more consumers to purchase their tests by claiming to provide information that is not currently scientifically backed. Therefore, it is important that guidelines and regulations be established regarding the marketing of these products. In the future, it will be important that these guidelines are set so that consumers are fully aware of the limitations of the product they are purchasing.

Incorporating Genetic Data into Electronic Health Records

Some of the greatest potential for genetic data lies in its future incorporation in the healthcare field. One way in which genetic data can be integrated into healthcare is if it is stored in electronic health records (EHRs). An EHR is a real-time, patient-centered digital record that stores information about a patient's medical history, diagnoses, medications, treatments, and laboratory test results ("Electronic Health Records," 2013). Because all of this information is stored digitally, one of the greatest features of an EHR is that patient health information can be shared and modified by providers across multiple healthcare systems. This allows for more efficient and streamlined patient care. Researchers conducting a study of over 500 DTCGT consumers found that 63% of participants reported that they would want genetic information to be included in their medical records (Gollust et al., 2017).

As the incorporation of genetic data into EHRs expands, physicians will be able to utilize electronic clinical-decision support (CDS) systems to determine the best treatment plan for patients. These CDS systems provide physicians with more information about a genetic test by

linking to electronic resources such as websites and databases. These systems can also display pre- and post-test pharmacogenomics warnings that indicate any potentially adverse reactions to drugs due to a patient's genetic variants (Aronson & Rehm, 2015). Aronson and Rehm see CDS systems in the future being able to guide clinicians through complex scenarios that involve multiple sources of patient data, including a patient's genetic information. However, CDS systems rely on access to structured electronic data, which is currently unlikely to be reliably transferred from DTCGT. Since the potential for genetic data to be incorporated into EHRs and CDS systems is so great, it will be important for health IT professionals to develop effective methods in which to structure genetic data so that it can easily be transferred to these systems (Aronson & Rehm, 2015).

Although there is such great potential for the use of genetic data in healthcare, there are many challenges that need to be overcome before this becomes a reality. One of the major current challenges to advancing the use of genetics in healthcare is the careful balance of storing and sharing personal genomic and health data in an accessible manner while protecting the privacy of individuals (Rehm, 2017).

Another challenge lies in the storage of genetic data. Existing health information architectures are incapable of storing genetic data in a way that provides for easy searching and sharing of the information across healthcare systems over a patient's lifespan (Shoenbill et al., 2014). Therefore, standards will need to be established for the methods that are used to share genetic data. Additional standards will need to be established on how to compress these large volumes of data and how genetic data can be interfaced with EHRs (Shoenbill et al., 2014).

Current technology is not adequate for interpreting genetic data due to the sheer complexity of genetics (Green & Guyer, 2011). In order for genomic data to be more efficiently

incorporated into healthcare, fundamental changes are needed in the infrastructure and mechanisms for genetic data interpretation and sharing. These changes will allow for a continuously learning healthcare system and improvements in both clinical care and research (Aronson & Rehm, 2015). In their paper discussing the future of genetics in healthcare, Green and Guyer (2011) recognized that genomics has already begun to improve diagnostics and treatments. However, these authors argued that profound improvements in the effectiveness of genetics in healthcare cannot realistically be expected for many years.

Chapter 6

Conclusion

Increased research and knowledge in the genetics field, coupled with improved technology, has allowed for the fast growth of the DTCGT industry over the past decade. The public's perception of these DTCGT services is generally positive and many individuals are motivated to pursue genetic testing for health, ancestry, and curiosity reasons. Some of the greatest potential for these tests lies in the opportunity for genetic data to be incorporated into healthcare. Specific potential uses of genetic data involve improving patient treatment by using pharmacogenomic information and developing a more accurate prediction of disease risk due to genetic markers within an individual's DNA. Additionally, these DTCGT services have the great opportunity of contributing consumer genetic data for use in research that discovers gene-disease associations. Despite the multiple benefits of DTCGT, there are currently many concerns regarding this industry. Most criticism involves the potential misunderstanding of results by consumers and the lack of clinical utility of the results. Other major concerns focus on the privacy and security of such highly personal information and the current lack of regulation on the validity of these tests and how they are marketed to consumers.

Because DTCGT is such a controversial topic, there is a need for major changes to be made within the industry in order for it to be more widely accepted in the general public and in the healthcare field. These changes involve providing more training to both consumers and healthcare professionals. Additionally, a balance between regulation and open access to consumers will need to be achieved. This will ensure that the results are reliable without

hindering consumer ability to access their genetic data. The major potential improvement in the future of the DTCGT industry is developing a way to incorporate genetic data into EHRs. If genetic data can reliably be transferred to EHR systems, patient treatment can be more informed and targeted.

Overall, the DTCGT industry has provided an exciting opportunity for consumers to learn more about themselves through their genome. However, the current level of genetics knowledge and the many concerns regarding this industry have left many dubious of these services and the information they provide to consumers. As more is uncovered through genetics research, the utility of these tests is expected to greatly improve. Additionally, if the current concerns are addressed and remediated, the DTCGT industry has the potential to become even more widespread. As a whole, genetics is a growing field that has the potential to greatly improve medical care. Since the use of genetics in healthcare is still relatively new and uncommon, it will be exciting to see its impact in the future.

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