

Direct-to-Consumer Genetic Testing and the Legalities, Ethical, and Current Issues

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Submitted to the Graduate Faculty of the
Department of Health Policy and Management
School of Public Health in partial fulfillment
of the requirements for the degree of
Master of Public Health

University of Pittsburgh

2024

UNIVERSITY OF PITTSBURGH

SCHOOL OF PUBLIC HEALTH

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April 26, 2024

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2024

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Abstract

Consumer-driven genetic testing is a newly developed technology that allows consumers to purchase tests online through companies such as Ancestry, 23andMe, etc.; and have results sent to them without the consultation of any clinician. This is growing to be a public health issue with the increased drive and marketing tactics that are used by these companies having led to an increase in consumers purchasing these tests. The willingness of consumers to purchase these tests without the knowledge of what happens to their genetic information can lead to both privacy and data violations. After the results of the tests, there could be increased stress and anxiety, healthcare burdens, and complications that can happen due to individuals seeking out unnecessary appointments and treatments. This then leads to multiple ethical concerns including a risk for false positive or false negative results based on the type of testing and interpretation used. Additionally, privacy and data violations may occur that can affect an individual's genetic information. Unfortunately, this could then lead to individuals having and creating distrust of the science community and can lead further away from the belief of what science can do for the improvement and benefit of health and precision medicine. This paper reviews the current legal and ethical issues with DTC genetic testing and proposes policy solutions to address these current challenges.

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Preface

I'd like to thank my wonderful family for their continuous support as I pursued my Master's degree. Their endless support, guidance, and love; have been priceless throughout this journey. I'd also like to thank the numerous faculty and staff throughout the University of Pittsburgh that I had the pleasure of meeting and connecting with, their support and guidance were immensely valuable. To my friends I have made in my cohort, thank you for a wonderful two years of numerous memories, jokes, and laughter that were shared both in and out of the academic settings.

To my essay readers, Professor Hughes, and Dr. Durst, thank you for your support as I write this complex topic. Your encouragement was the utmost treasured.

Thank you!

1.0 Background on Direct-to-Consumer Genetic Testing

Direct-to-consumer genetic testing is an innovative advancement of technology in the world of genetics. With DTC genetic testing, there are three different types. The three different types are tests that are completely consumer-driven, tests that have medical reviews, and third-party services. Fully consumer-driven testing companies focus on DTC testing kits that are often televised and commercialized so that individuals partake in test-kit ordering, testing, and receiving results without the involvement of their healthcare providers. DTC tests with medical reviews are those that individuals take with some level of provider oversight including their provider or a company-employed physician. With third-party services, individuals usually conduct a DTC test and then upload their genetic testing information/results on third-party websites, which can provide either additional information or help find additional relatives.

Genetic privacy is a term that is used in talking about the importance and need of keeping individuals' genetic information private whether that is from third parties, insurance, workplace, or even with their family and friends.¹³ With modern technology and the increase in modern healthcare delivery, the issue of genetic privacy has risen.¹³ The Health Insurance Portability and Accountability Act (HIPAA) and the Genetic Information Discrimination Act (GINA), were passed to address the potential impact of modern technology on the privacy of individual's health information.¹³

Throughout my essay, I will be discussing the need for more confidentiality, privacy, and security for genetic test information when it comes to DTC testing, as well as the need for provisions to be added to current legislation when it comes to DTC testing. I will also be discussing the importance of the legal, ethical, and policy issues related to this topic.

1.1 Direct-to-Consumer Testing (DTC)

DTC genetic testing has become increasingly popular since its introduction in the early 2000s, with the increasing demand leading many companies to create such products to market to individuals. There are currently more than 30 companies that offer genetic testing through the internet in ways that are testable for both health-related and non-health-related issues.¹⁷ There are numerous critics of DTC and many of their arguments are on the accuracy and quality of these tests and how they are self-mediated.³

The term DTC genetic testing has referred to the profitability, marketing, and sales of a variety of different tests; health, or non-health related.⁸ The way that DTC genetic testing works is that the test gets marketed out, then is ordered by the consumer, and delivered. The individuals then return the test kit with their genetic sample by mail to a laboratory.⁸ The issue with this is that all marketing that goes towards DTC has not been approved and is unregulated, however, some entities can step in regarding this but have yet to.⁸ In addition, while all this is at the consumer's disposal, even the providing of the results of the genetic information is given without a healthcare provider or genetic counselor.⁸

DTC testing often tests for a large number of traits instead of specifically testing for a single mutation or trait.¹⁷ These companies allow consumers to have full autonomy to partake in genetic testing to gain knowledge regarding their genetics, whether it would be current or for the future, which can have either positive or negative results.¹⁷ One identified issue with DTC testing has been the concept that consumers can have little knowledge regarding genetics and biology, and this may cause unnecessary stress and anxiety to consumers when their results are received.¹⁷ In a study that was conducted in which a health technology company recruited 3,640 employees to take a DTC test at a reduced cost, almost half of the individuals had expressed concerns with

the testing.¹⁷ The range of their concerns involved learning of their disease risk, emotions they would feel based on their results, and the privacy issues regarding the data.¹⁷

1.1.1 Advertising

There are many advertisements regarding some form of DTC genetic testing, whether it is from 23andMe, Ancestry, MapMyGenome, etc. On television, numerous commercials also appear with anecdotal stories of individuals who have taken these genetic tests and the joy and happiness they have received from their results. However, these do not address the negative impacts that DTC genetic testing can have or what the fine print on the DTC test kit boxes says. The anecdotes that commercials provide are all supposed to be emotional, light-hearted, and touching in the aspect of capturing and gathering the audience's attention to promote their products and have people use their products without a second thought. Companies, like 23andMe, use great marketing tactics to encourage individuals to purchase their items and submit their genetic information from either a saliva sample or a couple of swabs of their cheek to a variety of different types of information about their genetic makeup such as their ancestry, traits, health predisposition, carrier status, wellness, pharmacogenetics, etc.¹

Advertisements of genetic testing through the media, internet, or physical forms usually tend to have clinical information missing.¹² Sometimes throughout the advertisements they state the validity or the intended usage of those tests, and the results need to be confirmed clinically since they are usually for entertainment purposes.¹² Also, most of the consumers who are using these products have limited to no literacy or understanding of the results that they have received from these companies.¹²

Not only do DTC companies use televised commercials and other methods of advertisement, but so do diagnostic labs such as Myriad. The most common one in the United States was Myriad's advertising campaigns for its BRAC-Analysis test.⁸ Myriad's advertising campaign was aired, advertising a diagnostic test that they owned regarding both breast and ovarian cancer.⁵ In 2002, the advertisement aired which was launched only in two cities.⁸ The advertisement was aired on television, radio, and print media, in which it was quoted that Myriad advertised this to "alert women with a family history of cancer to recent advances in cancer prevention and early disease detection."⁸ The overall goal and main objective of the advertisement was to persuade consumers to go and consult their physician and ask to get the genetic test conducted.^{5,8} Studies were then conducted to see the popularity and impact the advertising campaign had regarding these genetic tests.^{5,8} The data indicated that there was an increased awareness of testing both for providers and patients, an increase in referring low-risk women for genetic counseling services, and an increase in the number of tests that were conducted overall.^{5,8} Then five years later, in 2007, Myriad had another controversial campaign that had launched for a BRAC-Analysis test for the states of New York, Connecticut, Rhode Island, and Massachusetts.^{5,8} Throughout the ads, it promotes the need for women to consult with their physicians and use the genetic tests, and although some physicians had said that this helped promote the awareness of getting tested for BRACA, numerous critics had stated the opposite, that it caused unnecessary stress and over usage of the test.^{5,8}

The overall goal of advertisements is to influence viewers of these commercials to try out their product to gain beneficial results regarding their health or even sheer curiosity of learning more about an individual's ancestry.¹⁸ This is a manipulative tactic used to convince recipients to do these to have autonomy over their health and influence their decision-making with purchasing

their products.¹⁸ A study by Schaper and Schick Tanz¹⁸ utilized a case study approach reviewing three different DTC genetic testing websites (one each from the US, Europe, and Asia) to analyze their advertisement and check to see if there is any communication that seems to be.¹⁸ There are 3 areas of criteria that were used to evaluate the advertisements; whether the material was establishing that there is medical professional reliability, the persuasion tactics of using the services, and the morality of self-conception.¹⁸

Schaper and Schick Tanz¹⁸ presented several images of genetic testing advertisements that they were able to analyze and take a look through. One specific advertisement was in capital letters stating how a viewer's health check is incomplete and that by taking a genetic profile test they can take control of their health.¹⁸ In smaller print, the advertisement states how this genetic profile test can help know the consumer's risk for various conditions such as heart disease, hypertension, diabetes, and obesity and how taking the genetic test can help protect against these conditions before they happen.¹⁸ This show advertisement appeals to the audience by implying that there have been numerous objectives probably overlooked during a normal physical and that they should get a genetic test done to have them take control of their health and aim for preventative measures against the conditions that are mentioned in the advertisement.¹⁸ Schaper and Schick Tanz¹⁸ also emphasize, the usage of the magnifying glass amplifying DNA, in the advertisement, and how that gives almost an "investigative" feel to consumers who may want to take control and uncover a mystery, which in this case would be their genetic information.¹⁸ What is very unique and intriguing about the wording of this advertisement is about how genetic testing should be conducted to take preventative measures, but in reality, preventative measures can be taken even without the need for genetic testing.¹⁸ Therefore, the use of both keywords and imagery persuades and convinces consumers to partake in DTC testing.¹⁸

2.0 Ethical Issues

Issues of ethical issues arose when the US Government Accountability Office (GAO) investigated genetic testing firms that sold directly to consumers and had misleading test results.²² The GAO had expressed concerns over the fact that these companies explicitly stated that they had to receive consent from consumers at first, however to a fictitious consumer that was planted by the GAO, stated that when they asked the companies if she could send her fiancé's DNA in to secretly surprise him with results, the companies had agreed for her to do that.²² Therefore, this poses both a risk of privacy and ethical concerns to individuals who have no idea who could gather their DNA and then send it to these companies.²²

A concern that has also plagued ethics issues is what would happen to the DNA samples they collected if these DTC genetic testing companies went out of business.²² There is fear that third parties would have access to this DNA, especially because businesses do not explicitly state what happens to the DNA samples they collected.²²

When discussing ethical issues, the four principles of bioethics (Autonomy, Nonmaleficence, Beneficence, and Justice) need to be taken into consideration when talking about DTC genetic testing. Table 1 highlights these four principles of bioethics, their definitions, and their relations to DTC.

4 Principles of Bioethics Table

Principles	Definitions	In relation to DTC
Autonomy	Emphasizes on an individual's right to make informed decisions regarding their health care.	Making sure that individuals have the knowledge and clear access to accurate information and understanding all that entails genetic testing such as risks, benefits, and limits...but also having consent.
Beneficence	Healthcare professionals/institutions have to act in the best interests of their patients	This is about making sure that there are valid and accurate test results and avoiding unnecessary distress or harm based on these test results. It can also include making sure that genetic data does not become misused.
Nonmaleficence	Healthcare providers must not harm their patients.	This can include providing care to help overall health outcomes, such as offering genetic counseling and services to help individuals understand and interpret their results.
Justice	When there is distribution of healthcare resources, there is a fair and equal care level	Making sure that everyone has access to genetic testing and also advocating policies that protect against genetic discrimination, but also valid and accurate results.

Table 1

Taking a look through all four principles of bioethics, their definitions, and how they are related to DTC, can help emphasize the ethical issues that DTC genetic testing has when these are not implicated or are broken.

Individuals who have an interest in taking their health into their own hands through DTC are more than likely to interpret results inaccurately, leading to increased stress and anxiety.¹⁶ This has been seen with individuals who use DTC to identify potential risks of specific conditions they may have.¹⁶ An example with 23andMe, is that they have this with the genetic markers for BRCA which identifies variants that are common in Ashkenazi Jewish individuals, and now have been updated to include more mutations, but certainly not all.¹⁶ In an example given, variants of BRCA to help identify if the condition is present in approximately 2% of Ashkenazi Jewish females and 0.001% of the overall general population. Therefore, based on these statistics, the results would miss the BRCA variant in about 80% of the population and can lead those who have received negative results outside the Ashkenazi Jewish population to have false results about their risk.¹⁶ What this means is that when variants of this condition are found in 2% of Ashkenazi Jewish females, but only 0.0001% in the overall population, therefore these mutations are much rarer.¹⁶ Since there is a much rarer chance of the general population having these variants, the DTC genetic tests may miss detecting this variant in about 80% of the population.¹⁶ Therefore, since it may miss detecting these variants, it can lead to an increase of false negative results.¹⁶ This continues to be a concern and has been a concern for older versions of these tests but highlights how there is a chance for false results when it comes to DTC testing.¹⁶

3.0 Privacy Issues

Privacy policies for genetic testing companies help consumers understand what protections they have when submitting a sample and how their data will (or will not) be protected.²¹ 23andMe is a popular DTC genetic testing company that Sklar²¹ discusses when talking about the privacy policies of these companies.²¹ Sklar²¹ states that the company, 23andMe, explains “23andMe will not sell, lease, or rent your individual-level information to any third party or a third party for research purposes without your explicit consent”.²¹ However, then if the individual does not consent to the research in which 23andMe participates, the website states “Genetic Information and Self-Reported Information may still be used by us and shared with our third-party service providers”.²¹ Therefore, the company does have the right and the ability to share the information of their samples and also what information would be shared as well.²¹

In a study conducted by Skeva et al²⁰, the authors reviewed 22 companies and their databases’ policies that showed what information they particularly seek out, to whom they share it, and what their overall policies are.²⁰ The results of the 22 companies state that the data information can be accessed by law enforcement and can be shared.²⁰ However, this does raise a concern when law enforcement does not necessarily follow official procedures and can access this data.²⁰ Law enforcement officials could pose as consumers and submit a sample to see if any matches can be hit on the database.²⁰ For example, a popular genealogical database in which people can upload their DNA raw data is called GEDmatch, a site where individuals can upload their DNA information to find their relatives, and anyone can access and find individuals through this site.²⁰

When it comes to privacy, consumers should have the knowledge and the right and the ability to know to whom their information is being shared and how.²⁰ Since this creates distrust and fear in DTC genetic testing and research, it is crucial for these companies to create transparency. This enables the company to help promote research and data sharing while also meeting the needs of the consumers to get what they originally had conducted the tests for.²⁰

Data privacy is a huge concern when it comes to the focus of the disclosure of privacy, third-party usage of the data, and confidentiality. In 2017, Laestadius et al¹¹, conducted a framework analysis with 30 DTC genetic testing companies and whether or not they explicitly state what is done to consumers' genetic testing and their samples.¹¹

The Laestadius et al¹¹, framework analysis takes a look at the 30 DTC genetic testing companies and states what is written in the privacy laws of their tests. ¹¹ Of the 30 companies that were analyzed, 20% of them had discussed the specific risks related to the disclosure of results from purchased services to third parties such as employers or insurers, which leaves 80% of the companies not having discussed this at all.¹¹ Similarly, when assessing the discussion of research usage and if/how the company plans to use this genetic data for it to be health-related research, only 3.3% of the 30 companies discussed this, and 3.3% partially discussed this, which leaves about 93.3% failed to discuss either at all or completely.¹¹ There is a difference between an individual who submits their genetic information after they've read the full disclosure and still decided to go through with it and another when consumers have no idea that this is even a possible outcome.¹¹ When it comes to the specific length of time for storage of the data for research, all 30 of the companies did not disclose this information.¹¹ In addition, only 40% of the companies had disclosed the plans of using genetic information for something that is unhealth-related.¹¹ This is all very interesting but disturbing to know because this indicates that consumers who are using

these DTC genetic testing products are likely not aware of what is exactly happening to their genetic information.¹¹ The overall common theme is that there is limited transparency and more clarity is needed regarding having consumers acknowledge what they are signing up for.¹¹ Laestadius et al¹¹ ., mentioned how even though companies would mention the disclosure to insurance companies, they fail to mention that the Genetic Information Nondiscrimination Act (GINA) fails to cover discrimination by insurance providers for life, disability, and long-term care insurance.¹¹ The need for increased protection of consumer's genomic data is a crucial need and privacy needs to be a must. At a minimum, having transparency about privacy policies is essential to inform consumers about what their data would be used for once it is submitted.¹¹

4.0 Genetic Exceptionalism

Throughout the expansion of genomics and policies regarding genetic policy, genetic exceptionalism is a term that refers to how genetic information is special or unique.⁴ This viewpoint leads to the overall concept and need that there should be specific policies that are required for genetic testing and genetic information to keep up with the constant updates and modifications of using genetic-related technologies whether that is in research, fun, or clinical care.⁴ Numerous scientists have argued that since genomic information is considered special and unique, there should be more policies and regulations placed besides GINA, to have special protections against genetic discrimination and need special protections.⁴

There have been numerous arguments and cases that have emerged regarding the idea that genetic information is special/unique.³ These arguments tend to be on how to fix the important considerations of whether genetic tests and the results from these tests should be handled differently than other types of medical information.³

Genetic exceptionalism began to appear in debates in the 1990s, regarding specific regulation of relevant medical tests and health information because of the risks of discrimination against patients.¹⁹ Genetic exceptionalism was debated due to the notion that genetic information is unique, however, all arguments that were in favor of this view were dismissed.¹⁹ In 1993, the Task Force on Genetic Information and Insurance talked about how genetic information is not important to the patient's future, in terms of its importance to other family members, or in terms of possible stigma within genetics.¹⁹ Scholars, *Lawrence Gostin* and *James Hodge*, legally agreed with what the Task Force had decided, but they did state that risks are being posed by the use of genetic information in a nonmedical way by educational institutions, the police, employers, etc.¹⁹

Garrison et al^{4,19}, criticized the term genetic exceptionalism and wanted to introduce the new notion of genomic contextualism which replaced genetic exceptionalism and started with the types of stigma and exclusion that could be associated with genomic tests.^{4,19} This is stating how guidelines, meticulous or specific, should be proposed for them since some tests and counseling situations could pose ethical concerns.¹⁹

There have been attempts at creating special legal protections for genetic information, however, there is criticism when classifying genetic information as unique. Numerous critiques have suggested that this brings forth ethical, social inclusion, and stigmatization issues when the overall goal is to prevent these issues from arising. When considering and implementing new policies, geneticists, and policymakers need to work together to ensure that there are proper privacy laws put into place to protect consumers, but also not create issues and further stigmatize individuals, when we are currently trying to put a stop to it for the ever-continuous growth of modern genetic technology.

5.0 Current Legal Landscape

Internationally and in the United States, there have been numerous current legal changes that have been made that impact genetic testing, and more specifically DTC genetic testing. However, there are differing definitions and perceptions of what exactly genetic privacy and discrimination are, as well as the impacts they have on politics and social issues.¹⁰

5.1 Legal Protection

5.1.1 Health Insurance Portability and Accountability Act (HIPAA)

The Health Insurance Portability and Accountability Act (HIPAA) was enacted in 1996 to protect health information and keep patient-doctor confidentiality but also so that only the patient individually has the right to access their health information and no other individual unless they were given responsibility.¹⁴ Unfortunately, even though employees hold several, training events throughout the year, HIPAA violations are still occurring by the employees that are handling this protected data.¹⁴ Five titles within HIPAA contribute to the Act to maintain the confidentiality of medical records of patients.¹⁴

The first title of HIPAA focuses on the ability of healthcare coverage to continue and transfer for American families who either lose or change their jobs but also prohibits the ability of healthcare coverage to be denied because of preexisting conditions.¹⁴ Title Two of HIPAA focuses on the need to help establish electronic transactions and identifiers for providers, health plan

providers, and employers to prevent fraud and abuse but also to enforce more health security and privacy.¹⁴ The third title revolves around the change of health insurance laws and deductions for medical insurance, while the fourth title talks about the specific conditions on group health plans about coverage for individuals with preexisting conditions.¹⁴ The last title focuses on company-owned life insurance and the treatment of individuals who are not US citizens.¹⁴

The common title that is most familiar to everyone is the privacy law regarding the protection and security of patient's medical records. There have been numerous talks regarding changing and implementing new regulations to help maintain the safety of health records, however, they have been deferred by the Department of Health and Human Services.¹⁴ With that, there are exceptions to the privacy rule by HIPAA in which protected health information (PHI) could be released without authorization.¹⁴ Providers can share private health information with companies they bill to receive money from the services that they have to provide, another is for public health activities they have to report to prevent or control the spread of diseases and also in request by court orders as well.¹⁴

HIPAA states to only protect the information of patients through a covered entity.²¹ Covered entities can be defined as hospitals, nursing facilities, clinics, etc.²¹ Therefore, clinical genetic test results are protected by HIPAA, but DTC testing companies are not considered covered entities, and therefore the results they generate are not protected by HIPAA.²¹

5.1.2 Genetic Information Nondiscrimination Act (GINA)

In 1995, the first federal legislation ever presented to prevent the misuse of genetic information was introduced.⁹ At this time, only about 300 genetic tests existed and they were most specifically only used for very rare diseases and conducted in research-supervised areas.⁹

However, this had sparked worry among Americans, that now health insurance companies would be able to use their genetic information, either to increase premiums or in the workplace as well.⁹ This debate lasted for about 13 years, until May 21, 2008, when the Genetic Information Nondiscrimination Act was passed.⁹ The Genetic Information Nondiscrimination Act is a federal law that protects unaffected individuals from discrimination by either employers or health insurance based on their genetic information.⁹ The goal of the act is to prevent health insurance companies from accessing and using an individual's genetic health to raise premiums or even determine eligibility for coverage.⁹ Similarly, this prevents insurance companies, from either requiring or requesting their customers undergo a genetic test because as mentioned it can be used to influence their eligibility for their insurance or raise their premiums.⁹ Also, the Genetic Information Nondiscrimination Act prevents employer discrimination based on genetic information.⁹ This prohibits employers from accessing and using a person's genetic results and information to make decisions when it comes to employment such as hiring, firing, job responsibilities, or anything else that is about employment.⁹ In addition, employers are prohibited from requesting, requiring, or simply purchasing any genetic information testing and asking for their employees and/or asking for their family members to take them.⁹

However, several flaws came out of this Act.⁹ GINA did not mandate that health insurers cover any specific genetic tests, so if an individual wants to get a genetic test completed that is not covered by their insurer, they may have to pay out of pocket for that test.⁹ In addition, GINA does not apply to the members of the military.⁹ Since GINA was enacted in 2008, which was approximately 16 years ago, the evolution of genetic testing and information has grown. Another flaw that comes with GINA is that it does not prevent discrimination from individuals who already are affected by a genetic condition.¹⁰ Therefore, a GINA violation can happen if the insurer decides

to not give health insurance to an individual who has an increased risk for a disease but is asymptomatic.¹⁰ The moment that an individual is symptomatic for the conditions, GINA would no longer be applied to them.¹⁰ In 2010, the Patient Protection and Affordable Care Act (ACA), prohibits any discrimination in health insurance for any conditions, therefore prohibiting discrimination for any health-related reasons and extends to individuals with pre-existing conditions.¹⁰

5.2 Regulations in the United States and Internationally

5.2.1 State Level in the U.S.

States in the US have passed privacy laws for citizens to gain better privacy protections that protect individuals' genetic information.⁸ States also can dictate whether healthcare provider authorization is required to order a laboratory test, while others allow laboratories to accept samples without authorization from a healthcare provider.⁸ Some states that prohibit DTC genetic testing may also have difficulties when it comes to not allowing the sales of DTC testing to consumers, especially with the easiness of ordering through the Internet.⁹ New York and Maryland are states that prohibit all DTC testing, while 25 states + D.C. allow DTC laboratory testing without any restrictions, and 13 states prohibit it.⁸ In 2021 Consumer Data Privacy Legislation, only seven states had brought forth legislation to protect consumer genetic information, and all of them had passed.⁶ In 2022 with Consumer Data Privacy Legislation; Kentucky, Maryland, and Wyoming were all states that had enacted consumer genetic privacy laws for their states.⁷ In 2023, Minnesota, Montana, Tennessee, Texas, and Washington all enacted Genetic Privacy Protection in their states.

In all 50 states, there is at least 1 statute that prevents some form of genetic discrimination from health insurance companies.^{10,15} As mentioned, consumers are increasingly becoming worried about their test results and whether the results of their tests could be used or shared in ways they never would have known. Some provisions that were added include the requirement of the company's policies and procedures to notify consumers and also state the usage and disclosure of their genetic material before receiving consumers' consent to collect their genetic data.^{6,7,15} This also includes the prohibition of insurance companies from using consumer genetic information, however, this does not include any legislation with regards to prohibiting discrimination based on genetic information collected by health providers.^{6,7,15}

5.2.2 Federal Level in the U.S.

At the statutory level, the federal government has limited oversight of the laboratories that conduct genetic testing.⁸ The Centers for Medicare and Medicaid Services (CMS) has implemented and enforced the Clinical Laboratory Improvement Amendments of 1988 (CLIA).⁸ This applies to every laboratory that operates in the United States.⁸ This statute prohibits the acceptance of any "materials that have derived from the human body for laboratory examination or other procedure" unless the CMS or other authorized entity gives the laboratory a certificate.⁸ This is very interesting considering that DTC doesn't necessarily have to be done in a CLIA lab if it is for entertainment purposes, as most consumer-driven are...however many labs are CLIA. Even with modern technology and the advancement of genetic testing, CMS has denied any petition of issuing regulation and has caused harm when it comes to DTC testing.⁸ This also leaves consumers not knowing whether their results have come from a lab that is CLIA certified and has done the necessary proficiency testing or has passed inspection.⁸ This is public information; however, it is

currently backed up and is more than two years out of date.⁸ Therefore, it is really difficult to even know if the test results and analytics are valid.⁸

In recent years, 23andMe has been working with the Food and Drug Administration (FDA), to undergo a premarketing review of their tests.² In 2013, however, the FDA sent a letter to the FDA regarding a cease-and-desist letter telling the company that they did not have any approval for them to offer any interpretation of their health-related genetic tests to new consumers.² The FDA had not been convinced regarding the test data and their validation, analytically and clinically.² After this, 23andMe then launched a \$5 million advertising campaign, first appearing on As Seen on TV.² They launched and put together a stronger regulatory team and launched a regulatory strategy to promote its Bloom Syndrome carrier screening test.² This set the stone for marketing and was able to show analytical and clinical validity to the FDA since this is a carrier screening test and does not provide consumers with a disease diagnosis or information about how the drug may impact their lives.² To receive approval for carrier screening or susceptibility tests, the FDA requires that there is a demonstration of genetic understanding before consumers can acknowledge and access their genetic information.²

To satisfy the needs of the FDA, 23andMe submitted information about the collection device, software, and instruments used, as well as how the test's analytical performance was conducted.² Meeting the clinical and validity requirements of the tests helped fulfill the regulatory controls that the FDA had allowed for 23andMe to launch their Bloom Syndrome carrier screening tests.² The FDA has also stated for over-the-counter DTC, that the manufacturer has to provide information on how they can find a qualified professional genetic counselor. The companies must also label the limitations of this test and include warnings that these tests are not considered as substitution from visiting healthcare providers, these tests don't diagnose a health condition, the

lab may have difficulties processing samples, and ethnicities may affect how genetic health results are interpreted.²

5.2.3 Internationally

When taking a look through international policies and their regulations and laws regarding DTC genetic testing, it is interesting to see the differences throughout all regions as shown below in Table 2.

INTERNATIONAL POLICIES WITH DTC

Region/Location	Countries	Laws/Regulations
European Union	Belgium, France, Italy, Germany, United Kingdom	Provisions were added to the Charter of Fundamental Rights of the EU (2000) and the Convention on Human Rights and Biomedicine (1997). The provisions added were about insurers and how they are not allowed to require genetic tests for insurance purposes and genetic info, that exists from family members is not allowed to be processed by insurers. Laboratories have voluntary accreditation and get inspected every five years.
Asia	South Korea, Taiwan, India	South Korea has applied regulations prohibiting discrimination when it comes to a person's genetic components, and this applies to education, insurance, and employment. Taiwan passed the Personal Information Protection Act in 2012. India has acts that apply to individual data and insurance companies cannot have exclusions that are about genetic disorders.
South America	Chile, Argentina	Chile has regulations regarding clinical settings, but nothing when it comes to education, insurance, or employment. Argentina discusses the prohibition of using genetic information for discriminatory purposes but does not include either physical or blood tests.
Middle East	Israel	DNA testing is required in Israel for couples to either seek legal marriages or citizenship applicants. They are the only Middle Eastern Country to have anti-genetic discrimination legislation and prohibit insurers from requesting genetic testing or even asking if their customers have done genetic testing.
Africa	Morocco and Tunisia	<i>"Policy Requirements, Procedures and Guidelines for the Conduct and Review of Human Genetic Research in Malawi."</i> This states how genetic information poses risks to harmful consequences in social life such as education, health insurance, and immigration, and that this could lead to further separation and discrimination for individuals and families.

Table 2

Author's analysis of international policies in Joly et al.

Overall, throughout the regions, it is seen that most of them have a common theme, in which insurers are not allowed to either ask their customers to take a genetic test or use either family or users' genetic information and put it against them to raise their premiums. In addition, we do see that in some countries, anti-genetic discrimination is applied and expanded to even education, employment, and insurance, too.

5.3 Gaps and Relation with Claims Regulation

5.3.1 Federal Trade Commission

The Federal Trade Commission prevents “unfair or deceptive acts or practices in or affecting commerce”, and this prohibits the spread of false claims and false advertisement when it comes to medicine, devices, food, or cosmetics.⁸ DTC genetic testing companies state that they have validity without actual scientific evidence and indeed promote false advertisements, but yet the Federal Trade Commission has not brought any legal action against these companies in this regard.⁸ They are within their rights to do so, however, they have yet to bring forth legal authority, even when they have received complaints about specific tests.³ What the Federal Trade Commission has done, however, was send out a warning to consumers about claims these companies make and advertising that they have, but yet they never have conducted legal action against these companies.⁸

5.3.2 Congress

The overall goal of the United States Congress is to not only help draft legislation but also conduct investigations and hearings over deceptive practices.⁸ In 2006, the Senate Special Committee on Aging had a hearing regarding a report by the Government Accountability Office (GAO) regarding companies that offer nutrigenetic tests over the Internet.⁸ The GAO investigated four companies that submitted DNA samples with fictitious profiles and analyzed the reports from the companies.⁸ They found that all four companies said they were not able to diagnose diseases, but they sent them back stating that the fake consumers were at risk for type 2 diabetes, cancer, heart disease, osteoporosis, and brain aging.⁸ These companies then recommended dietary supplements that had been “personalized” for them, but the GAO had found out that they were vitamins that were normally purchased at drug stores, but costed way more.⁸ However, the FDA sent out numerous letters to several of these companies to subject them to the FDA regulation, however, it is not known what has transpired, because as of right now these companies still offer their DTC testing.⁸

6.0 Risks Associated with Direct-to-Consumer

There are numerous risks posed and associated with consumer-driven DTC genetic testing. There is overall fear and weariness when it comes to the overall accuracy, validation, and interpretation of the results of the tests. Receiving inaccurate results causes unnecessary anxiety, stress, and healthcare spending because this could lead individuals to seek care and treatments that may not be needed, especially in the case of a false positive result. In addition, there is the risk of privacy breaches and insufficient knowledge of individuals who submit their DNA samples about what these companies do with the samples. There are not any known laws or regulations that specify what these companies are allowed to do with samples that are submitted. They could be kept in the lab forever, can be sold, used in research that is not known to the consumers, etc. This raises issues of privacy for individuals but can also create confusion and conflict for those who are unaware of these predicaments. A great way to help address these risks is by implementing stronger regulatory committees within these DTC companies to help strengthen the accuracy, validation, and interpretation of the results of these tests.

As mentioned, since these genetic tests are consumer-driven, there are often no clinician aides, help, or interpreters when it comes to reviewing the results of these tests. To help stray away from incorrect interpretations, access to genetic counselors should be proposed in legislation, or by companies to help consumers address the results that they receive. Overall, there is a concern and risk of the possibility of inaccurate/invalid results and misinterpreting results that can lead to a domino effect on the consumer's plan of action. Taking these DTC tests at home without the referral of a provider is convenient, however, what is not taken into account is also the risks that can be associated with that, which has been mentioned.

7.0 Reform/Policy Options

Numerous considerations can be taken into account when suggesting policy options or reformation. One I would implement would be the need and usage of informed consent. I think it is very important for these consumer-driven companies to explicitly have written out everything regarding their product, the risks, conflicts, stating the possibility of inaccurate results, suggestions of consulting their PCPs regarding the results, and overall, what the usage of their samples will be. This could go about spreading the overall education and awareness of what these tests are, and what it necessarily means to submit DNA samples to these companies. Raising awareness and education can help individuals not get blindsided by what is happening to their information but also plan and know the proper resources to seek out help to get accurate interpretations of the results. In addition, raising awareness and education could also lessen fears that individuals may have about submitting their DNA samples and can also help contribute to research and allow for studies to help gain more information that can be utilized towards precision medicine.

A suggestion that could be implemented would be the requirement for genetic counselors to be provided based on the results of individuals. With numerous of those who will receive their results not being proficient in genetic literacy, there must be genetic counselors would be available to help with the interpretation of results, and also help with what would be the next course of action.

There could also be further strict regulations when it comes to data privacy and the protection of individuals' DNA when it comes to them submitting their information to these companies. With these regulations, it is important to include that companies are required to explicitly state what the DNA would be used for, if the information would be sold and to whom,

how long would the information be stored in their database, how long the sample would be kept, and providing genetic counseling. With these restrictions, I believe that consumers would feel more comfortable but also protected regarding their genetic information.

GINA could also have provisions added to which there would be changes on the definition of genetic testing, what qualifies as a gene test, and the need to have privacy regarding the tests and their results. This law was created in 2008, at a time when consumer-driven genetic tests were yet to be on the rise or have gained as much popularity as they have currently. With the ever-surging growth of DTC, the need to go back and revise GINA is a must and needs, to accommodate today's current events. There should be an establishment of genetic laws or data privacy laws that consist of these genetic laws to be updated every several years, to keep up with current events. Perhaps a committee can be created at a governmental board, that is full of expert geneticists to help aid policymakers in making sure laws/regulations are kept up to date.

8.0 Conclusion

Overall, when it comes to consumer-driven DTC, numerous issues arise regarding the gaining popularity and advertisement constantly through commercials, social media, billboards, flyers, stores, etc. With the ever-growing marketing growth of such products, it is drawing in consumers to conduct these tests to fulfill their own needs, which can then lead to a backfire of ethical and privacy issues. DTCs are not fully accurate with their results and therefore have a shaky validity, which can lead to false positives, and have consumers be in fear, anxious, and stressed regarding their results and can place an emotional burden but also a financial burden since they would more than likely be seeking out health and clinical care when these appointments and treatments would be unnecessary. In addition, with regards to privacy, as mentioned throughout, DTC companies rarely state what they do with the samples that they receive from consumers, therefore causing further unease to individuals on what is done. Some limited legal efforts and provisions are currently being made to protect an individual's genetic information, which leads to the idea and criticism of genetic exceptionalism and that the treatment of genetic information can be debated as considered different than other medical information. To improve and suggest policies to help protect the privacy of individuals' genetic information, there should be strict regulations regarding data privacy and informed consent. Requirement of genetic counselors should be present after each distribution of genetic results and education/awareness of what is happening when it comes to these tests. Also, there should be provisions added to GINA that can accommodate modern technology and a committee full of well-versed and experienced individuals who can update these laws/regulations as modern technology develops. Genetic policy is

unfortunately very new and yet limited to none has been done, but one step at a time will go a long way in the future.

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