VOLUME XIV, ISSUE 2 SPRING 2020

Articles

AMERICAN UNIVERSITY
WASHINGTON COLLEGE OF LAW

* * *

AMERICAN UNIVERSITY
HEALTH LAW & POLICY BRIEF
VOLUME 14 • ISSUE 2 • SPRING 2020

TABLE OF CONTENTS

Articles

* * *

2019-2020

Editor-in-Chief
MIKA SHARPE

Executive Editor
ERIN DONNELLY

Managing Editor
SHELBY TAYLOR

Marketing Director
ELIZABETH RATERMAN

Symposium Director
COURTNEY THOMAS

Articles Editors
ELLE BAILES
REGINA DESANTIS
ANDREW FLORA
KATIE WAHL

Staff
JAMES CAMPBELL
CALE COPPAGE
ALICIA ESPINOSA
NINA FAINMAN-ADELMAN
KATHERINE FREITAS
LAUREN HEIDE
SARAH KNARZER
TED NORTH
SARAH SPARDY
MARIANA TERAN
MATTHEW THOMAS
JULIO TORRES
CAROLINE TRABUSCO

Faculty Advisor
LINDSEY WILEY

* * *

Letter from the Editors

Dear Reader:

On behalf of the Editorial Board and Staff, we proudly present Volume 14, Issue 2 of the *Health Law & Policy Brief* (HLPB). HLPB is an online publication run entirely by law students at American University Washington College of Law (WCL). Since its formation in 2007, HLPB has published articles on a wide variety of topics in the areas of health law, food and drug law, and emerging health technologies. Such topics include health care privacy and data security, health care fraud and abuse, medical malpractice, bioethics and regulation of human subjects research, global health law, and others. HLPB also maintains a blog on emerging health law issues that can be found on our website at www.healthlawpolicy.org. Furthermore, HLPB organizes an annual symposium on an emerging health law topic featuring distinguished speakers.

Volume 14.2 features a novel article titled *The Clinical and Economic Impact of The Early Detection and Diagnosis of Cancer* by Dr. Peter J. Deckers, Dr. Richard Manning, Ms. Tricia Laursen, Ms. Stacey Worthy, and Ms. Shruti Kulkarni. The authors advocate for an adoption of the "*Early Detection and Diagnosis*" (EDD) method that would increase early detection rates of cancer by teaching patients to self-assess and accurately describe their symptoms, which would then enable the practitioners to make earlier and more accurate diagnoses. The authors analyze the EDD's potential economic and clinical impact and challenges associated with its adoption in the United States.

Our second article, written by a WCL student, Juan Pablo Sarmiento Rojas, analyzes the potential privacy concerns associated with Direct-to-Consumer (DTC) Genetic Testing. Rojas considers current privacy laws in the United States and Europe and proposes changes to the existing legislation that would provide greater privacy protection for consumers and increase oversight and regulation of DTC genetic testing companies.

We would like to thank the authors for their hard work and cooperation in writing, researching, and editing their work. We would also like to thank HLPB's article editors and staff members who worked diligently on this issue. Their efforts are greatly appreciated, and we are proud of their work.

For more information about HLPB, or for questions on how to subscribe to our electronic publication, please visit our website at www.healthlawpolicy.org.

Sincerely,
Mika & Erin

Mika Sharpe
Editor-in-Chief

Erin Donnelly
Executive Editor

* * *

The Clinical and Economic Impact of the Early Detection and Diagnosis of Cancer

Peter J. Deckers,[*] Richard Manning,[*] Tricia Laursen, [*] Stacey Worthy,[*] and Shruti Kulkarni[*]

Many detectable cancers are often undiagnosed, misdiagnosed, or diagnosed too late. Recently developed early detection technologies can be costly and inaccessible. In contrast, low-cost, practical solutions are needed now. This article advocates for "Early Detection and Diagnosis (EDD)," a method where practitioners 1) teach patients to self-assess abnormal symptoms, see their practitioner promptly, and accurately describe symptoms; and 2) make diagnoses in response. Despite EDD's effectiveness, barriers including low health literacy, symptom misinterpretation and minimization, and inadequate patient-provider communication have impeded wide adoption in the United States. Widespread education and awareness efforts, including state legislative and regulatory activity, can overcome these barriers.

This article explains EDD's clinical and economic impact and challenges associated with its adoption in the United States. It recommends state legislatures enact laws requiring that 1) medical boards develop guidelines on how providers can train patients to detect cancer signs early and promptly seek a diagnosis; 2) medical boards offer continuing medical education courses on the training guidelines; and 3) state health plans cover these services. These actions make patients more likely to detect their cancer symptoms and promptly report relevant information to their provider. In turn, providers will be able to make an earlier and more accurate diagnosis.

* Dr. Deckers serves on the board of directors of 15-40 Connection, a 501(c)(3) not-for-profit organization whose mission is to educate and empower people about early cancer detection. Dr. Deckers also serves as a surgical oncologist at *UConn Health* and as Professor of Surgery and Dean Emeritus at *UConn School of Medicine*.

* Dr. Manning is a Partner in the Life Sciences Practice at Bates White, LLC in Washington, DC. He provides consulting services and expert testimony on matters relating to economics for clients in government, law firms and other private enterprises.

* Ms. Laursen is the President of 15-40 Connection, a 501(c)(3) not-for-profit organization whose mission is to save lives by teaching people how to detect cancer early and be active partners in their healthcare.

* Ms. Worthy is a partner at DCBA Law & Policy LLP in Washington, DC. She provides counsel to not-for-profit health policy and patient advocacy organizations, health care programs and providers, and pharmaceutical and biotechnology companies in various aspects of their legal, regulatory, and business affairs.

* Ms. Kulkarni is an associate at DCBA Law & Policy LLP in Washington, DC. She provides counsel to pharmaceutical, biotechnology, and medical device companies. She offers insight on federal and state legal, legislative, regulatory, and enforcement matters.

TABLE OF CONTENTS

INTRODUCTION

Cancer is currently the leading cause of life years lost in the United States, and many easily detectable forms of cancer are often undiagnosed, misdiagnosed, or diagnosed too late, leading to avoidable deaths.[1] Moreover, cancer is one of the most expensive diseases to treat and can result in considerable financial burden.[2] Patients and their families can experience financial hardship from high out-of-pocket expenses, which can significantly diminish quality of life and even interfere with delivery of quality care.[3]

Ahead of the 2020 presidential election, improving the detection and treatment of cancer and lowering the cost of health care have been two bipartisan focal points for elected officials and presidential candidates alike.[4] In June 2019, former Vice President Joe Biden stated that he wanted to "cure cancer" during his presidency.[5] Similarly, in March 2018, President Trump called for Americans to speak to their health care providers to learn more about cancer prevention measures that can save their lives.[6] As the President encouraged various stakeholders, including government agencies, to increase awareness to help more Americans survive cancer, the American people have called on the government to lower health costs.[7] Twenty-eight percent of respondents in a recent Kaiser Family Foundation poll reported that lowering health care costs should be a top health care priority for elected Democratic officials.[8] While these two goals are seemingly at odds with each other, it is possible to achieve them both at once. State legislators and medical boards have the opportunity to put policies in place that encourage low cost, practical solutions to detect and diagnose cancer earlier, thereby improving care and saving the health system money.

Diagnosing cancer at earlier stages, before it metastasizes, can significantly increase survival rates and reduce costs to both the patient and the general public.[9] Yet, while

---

[1] *Years of Life Lost*, Nat'l Cancer Inst., https://progressreport.cancer.gov/end/life_lost#field_additional_information (last updated Feb. 2019); Cancer Research UK, Saving Lives, Averting Costs: An Analysis of The Financial Implications Of Achieving Earlier Diagnosis Of Colorectal, Lung And Ovarian Cancer (2014), https://www.cancerresearchuk.org/sites/default/files/saving_lives_averting_costs.pdf.

[2] S. Yousuf Zafar & Amy P. Abernethy, *Financial Toxicity, Part I: A New Name for a Growing Problem*, 27 Oncology 80, 80 (2013).

[3] *Id.* at 81.

[4] Shefali Luthra, *Promising to Cure Cancer is Easy Politics. The Science is Much More Difficult,* LA Times (June 22, 2019, 7:00 AM), https://www.latimes.com/science/la-sci-cure-cancer-politicians-science-20190622-story.html.

[5] Tal Axelrod, *Biden says as president he wants to 'cure cancer,'* The Hill (June 11, 2019, 3:04 PM), https://thehill.com/homenews/campaign/447982-biden-says-as-president-he-wants-to-cure-cancer.

[6] *President Donald J. Trump Proclaims April 2018 as Cancer Control Month*, Executive Off. of the President (Mar. 29, 2018), https://www.whitehouse.gov/presidential-actions/president-donald-j-trump-proclaims-april-2018-cancer-control-month.

[7] *Id.*

[8] Ashley Kirzinger et al., *KFF Health Tracking Poll–June 2019: Health Care in the Democratic Primary and Medicare-for-All*, KFF (June 18, 2019), https://www.kff.org/health-reform/poll-finding/kff-health-tracking-poll-june-2019.

[9] Hyunsoon Cho et al., *When Do Changes in Cancer Survival Mean Progress? The Insight from Population Incidence and Mortality*, 2014 J. Nat'l Cancer Inst. Monographs 187, 187–97 (2014); Zafar & Abernethy, *supra* note 2.

diagnostic tests exist to detect cancer early on, such tests can be costly or inaccessible for certain patients.[10] Additionally, these tests are only available for a limited number of cancers, such as cervix, colon, breast, prostate, endometrial, and lung cancer, even though more than one hundred types of cancer exist.[11] While investors continue to fund innovative technology, practitioners need a low-cost, practical solution that they can use now. One practical solution is a method referred to herein as "Early Detection and Diagnosis" (EDD)—a combination of teaching patients the Three Steps to Early Detection ("Three Steps") and the provider making an early and accurate diagnosis based on the information received.[12] The Three Steps are: 1) establishing a personal health baseline; 2) detecting health changes that last for more than two weeks; and 3) reporting signs and symptoms to a practitioner.[13]

EDD has been widely adopted in the United Kingdom but not in the United States, partly due to barriers such as low health literacy, misinterpretation and minimization of symptoms, inadequate patient-provider relationship and communication, cognitive biases, and insufficient time spent with patients.[14] These barriers can be overcome with widespread education and awareness efforts, including through state legislative activity and regulatory activity by medical boards.

This article explains the clinical and economic impact of EDD. It identifies challenges that patients and practitioners face in implementing EDD. Finally, it proposes legislative and regulatory solutions that promote EDD. These solutions are intended to improve care for the patient while reducing costs to the healthcare system.

---

[10] Susan G. Komen, Understanding Cost and Cost Coverage Issues with Diagnostic Breast Imaging 3 (2019), https://ww5.komen.org/uploadedFiles/_Komen/Content/What_We_Do/Advocacy/komen-understanding-cost-coverage-with-dbi-final-report.pdf.

[11] *Guidelines for the Early Detection of Cancer*, Am. Cancer Soc'y (last revised May 30, 2018), https://www.cancer.org/healthy/find-cancer-early/cancer-screening-guidelines/american-cancer-society-guidelines-for-the-early-detection-of-cancer.html.

[12] *3 Steps Detect*, 15-40 Connection, https://www.15-40.org/3-steps-to-early-detection/ (last visited Mar. 27, 2020).

[13] *Id.*

[14] Thomas E. Kottke, *Overcoming the Barriers to Cancer Screening*, 73 Mayo Clinic Proc., 386, 387 (1998); Minjoung M. Koo et al., *Symptom Signatures and Diagnostic Timeliness in Cancer Patients: A Review of Current Evidence*, 2 Neoplasia 165, 166 (2018). *See generally* Claire Jones et al., *A Systematic Review of Barriers to Early Presentation and Diagnosis with Breast Cancer Among Black Women* 4 BMJ Open 1, 2, 7–8 (2014).

I. BACKGROUND

A. Overview of EDD

EDD allows practitioners to identify cancer at an early stage and administer effective treatment sooner than would have otherwise been possible.[15] There are two generally accepted methods of early detection. First, practitioners can screen asymptomatic individuals for the presence of cancer.[16] Second, patients can identify signs and symptoms that suggest a change to their health, promptly seek care, and obtain a clinical diagnosis.[17] While screening interventions can detect asymptomatic cancer, they are only available for a limited number of cancers and their effectiveness is often constrained by cost and lack of use due to low patient adherence and unwillingness to undergo screening.[18] Given that most patients are diagnosed with cancer after they present with symptoms, this article focuses on the second method of detection—self-identification.[19]

1. The role of the patient

Patients with cancer who are aware of persistent health changes and promptly seek an evaluation of such changes are more likely to receive an early diagnosis and treatment.[20] "Persistent health changes" are subtle changes in an individual patient's normal health that last longer than two weeks.[21] Many forms of cancer are at least subtly symptomatic.[22] Though patients present with different symptoms, some of the most common cancer symptoms include persistent cough or hoarseness, an unexplained lump, unexplained weight loss, change in the appearance of a mole, persistent change in bowel habits, persistent change in bladder habits, abdominal bloating, unexplained pain, extreme fatigue, fever, difficulty swallowing, blood in urine, rectal bleeding, other unexplained bleeding, changes to the breast, or a sore that does not heal.[23]

Not all patients experience the same symptoms, and some may decide not to report experiencing a symptom if the symptom is not one that is commonly associated with

---

[15] *Guide to Early Cancer Diagnosis* 8, World Health Org., (2017), http://apps.who.int/iris/bitstream/handle/10665/254500/9789241511940-eng.pdf;jsessionid=1E6CC8FBDE89690115E632DEB7BCCB1D?sequence=1;%20; *Cancer Screening*, Nat'l Cancer Inst. (last updated Apr. 9, 2018), https://www.cancer.gov/about-cancer/screening.

[16] *Guide to Early Cancer Diagnosis*, *supra* note 15, at 9.

[17] *See 3 Steps Detect*, *supra* note 12 (suggesting that if a patient experiences a subtle change in their health that lasts two weeks or more, they should contact their doctor).

[18] R.N. Battista & S.A. Grover, *Early Detection of Cancer: An Overview*, 9 Ann. Rev. Pub. Health. 21, 22, 39–40 (1988).

[19] Katriina L. Whitaker et al., *What Prompts Help-Seeking for Cancer 'Alarm' Symptoms? A Primary Care Based Survey,* 114 Brit. J. Cancer 334, 334 (2016).

[20] Koo et al., *supra* note 14, at 165–66.

[21] *Use the Two-Week Rule: Know When It's Time to Talk to Your Doctor*, 15–40 Connection, https://www.15-40.org/3-steps-to-early-detection/2-week-rule (last visited Mar. 27, 2020).

[22] Whitaker et al., *supra* note 19, at 334, 336.

[23] Kelly Winstanley et al., *The Impact of Body Vigilance on Help-Seeking for Cancer 'Alarm' Symptoms: A Community-Based Survey*, 16 BMC Pub. Health, 1172, 1173 (2016).

a particular condition.[24] Without practitioner-patient dialogue and ongoing education about EDD, patients may mistakenly rely on common symptoms lists as comprehensive and decide to ignore or not report subtle and persistent health changes that are not listed as a common cancer symptom. This issue further emphasizes the need for improved ongoing patient education and practitioner-patient dialogue.

2. The role of the physician

Once a patient has identified a persistent health change, the practitioner and patient must work together so that the patient receives a timely and accurate diagnosis. Such diagnoses depend on sufficient communication and accurate patient history, patient-reported symptoms, and a physical examination.[25] The practitioner must have "an appropriate index of suspicion" and conduct a clinical evaluation of any screening tests, procedures, and other clinical data before the cancer progresses.[26] Practitioners who are trained to detect less obvious signs of certain cancers are more likely to make an earlier diagnosis.[27] Trained practitioners acting alongside engaged patients can improve care beyond what either could have achieved alone.

II. ECONOMIC IMPLICATIONS OF EDD VS. DELAYS IN DETECTION AND DIAGNOSIS

A. The Direct Economic Impact of EDD

EDD of cancer can significantly reduce both direct and indirect costs to patients and the health system. In 2014, the U.S. health system spent roughly $87.8 billion on direct cancer-related health care, $4 billion of which was spent directly by patients and their families.[28] Such spending included approximately 58% for hospital outpatient or office-based provider visits, 27% for hospital inpatient stays, 12% for prescribed medicines, 2% for home health, and 1% for emergency room visits.[29] Much of this cost is undoubtedly associated with expensive multi-disciplinary treatments for advanced stages of cancer.

---

[24] Tracy L. Finlayson et al., *Assessing Symptoms, Disease Severity, and Quality of Life in the Clinical Context: A Theoretical Framework*, 10 Am. J. Managed Care 336, 336 (2004). *See generally* Minjoung M. Koo et al., *Typical and atypical presenting symptoms of breast cancer and their associations with diagnostic intervals: Evidence from a national audit of cancer diagnosis,* Cancer Epidemiology 140, 140–46 (2017).

[25] Jerome P. Kassirer, *Imperatives, expediency, and the new diagnosis,* 1 Diagnosis 11, 12 (2014).

[26] *Guide to Early Cancer Diagnosis*, *supra* note 15, at 14.

[27] National Academy of Sciences, Fulfilling the Potential of Cancer Prevention and Early Detection 294 (2003).

[28] *The Costs of Cancer: Addressing Patient Costs*, Am. Cancer Soc'y: Cancer Action Network 1, 1–2 (2017), https://www.fightcancer.org/sites/default/files/Costs%20of%20Cancer%20-%20Final%20Web.pdf.

[29] *Id.*

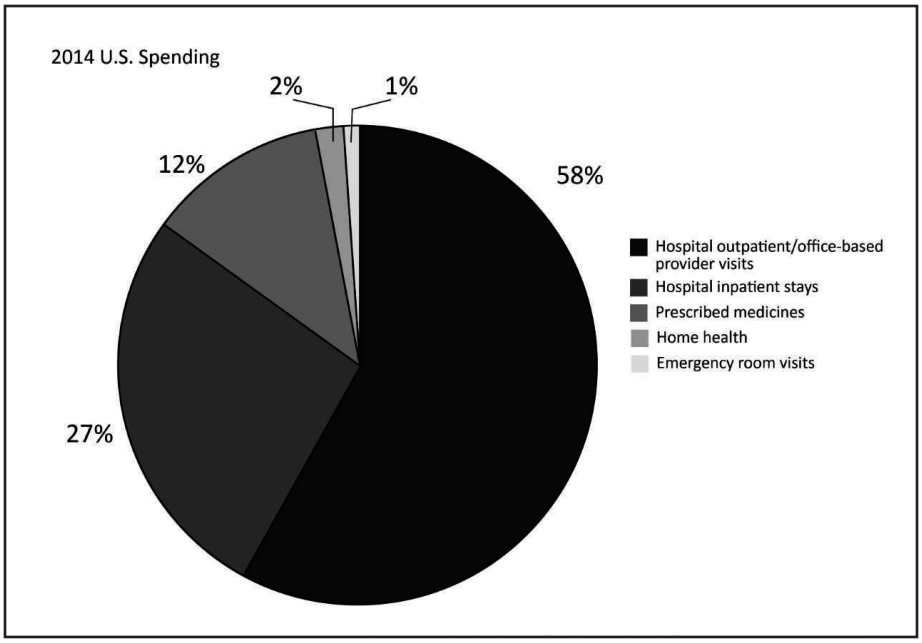

Fig. 1. 2014 Patient costs for cancer related health care in the U.S. Image shows a pie chart depicting the distribution of patient costs for cancer related health care in the U.S. in 2014.

With the cost of cancer care continuing to rise, EDD can ease financial strain and result in significant cost savings.[30] A 2017 study estimated that earlier diagnosis of all cancer types could save an estimated $26 billion annually on treatment costs alone in the U.S.[31] The study also noted that early diagnosis of the top five cancers—breast, lung, prostate, colorectal, and melanoma—could result in cost savings of over $10.7 billion a year.[32] Moreover, early diagnosis may reduce the need for expensive novel drug therapies for advanced stages.[33]

Treatment in later stages of many types of cancer are associated with much higher treatment costs, despite diminished survival rates. For example, treatment costs for lung cancers diagnosed at stage I averaged $7,239 a month, compared to $21,441 for those diagnosed at stage IV.[34] A 2018 study found that costs for the first year of treatment for gastric cancers diagnosed at stage I averaged $8,900, while treatment for such cancers

---

[30] Elizabeth Goss, *The State of Cancer Care 2018*, Nat'l Coalition for Cancer Survivorship (2018).

[31] Zura Kakushadze et al., *Estimating Cost Savings from Early Cancer Diagnosis*, Data 1, 2, 13 (Sept. 4, 2017).

[32] *Id.* at 13.

[33] *See Early cancer diagnosis saves lives, cuts treatment costs*, World Health Org., https://www.who.int/news-room/detail/03-02-2017-early-cancer-diagnosis-saves-lives-cuts-treatment-costs (last visited Mar. 27, 2020) (asserting that the cost of cancer treatment is much less in cancer's early stages).

[34] Thomas R. Gildea et al., *A Retrospective Analysis of Delays in the Diagnosis of Lung Cancer and Associated Costs*, 9 ClinicoEconomics & Outcomes Res. 261, 261 (2017).

diagnosed at stage IV averaged over $25,000.[35] A breast cancer study published in 2017 found that the average per-patient cost during the first two years following diagnosis was $71,909 and $97,066 when diagnosed at stage 0 and I/II, respectively, but rose to $159,442 for a stage III diagnosis and $182,655 for a stage IV diagnosis.[36] Similarly, another study revealed average treatment costs in the first year following colorectal cancer diagnosis to be $49,189, $66,613, $83,980, and $108,599 when diagnosed at stage I, II, III, and IV, respectively.[37] The same study found similar, yet less dramatic, cost trends for breast, prostate, and lung cancer.[38]

B. The Indirect Economic Impact of EDD

Delayed cancer diagnoses also impose substantial indirect costs on society. Although these costs are not as directly observable as the direct costs of care, they are nonetheless real. While the largest indirect costs of cancer are associated with years of life lost and diminished quality of life, EDD may result in improved survival, quality of life, patient experience, and reduced costs.[39]

The potential impact of early diagnosis is particularly illustrative for breast, skin, colon and rectal, lung, and prostate cancers. Symptoms of breast cancer (e.g., nipple discharge) can appear as early as stage I.[40] A patient with a diagnosis at stage I has a five-year survival rate of approximately 99%, compared to 27% during stage IV.[41] Skin cancer also presents with visible changes to the skin in early stages.[42] According to one study, the five-year survival rate for early stage melanoma is 99% compared to 20% at stage IV.[43] The five-year survival rate for early stage colon and rectal cancer is 90% compared to 14% for late stages, 56% for early stage lung cancer compared to 5% in late stages, and 99% for early stage prostate cancer compared to 30% in late stages.[44]

---

[35] Jie-Hyun Kim et al., *Early Detection is Important to Reduce the Economic Burden of Gastric Cancer*, 18 J. Gastric Cancer 82, 86 (2018).

[36] Helen Blumen et al., *Comparison of Treatment Costs for Breast Cancer, by Tumor Stage and Type of Service*, 9 Am. Health & Drug Benefits 23, 23 (2016).

[37] *Cancer Care Spending in California: What Medicare Data Say,* Cal. Healthcare Found. 1, 10 (2015), https://www.chcf.org/wp-content/uploads/2017/12/PDF-CancerCareSpendingMedicare.pdf.

[38] *Id.*

[39] K. Robin Yabroff et al., *Economic Burden of Cancer in the United States: Estimates, Projections, and Future Research,* 20 Cancer Epidemiology, Biomarkers & Prevention 2006, 2006, 2008 (2011).

[40] *Cancer Facts & Figures*, Am. Cancer Soc'y, 1, 10 (2018), https://www.cancer.org/content/dam/cancer-org/research/cancer-facts-and-statistics/annual-cancer-facts-and-figures/2018/cancer-facts-and-figures-2018.pdf.

[41] *Id.* at 11–12, 21.

[42] *Id.* at 23.

[43] *Id.* at 24.

[44] *Id.*

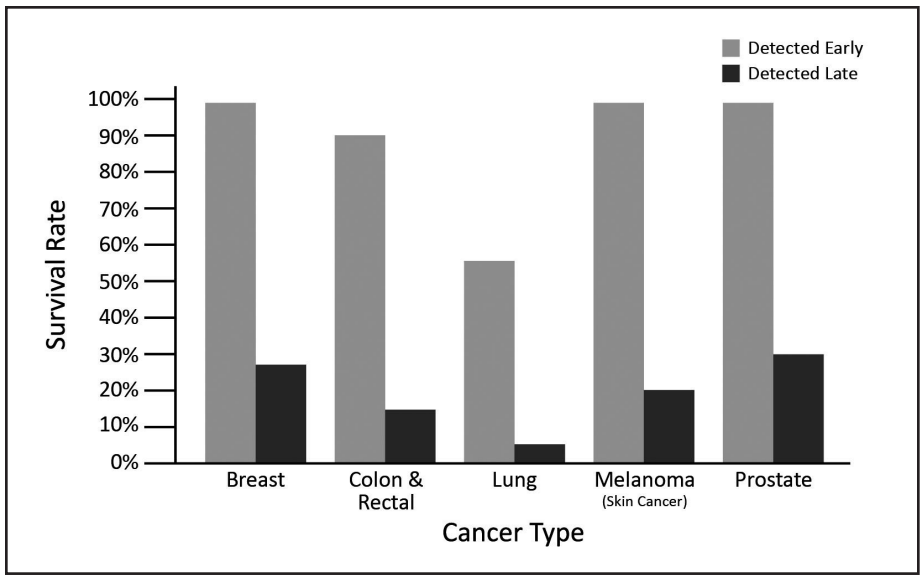

Fig. 2. Comparison of survival rates for cancer that was detected early and cancer that was detected late.[45] Data sourced from studies on survival rates for breast, colon and rectal, lung, melanoma, and prostate cancers from 2017 to 2018.[46]

Other indirect costs of cancer include reduced productivity and absenteeism. One study estimated that annual productivity lost due to cancer mortality (which increases with delayed diagnosis) was estimated to rise to $148 billion by 2020.[47] An additional study noted that the cost of cancer-related employee absenteeism between 2004 and 2008 was $8.1 billion, with a median cost per state of nearly $116 million.[48]

Additionally, delayed diagnosis can increase the risk of costly malpractice lawsuits.[49] Medical malpractice claims may arise out of delayed diagnosis or misdiagnosis, resulting from a physician's conduct or inaction, that increases the risk of recurrence or spread of the patient's cancer and results in sustained injuries.[50] A 2015 study showed that a majority of medical malpractice lawsuits involving breast cancer were due to delayed diagnosis stemming from inadequate patient self-assessment.[51] EDD can improve assessments and reduce these risks and expenses.

---

[45] *Id.*

[46] *Id.* at 21.

[47] Alison Pearce et al., *Projecting Productivity Losses for Cancer-Related Mortality 2011–2030*, 16 BMC Cancer 1, 2 (2016).

[48] Florence K. Tangka, *State-Level Estimates of Cancer-Related Absenteeism Costs*, 55 J. Occupational Envtl. Med. 1015, 1015, 1019 (2016).

[49] Alicia Gallegos, *Delayed Diagnosis Tops Breast Cancer Malpractice Claims*, MDEdge (Oct. 20, 2015) https://www.mdedge.com/obgyn/article/103656/breast-cancer/delayed-diagnosis-tops-breast-cancer-malpractice-claims.

[50] *Id.*

[51] Penny Greenberg, *Navigating Risks in Breast Cancer Diagnosis and Treatment: How Physicians Can Enhance Patient Safety*, Patient Safety and Quality Healthcare (Oct. 12, 2015), https://www.psqh.com/analysis/navigating-risks-in-breast-cancer-diagnosis-and-treatment.

III. BARRIERS TO EDD

Cancer is one of the most commonly missed diagnoses in the US.[52] Inaccurate or delayed cancer diagnoses have been described as "a blind spot in the delivery of quality care."[53] Both patients and practitioners contribute to this phenomenon.

A. Patient Barriers

Though some patients may recognize persistent and subtle health changes, many delay seeking medical help.[54] According to one study, over a third of individuals with cancer who reported a persistent health change in the past three months chose not to seek help from a practitioner.[55] These delays are due to a number of factors, including misinterpretation and minimization of symptoms in which patients do not recognize they need medical intervention, poor health literacy, psycho-social factors, and lack of access to a practitioner.[56]

1. Misinterpretation and minimization of symptoms

Two barriers to timely diagnosis of cancer include misinterpretation and minimization of symptoms.[57] Patients may dismiss ambiguous symptoms of cancer as less serious ailments, or simply feel that they do not rise to the level of requiring medical intervention.[58] Women with ovarian cancer may mistakenly attribute early symptoms, such as abdominal distension, pelvic or abdominal pain, and frequent urination, to irritable bowel syndrome, aging, or stress.[59] Patients may confuse symptoms of lung cancer (e.g., coughing, shortness of breath, and tiredness) with

---

[52] Laura Landro, *The Key to Reducing Doctors' Misdiagnoses,* Wall St. J. (Sept.12, 2017), https://www.wsj.com/articles/the-key-to-reducing-doctors-misdiagnoses-1505226691.

[53] *Id.*

[54] S.L. Quaife et al., *Recognition of Cancer Warning Signs and Anticipated Delay in Help-Seeking in a Population Sample of Adults in the UK.,* 110 Brit. J. of Cancer 12, 12–16 (2014).

[55] Whitaker et al., *supra* note 19, at 1.

[56] *See, e.g.*, Nancy S. Morris et al., *The Association Between Health Literacy and Cancer-Related Attitudes, Behaviors, and Knowledge*, 18 J. Health Comm. 223, 225 (2013); Stephanie Smith et al., *'I know I'm not invincible': An interpretative phenomenological analysis of thyroid cancer in young people*, 23 Brit. J. Health Psychol. 352, 361, 363 (2018); Jascha de Nooijer et al., *A Qualitative Study on Detecting Cancer Symptoms and Seeking Medical Help: An Application of Andersen's Model of Total Patient Delay*, 42 Patient Educ. & Counseling 145, 148, 152–55 (2001); *Five Ways Tech-Savvy Millennials Alter Health Care Landscape*, Cision PR Newswire (Mar. 23, 2015), https://www.prnewswire.com/news-releases/five-ways-tech-savvy-millennials-alter-health-care-landscape-300054028.html.

[57] Grace McCutchan et al., *Barriers to Cancer Symptom Presentation Among People from Low Socioeconomic Groups: A Qualitative Study*, 16 BMC Pub. Health 1052, 1054–55 (2016).

[58] *See generally* Robert A. Simmons et al., *Health Literacy: Cancer Prevention Strategies for Early Adults,* 53 Am. J. Preventative Med., 73, 73–74, 76 (2017). *See also* Jennifer Ann Fish et al., *Understanding Variation in Men's Help-Seeking for Cancer Symptoms: A Semistructured Interview Study,* Psychol. of Men & Masculinity 1, 4 (Apr. 2018).

[59] Kate E. Brain et al., *Ovarian Cancer Symptom Awareness and Anticipated Delayed Presentation in a Population Sample*, 14 BMC Cancer 1, 2 (2014).

other ailments, particularly if they have a history of smoking, thereby delaying the diagnosis of lung cancer.[60]

Even when a patient properly identifies symptoms, psycho-social factors such as feelings of fear, worry, shame, and embarrassment can cause a delay in seeking treatment.[61] For example, studies have shown that patients may wait to seek medical advice for symptoms associated with sex organs due to the private nature of the symptoms or feelings of embarrassment and shame.[62] Others conform to a cultural mindset that if they simply "tough it out," their symptoms will subside.[63] They may wait for their symptoms to worsen or accumulate before seeking medical attention.

Individuals experiencing persistent health changes may downplay the seriousness of those symptoms because they do not know that these changes could indicate a serious health condition or because patients do not know about or apply the two-week rule to evaluate causes such as diet or stress.[64] A study of individuals diagnosed with colorectal cancer found that 52% displayed at least one cognitive barrier to diagnosis, and of those, 40% minimized the seriousness of those symptoms, attributing the symptoms to aging, diet, stress, or ulcers rather than to cancer.[65] This downplaying of seriousness delayed diagnosis by an average of two months.[66]

2. Health literacy

Evidence suggests that individuals with lower health literacy may be more likely to forego cancer screenings, avoid physician visits, and have higher mortality rates.[67] Health literacy is "the degree to which individuals have the capacity to obtain, process, and understand basic health information and services needed to make appropriate health decisions."[68] Health literacy includes the ability to read, understand spoken information, and understand numbers.[69]

---

[60] *Signs and Symptoms of Lung Cancer*, Am. Cancer Soc'y (last revised Oct. 1, 2019), https://www.cancer.org/cancer/lung-cancer/detection-diagnosis-staging/signs-symptoms.html.

[61] *See* Jascha de Nooijer et al., *supra* note 56, at 148, 152, 153–55.

[62] Mohamadreza Neishaboury et al., *Does Embarrassment Contribute to Delay in Seeking Medical Care for Breast Cancer? A Review,* 2 Archives of Breast Cancer 75, 75–77 (2015); Chantal Balasooriya-Smeekens et al., *The Role of Emotions in Time to Presentation for Symptoms Suggestive of Cancer: A Systematic Literature Review of Quantitative Studies*, 24 Psycho-Oncology 1594, 1594–97 (2015).

[63] Lee M. Ellis et al., *Losing "Losing the Battle with Cancer,"* 1 JAMA Oncology 13, 13 (2015).

[64] *See supra* notes 12–13 and accompanying text.

[65] Janette L. Vardy et al., *Cognitive Function in Patients with Colorectal Cancer Who Do and Do Not Receive Chemotherapy: A Prospective, Longitudinal, Controlled Study,* 33 J. Clinical Oncology 4085, 4088 (2015).

[66] *Id.*

[67] Nancy S. Morris et al., *The Association Between Health Literacy and Cancer-Related Attitudes, Behaviors, and Knowledge,* 18 J. Health Comm. 223, 225 (2013).

[68] *Id.*; Levent Dumenci et al., *Measurement of Cancer Health Literacy and Identification of Patients with Limited Cancer Health Literacy,* 19 J. Health Comm. 205, 206 (2014).

[69] *Health Literacy*, Nat'l Inst. Health, https://nnlm.gov/initiatives/topics/health-literacy (last visited Mar. 27, 2020).

Patients with low health literacy may not recognize signs and symptoms of cancer, may delay screenings, and may have difficulty distinguishing credible scientific and medical evidence from misconceptions.[70] For example, low health literacy has been associated with cancer fatalism, or the general belief that cancer outcomes are predetermined by fate.[71] Lower awareness of and negative beliefs about cancer have been associated with delays in reporting cancer symptoms, advanced stage diagnosis, and lower survival rates.[72]

3. Barriers more common among young adults

Young adults may delay seeking a diagnosis due to the perception of invincibility.[73] A 2018 study that interviewed young adults aged nineteen to thirty-four with thyroid cancer found that a majority stated that they were shocked by their diagnosis because they felt they were too young to have cancer.[74] Another study concluded that the youth of individuals aged twenty to forty-three contributed to delays in cancer diagnosis because they experienced a feeling of invincibility or a state of denial.[75]

Lack of strong patient-provider relationships could also lead to delayed or inaccurate diagnoses in young adults. Millennials tend to view health care as a transaction that should occur quickly and efficiently.[76] They are twice as likely as baby boomers to prefer walk-in clinics or retail health facilities over traditional primary care physicians because of their efficient health care delivery.[77] Consequently, millennials may be less prone to develop a strong patient-provider relationship, and providers may not have a comprehensive understanding of the patient's health, resulting in a delayed or inaccurate diagnosis.

B. Clinical Barriers

The diagnostic process is fundamentally dependent on the personal interaction between the provider and the patient, the sufficiency and accuracy of information shared and gathered through the patient's history and exams, and the practitioner's clinical

---

[70] Julia L. Halverson et al., *Health Literacy and Health-Related Quality of Life Among a Population-Based Sample of Cancer Patients,* 20 J. Health Comm.1320, 1322 (2015).

[71] McCutchan et al., *supra* note 57, at 1058.

[72] *Id.* at 1053 (suggesting that poor knowledge, negative beliefs, and barriers to help-seeking result in a long-patient interval). *See also* L.S. Karliner et al., *Language Barriers, Location of Care, and Delays in Follow-Up of Abnormal Mammograms,* 50 Med. Care 171, 172 (2012) (contending that poor communication between minority women and physicians leads to less knowledge and subsequently a barrier to follow-up care).

[73] *See* Smith et al., *supra* note 56, at 361, 363.

[74] *Id.* at 355, 363.

[75] *See* Baukje B. Miedema et al., *Young Adults' Experiences with Cancer: Comments from Patients and Survivors*, 52 Can. Fam. Physician 1447, 1449 (2006).

[76] Kristin Kovesdy, *7 Ways Millennials are Changing the Healthcare Industry (and What it Means to You)*, HFA, https://teamhfa.com/insights/7-ways-millennials-are-changing-the-healthcare-industry-and-what-it-means-to-you/ (last visited Mar. 12, 2020).

[77] *Five Ways Tech-Savvy Millennials Alter Health Care Landscape*, *supra* note 56.

evaluation of that information.[78] Various barriers can prevent practitioners from promptly diagnosing cancer, such as diagnostic errors, inadequate practitioner-patient communication, and insufficient time with the patient.

1. Cognitive biases

Diagnostic errors, or inaccurate or delayed diagnoses, "persist throughout all settings of care and continue to harm an unacceptable number of patients."[79] According to one study, 75% of diagnostic errors are due to cognitive biases such as 1) confirmation bias, or the tendency to seek only as much information as necessary to form an initial clinical impression; and 2) anchoring, or the tendency to stick with initial impressions even as new information becomes available.[80] In other cases, the selection of the first "diagnosis that comes to mind because it is common, serious, recently encountered, or otherwise noteworthy;" and unpacking, or the "failure to elicit all relevant information," prevents a timely diagnosis.[81]

Just as a patient may dismiss a subtle health change associated with cancer, a practitioner may not accurately identify such a change. Cancer is one of the most frequently missed diagnoses, often due to commonality of symptoms combined with cognitive biases.[82] These misdiagnoses have been attributed to anchoring and other cognitive biases.[83]

2. Inadequate practitioner-patient communication

Inadequate practitioner-patient communication can prevent an accurate diagnosis. Practitioners may not educate or train patients on identifying subtle and persistent health problems or convey the need for them to seek medical intervention promptly. As a result of this lack of training, patients may not deem subtle and persistent health changes important enough to bring to their physicians' attention.

Practitioners may confuse patients by using medical jargon that patients are unable to comprehend.[84] Individuals with a limited understanding of the information that their practitioners convey to them are consequently more likely to experience treatment delays.[85] Additionally, practitioners may not adequately convey the risks of a cancer

---

[78] Erin P. Balogh et al., Improving Diagnosis in Health Care, The National Academies Press 37 (2015).

[79] *Id.* at 1.

[80] Edward Etchells, *Anchoring Bias with Critical Implications*, Agency for Healthcare Res. & Quality, Patient Safety Network (June 2015), https://psnet.ahrq.gov/webmm/case/350/anchoring-bias-with-critical-implications.

[81] Alexis R. Ogdie et al., *Seen Through Their Eyes: Residents' Reflections on the Cognitive and Contextual Components of Diagnostic Errors in Medicine*, 87 Acad. Med., 1361, 1363, 1365 (2012).

[82] *Id.* at 1361.

[83] *Id.*

[84] Suzanne Graham & John Brookey, *Do Patients Understand?*, 12 Permanente J. 67, 67–68 (2008).

[85] B. Noonan, *Understanding the Reasons Why Patients Delay Seeking Treatment for Oral Cancer Symptoms from a Primary Health Care Professional: An Integrative Literature Review,* 18 Eur. J. Oncology Nursing *118 (2014). See* Halverson et al., *supra* note 70, at 1328.

diagnosis or the benefits of certain procedures and treatments.[86] A study on patients' perspectives of colorectal cancer screenings found that at least 77% of eligible patients said it was important for physicians to explain the purpose of screening, risks and benefits, test accuracy, and alternatives when considering whether to participate in screening.[87] Yet, not enough patients received such information.[88]

3. Lack of sufficient time

Practitioners may be unable to make an early diagnosis because they do not have enough time during patient visits to properly assess symptoms.[89] Many practitioners, under pressure to be efficient, multitask when patients are speaking and may miss important information.[90] Failure to sufficiently investigate patient symptoms and complaints correlated with more than a six-month delay in the diagnosis of colorectal cancer.[91] Yet, it is not unusual for primary care doctors' appointments to be scheduled at fifteen-minute intervals; during that short period of time, the patient may not even have the practitioner's undivided attention.[92]

IV. RECOMMENDATIONS TO IMPROVE RATES OF EDD

In light of recent statements from the current Administration encouraging improved cancer care, it is important for states to retain control over the practice of medicine and find their own ways to reduce health care costs while also improving cancer care.[93] As such, states should take steps to encourage practitioners to implement EDD education and training programs for patients. To overcome adoption barriers, states should require medical boards to develop guidelines for training practitioners on EDD in primary care settings. Additionally, states should require medical boards to ensure that continuing medical education (CME) courses are available, which would educate primary care physicians and other practitioners on the guidelines and encourage them to adopt practical, low-cost solutions to detect cancer sooner and more accurately. Finally, states should require health plans to provide coverage of these services as an essential

---

[86] M Finch, et al., *Women's Experiences With Ovarian Cancer: Reflections on Being Diagnosed,* 12. J. Oncology Nursing 152, 158 (2002).

[87] M. K. Barton, *Physician-Patient Communication Regarding Colorectal Cancer Screening is Lacking,* 62 Cal. Cancer J. Clinicians 1, 1 (2012).

[88] *See id.* (explaining that, of patients valuing test accuracy information, only seven percent received that information).

[89] *See generally* Roni Caryn Rabin, *15-minute Doctor Visits Take a Toll on Patient-Physician Relationships*, Kaiser Health News (Apr. 21, 2014), https://khn.org/news/15-minute-doctor-visits/ (suggesting that shorter, rushed interactions between practitioners and patients is increasingly common).

[90] *Id.*; G.P. Guy, *Visit Duration for Outpatient Physician Office Visits Among Patients with Cancer*, 8 J. Oncol Prac. 2 (2012).

[91] *See* Amanda L. Thorne et al., *Reduction in Late Diagnosis of Colorectal Cancer Following Introduction of a Specialist Colorectal Surgery Service,* 88 Annals Royal C. Surgeons Eng. 562, 563 (2006) (showing that some patients were delayed in diagnosis because of their own refusal or delay in participating in investigations).

[92] Guy, *supra* note 90.

[93] *See* Exec. Office of the President, *supra* notes 6–7 and accompanying text.

health benefit—encouraging patient-practitioner collaboration by ensuring practitioners receive adequate compensation and additional time to implement EDD. These legislative and regulatory efforts would not only decrease cost of treatment and life years lost, but also lower the risk of malpractice lawsuits stemming from missing or delaying such diagnoses.

A. Guidelines and Professional Education

Patients often play a key role in diagnosing their cancer. Unless a patient is able to detect signs and symptoms of cancer and share that information with a health care provider promptly, a provider may not be able to make a diagnosis or begin treatment. As such, practitioners must start the process that leads to diagnosis by training patients to identify subtle and persistent health changes and seek prompt medical attention. Consequently, it is imperative that health care providers are aware of the barriers to EDD of cancer and how to overcome them. Pursuant to state legislation requiring such, state medical board and voluntary medical associations should develop guidelines and offer educational courses for primary care providers and others aimed at increasing adoption rates of EDD. Guidelines and educational courses should identify the treatment barriers discussed herein and offer training in treatment strategies that will help patients to recognize and understand persistent health changes and practitioners to be mindful of cognitive biases that may unnecessarily delay diagnoses.

One such educational tool is the Three Steps.[94] Practitioners and other medical professionals, such as nurse practitioners, medical assistants, patient advocates, and even first responders, can utilize the Three Steps to help patients engage more meaningfully with their health and recognize symptoms of cancer quickly.[95] First, patients must establish a personal health baseline by being aware of when they feel "normal" or at their best.[96] This includes understanding normal energy levels, presence and intensity of pain, weight, sleep patterns, motor control and reflexes, bowel habits, and appearance.[97] Patients can establish their baseline health by conducting self-examinations; taking photographs, making notes, or using a calendar to track changes to their health, pain, and energy levels; and undergoing regular physical examinations.[98]

---

[94] *3 Steps Detect*, *supra* notes 12–13 and accompanying text.

[95] *See generally id.* (explaining the Three Steps method).

[96] *Remember What Great Feels Like*, 15-40 Connection, https://www.15-40.org/3-steps-to-early-detection/remember-your-great/ (last visited Mar. 27, 2020).

[97] *Id.*

[98] *Id.*

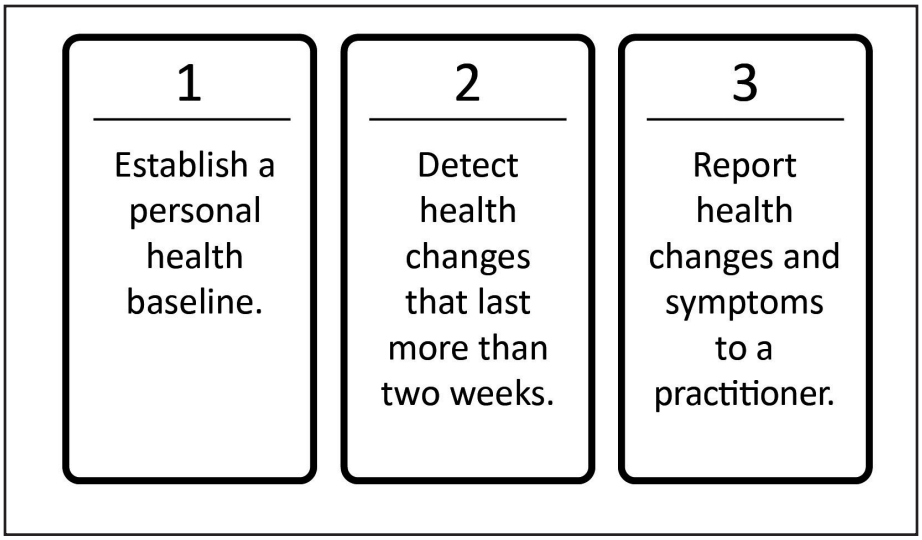

Fig. 3. Outline of the three steps of early detection of cancer.[106]

Second, patients must detect changes to their health that last for longer than two weeks.[99] Patients should be educated to understand what potential cancer symptoms can look and feel like.[100] Rather than dismissing ambiguous symptoms, patients should learn to keep track of any persistent health change, no matter how subtle.[101]

Third, patients must promptly and thoroughly report signs and symptoms that last longer than two weeks to a practitioner.[102] To facilitate this process, they should bring the practitioner a list of health changes or items for discussion, including the dates and duration of which the signs and symptoms were present.[103] Patients should trust their instincts, even if their practitioners dismiss their concerns, and ask for help in determining the underlying reason for the health change or obtain a second opinion.[104] Once the patient receives a diagnosis and a treatment plan, he should 1) ask the practitioner what to expect; 2) continue to monitor his health; and 3) follow up with the practitioner if his health deviates from what is expected.[105]

---

[99] *Use the Two-Week Rule*, 15-40 Connection, https://www.15-40.org/3-steps-to-early-detection/2-week-rule/ (last visited Mar. 27, 2020).

[100] *Id.*

[101] *Id.*

[102] *Share With Your Doctor*, 15-40 Connection, https://www.15-40.org/3-steps-to-early-detection/share-with-your-doctor/ (last visited Mar. 27, 2020).

[103] *Id.*

[104] *Id.*

[105] *Id.*

[106] *3 Steps Detect*, *supra* notes 12–13.

Training medical school students, residents, and fellows can help ensure that future practitioners are aware of how to teach patients the Three Steps.[107] According to the National Cancer Institute, "[p]rimary care physicians, physician assistants, and nurse practitioners are the major sources of health information related to prevention of cancer in health care settings for [most] patients."[108] Research also shows that improved communication between practitioners and patients with breast cancer has been associated with "a sense of choice, improved actual treatment, and patient satisfaction with care."[109] Because health care professionals and organizations are responsible for creating environments in which patients and their families can learn about and engage in the diagnostic process, professional education resources on improving the quality of patient-provider communication, the patient-provider relationship, and patient engagement will be imperative in improving rates of EDD.[110]

For example, programs may focus on how practitioners can tailor their communications for low health-literacy adults to ensure that patients know the value of the information they share and that they are able to comprehend the information that they receive.[111] To counter patient tendencies to forget information or retain incorrect information, training may be offered on information retention and comprehension tools, such as the teach-back method, in which practitioners ask the patient questions to determine whether the patient recalls and understands any information or instructions conveyed.[112]

Education should also extend to printed and digital patient materials. This would ensure that patient educational materials, instructions, and consent forms are written in plain language, for a sixth-grade reading level or less, without using medical jargon.[113] Programs can highlight the use of plain English such as "cancer-causing" rather than

---

[107] *See generally* Anjali Choudhary & Vineeta Gupta, *Teaching Communications Skills to Medical Students: Introducing the Fine Art of Medical Practice*, 5 Int'l J. Applied Basic Med. Res. 41 (2015) (suggesting that teaching medical students communication skills will improve practitioner-patient interactions, which correlate with improved healthcare outcomes).

[108] Ronald M. Epstein & Richard L. Street Jr., *Patient-Centered Communication in Cancer Care: Promoting Healing and Reducing Suffering*, Nat'l Cancer Inst., 71 (2015).

[109] Daniel J. Oates & Rebecca A. Silliman, *Health Literacy: Improving Patient Understanding*, 23 Oncology J. (2009).

[110] *The Patient-Provider Relationship Study: The Ripple Effect Starts with Boomers*, Solutionreach, https://www.solutionreach.com/rethinking-the-patient-provider-relationship (last visited Mar. 27, 2018) (emphasizing the importance of communication for the patient-provider relationship and patient satisfaction).

[111] *Quick Guide to Health Literacy*, Dep't of Health and Human Servs., https://www.centralwestgippslandpcp.com/assets/files/pre-2019/projects/health-literacy/guide/Quickguide.pdf (last visited Mar. 27, 2020) [hereinafter "*Quick Guide*"]; Shaghayegh Vahdat et al., *Patient Involvement in Health Care Decision Making: A Review*, 16 Iran Red Crescent Med. J. 1, 1–3 (2014).

[112] *Quick Guide*, *supra* note 111; Epstein & Street, *supra* note 108.

[113] Richard S. Safeer & Jann Keenan, *Health Literacy: The Gap Between Physicians and Patients*, 72 Am. Fam. Physician 463, 467 (2005); Adam E. M. Eltorai et. al., *Readability of Patient Education Materials on the American Association for Surgery of Trauma Website*, 3 Archives of Trauma Res. 1, 2 (2014).

"carcinogen," "into your vein" rather than "intravenous," or "related to the lungs" rather than "pulmonary."[114]

Treating patients as "equal partners" in their care leads to better adherence to recommended prevention and treatment processes and improved clinical outcomes.[115] It is crucial that primary care practitioners establish strong relationships with patients to build trust and motivate patients to report symptoms. Educational programs on participatory decision-making can help practitioners develop protocols to establish such relationships. Participatory decision-making establishes "a partnership among practitioners, patients and their families" that helps patients to understand their health care "wants, needs and preferences" and make informed decisions.[116] Training on such practices will be beneficial to increasing rates of early diagnosis because these practices build trust between patients and providers and empower patients to understand their health. Practitioners with a participatory decision-making style are thirty percent less likely to have patients leave their care.[117] This may be particularly beneficial for young adults, who are more prone to go to urgent care clinics or frequently change primary care providers.

Professional education programs should also focus courses on cognitive biases and the associated risks of diagnostic errors. Courses should train practitioners on how to recognize cognitive biases and how they can broaden their diagnostic thinking process. For example, practitioners can be trained on how to broaden differential diagnoses by participating in case discussions with colleagues that work through their thought processes, sharing uncertainty and techniques to avoid narrowly framing cases.[118] Clinicians can be trained to reduce confirmation bias by actively seeking information that could diverge from the current impression.[119] They can seek additional information during the physical examination or about a patient's history that may lead to an alternative diagnosis.[120]

Moreover, practitioners can utilize patient communication training to avoid cognitive biases by asking their patients how their health has changed since their last visit and whether those changes have lasted longer than two weeks. They can also encourage their patients to report symptoms by explaining that they cannot test for all changes in the patient's health and that consequently, it is critical for patients to accurately report persistent health changes.

---

[114] *See generally Quick Guide*, *supra* note 111.

[115] *See generally* Epstein & Street, *supra* note 108; Institute of Medicine, Improving the 21st Century Healthcare System, Crossing the Quality Chasm: A New Healthcare System for the 21st Century (2001).

[116] Kristin L. Carman et. al., *Patient and Family Engagement: A Framework for Understanding the Elements and Developing Interventions and Policies*, 32 Health Affairs 223, 224 (2013).

[117] *Id.*

[118] *See* Ogdie et al., *supra* note 81, at 1367 (encouraging discussion among physicians about cognitive biases and diagnostic error).

[119] *See generally id.* at 1365 (explaining confirmation bias).

[120] *Id.*

Programs may also highlight the benefits of increasing physicians' time spent with patients. While shorter patient visits may increase the number of patients seen in a practice, longer visits may be more effective by allowing time for cancer screenings and health education discussions, potentially lowering future treatment costs.[121] Conversations between physicians and patients build trust and can uncover helpful information in making a diagnosis and improving patients' overall health.

B. Coverage of EDD

To further encourage the adoption of EDD, including the training that practitioners must offer to patients, it is imperative that practitioners receive adequate reimbursement for their time and efforts. Given that short patient visits, often influenced by health plans, are already a barrier to accurate and timely diagnosis, providing sufficient coverage of such services may reduce this barrier. Qualified health plans governed by the Patient Protection and Affordable Care Act are required to offer coverage of essential health benefits, including preventive services.[122] Additionally, states may add additional services to the list of essential health benefits that plans must cover.[123] As such, states should add EDD to their list of essential health benefits. The increase in patient satisfaction, quality of life, and cost-savings actualized by this low-cost method should offset the cost of covering this benefit.[124]

CONCLUSION

Detecting and diagnosing cancer at earlier stages can increase survival rates and reduce costs to the patient and the general public. To improve rates of EDD, state legislatures should require medical boards to develop guidelines that encourage patient-practitioner collaboration and education on EDD, ensure CME courses are available to incentivize practitioners to adopt such practices, and require health plans to cover such services. These legislative solutions can facilitate partnerships between practitioners, patients, caregivers, and communities. In turn, such actions will improve education, patient satisfaction, and quality of care; lower health care costs; and reduce the risk of medical malpractice for providers.

---

121 Guy, *supra* note 90.

122 *Health Coverage Rights and Protections*, HEALTHCARE.GOV, https://www.healthcare.gov/health-care-law-protections/ (last visited Mar. 27, 2020).

123 *Information on Essential Health Benefits (EHB) Benchmark Plans*, CTR. FOR MEDICARE AND MEDICAID SERV., https://www.cms.gov/CCIIO/Resources/Data-Resources/ehb (last visited Mar. 27, 2020).

124 *See* Kakushadze et al., *supra* note 31, at 13 (discussing cost savings from early diagnosis of cancer).

DISCLOSURE STATEMENTS

Dr. Deckers serves on the board of directors of 15-40 Connection, a 501(c)(3) not-for-profit organization whose mission is to educate and empower people about early cancer detection. A list of 15-40's funders can be found on its website. Dr. Deckers also serves as a surgical oncologist at UConn Health and as Professor of Surgery and Dean Emeritus at UConn School of Medicine.

Ms. Laursen is the President of 15-40 Connection, a 501(c)(3) not-for-profit organization whose mission is to save lives by teaching people how to detect cancer early and be active partners in their healthcare.

Dr. Manning is a Partner in the Life Sciences Practice at Bates White, LLC in Washington, DC. He provides consulting services and expert testimony on matters relating to economics for clients in government, law firms, and other private enterprises.

Ms. Worthy is a partner at DCBA Law & Policy LLP in Washington, DC. DCBA's clients include not-for-profit health policy and patient advocacy organizations, health care providers and programs, and biotechnology manufacturers, and not-for-profit organizations. To avoid conflicts of interest, DCBA adheres to the District of Columbia Rules of Professional Conduct.

Ms. Kulkarni is an associate at DCBA Law & Policy LLP in Washington, DC. DCBA's clients include health care providers, device and drug manufacturers, and not-for-profit organizations. To avoid conflicts of interest, DCBA adheres to the District of Columbia Rules of Professional Conduct.

* * *

Direct-to-Consumer Genetic Testing: Rethinking Privacy Laws in the United States

*Juan Pablo Sarmiento Rojas**

TABLE OF CONTENTS

* Juan Pablo Sarmiento Rojas is a 2020 Juris Doctor and LLM Candidate at American University Washington College of Law. His passion for health law originated from his parents' love and dedication to dentistry and the underprivileged communities they serve. Since beginning law school Juan Pablo has had the opportunity to serve the health law field through his participation in the Disability Law Clinic at WCL, where he was able to learn and practice how to become a better attorney and a more understanding individual.

INTRODUCTION

Direct-to-consumer (DTC) genetic testing companies have existed since 1996, but it was not until 2017 that the market began to materialize into a booming industry.[1] In 2019, economists projected that by 2024 the DTC genetic testing industry will grow by almost twelve percent worldwide and the market will reach twenty-two billion dollars.[2] In the United States, the number of companies providing consumers with genetic testing is at an all-time high and continues to grow. Some of the most recognizable names in the industry include 23andMe, AncestryDNA, FamilyTreeDNA, LivingDNA, The Genographic Project, and MyHeritage.[3]

The number of people who used a DTC DNA testing kit doubled in 2017, and the total number of users exceeded twelve million in 2018.[4] Most consumers are located in the United States,[5] which some hypothesize is due to the United States' long history of immigration.[6] DTC genetic testing has become so prevalent that it is now a popular gift for holidays.[7] Unfortunately, the reality is that millions of people handing over their DNA to a private company are completely unaware of the potential legal ramifications and privacy concerns that accompany DTC testing.

This Article will analyze the current DTC genetic testing trend and the potential privacy concerns it creates. It will then review current legislation, both domestic and international, to determine how individuals are protected and where protection lacks. Lastly, the Article will propose solutions and improvements to current United States legislation in order to develop a comprehensive federal regulation that will protect individuals' genetic information without hindering the lucrative DTC genetic testing

---

[1] Scott Bowen & Muin J. Khoury, *Consumer Genetic Testing Is Booming: But What are the Benefits and Harms to Individuals and Populations?,* Ctr. for Disease Control & Prevention (June 12, 2018), https://blogs.cdc.gov/genomics/2018/06/12/consumer-genetic-testing [hereinafter CDC].

[2] Sumant Ugalmugle & Rupali Swain, *Genetic Testing Market Size By Test Type (Predictive Testing, Carrier Testing, Prenatal And Newborn Testing, Diagnostic Testing, Pharmacogenomic Testing, Nutrigenomic Testing), By Application (Cancer, Genetic Disease, Cardiovascular Disease), Industry Analysis Report, Regional Outlook, Application Potential, Competitive Market Share & Forecast, 2020 -2026*, Global Mkt. Insights (Feb. 2020), https://www.gminsights.com/industry-analysis/genetic-testing-market; *Genetic Testing Market Will Register 11.6% Growth to cross $ USD 22.5 billion by 2024,* Market Watch, (Feb. 26, 2019, 3:49 PM), https://www.marketwatch.com/press-release/genetic-testing-market-will-register-116-growth-to-cross-usd-225-billion-by-2024-2019-02-26 [*hereinafter* "Market Watch Press Release"].

[3] Molly McLaughlin, *The Best DNA Testing Kits for 2020*, PC Mag., https://www.pcmag.com/roundup/356975/the-best-dna-testing-kits (last updated Jan. 22, 2020). *See also* Market Watch Press Release, *supra* note 2.

[4] Antonio Regalado, *2017 Was the Year Consumer DNA Testing Blew Up*, MIT Tech. Rev. (Feb. 12, 2018), https://www.technologyreview.com/s/610233/2017-was-the-year-consumer-dna-testing-blew-up.

[5] *Id.*

[6] Passant Rabie, *Will America's Obsession With Genetic Testing Ever Fade?,* Sci. Line (Feb. 20, 2019), https://scienceline.org/2019/02/will-americas-obsession-with-genetic-testing-ever-fade.

[7] Ed Cara, *Don't Take the DNA Test You'll Probably Get for Christmas*, Gizmodo (Dec. 19, 2018, 9:10 AM), https://gizmodo.com/dont-take-the-dna-test-youll-probably-get-for-christmas-1831068871.

market. This Article proceeds in six parts. Part I is an introduction to DTC genetic testing and an overview of the existing concerns associated with these services. Part II provides factual background about how the DTC genetic testing process works, the science behind it, and the potential privacy issues associated with it. In Parts III and IV, this Article examines United States and European privacy legislation to determine how various laws apply to DTC genetic testing and how more comprehensive privacy legislation may be developed. Lastly, Parts V and VI provide a policy recommendation and a conclusion based on transparency, increased access to information, and greater consumer protection, while balancing the interests of the parties.

A. Concerns Associated with DTC Genetic Testing

Getting your DNA tested may seem harmless, but it could possibly lead to your arrest—which is exactly what happened to Joseph James DeAngelo in April of 2018.[8] Police located DeAngelo through GEDmatch, an open source genetic information database that stores consumer DNA.[9] The database consists of genetic information that genetic testing companies provide so that participants can find estranged family members.[10] Police found DeAngelo based on DNA information one of his relatives provided.[11] Two years after his arrest, DeAngelo awaits trial on thirteen counts of murder and kidnapping, and is expected to plead guilty if the prosecutors agree to a life sentence and forego the death penalty.[12] This case, although particularly unique, highlights potential privacy, legal, and social concerns that may arise from DTC genetic testing and the need for regulation.

Currently, DTC genetic testing companies in the United States are largely unregulated, which means that they are not subject to strict privacy standards.[13] Additionally, although DTC genetic testing companies assure consumers that they will do their best to protect consumer data, companies are still required to disclose the collected information if

---

[8] Gabriella Borter, *DNA Test Clears Golden State Killer Suspect of 1975 Murder: Prosecutor*, Reuters (Jan. 9, 2019, 10:13 AM), https://www.reuters.com/article/us-california-goldenstatekiller/dna-test-clears-golden-state-killer-suspect-of-1975-murder-prosecutor-idUSKCN1P31P9.

[9] Susan Scutti, *What the Golden State Killer Case Means for Your Genetic Privacy*, https://www.cnn.com/2018/04/27/health/golden-state-killer-genetic-privacy/index.html (last updated May 1, 2018, 12:01 AM).

[10] *Id.*

[11] Ryan Lillis et al., *'Open-Source' Genealogy Site Provided Missing DNA Link to East Area Rapist, Investigator Says,* Sacramento Bee (Apr. 27, 2018, 11:03 AM), https://www.sacbee.com/news/local/crime/article209987599.html.

[12] Breeanna Hare & Christo Taoushiani, *What We Know About the Golden State Killer Case, One Year After a Suspect Was Arrested*, CNN (Apr. 24, 2019), https://edition.cnn.com/2019/04/24/us/golden-state-killer-one-year-later/index.html; Michael Levenson & Heather Murphy, *Golden State Killer Suspect Offers to Plead Guilty,* NY Times, (Mar. 4, 2020), https://www.nytimes.com/2020/03/04/us/golden-state-killer-trial.html.

[13] Inst. of Med. and Nat'l Research Council of the Nat'l Acad., Direct-to-Consumer Genetic Testing: Summary of a Workshop, https://www.ncbi.nlm.nih.gov/books/NBK209639 [*hereinafter* "DTC Genetic Testing: Summary of a Workshop"]; Megan Molteni, *The US Urgently Needs New Genetic Privacy Laws,* WIRED (May 1, 2019 08:00 AM), https://www.wired.com/story/the-us-urgently-needs-new-genetic-privacy-laws.

legally compelled pursuant to subpoenas, search warrants, and other legally enforceable actions.[14] Further, there are ongoing issues concerning false positives and insurance coverage disclosures.[15] An understanding of how genetic testing is done, what the science behind it is, and what current privacy statements do to protect consumers and their DNA is essential to creating comprehensive privacy legislation.

I. BACKGROUND

A. The Genetic Testing Process

The genetic testing process is fairly standardized across all DTC genetic testing companies. The consumer begins by deciding what type of information they want to receive, which determines the cost of the service.[16] The most extensive report available from 23andMe, which costs close to two hundred dollars, provides information about health predispositions, geographical ancestry, wellness analysis, carrier status, and specific DNA traits.[17] After the consumer pays for the service, the DTC genetic testing company sends the consumer a testing kit.[18] Once the kit arrives, the consumer provides his DNA by spitting into a test tube containing an identifying barcode, and then sends the sample back to the company.[19] Three to five weeks later, the company provides the consumer access to the report via email.[20]

The main method of genetic testing DTC companies use is called genotyping.[21] Genotyping is the process by which an individual's DNA is sequenced and then specific locations throughout the sequence are analyzed to identify variants.[22] A variant is the difference in a DNA sequence between individuals, which gives people their unique traits.[23] Variants are associated with an individual's predispositions and specific physical

---

[14] *23andMe Guide for Law Enforcement*, 23andMe, https://www.23andme.com/law-enforcement-guide (last visited Mar. 5, 2020).

[15] Carolyn Crist, *Direct-to-Consumer Genetic Test Results May be Unreliable*, Reuters (Oct. 25, 2019, 5:01 PM), https://www.reuters.com/article/us-health-genetic-tests/direct-to-consumer-genetic-test-results-may-be-unreliable-idUSKBN1X42EI; Brandy D. Gunsolus, *Implications of Direct-to-Consumer Genetic Testing; Improving Patient Autonomy or Risking Patient Safety?*, Am. Soc'y Clinical Laboratory Sci., https://www.ascls.org/communication/ascls-today/336-ascls-today-volume-33-number-3/558-implications-of-direct-to-consumer-genetic-testing (last visited Mar. 5, 2020); *Genetic Testing Threatens the Insurance Industry,* economist (Aug. 3, 2017), https://www.economist.com/finance-and-economics/2017/08/03/genetic-testing-threatens-the-insurance-industry.

[16] *What Is The Cost Of Genetic Testing, And How Long Does It Take To Get The Results?*, U.S. Nat'l Library Med. (Mar. 17, 2020), https://ghr.nlm.nih.gov/primer/testing/costresults.

[17] *Health + Ancestry Service*, 23andMe, https://www.23andme.com/dna-health-ancestry (last visited Mar. 19, 2020).

[18] *How it Works*, 23andMe, https://www.23andme.com/howitworks (last visited Mar. 19, 2020).

[19] *Id.*

[20] *Id.* The previous and any subsequent descriptions of direct-to-consumer genetic testing focuses on 23andMe but is roughly the same for many of the main competitors. *Id.*

[21] Rachel Horton et al., *Direct-to-consumer genetic testing*, BMJ (Oct. 16, 2019), https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6829432; *Difference Between DNA Genotyping & Sequencing,* 23andMe, https://customercare.23andme.com/hc/en-us/articles/202904600-Difference-Between-DNA-Genotyping-Sequencing (last visited Mar. 19, 2020).

[22] *Our Science*, 23andMe, https://www.23andme.com/genetic-science (last visited Mar. 19, 2020).

[23] *Id.*

traits, the genes they carry, and the geographic center where such variants are usually found.[24] Companies generate a report by comparing the consumer's DNA sequence to other DNA sequences the company has access to and looking for patterns that match the consumer's variants.[25] One drawback to this process is that its reliability is dependent on the size of the sample the company has access to.[26] For this reason, consumers of European ancestry (a group with a larger sample size) generally receive more accurate results than Native Americans and those of African descent.[27]

DTC genetic testing companies determine genetic predispositions and carrier statuses and identify different types of variants similarly; thus, the results may be just as inconsistent if based on a small sample size and if the results fail to consider other factors that may lead to the development of a disease.[28] Unfortunately, the results are often inconsistent across companies because different companies look at different variants for the same type of predisposition.[29] These tests do not actually give consumers much information about their health or where they come from, and may create privacy concerns and raise ethical considerations.[30] For example, one such issue arises when a couple becomes aware that they are both carriers of a certain disease. In such situations, the couple may be put in the difficult position of having to decide whether to forgo having children or otherwise risk passing the gene onto their offspring.[31] Additionally, consumers often do not realize that the companies have rights to future use of their DNA.[32] Genetic testing companies may even sell such information;[33] since DNA tells us so much about an individual, it is conceivable that insurance companies, pharmaceutical companies, and others would be willing to pay for it.[34] As previously noted, current laws do not provide sufficient protection, and even though DTC genetic testing companies have privacy

---

[24] *Id.*

[25] Rafi Letzter, *How Do DNA Ancestry Tests Really Work?,* Live Sci. (June 4, 2018), https://www.livescience.com/62690-how-dna-ancestry-23andme-tests-work.html.

[26] *Id.*

[27] Elizabeth Weise, *Looking for Your Roots? For Asians, Blacks and Latinos, DNA Tests Don't Tell Whole Story*, USA Today, https://eu.usatoday.com/story/news/2018/12/02/asians-blacks-latinos-genealogical-tests-dont-tell-full-story/2132681002 (last updated Dec. 3, 2018, 11:42 AM).

[28] *What Do The Results Of Direct-To-Consumer Genetic Testing Mean?*, U.S. Nat'l Library Med. (Mar. 17, 2020), https://ghr.nlm.nih.gov/primer/dtcgenetictesting/dtcresults.

[29] Tina Hesman Saey, *Consumer DNA Testing Promises More Than It Delivers*, ScienceNews (May 22, 2018, 12:00 PM), https://www.sciencenews.org/article/consumer-genetic-testing-dna-genome?tgt=nr.

[30] *Id.*

[31] *Id.*

[32] Erin Brodwin, *DNA-Testing Companies Like 23andMe Sell Your Genetic Data to Drugmakers and Other Silicon Valley Startups*, Bus. Insider (Aug. 3, 2018, 11:45 AM), https://www.businessinsider.com/dna-testing-ancestry-23andme-share-data-companies-2018-8?IR=T; Who's *Making Money From Your DNA?,* BBC, https://www.bbc.com/worklife/article/20190301-how-screening-companies-are-monetising-your-dna (last visited Mar. 19, 2020).

[33] *Id.*

[34] Charles Seife, *23andMe is Terrifying, But Not for the Reasons the FDA Thinks*, Sci. Am. (Nov. 27, 2013), https://www.scientificamerican.com/article/23andme-is-terrifying-but-not-for-the-reasons-the-fda-thinks.

statements that aim to protect consumers, the reality is that companies may change them and that they are not as comprehensive as consumers may think.[35]

B. Privacy Statements & Terms of Service

Before purchasing a service from a DTC genetic testing company, a consumer is often asked to read and accept a privacy statement and terms of service, but he may not understand the terms of these contracts.[36] Genetic testing companies' privacy statements often contain complex language that readers without a legal education or heightened interest in privacy protection are likely to ignore.[37] Consumers are often quick to click "agree" and consent to all of the company's terms and conditions without understanding what duties the company has, what rights the consumer has, and that the terms and conditions may change.[38]

On its website, 23andMe notes five key ways it protects consumers' privacy.[39] The policy states that consumers have control over how their information is used, stored, and shared.[40] Specifically, a consumer can decide whether the DNA sample can be stored, whether it is accessible to other consumers, and whether he wishes to be connected with distant relatives.[41] Next, the policy informs the consumer of the type of information the company collects, how it is stored, and how the company protects the data by stripping it of personally identifying information—such as one's name, birthdate, address, email, credit card number, and web behavior.[42]

After the DTC genetic testing company receives the consumer's personal data, the data is assigned a randomized identification number and the genetic information is identified using a barcode system.[43] Further, the company informs the user of its policy regarding third party sharing.[44] It states that identifiable genetic data will not be sold, leased, or rented to third parties without the consumer's explicit consent, but that aggregate data, which has been stripped of personally identifying information, will be used and shared with third parties regardless of consent.[45] 23andMe's website also explains the company's policy concerning minors and incapacitated individuals, its obligation to comply

---

35 *Id.*

36 *Id.* As previously noted, this article focuses on 23andMe's privacy statement and procedures, with the understanding that other direct-to-consumer genetic testing companies operate in a similar but not identical manner.

37 Cassie Martin, *Privacy and Genetic Consumer Testing Don't Always Mix*, ScienceNews (June 5, 2018, 7:00 AM), https://www.sciencenews.org/blog/science-public/privacy-and-consumer-genetic-testing-dont-always-mix.

38 *Id.*

39 *Privacy is in our DNA*, 23andMe, https://www.23andme.com/privacy (last visited Mar. 19, 2020).

40 *Id.*

41 *Id.*

42 *Id.*

43 *Id.*

44 *Id.*

45 *Id.*

with law enforcement requests, and its policy of not providing insurance companies or employers with information based on the Genetic Information Nondiscrimination Act (GINA) and California's version of GINA (CalGINA).[46] The policy then discusses the company's security practices and how it uses technology to prevent hacking, and informs consumers that they have the right to consent to participate in research.[47] On its face, the information 23andMe provides is fairly user-friendly; however, a more in-depth analysis of the actual privacy statement highlights real concerns which indicate that the initial overview is not as informative and consumer friendly as it first appears.

The first major concern is that the opt-out clause does not actually give consumers the option to fully opt out. The consumer may be able to opt out of the company sharing non-anonymized data, but according to its privacy statement, 23andMe may still share aggregate consumer data once it has been stripped of personally identifying information.[48] Unfortunately, stripping the data of personal identifiers does not always completely protect consumer privacy, as it may still be possible to identify an individual by comparing their genetic data with the genetic data submitted by their close relatives or with data that is otherwise publicly available.[49]

Another privacy concern arising out of 23AndMe's terms of service and privacy statement concerns the opt-in clause. The opt-in provision allows the consumer to opt in to participate in scientific research conducted on behalf of academic, nonprofit, and industry organizations.[50] Around eighty percent of consumers decide to opt in, likely reasoning that doing so may advance medical research; further, consumers may assume that the companies will protect their DNA information since the company's financial success is dependent on remaining trustworthy to consumers.[51]

Unfortunately, one big concern is that DTC genetic testing companies are focused on making a profit and not necessarily on furthering medical research. For example, in 2018, 23andMe sold exclusive rights to its consumer data to one of the world's biggest drug manufacturers for 300 million dollars.[52] This transaction granted a large pharmaceutical company access to the genetic information of millions of consumers, which could have devastating effects on consumer privacy. Potentially, the company could decide to increase its profit by selling a drug, developed with the use of uncompensated consumers'

---

[46] *Id.*

[47] *Id.*

[48] *23andMe, Ancestry, and Selling Your DNA Information*, DNAeXplained (Dec. 30, 2015), https://dna-explained.com/2015/12/30/23andme-ancestry-and-selling-your-dna-information.

[49] Dan Vergano, *DNA Detectives Seek Origins of You*, USA Today (June 13, 2013, 4:58 PM), https://www.usatoday.com/story/news/nation/2013/06/13/dna-detectives-seek-origins-of-you/2420071.

[50] *Research*, 23andMe, https://www.23andme.com/research (last visited Mar. 19, 2020).

[51] Eric Rosenbaum, *5 Biggest Risks of Sharing Your DNA With Consumer Genetic-Testing Companies*, CNBC (June 16, 2018, 2:18 PM), https://www.cnbc.com/2018/06/16/5-biggest-risks-of-sharing-dna-with-consumer-genetic-testing-companies.html.

[52] Daniel Oberhaus, *23andMe Sold Access to Your DNA Library to Big Pharma, But You Can Opt Out*, Vice (July 26, 2018, 3:20 PM), https://www.vice.com/en_us/article/xwkaz3/23andme-sold-access-to-your-dna-library-to-big-pharma-but-you-can-opt-out.

DNA, at ridiculous prices. Considering the pharmaceutical industry's track record, including the recent increase in the cost of EpiPens from about fifteen dollars to over seven hundred dollars, it is quite possible that consumers' altruistic ideals will conflict with the profit-maximizing reality of running a major pharmaceutical company.[53] Even if pharmaceutical companies sell the medication that was developed from consumers' DNA at an affordable price, there is no way for consumers to know how that company uses and stores their genetic data or with whom their data was shared.

DTC genetic testing companies appear transparent at first; however, the reality is that the fine print often contains important information that obscures the fact that consumers may waive important rights regarding access to their genetic information. Furthermore, privacy policies often reference federal laws, such as the Genetic Information Nondiscrimination Act of 2008 (GINA), Health Insurance Portability and Accountability Act (HIPAA), and European Union's General Data Protection Regulation (GDPR), to appear compliant with laws and regulations and make customers more comfortable sharing their genetic information. However, this practice may appear deceiving since the laws regarding DTC genetic testing remain vague.[54]

II. UNITED STATES PRIVACY LEGISLATION

A. Genetic Information Nondiscrimination Act of 2008 (GINA)

DTC genetic testing companies rely on GINA, a federal law that sets the minimum standards insurance companies and employers need to comply with.[55] GINA, enacted in 2008, prohibits discrimination by insurance companies and employers based on genetic information.[56] Despite the usefulness of genetic sequencing for advancing medical progress and early detection of illness, Congress actually passed GINA in response to a rise in discrimination and forced sterilization of individuals who are likely to be carriers of certain genes.[57] Examples include the forced sterilization of women who were presumed to have genetic "defects" based on mental disabilities such as Down syndrome

---

[53] *Id.*; Linnea I. Laestadius et al., *All Your Data (Effectively) Belongs to Us: Data Practices Among Direct-to-Consumer Genetic Testing Firms*, Genet Med. 19, 513–20 (2017), https://doi.org/10.1038/gim.2016.136 (stating that most direct-to-consumer genetic data companies failed to consistently meet international transparency guidelines concerning confidentiality, privacy, and secondary use of data).

[54] Laestadius et al, *supra* note 53. Journal Nature research reported that most direct-to-consumer genetic data companies failed to consistently meet international transparency guidelines concerning confidentiality, privacy, and secondary use of data. *Id*.

[55] *What is GINA?*, 23andMe, https://customercare.23andme.com/hc/en-us/articles/202907820-What-is-GINA- (last visited Mar. 19, 2020).

[56] Genetic Information Nondiscrimination Act of 2008, Pub. L. No. 110-223, §§ 201–223, 122 Stat. 881 (2008).

[57] *Id.*; Jessica L. Roberts, *Preempting Discrimination: Lessons from the Genetic Information Nondiscrimination Act*, 63 Vand. L. Rev. 439, 463 (2019); Amy Harmon, *Congress Passes Bill to Bar Bias Based on Genes*, NY Times, (May 2, 2008), https://www.nytimes.com/2008/05/02/health/policy/02gene.html?auth=login-email&login=email.

and the stigmatization of African Americans based on their genetic predisposition to be carriers of sickle cell anemia.[58]

GINA prohibits health insurance groups and employers with more than fifteen employees from requesting or requiring genetic testing for the purposes of making eligibility and coverage decisions.[59] Further, insurance companies are prohibited from adjusting premiums or determining costs based on genetic information.[60] Insurance companies are not allowed to use predictive genetic test results, such as those provided by DTC genetic testing companies, to make eligibility decisions; however, they are allowed to adjust premiums once the disease manifests.[61] Once an illness begins to physically manifest, the insurance company can take those symptoms into consideration without violating GINA.[62] Additionally, incidental disclosure of genetic information to insurance companies or employers and voluntary participation in genetic testing for research purposes do not constitute violations of GINA.[63] The statute permits the Secretary of Labor to enforce its provisions by imposing sanctions on any health group plan or employer that fails to meet its requirements.[64] However, the Secretary has discretion to waive all or part of the penalty for reasonable cause.[65]

The most notable limitation of GINA is that it does not cover life insurance, long-term care insurance, or disability insurance providers, which means that these companies can take an individual's family history and genetic information into account when deciding whether to provide coverage.[66] Even though insurance companies do not require consumers to take genetic tests, they periodically ask insureds to disclose any new medical information that they are aware of, which can include information learned through genetic testing.[67] If insureds fail to disclose such information, the insurance company may decide to terminate or deny coverage.[68] Thus, it is important that consumers are fully aware that the information they learn from DTC genetic testing could be used against them to determine insurance rates and coverage eligibility. Life, long-term, and disability insurance companies argue that the denial of coverage or the change in rates based on new information is the only financially logical approach.[69]

---

58 Roberts, *supra* note 57, at 463–65.

59 Genetic Information Discrimination Act §§ 201–202.

60 *Id.*§§ 101–102.

61 42 U.S.C.A. § 300gg-53 (West).

62 *Id.*

63 Genetic Information Discrimination Act § 301 (stating that the penalties are $100 for each day of noncompliance per individual, no less than $2,500 per day of noncompliance if the party fails to correct by a date established by notice, and $15,000 if over a year of non-compliance).

64 *Id.*

65 *Id.*

66 Christina Farr, *If You Want Life Insurance, Think Twice Before Getting a Genetic Test*, Fast Company (Feb. 17, 2016), https://www.fastcompany.com/3055710/if-you-want-life-insurance-think-twice-before-getting-genetic-testing.

67 *Id.*

68 *Id.*

69 *Id.*

This argument is partially supported by research that has shown that when consumers learn about their medical predispositions, they are more likely to enroll in one of these types of insurance plans that GINA does not cover.[70] Critics, and those concerned with maintaining the privacy of their genetic information, counterargue that the number of people enrolling in these types of insurance plans at a much younger age based on what they learn from their genetic test results should be enough to counterbalance the costs.[71]

GINA suffers from other limitations, including that it does not extend to education, housing, or mortgage lending.[72] If the statute were to provide greater privacy protections, the risk of discrimination based on genetic test results could be greatly diminished. Overall, while GINA provides some protection to consumers against such discrimination, it does not apply to certain types of insurance and it discourages individuals from participating in medical research.

B. Health Insurance Portability and Accountability Act (HIPAA)

When Congress enacted HIPAA in 1996, its goal was to improve the administration of health insurance and to protect sensitive patient information from unauthorized disclosure.[73] The Act requires that "covered entities" follow strict regulations and grants patients certain rights.[74] Covered entities include health plans such as insurance companies, healthcare providers such as doctors and hospitals, and business associates such as lawyers, consultants, and billing companies.[75] Covered entities must protect patients' medical information, privileged conversations between doctors and patients, insurance information, billing information, and other health-related information.[76] Additionally, HIPAA gives a patient the right to obtain a copy of his medical records, the right to correct his records, and the right to know how and to whom his protected health information is disclosed.[77]

HIPAA creates requirements that certain covered entities must follow.[78] The Privacy Rule sets the standard for protecting individually indefinable health information,[79] while the Security Rule sets standards to maintain the confidentiality, integrity, and availability of electronically protected health information.[80] Lastly, the Breach Notification Rule

---

[70] *Id.*

[71] *Id.*

[72] Sarah Zhang, *The Loopholes in the Law Prohibiting Genetic Discrimination*, Atlantic (Mar. 13, 2017), https://www.theatlantic.com/health/archive/2017/03/genetic-discrimination-law-gina/519216.

[73] *HIPAA Privacy and Security for Beginners*, Wiley Rein LLP (July 2014), https://www.wiley.law/newsletter-5029.

[74] 45 C.F.R § 164.502.

[75] *HIPAA Privacy and Security for Beginners, supra* note 73.

[76] *Id.*

[77] *Id.*

[78] *Id.*

[79] Office for Civil Rights, *HIPAA for Professionals*, U.S. Dep't Health & Hum. Servs. (June 16, 2017), https://www.hhs.gov/hipaa/for-professionals/index.html [hereinafter HHS].

[80] *Id.*

requires covered entities to notify patients and to follow certain protocols in the event of a data breach.[81]

The main goal of the Privacy Rule is to protect individuals' health information while allowing the flow of information to promote high quality healthcare.[82] The rule balances covered entities' need to have flexibility in how they use the information with the need to protect patients' privacy.[83] Specifically, the Privacy Rule protects individually identifiable health information, also known as Protected Health Information (PHI).[84] PHI is any information relating to an individual's physical or mental condition, the health care services provided to the individual, or payment for such services which may be used to identify the individual.[85] PHI includes, but is not limited to, name, date of birth, address, and Social Security number.[86] The rule allows disclosure under certain conditions¾with patient consent or if the data is stripped of personal identifiers.[87] Further, the covered entity may, without permission, disclose PHI for payment and treatment operations, research, protecting public health, or healthcare operations.[88] Additionally, the Privacy Rule sets guidelines for the use of PHI for marketing purposes.[89] Generally, the law requires that the patient authorize the covered entity to use their PHI for a marketing purpose unless one of the two exceptions applies: if there is face-to-face communication or if the covered entity provides a promotional gift of nominal value.[90] The rule makes clear that notification of health-related products or services, communications for treatment of an individual, and case management and care coordination do not constitute marketing; thus, they are not protected under the marketing provision.[91]

HIPAA's Security Rule focuses primarily on protecting electronically stored data.[92] The rule requires covered entities to maintain reasonable and appropriate administrative, technical, and physical safeguards for protecting PHI.[93] The application of the Security Rule becomes more important as the amount of electronically stored patient data continues to grow. The advent of the cloud and other electronic services for storing information has allowed easier access for doctors and patients, but has also made patient data more vulnerable to unauthorized access.[94] Accordingly, the Security Rule ensures

---

81 Office for Civil Rights, *Breach Notification Rule*, HHS (July 26, 2013), https://www.hhs.gov/hipaa/for-professionals/breach-notification/index.html.

82 Office for Civil Rights, *Summary of the HIPAA Privacy Rule*, HHS (July 26, 2013), https://www.hhs.gov/hipaa/for-professionals/privacy/laws-regulations/index.html.

83 *Id.*

84 *Id.*

85 *Id.*

86 *Id.*

87 *Id.*

88 *Id.*

89 *Id.*

90 *Id.*

91 *Id.*

92 Office for Civil Rights, *Summary of the HIPAA Security Rule*, HHS (July 26, 2013), https://www.hhs.gov/hipaa/for-professionals/security/laws-regulations/index.html.

93 *Id.*

94 *Id.*

that covered entities maintain reasonable and appropriate administrative, technical, and physical safeguards for protecting electronic PHI.[95] Under the rule, covered entities must assure confidentiality, integrity, and availability of PHI, and must identify and protect against reasonably anticipated risks.[96] Lastly, the rule calls for risk analysis and risk management by requiring an evaluation of the likelihood and impact of potential risks to PHI and the implementation of appropriate security measures, including administrative, physical, and technical safeguards.[97]

The last major component of HIPAA is the Breach Notification Rule.[98] This rule requires covered entities and their business associates to provide notification following a breach involving PHI.[99] A breach consists of an impermissible acquisition, access, use, or disclosure of PHI that compromises its security or privacy.[100] An unauthorized use or disclosure is assumed to be a breach unless the covered entity or business associate can demonstrate a low probability that the breach compromised the PHI.[101] The rule creates three exceptions to what constitutes a breach: (1) when a person acting under authority from a covered entity or business associate accesses PHI in good faith; (2) when a person associated with the covered entity or business associate discloses PHI to another covered entity or business associate and the information is no longer used or disclosed; and (3) when a covered entity or business associate has a good faith belief that the unauthorized person to whom the information was disclosed is unable to retain the information.[102]

If none of the exceptions apply and there was a breach, then HIPAA outlines notification requirements in the Breach Notification Rule.[103] Under this rule, affected individuals must be notified upon the discovery of the breach.[104] Notification must be through a written notice either through first-class mail or email.[105] If there is no way to notify the affected individual, the covered entity must post the notice on its website or in a major print publication or broadcast media where affected individuals are likely to reside.[106] The notice must include a description of the breach, the type of information involved, steps affected individuals should take, and steps the covered entity is taking to investigate, mitigate, and prevent further disclosure of PHI.[107] If a business associate suffers a breach, the business associate must notify the covered entity of the breach

---

95 *Id.*

96 *Id.*

97 *Id.*

98 *Breach Notification Rule, supra* note 81.

99 *Id.*

100 *Summary of the HIPAA Security Rule*, *supra* note 92.

101 *Id.*

102 *Id.*

103 *Id.*

104 *Id.*

105 *Id.*

106 *Id.*

107 *Id.*

without unreasonable delay and no more than sixty days after the breach.[108] If a breach affects more than five hundred individuals, the rule requires the entity to issue a notice to a prominent local media outlet, as well as a notice to the Secretary of Health and Human Services (HHS).[109] To clarify, these provisions only apply if the compromised data qualifies as unsecured PHI and the incident involved a covered entity or a business associate.[110]

Genetic data falls within HIPAA's definition of PHI, but because the statute narrowly defines business associates and covered entities, DTC genetic testing companies are unlikely to qualify as such.[111] As a result, HIPAA minimally protects consumers from privacy violations and dangers associated with DTC genetic testing. DTC companies allow their consumers to assume that since they make consumer data anonymous before sharing it with third parties, they are compliant with HIPAA, despite not being required to do so. This practice minimizes the fact that genetic data can be re-identified with the use of genetic samples supplied by an individual's relatives, making the process of de-identifying insufficient to protect the privacy of an individual's genetic information.[112]

In addition to providing data regarding where one's ancestors originated from, DTC genetic testing companies provide information regarding an individual's predisposition to certain illnesses and carrier status for identified diseases.[113] These services may blur the line of what is considered a covered entity, since the definition of a covered entity—a healthcare provider, health plan, or healthcare clearinghouse that handles patient information electronically—could not have contemplated the existence of DTC genetic testing companies in 1996.[114] In other words, when HIPAA was originally passed, the legislature could not have anticipated that twenty-five years later companies would begin providing quasi-diagnostic services directly to consumers and selling de-identified consumer health information.

DTC genetic testing companies store large amounts of consumer health information which, due to lack of regulation, makes such data vulnerable to unauthorized exposure.[115] For example, in 2017, major DTC genetic testing company MyHeritage suffered an

---

[108] *Id.*

[109] *Id.*

[110] *Breach Notification Rule, supra* note 81.

[111] Linnea Laestadius, *Transparency and Direct-to-Consumer Genetic Testing Companies*, Petrie-Flom Ctr. Harv. L. Sch. (Nov. 22, 2016), http://blog.petrieflom.law.harvard.edu/2016/11/22/transparency-and-direct-to-consumer-genetic-testing-companies.

[112] Dana A. Elfin, *DNA Testing? You Might Want to Wait for More Legal Protection*, Bloomberg L. (Jan. 7, 2019, 5:40 AM), https://news.bloomberglaw.com/pharma-and-life-sciences/dna-testing-you-might-want-to-wait-for-more-legal-protection.

[113] *What Health Related Information Can I Learn From 23andMe?,* 23andMe, https://customercare.23andme.com/hc/en-us/articles/115013843028-What-health-related-information-can-I-learn-from-23andMe- (last visited Mar. 19, 2020).

[114] Peter Pitts, *The Privacy Delusions of Genetics Testing,* Forbes (Feb. 15, 2017, 1:26 PM), https://www.forbes.com/sites/realspin/2017/02/15/the-privacy-delusions-of-genetic-testing/#480fae0b1bba.

[115] *See, e.g.,* Makena Kelly, *MyHeritage Breach Leaks Millions of Account Details*, Verge (June 5, 2018, 2:08 PM), https://www.theverge.com/2018/6/5/17430146/dna-myheritage-ancestry-accounts-compromised-hack-breach.

electronic privacy breach that affected over ninety-two million accounts.[116] The breach exposed consumers' email and password information.[117] Fortunately, no genetic data was compromised during the breach, but such a risk remains a concern.[118] Under current laws, the company would not have been held liable for failing to adequately protect personally identifiable information.[119]

Additionally, without HIPAA's application in this context, DTC genetic testing companies rely on their defined privacy policy terms and ability to use the information as they find "appropriate" without the force of law defining what "appropriate use" is or what is "sufficient" to protect consumer privacy interests.[120]

HIPAA's Privacy Rule protects patients' health information while ensuring a flow of information to provide quality healthcare and protect the public's overall wellbeing.[121] Permitting companies to continue operating in a manner analogous to covered entities without subjecting them to HIPAA's requirements undermines the purpose of the statute. Fortunately, even though federal regulation is lacking, some states have developed their own legislation to increase privacy protections for consumers' genetic data and electronically stored information.

C. State Privacy Laws—Alaska Genetic Privacy Act, California Genetic Information Nondiscrimination Act (CalGINA), & California Consumer Privacy Act (CCPA)

At least seventeen states have enacted laws designed to provide protections against privacy violations and discrimination to genetic testing consumers.[122] Alaska and California are two such states with exemplary laws. Alaska passed the Alaska Genetic Privacy Act in 2004, which strictly limits access, retention, and disclosure of genetic data without informed written consent from the consumer.[123] California's Genetic Information Nondiscrimination Act (CalGINA), which took effect in 2012, extends the federal GINA beyond employment and health insurance to include housing, mortgage lending, education, and public accommodation.[124] Both laws provide important protections for consumers, and any new federal law should incorporate elements from these and other similar state laws.

---

116 *Id.*

117 *Id.*

118 *Id.*

119 *Summary of the HIPAA Privacy Rule, supra* note 82.

120 Mary Fraker & Anne-Marie Mazza, *Direct-To-Consumer Genetic Testing: Summary of a Workshop*, Nat'l Acads. Press Nat'l Ctr. Biotechnology Info. (2010), https://www.ncbi.nlm.nih.gov/books/NBK209632/.

121 *Summary of the HIPAA Privacy Rule, supra* note 82.

122 DTC Genetic Testing: Summary of a Workshop, *supra* note 13.

123 *State Genetic Privacy Policy*, Elec. Privacy Info. Ctr., https://epic.org/state-policy/genetic-privacy/ (last visited Mar. 19, 2020).

124 Cal. S.B. 559 ch. 261 (2011).

On January 1, 2020, California added to its protection of consumer privacy by putting into effect the California Consumer Privacy Act (CCPA).[125] The CCPA grants California consumers the right to know how their personal information is being collected, used, shared, or sold.[126] Consumers also have the right to compel businesses to delete their personal information.[127] Lastly, consumers have the right to opt out, which prohibits the selling of their information by businesses to third parties.[128] The businesses that are subject to the regulation, mainly big companies that store and process large amount of personal information, are obliged to create procedures and respond to consumers exercising their new rights.[129] The new law has forced DTC genetic testing companies to provide the required information to California residents and to create the necessary procedures.[130] Legislators should look at the CCPA and consider a federal regulation that would provide similar protections to consumers nationwide.

While these State laws increase privacy protections, they suffer from a number of limitations. For example, Alaska's Genetic Privacy Act creates an exception to the written informed consent requirement for genetic data collected for law enforcement purposes.[131] Although not directly applicable to direct-to-consumer genetic testing companies, having such exceptions weakens consumer protection overall since it provides a means for access. Further, CalGINA creates an exception in cases where the presence of a disease has already begun to manifest in an individual.[132] The difficulty then becomes determining when exactly the disease began to manifest, and whether the presence of identifiable gene variations constitutes manifestation of the disease for the purposes of the Act.[133] Lastly, instead of automatically protecting consumers, the CCPA places a burden on them to exercise their rights, while also increasing the cost of compliance for companies.[134] These types of weaknesses in regulations need to be addressed in any subsequent legislation.

---

125 Cal. Civ. Code § 1798.100 (West 2020).

126 *Id.*

127 *Id.* § 1798.105.

128 *Id.* § 1798.120.

129 *Id.* §§ 1798.125, 1798.130, 1798.135.

130 *See e.g.*, *Privacy Notice for California Residents*, 23andMe (Jan. 1, 2020), https://www.23andme.com/about/california-privacy.

131 *State Genetic Privacy Policy*, *supra* note 123.

132 *Genetic Information Databases and Privacy Concerns*, Law Shelf, https://lawshelf.com/blog/post/genetic-information-databases-and-privacy-concerns (last visited Mar. 19, 2020).

133 *Id.*

134 Sara Morrison, *California's New Privacy Law, Explained*, Vox (Dec. 30, 2019, 6:50 PM), https://www.vox.com/recode/2019/12/30/21030754/ccpa-2020-california-privacy-law-rights-explained.

III. EUROPEAN PRIVACY LEGISLATION

A. General Data Protection Regulation (GDPR)

In 2018, the European Union (EU) adopted the General Data Protection Regulation (GDPR), which remains the most comprehensive privacy legislation worldwide.[135] The goal of the GDPR is to harmonize privacy laws across Europe while giving EU citizens more control over their data.[136] Protection of data and privacy are at the core of the EU and are considered fundamental freedoms under the European Union Charter.[137] To assure maximum consumer protection, the United States should model its privacy laws after the GDPR.

The GDPR provides some of the key rights and duties that the United States should adopt to expand protections for individuals' genetic data. First, the GDPR has a vast territorial reach, as it applies to any company that processes data of EU citizens, regardless of where the organization is physically based.[138] This means that DTC genetic testing companies must remain compliant with the GDPR if they offer services to EU citizens, even if the businesses are based outside of the Union. Further, the GDPR requires companies to process personal data lawfully, fairly, and transparently.[139] Businesses may only collect data for specified and limited legitimate purposes unless one of the exceptions applies—the data is processed for the public good or for historical and scientific research purposes.[140] The data must also be accurate and kept up to date; therefore, companies are responsible for ensuring accuracy and allowing for correction or deletion of data without undue delay.[141] Further, businesses must process data in a manner that preserves its integrity and confidentiality and protects against unauthorized possession, use, destruction, or damage.[142] Lastly, businesses must keep data in a form that permits identification of data subjects for no longer than necessary for the purposes for which the personal data is processed.[143]

The GDPR makes all processing and use of genetic data unlawful unless the consumer consents or it is necessary for performance of a contract, complying with legal obligations, protecting vital interests of the individual or a third party, carrying out a public interest task, or carrying out legitimate interests when such interests are not outweighed by the interests of the consumer.[144] With regard to consent, a company must

---

135 Mark Scott & Laurens Cerulus, *Europe's New Data Protection Rules Export Privacy Standards Worldwide*, Politico, https://www.politico.eu/article/europe-data-protection-privacy-standards-gdpr-general-protection-data-regulation (last updated Feb. 6, 2018, 4:50 AM).

136 *Id.*

137 *Id.*

138 *Data Protection*, 23andMe, https://www.23andme.com/gdpr (last visited Mar. 19, 2020).

139 General Data Protection Regulation, 2016/679 Official J. Eur. Union Ch. 2, Art. 5(1)(a), (2016).

140 *Id.* Art. 5(1)(b).

141 *Id.* Ch. 1, Art 4(d).

142 *Id.* Ch. 2, Art. 5(1)(f).

143 *Id.* Ch. 2, Art. 5(e)-(f).

144 *Id.* Ch. 2, Art. 6(1)(a)-(f).

be able to demonstrate that the consumer has consented and was provided an intelligible and clearly written declaration.[145] Finally, the consumer has the right to withdraw his consent at any time.[146]

The legislation also grants consumers the right to receive transparent information from and communication with those that have access to their personal data,[147] which includes the right to know the identity and contact information of all parties who have access to that data.[148] Further, consumers have a right to request deletion or make changes to personal information and to prohibit or restrict processing of personal data.[149]

In addition to expanding rights for European citizens, the GDPR also places duties on those who process their data.[150] When data is transferred to another country, that country must have laws in place that create legally enforceable paths that consumers can take if their privacy is violated.[151] If the company has not placed appropriate legal safeguards, then the company needs to ensure through other means that the individual's privacy will be protected or that the consumer will have sufficient legal recourse.[152] Such recourse may include a legally binding and enforceable contract, binding corporate rules, or an approved code of conduct.[153] Additionally, the "controllers," which are those organizations possessing the personal data, must implement appropriate technical and organizational measures to ensure that processing is performed in accordance with the regulation.[154] This includes a requirement that the company holding the information takes the necessary steps to safeguard the processing of personal data, such as performing pseudonymization, a de-identification procedure.[155] In the case of a breach, the controller must notify a supervisory authority within seventy-two hours of discovering the breach unless the breach is unlikely to result in a risk to the rights and freedoms of the individuals affected.[156]

In the event of a breach, consumers have the right to file a complaint and pursue a judicial remedy to seek damages.[157] Non-compliant companies can be fined to a degree that is effective, proportionate, and dissuasive.[158] The penalizing body considers the nature, gravity, and duration of the infringement to determine the appropriate fine.[159] The

---

145 *Id.* Ch. 2, Art. 7(1)-(2).

146 *Id.* Ch. 2, Art. 7(3).

147 *Id.* Ch. 3, § 1 Art. 12.

148 *Id.* Ch. 3, § 2 Art. 13(1)(a).

149 *Id.* Ch. 3, § 3 Art. 16, 17(1), 18(1).

150 *Id.* Ch. 5, Art. 45(1).

151 *Id.*

152 *Id.* Ch. 5, Art. 46(1).

153 *Id.*

154 *Id.* Ch. 4, Art. 24.

155 *Id.* Ch. 4, Art. 25(1).

156 *Id.* Ch. 4, Art. 33(1–2).

157 *Id.* Ch. 8, Art. 77–79, 82(1).

158 *Id.* Ch. 8, Art. 83(2).

159 *Id.*

GDPR's strict enforcement mechanisms and severe penalties distinguish it from other privacy laws and incentivize businesses to comply, which results in greater consumer protection overall.

Despite its strengths, the GDPR is not without criticism. For example, commentators criticize the GDPR provisions regarding when exceptions may apply as vague,[160] which weakens its overall effects and creates potential loopholes that companies may exploit to circumvent complying with regulations. For example, consider a company offering services or goods to EU citizens but processing their data at the point of transaction with a different company located outside of the EU.[161] In this scenario, the EU citizen's personal data is not protected under the GDPR.[162] For the purposes of DTC genetic testing companies, it is conceivable that 23andMe or another similar company may offer their services to individuals in the EU, but when those individuals submit payment the company processing the transaction is an independent company. Since the company is not operating in the EU, the individual's identifiable personal data held by the third-party company would not be protected. The consumer's genetic information may remain protected if third party companies are acting in good faith, but other personally identifiable information that is attached to the genetic health information would not be subject to the GDPR protections, thereby increasing the risks of unauthorized access and the possibility of re-identification of genetic information if the data is stored together.

Further, the GDPR provides several exceptions to compliance which could potentially create more loopholes that companies might exploit. Public interest, scientific, and historical research purposes are common exceptions to many of the requirements set by the regulation.[163] It is conceivable that some DTC genetic testing companies will take advantage of this ambiguity and attempt to characterize their services as exempt from requirements because their work furthers scientific advancement.

Lastly, the GDPR prohibits processing of genetic data for the purpose of identifying an individual; however, a number of exceptions weaken the effect of this provision.[164] These exceptions allow identification under certain circumstances—if an individual provides explicit consent, if legally compelled, if necessary to protect vital interests of the data subject or other natural person, if required to protect public interest or health, if medically necessary, and for research purposes, among others.[165] The vagueness and inherent breadth of these exceptions make genetic data and certain categories of personal data less protected than other data. These exceptions also provide extensive leeway for DTC genetic testing companies to continue operating with limited oversight and regulation.

---

[160] *Top Five Concerns with GDPR Compliance*, Thomson Reuters, https://legal.thomsonreuters.com/en/insights/articles/top-five-concerns-gdpr-compliance (last visited Mar. 19, 2020).

[161] Robert Madge, *Five Loopholes in the GDPR*, Medium (Aug. 27, 2017), https://medium.com/mydata/five-loopholes-in-the-gdpr-367443c4248b.

[162] *Id.*

[163] General Data Protection Regulation 2016/679 Official J. Eur. Union Ch. 2, Art. 5(1)(b), (e) (2016).

[164] *Id.* Art. 9 (1)–(2) (2016).

[165] *Id.*

IV. POLICY RECOMMENDATION

The foregoing discussion evidences a clear need for comprehensive federal regulation to protect the privacy of consumers' personal and genetic information in the United States. Any such legislation must take into consideration the successes and failures of existing laws. Healthcare and medical decisions made by insurers, healthcare providers, and patients are increasingly informed and influenced by DTC testing results.[166] Therefore, any new legislation must incorporate a requirement that consumers are fully informed of the risks and benefits of genetic testing.[167] Further, it is vital that regulations are written in language that an average consumer is able to understand and appreciate.[168]

Additionally, it is important that DTC genetic testing companies are involved in the conversation regarding new legislation. Giving these companies a say will help increase transparency and incentivize them to act in good faith and comply with the resulting legislation. So far, DTC genetic testing companies have been willing to cooperate and improve privacy protection, as evidenced by their recent agreement to adopt the best practices guidelines that aimed to address concerns with consumer privacy.[169]

A. Priorities

A careful balancing of priorities should be an essential component of any future regulation. A comparison of the United States and European Union privacy and data protection laws indicates that the United States is more concerned with the integrity of data as a business asset, while the European Union prioritizes individual rights before business interests.[170] The United States should strive to enact legislation that increases the protection of an individual's right to privacy, rather than wholly prioritizing business interests.

A thorough analysis of GINA, HIPAA, CCPA, and the GDPR provides a valuable framework for the legislature to expand upon. Further, a thorough analysis of DTC genetic testing companies' privacy statements and terms of service can help Congress understand how such companies operate and determine issues that need to be addressed.

The biggest issues with companies' privacy statements and terms of service are that they provide inadequate consumer protection and are heavily one-sided since the companies can change the terms at any time. For this reason, Congress should include specific provisions providing guidance concerning privacy statements and terms of service. First, the regulation should ensure that the documents are written in plain English and are concise enough that the everyday consumer would be willing and able to read and understand them. By removing the legal jargon and simplifying the documents overall,

---

166 Fraker & Mazza, *supra* note 120.

167 *Id.*

168 *Id.*

169 Mallory Locklear, *23andMe, Ancestry and Others Agree to Genetic Privacy Guidelines*, Engadget (July 31, 2018), https://www.engadget.com/2018/07/31/23andme-ancestry-genetic-privacy-guidelines.

170 Andrada Coos, *EU vs US: How Do Their Data Protection Regulations Square Off?*, EndPoint Protector (Jan. 17, 2018), https://www.endpointprotector.com/blog/eu-vs-us-how-do-their-data-protection-regulations-square-off.

consumers could be better informed about what rights they are giving up and about the companies' duties regarding the use and protection of their genetic information. Second, the legislation should include a provision mandating that the privacy statement and terms of service the consumer had already agreed to will not be subsequently altered without the consumer's explicit consent. Essentially, the regulation should prohibit DTC genetic testing companies from rolling back protections that the consumers had when they agreed to the original terms of the contract. In practice, this would mean that the only changes the companies would be allowed to make would be changes that improve the privacy and security of consumers' genetic and personal data.

The purpose of GINA, to protect individuals from discrimination based on genetic information, is key to protecting privacy; therefore, new legislation must also prohibit such discrimination. Accordingly, the new law should ensure that any discriminatory use of genetic information is a violation of the law, and that consumers have a private cause of action to pursue claims and exercise their rights and protections. Further, the law must apply to the sectors that are excluded under GINA. CalGINA attempted to expand such protections by applying the regulation to education, public services, housing, and mortgage lending.[171] The new law should go even further by placing a total prohibition on the use of genetic information for any application or eligibility purpose other than scientific research. The law should also clearly state that the detection of markers in genetic sequences of a possible disease does not constitute a manifestation of a disease. This statement is necessary because GINA and CalGINA only apply before a disease physically manifests, which greatly diminishes the protections that these laws provide.

Additionally, future legislation must balance the interests of businesses with the need to protect individuals' privacy. Considering HIPAA, the law must clarify whether DTC genetic testing companies can be considered covered entities or business associates. By focusing on the type of services that an entity provides, the legislation could expand the definition of a covered entity to include DTC genetic testing companies because they provide health-related services. Alternatively, the legislation could focus on the type of information an entity holds and thus include DTC genetic testing companies because they collect health-related information. Lastly, the law could require a physician referral before consumers can purchase genetic testing services—this option would likely hurt the DTC genetic testing businesses but would benefit the consumers. Though consumers might be less inclined to take the extra step of visiting a physician for a referral, if they do, the physician may educate them about the potential consequences of genetic testing.

Further, Congress should expand requirements regarding consumer consent and data de-identification. The law should incorporate elements of the Alaska Genetic Privacy Act and require consumer's written intelligible consent, even if the data is anonymized. Additionally, the law should eliminate exceptions relating to law enforcement and instead require a valid search warrant for access to genetic databases. As for de-identification requirements, Congress should demand a study in which various techniques are evaluated to determine the most effective way to make genetic information anonymous.

---

[171] Samual A. Garner & Jiyeon Kim, *The Privacy Risks of Direct-to-Consumer Genetic Testing: A Case Study of 23andMe and Ancestry*, 96 Wash. U.L. Rev. 1219, 1232 (2019).

This is important since currently, even if genetic information is de-identified, it is still possible to re-identify an individual based on genetic information provided by their close relatives.[172] Lastly, HIPAA's notice requirements are fairly exhaustive and thus should be incorporated into new legislation, though Congress should eliminate the exception that organizations do not need to give notice if it is determined that there is a low risk of PHI exposure.

Giving the individual the right to make determinations regarding use of their personal data is one of the key aspects of the GDPR. Legislatures should model any new regulation after the GDPR and further expand on the rights and duties the GDPR created. The new legislation should specifically improve on the GDPR's fining system, long-arm provision, and regulation of data transfer. The GDPR's steep financial penalties create strong incentives for companies to become and remain compliant. Similarly, the new law should set rigid minimum penalties, but allow for some flexibility when determining the appropriate fine for non-compliant companies based on a number of factors.[173] Some of these factors include the nature of the infringement, whether the infringement was intentional or negligent, whether there are any mitigating factors, whether the company had preventive measures in place, and the type of data that was subject to the breach.[174]

An independent quasi-judicial body should be charged with enforcing the penalties against non-compliant companies, similarly to how different regulatory agencies are in charge of enforcing different areas of law in the United States, or how HHS's Office of Civil Rights (OCR) enforces penalties for non-compliance with HIPAA and other regulations.[175] OCR has imposed over $116 million in civil penalties since 2003.[176] The office plays a quasi-judicial role in determining compliance with HIPAA and then enforcing civil fines. This proposed body will be a direct way to hold companies responsible—a preferable alternative to having to go to court in order to force companies to change how they operate. The effectiveness of any new legislation would be greatly diminished without a strict enforcement mechanism that includes monetary penalties. If a company fails to comply even after monetary penalties are imposed, then the new law should give consumers a private right of action so that they can pursue their claims in court.

Further, the GDPR has a vast territorial reach that protects individuals regardless of where a company is located. This long-arm provision should be incorporated into U.S. legislation and include a requirement that any third-party providing services to the companies in the U.S. is also subject to the regulation. Additionally, there should be a requirement that whenever an individual's genetic data is transferred to a third party, the organization must provide notice to the individual regardless of consent. By

---

172 Julian Segert, *Understanding Ownership and Privacy of Genetic Data*, Harv. U. (Nov. 28, 2018), http://sitn.hms.harvard.edu/flash/2018/understanding-ownership-privacy-genetic-data.

173 *Administrative Fines*, GDPR EU, https://www.gdpreu.org/compliance/fines-and-penalties (last visited March 19, 2020).

174 *Id.*

175 *About Us*, HHS, https://www.hhs.gov/ocr/about-us/index.html (last visited March 22, 2020).

176 *Enforcement Highlights*, HHS, https://www.hhs.gov/hipaa/for-professionals/compliance-enforcement/data/enforcement-highlights/index.html (last visited March 22, 2020).

including this requirement, the legislation would ensure that the individual remains informed of who has their information, where it is going, and how it is being used. This requirement would require a lot of effort on the part of businesses; however, any financial and logistical burden would be outweighed by the interest of protecting the individual's property and privacy.

CONCLUSION

Direct-to-consumer genetic testing is a growing industry. The companies offering these services provide individuals with information about where their DNA originated from and inform them of potential health issues that they are at risk of developing. Numerous privacy concerns have already arisen from access to such information. The blurred line of what constitutes health services has shielded these companies from regulation, therefore failing to provide adequate safeguards concerning genetic privacy. This Article has suggested several regulations for United States lawmakers to consider when developing a comprehensive federal law that will provide protection for consumers while allowing genetic testing and any resulting scientific benefits to continue. Now, it is up to Congress to address these issues and protect the American people and their privacy.

AMERICAN UNIVERSITY
WASHINGTON, DC

American University Washington College of Law

4300 Nebraska Avenue, NW

Washington, D.C. 20016

To submit an original article for possible publication in future issues of *HLPB*, please email your article to healthlawandpolicybrief@wcl.american.edu, or mail a hard copy of your submission to *Health Law & Policy Brief*, American University Washington College of Law, 4300 Nebraska Avenue, NW Washington, D.C. 20016. Please submit your article in doubled-spaced paragraph format using Times New Roman, 12 pt. font. Additionally, please submit a short cover letter containing all authors' contact information, including home addresses, telephone numbers, and email addresses.

HEALTH LAW & POLICY BRIEF
EST. 2007
WASHINGTON COLLEGE OF LAW

AMERICAN UNIVERSITY
WASHINGTON
COLLEGE OF LAW