Introduction

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For the average person genetic testing has two very different faces: the rise of genetic testing is promoted as the democratization of genetics by enabling individuals to gain new insights into their unique makeup. At the same time, many regard genetic testing and sequencing as revealing something intensely personal and private about themselves. Genetic testing raises legal and ethical questions that loom ever larger, especially as genetic testing is becoming more commonplace, affordable, and comprehensive. Already in 2018 the global genetic testing and consumer/wellness genomics market was valued at $3.4 billion, with market analysts in 2019 predicting that it will double in value by 2025.

Direct-to-Consumer (DTC) genetic testing kits have become an essential part of this global industry. Because DTC bypasses traditional trusted health care intermediaries, DTC genomics also raises the legal and ethical stakes. For example, what standards should the regulators like the Food and Drug Administration apply in determining which tests are available to consumers? How should we mediate privacy and other ethical concerns around genetic databases, especially considering that genetic information is at once both individualistic and communal? In an age where serial killers are caught because their relatives chose to submit DNA to a consumer genealogy database, is genetic privacy for individuals really possible?

This volume will consider the ethical, legal, and regulatory challenges presented as genomics become commonplace, easily available consumer products. The most significant focus of the volume is to articulate regulatory solutions to protecting consumer safety and privacy while still promoting innovation. Throughout the volume, our authors evaluate the myriad of models used to deliver consumer genetics, including the traditional health care setting, life insurance, Do-It-Yourself (DIY) Bio, and, most notably, DTC. The DTC delivery model, in particular, requires us to reopen questions that have already been answered for more traditional health care delivery models. The book’s goal is to provide guidance for

medical providers, patients, policymakers, and other stakeholders interested in this field.

The book is divided into five parts. Part I, Consumer Genetic Technologies: Rights, Liabilities, and Other Obligations, introduced by I. Glenn Cohen, orients the reader both to the structure for delivering genetic technologies to consumers and also to the ethical questions raised by the widespread availability of these products. These chapters show how the chimeric nature of consumer genetic technologies make them challenging to regulate and how the diversity in their structure and delivery defeats any attempt at one-size-fits-all regulatory design.

Gary Marchant, Mark Barnes, Ellen Clayton, and Susan Wolf, in their chapter, Liability Implications of Direct-to-Consumer Testing, introduce the reader to direct-to-consumer genetic testing, one of the most common delivery models for genetic technologies. Marchant et al. explore the liability questions raised when genetic tests produce erroneous results. This chapter illustrates the tension between the health care system and the DTC model. When patients give their physicians DTC genetic data, they struggle with whether to use and rely upon this information or to disregard it. By examining the challenges that health care providers face when incorporating DTC products into their work, Marchant et al. help the reader to begin to understand how different DTC genetic technologies can come from genomics delivered more traditionally within the health care system. This chapter is part of a central dialogue of the book – how to choose between self-regulation, increased traditional regulation, or other solutions to governing this space.

Anya Prince, in her chapter, Consuming Genetics as a Life Insurance Consumer, considers a very different delivery model. Prince focuses on the development of life insurers as genetic consumers, initiating testing for potential enrollees. While Marchant et al. are concerned with the impact that consumer-initiated testing will have on medical professionals, Prince wrestles with the opposite scenario: How can individuals be properly protected when they are compelled to undergo genetic testing by a third party?

Jessica Roberts argues for personal property rights as a necessary protection for consumers in her chapter, In Favor of an Action for Genetic Conversion. She notes that consumers who use DTC genetic tests rarely enter into the kind of fiduciary or research relationships with the DTC companies that would provide consumers with protections against breach of fiduciary duty, lack of informed consent, invasion of privacy, or unjust enrichment. Therefore, genetic conversation claims, although historically disfavored by courts, could be a useful tool to protect the rights of consumers.

The next chapter, Jorge Contreras’s Direct to Consumer Genetics and Personal Health Data, offers a very different answer to the same question for consumer genetic technologies, such as “who owns this data?” He recognizes that many consumers expect some sort of personal property rights in their genetic data, and that many DTC-GT companies imply such a right in their advertisements.
Nevertheless, a personal property right in genetic data would be a significant departure from current US law and would cause significant barriers in using genetic data for biomedical research and public health purposes. Therefore, he argues, protecting consumers’ privacy interests can be better accomplished through contractual arrangements. This demands more focus and regulation of DTC providers’ privacy policies and practices, along with regulatory oversight of data governance. (A chapter in the last section of this book, by James Hazel, complements Roberts and Contreras’ work by providing a deep-dive into the current privacy policies of DTC companies.)

Scott Schweikart closes Part I with his chapter, *Governance in the Era of CRISPR and DIY-Bio: Regulatory Guidance of Human Genome Editing at the National and Global Levels*.

Schweikart reminds the reader of the diversity of delivery models for these technologies by comparing the regulation of consumer genetic technologies via “Big-Bio” and “DIY-Bio.” He argues that the traditional top-down regulatory model, used to regulate pharmaceutical safety for example, can be successfully applied to consumer genetic products offered by Big-Bio. By contrast, DIY-Bio consumer genetic offerings will require a more creative bottom-up approach to successfully protect consumers, such as incentives to build proper ethical and regulatory consensus among DIY-Bio groups. Schweikart sets the stage for several later chapters, which make the argument that new delivery models compel novel approaches to regulation.

Part II of this volume, *Privacy in the Age of Consumer Genetics*, introduced by Nita A. Farahany, explores the ethical challenges brought about by increased genetic testing. The first two chapters consider how additional information will affect relationships between biological parents and potential children, introducing readers to another significant theme of the volume: that genetic information is at once intensely personal but also communal, shared between people who may otherwise be strangers to each other. This duality makes it challenging to regulate from a data privacy perspective.

In the first chapter of this Part, *Non-Invasive Prenatal Whole Genome Sequencing: Ethical and Policy Post-Birth Implications*, Vardit Ravitsky considers questions of privacy and right to access information at the start of life. Ravitsky explores the implications of Non-Invasive Prenatal Testing (NIPT), which is increasingly popular among potential parents. Many ethicists are uncomfortable with parent-initiated genetic testing of children, unless there is an immediate health need, in order to preserve that individual’s ability to choose whether or not to undergoing testing once they reach adulthood. On the one hand, this logic applies to NIPT, preventing potential parents from sequencing a fetus’s whole genome. On the other hand, unlike a child, many would argue that a fetus does not have a right to privacy. Ravitsky considers both perspectives, concluding that the wishes of the parent should govern, especially if we are to respect the reproductive autonomy of the pregnant
person. Ravitsky evaluates possible mechanisms to balance the reproductive autonomy of pregnant consumers with the interests of their prospective children.

Seema Mohapatra, in her chapter, *The Myth of “Anonymous” Gamete Donation in the Age of Direct-to-Consumer Genetic Testing*, likewise focuses on privacy in the context of reproduction, this time in the context of anonymous gamete donation. Mohapatra notes that the widespread use of DTC-GT has undermined the ability of gamete donors to be anonymous, despite any legal or regulatory promises. She rightfully argues that a façade of anonymity presents significant legal and ethical quandaries. Mohapatra concludes that the lack of anonymity is a reality that must be disclosed to potential donors, even if it ultimately may deter gamete donation. Privacy as anonymity may be unattainable in gamete donation, so the best we might hope for is informed decision-making by potential donors. Mohapatra’s chapter demonstrates to the reader a unique tension in genetic data privacy, that is, that our genetic material is shared with relatives and our individual privacy can be undermined by the choices of others. This tension is reflected in other chapters, most significantly in Kif Augustine-Adams’ very personal reflection on the topic in Part IV.

Kayte Spector-Bagdady, in her chapter, *Improving Commercial Genetic Data Sharing Policy*, demonstrates that there is a clear gap between the professional privacy standards for DTC consumer genetic testing and the privacy needs of consumers using these tests. Spector-Bagdady argues that the lack of clarity and transparency in the commercial genetic data research industry prevents individuals from making informed choices about using DTC tests and contributing their data to these databases. Spector-Bagdady flags two areas that need improvement in order to achieve “privacy best practices” for the DTC consumer genetics industry: (1) transparency in sharing deidentified health data; and (2) specific consent (as opposed to broad consent). Spector-Bagdady’s concerns, that our consumer genetic privacy standards and frameworks are inadequate, is a pervasive theme in this volume. She is one of many contributors, such as Jessica Roberts in Part I, grappling with the sense that genetic information requires heightened protections and that our delivery models for consumer genetic testing fail to meet these needs.

Yaniv Heled and Liza Vertinsky bring our privacy exploration into the near future with their warning regarding potential genetic paparazzi in their chapter, *Genetic Paparazzi*. They anticipate a future in which the genetic information of public figures will become a subject of fascination and media interest. Will the paparazzi bring swabs and sterile tools along with cameras as they follow celebrities and politicians? If so, Heled and Vertinsky note that the right to privacy exercised by these notable figures will conflict with the right of the public to share newsworthy information. Unfortunately for these celebrities, they argue, the current legal framework does not adequately guard against nonconsensual testing, sequencing, analysis, and publication of genetic material and information. While most of us are not famous enough to need to worry about the genetic paparazzi, Heled and Vertinsky...
Part III, Tinkering with Ourselves: The Law and Ethics of DIY Genomics, introduced by Henry T. (Hank) Greely, turns the focus of our volume from using consumer genetic technologies to the production and delivery of genomics. Called by a variety of names, including DIY gene editing, DIY-Bio, Community Bio, genetic biohacking, or nontraditional gene editing, this community moves genomics from more traditional settings and into people’s private homes. Aided by knowledge sharing communities online, the DIY community is a major player in how genetic technologies are delivered. As Schweikart argued in Part I earlier, this nontraditional system will require innovative approaches to regulation. Otherwise, we run the risk of smothering this novel delivery model and losing a valuable source of innovation in genetic technologies. The chapters in Part III all attempt to address the question: How do the regulatory and ethical questions and answers change when we move away from traditional delivery models into DIY genomics?

Barbara Evans, in her chapter, Programming Our Genomes, Programming Ourselves: The Moral and Regulatory Challenge of Regulating Do-It-Yourself Gene Editing, provides the reader with a solid overview of the difficulties of applying traditional regulations to a decidedly nontraditional scientific ecosystem. Evans documents the “Coordinated Framework,” a compilation of statutes and regulations that are intended to govern DIY gene editing and highlights both the micro-problems and the macro-issue that a DIY community structure does not naturally lend itself to regulation. Evans then challenges the reader to consider whether regulation of DIY gene editing is necessary or justified.

Max Mehlman and Ronald Conlon also consider an appropriate approach to the regulation of DIY gene editing in their chapter, Governing Non-Traditional Gene Editing. The reader will likely be surprised to learn that ham radio, sport rocketry, home brewing, and computer hacking are all useful historic analogies to DIY gene editing analogies that can help inform an appropriate regulatory response. Mehlman and Conlon argue that most of these activities rely heavily on self-regulation, facilitated by organizations that represent amateur’s interests, and include expectations of self-monitoring. They do note that traditional regulation of these types of activities can become appropriate as the community experiences significant growth and activities become riskier. Therefore, they suggest that self-regulation may be appropriate for DIY gene editing at the moment, but external regulation may become the better choice once DIY gene capabilities expand.

Patti Zettler, Christi Guerini, and Jake Sherkow close Part III with a very human focus on the people active in the DIY gene editing community in their chapter, Finding a Regulatory Balance for Genetic Biohacking. Their focus is still on articulating the appropriate regulatory mechanisms for DIY gene editing, but the authors add qualitative interviews with members of the DIY gene editing community.
Zettler, Guerini, and Sherkow argue for a range of regulatory approaches, moving beyond relying on FDA enforcement actions to include educational and engagement activities by other agencies such as the National Science Advisory Board for Biosecurity. They, along with Mehlman, Conlon, and Evans, recognize the valuable innovation that the DIY gene editing community contributes to public health and science, and argue that any regulatory approach should attempt to preserve that perspective.

Part IV, Consumer Genetics and Identity, introduced by Carmel Shachar, focuses on the very personal impact that consumer genetic technologies can have on users. The appeal of genetic testing is that we learn more information about ourselves, but that information is a double-edged sword. Genetic testing can be problematically conflated with race, as Jonathan Kahn describes, or predict an unhappy medical future as Emily Largent discusses. Genetic information is also closely tied to family as demonstrated from very different perspectives in the chapters Kif Augustine-Adams and Natalie Ram. It is becoming increasingly difficult to avoid being affected by genetic testing because it is becoming very likely that at least one of one’s genetic relatives will test, revealing communal genetic information.

Kif Augustine-Adams open Part IV with a powerful personal narrative, Generational Failures of Law and Ethics: Rape, Mormon Orthodoxy, and the Revelatory Power of Ancestry DNA. Augustine-Adams tells the story of her mother, a sexual assault survivor who gave birth in secrecy, pursued a closed adoption, and hoped to keep that chapter of her life secret from her subsequent children. Augustine-Adams also tells her own story, that of a woman discovering a sibling through a third relative’s use of DTC-GT. Augustine-Adams and her mother’s stories collide as Augustine-Adams shares her knowledge with her mother, to her mother’s great distress. Augustine-Adams understands that genetic testing has stolen her mother’s ability to control the narrative of her own life. This chapter focuses the reader on a central problem in the area: How can we preserve individual autonomy and privacy when biological relatives can make choices about our shared genetic information that undermine our personal preferences?

Jonathan Kahn takes us one step broader, away from the family and into questions of race and ethnicity in his chapter, Precision Medicine and the Resurgence of Race in Genomic Medicine. Kahn documents the desire of biomedical researchers over the past decade to explore the complexities of gene environment interactions as to racial groups and the unfortunate tendency to forget the environmental contributions in favor of drawing relationships between genes and race. Kahn notes that race is a useful and convenient category for use in biomedical and genetic projects, acting as a proxy not only for genetic/geographic ancestry but also for social circumstances, such as environment and class. Kahn warns the reader that diversity as a goal in genomics initiatives can introduce a potentially problematic focus on race. This chapter causes the reader to reflect both on the role of genetics and environment in
shaping people, and on how we import our own identities and understanding into genomics.

Emily Largent, in her chapter *Losing Our Minds? Direct-to-Consumer Genetic Testing and Alzheimer’s Disease*, considers the age-old question “is it always better to know or can ignorance be bliss?” in the context of genetic testing. She focuses on a test for a marker of Alzheimer’s Disease, a devastating condition with no treatment or cure, and considers whether the DTC setting is an appropriate one for delivering such significant information. Largent suggests that consumers largely are not harmed by this knowledge and that it is indeed better to know, so that appropriate preparations can be made. Like the other chapters in this Part, Largent focuses on how an individual’s identity may be affected and reshaped by genetic information.

Finally, in her chapter *Investigative Genetic Genealogy and the Problem of Familial Forensic Identification*, Natalie Ram returns the reader to the privacy challenges raised by shared genetic information. Ram considers recent high-profile arrests made on the basis of genetic genealogy, often using distant genetic relatives to identify suspects. Ram notes that genetic relatives share genetic data in ways that are immutable and involuntary. She is concerned with both the broad reach of genetic genealogy, which can be used to identify a majority of Americans of European descent already, and the lack of voluntariness, that is, that individuals are not actively choosing to participate in the databases used by law enforcement. By raising the ethical and legal concerns with the use of genetic genealogy to solve crimes, Ram complicates the media narrative presentation of law enforcement triumphs and reminds the reader that not all applications of technology should be allowed.

Melissa Uveges introduces Part V, *The Impact of Genetic Information*. In some ways this Part is a continuation of Part IV, as both focus on the implications of genetic testing for individuals and families. While Part IV concerns how genetic information informs identity, Part V explores the effects that individuals, families, and health care providers must grapple with as they engage in consumer genetic technologies. Part V introduces genetic counselors as important players in the exercise of consumer genetic technologies, with contributions both about and by genetic counselors. Part V then continues our exploration of the appropriate regulatory approaches to a technology that is often delivered outside of the hospital or clinic, with two chapters that consider the role of traditional regulatory action and self-regulation. By returning to this central theme, Part V invites the reader to consider which regulatory tools they find most likely to strike the right balance between protecting patient safety and privacy while harnessing the innovation and potential of genomics.

Leila Jamal, Will Schupmann, and Benjamin Berkman open this Part with their chapter, *An Ethical Framework for Genetic Counseling in the Genomic Era* by considering the standard of clinical care for genetic counselors and articulating a set of ethical principles that should be used to guide genetic counseling practice.
Many previous chapters have discussed self-regulation from the perspective of those developing and delivering consumer genetic products, but Jamal and her colleagues apply self-regulation to the relatively new profession of genetic counseling. The authors urge genetic counselors to de-emphasize non-directiveness in genetic counseling, arguing that it overprivileges patient autonomy to the detriment of other important ethical values. Genetic counselors should instead be guided by principles of individual and familial beneficence and nonmaleficence, they argue, incorporating factors such as: whether counseling occurs before or after testing, the information burden of testing, the magnitude and risk of the conditions tested for, relational considerations, role-based considerations, and familial considerations.

Emily Qian, Magalie Leduc, Rebecca Hodges, Bryan Cosca, Ryan Durigan, Laurie McCright, Doug Flood, and Birgit Funke continue the exploration of the role of genetic professionals in their chapter, *Physician-Mediated Elective Whole Genome Sequencing Tests: Impacts on Informed Consent*. Qian and her colleagues explore how genetic testing increasingly utilizes a physician-mediated consumer-driven model, in which consumers initiate a request for genetic testing that is then approved by a physician. They then consider the demographics of the providers participating in this model, noting that genetic professionals are rarely included in the initial ordering process and that primary care providers provide the majority of genetic test authorizations. This raises concerns that primary care providers may not be best equipped to counsel patients on their genetic test results and that genetic professionals should enter earlier in the process to ensure informed consent from patients.

Catherine Sharkey and Kenneth Offit then return us to the central question of the best tools to regulate DTC-GT with their chapter, *Regulatory and Medical Aspects of DTC Genetic Testing*. Sharkey and Offit flag the FDA’s recent marketing authorizations for testing for mutations associated with hereditary breast and colon cancer and pharmacogenetic susceptibilities to provide an opportunity to reconsider the regulatory framework for DTC-GT. Tracing the history of DTC-GT and its incorporation into precision/personalized medicine, they explore the FDA’s role as safety regulator and protector of patient privacy. Sharkey and Offit end with an interesting exploration of the FDA’s potential to regulate the big genetic databases potentially amassed by DTC-GT companies. While many previous chapters argued for an emphasis on self-regulation, Sharkey and Offit provide a counterpoint and argue that the FDA has an important role to play in the regulation of some DTC genetic tests.

In the book’s last chapter, *Privacy Best Practices for Direct-to-Consumer Genetic Testing Services: Are Industry Efforts at Self-Regulation Sufficient?*, James Hazel revisits the question of self-regulation. He evaluates the Future of Privacy Forum’s “Best Practices for Consumer Genetic Testing Services,” and considers the extent to which industry leaders have implemented these nonbinding guidelines. Of the eight DTC-GT companies involved with the Best Practices for Consumer Genetic
Testing Services, not one, he notes, had privacy documents that appeared to comply with all aspects of these voluntary guidelines. Hazel’s work is again a counterpoint to many of the calls for self-regulation found throughout this volume. Instead, he argues for the Federal Trade Commission to play a greater role in protecting consumer privacy.

CONCLUSIONS

Consumer genetic technologies are relatively new and still in a period of innovation and uncertainty. As would be expected in such a nascent field, many delivery models are used to allow access to genetic testing technologies, including incorporation into the traditional medical system, DTC delivery of products, and DIY genetic testing. Our volume explores the many different regulatory schemes that could be used to protect consumer privacy and safety. Some of our contributors are concerned that traditional regulation will stifle innovation or be inappropriate for DTC or DIY genetic testing and argue for thoughtful self-regulation. Other contributors worry that self-regulation will be insufficient and that regulatory agencies must think creatively about how to achieve privacy and safety goals in this field. Finding appropriate regulatory tools is an important task for all stakeholders because of the substantial impact that consumer genetic technologies can have on our identities, families, and personal choices.