

# TAKING RESPONSIBILITY: REGULATIONS AND PROTECTIONS IN DIRECT-TO-CONSUMER GENETIC TESTING

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Starting in November 2007, the New York State Department of Health mailed “cease and desist” letters to thirty-one companies, ordering them to stop providing genetic tests directly to consumers without the involvement of a licensed physician.<sup>1</sup> In June 2008, the California Department of Public Health sent similar letters to thirteen genetic testing companies, also involved in direct-to-consumer (DTC) genetic testing services.<sup>2</sup> These cease-and-desist letters echo public concern regarding laboratory testing standards, the need for physician involvement, and the use of misleading advertising.<sup>3</sup> California has since granted licenses to a number of these companies,<sup>4</sup> but discussions regarding the concerns of DTC testing continue. Among these concerns are the lack of standards used to demonstrate the validity of different genetic tests, uncertainty as to whether healthcare professionals must always be involved in the ordering of such tests to protect patients, and lack of consumer understanding regarding the use of genetic testing.<sup>5</sup>

This Note aims to address the debate surrounding genetic testing within the context of the DTC market. Part I describes the potential for genetic testing in the clinical setting as a result of recent scientific advances. Part II provides an introduction to the DTC genetic testing industry, including the business models being employed by various firms. Part II also pro-

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1. Sarah Lynch, *Should Genetic Testing be Regulated?*, TIME, July 22, 2008, available at <http://www.time.com/time/health/article/0,8599,1825539,00.html>.

2. *Id.*

3. *Id.*

4. See e.g., Andrew Pollack, *California Licenses 2 Companies to Offer Gene Services*, N.Y. TIMES, Aug. 19, 2008, at C3.

5. Andrew Pollack, *Gene Testing Questioned by Regulators*, N.Y. TIMES, June 26, 2008, at C1.

vides a more detailed account of the DTC genetic testing debate, with particular focus on the lack of adequate regulations, the lack of required involvement from a licensed physician or genetic counselor, and the lack of adequate consumer protection. Part III discusses solutions to address the risks associated with DTC genetic testing through clarified scientific standards, targeted regulations surrounding the quality of genetic testing, industry self-standardization regarding genetic test reliability and results, and assumption by DTC companies of a duty to warn consumers about potential risks of genetic testing.

## I. OVERVIEW OF GENETIC TESTING

Who we are as individuals is a result, primarily, of two principal factors: genetics and environment.<sup>6</sup> Understanding the influence these factors have on a particular trait, behavior, or as the basis for diseases such as cancer, has been a central theme of biological research for decades.<sup>7</sup> While strategies such as twin studies have led to key insights to the interaction between genetics and environment, until recently our understanding has been limited by technology.<sup>8</sup> However, with the completion of the human genome project and modern advances in genetic analytical techniques, rapid progress is being made in identifying which genes, and to what extent genetics guide who we are as individuals.<sup>9</sup>

Translating gene-trait association studies towards healthcare has become a cornerstone of what is termed “personalized medicine,” or the tailoring of medical treatments, both responsive and anticipatory, based in part on an individual’s genes.<sup>10</sup> The growing understanding of how our genetics influence us as people has also led to the marketing of genetic tests to consumers directly over the Internet.<sup>11</sup> This DTC commercial ge-

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6. See generally Muin J. Khoury et al., *Do We Need Genomic Research for the Prevention of Common Diseases with Environmental Causes?*, 161 AM. J. EPIDEMIOLOGY 799, 800 (2005) (discussing the importance of understanding genetic risk factors with known environmental causes).

7. See *id.*

8. Alan E. Guttmacher & Francis S. Collins, *Welcome to the Genomic Era*, 349 NEW ENG. J. MED. 996, 996-98 (2003).

9. *Id.*

10. See generally John Bell, *Predicting Disease Using Genomics*, 429 NATURE 453 (2004) (discussing the use of genetics in predicting disease, drug discovery, disease monitoring, and clinical practice).

11. Gail H. Javitt, *Policy Implications of Genetic Testing: Not just for Geneticists Anymore*, 13 ADV. CHRONIC KIDNEY DISEASE 178, 179 (2006).

netic testing industry has seen rapid growth in recent years, preceding not only federal regulations, but often the science itself.

#### A. The Science of Genetic Testing Has Advanced Rapidly in Recent Years

While a number of scientific events have advanced our understanding of human genetics, two in particular—the completion of the human genome and the increase of gene-trait association studies—have laid the groundwork for genetic testing as it is used today. The completion of the human genome in 2001 marked a landmark achievement in understanding human biology by providing a list of the genes and intergenic regions within human chromosomal DNA.<sup>12</sup> Subsequent drafts and annotations of the genome have given a refined list of 20,000-25,000 protein-coding genes and just over 3 billion nucleotide basepairs.<sup>13</sup> This completion provided researchers and doctors with a reference sequence from which further genetic study could take place.<sup>14</sup>

Following the completion of the human genome, researchers increasingly studied how genetic variation contributes to heritable traits and diseases.<sup>15</sup> Most aspects of human biology stem from either genetic (hereditary) or environmental (non-hereditary) factors. While some genetic traits, such as height and eye color, have little influence from environmental factors, others, such as obesity and some forms of cancer, only manifest in combination with certain environmental conditions.<sup>16</sup>

Identifying genetic variations and understanding their physiological manifestation (termed “phenotype”) has become a centerpiece in this new age of genetics. There are many different kinds of DNA sequence variations, ranging from complete, extra, or missing chromosomes down to single nucleotide changes.<sup>17</sup> Most studies of human genetic variation have focused on single nucleotide polymorphisms (SNPs), which are substitutions in individual bases along a chromosome.<sup>18</sup> Experts estimate that

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12. Guttmacher & Collins, *supra* note 8, at 996.

13. Int'l Human Genome Sequencing Consortium, *Finishing the Euchromatic Sequence of the Human Genome*, 431 NATURE 931, 943 (2004).

14. Guttmacher & Collins, *supra* note 8, at 996.

15. *See* Bell, *supra* note 10.

16. Khoury, *supra* note 6, at 802 (discussing approaches to identifying gene-environment interaction).

17. *See* Nicholas Wade, *Genetic Catalog May Aid Search for Roots of Disease*, N.Y. TIMES, Oct. 27, 2005, at A20.

18. *Id.*

SNPs occur on average somewhere between every 1 in 100 and 1 in 1,000 base pairs in the human genome.<sup>19</sup> By conducting familial studies or larger “genome-wide association studies,” researchers look to link genetic variation to phenotypes with statistical significance.<sup>20</sup> The linkage serves as the scientific basis for genetic tests: by testing for specific genetic variations, physicians can determine risk for disease, understand behavioral characteristics, or identify genetic causes of existing conditions.<sup>21</sup> These studies have led to genetic tests for approximately 1,400 genetic variations, with more than 1,000 additional tests currently in development.<sup>22</sup>

### **B. Advances in Genetic Testing Has Brought the Promise of “Personalized Medicine”**

Genetic testing represents a key component of the use of new methods of molecular analysis and bioinformatics to better manage a patient’s disease or predisposition to disease, otherwise known as “personalized medicine.” In a medical context, information about a patient’s genes, gene expression profile, or “genotype” could be used to tailor medical care to an individual’s needs.<sup>23</sup> Understanding the genetic profile of an individual would, in theory, assist clinicians in identifying predisposition for disease, carriers for disease, or drug sensitivities; would make newborn screening and prenatal testing more effective; and might even facilitate the drug design process. Already, for example, genetic testing has allowed for huge inroads in oncology, both in understanding the genetic makeup of cancer cells to allow for better prognosis and treatment, as well as screening individuals thought to be at risk for hereditary forms of cancer.<sup>24</sup>

### **C. Better Technology Has Led to Direct-to-Consumer Marketing of Genetic Tests**

Along with the advance of our understanding of genetics and heredity, there has been a recent proliferation of commercially available tests mar-

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19. Int’l HapMap Consortium, *A Haplotype Map of the Human Genome*, 437 NATURE 1299, 1301 (2005).

20. Thomas A. Pearson & Teri A. Manolio, *How to Interpret a Genome-Wide Association Study*, 299 JAMA 1335, 1335 (2008).

21. *Id.*; see also Khoury, *supra* note 6.

22. See Charles Schmidt, *Regulators Weigh Risks of Consumer Genetic Tests*, 26 NATURE BIOTECH. 145, 145 (2008) (quoting Steve Gutman, FDA’s director for *in vitro* diagnostics).

23. Khoury, *supra* note 6.

24. See *American Society of Clinical Oncology Policy Statement Update: Genetic Testing for Cancer Susceptibility*, 21 J. CLINICAL ONCOLOGY 2397, 2397-406 (2003) (discussing the use of genetic testing in clinical oncology).

keted directly to consumers. One manner of DTC genetic testing advertises to consumers but still requires the prescription of a physician. This is similar to the DTC advertising of prescription pharmaceuticals. For example, Myriad Genetics' BRACAnalysis test for susceptibility to hereditary breast and ovarian cancer became extensively advertised in 2002 using a variety of media outlets.<sup>25</sup> This advertisement saw a marked rise of sales among women within the age group of twenty-five to fifty-four years, despite being appropriate for only a small percentage of patients with a strong family history of breast cancer.<sup>26</sup>

Many DTC companies are marketing and selling genetic tests directly to consumers, completely circumventing the involvement of a trained physician. These companies provide genetic testing not only for health-related purposes, but also to provide information about a person's ancestry, behavior and personality, and for paternity testing and employment screening services. For example, one Internet-based dating service, genepartner.com, works to "match men and women by analyzing specific genes in their DNA."<sup>27</sup>

There are three principal, nonexclusive business models used by the DTC genetic testing market. The first model profits through the sale of the tests themselves. Costing between approximately \$400<sup>28</sup> and \$250,000,<sup>29</sup> these tests analyze a consumer's DNA, assess variability at 500,000 to 1 million SNP sites, or completely sequence the full genome of a customer.<sup>30</sup> These services typically store the data in an online private account, compare the results with "phenotype databases"<sup>31</sup> maintained by the com-

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25. Bryn Williams-Jones, *'Be Ready Against Cancer, Now': Direct-to-Consumer Advertising for Genetic Testing*, 25 *NEW GENETICS & SOCIETY* 89, 92 (2006).

26. Andrew Pollack, *A Genetic Test That Very Few Need, Marketed to the Masses*, *N.Y. TIMES*, Sept. 11, 2007, at C3; see also Melanie F. Myers, et al., *Genetic Testing for Susceptibility to Breast and Ovarian Cancer: Evaluating the Impact of a Direct-to-Consumer Marketing Campaign on Physicians' Knowledge and Practices*, 8 *GENETICS & MEDICINE* 361, 365-67 (2006).

27. Gene Partner Home Page, <http://www.genepartner.com> (last visited Dec. 10, 2008).

28. See 23andMe Home Page, <https://www.23andMe.com> (last visited Feb. 4, 2009).

29. See Knome Home Page, <https://www.knome.com> (last visited Feb. 4, 2009).

30. David J. Hunter, Muin J. Khoury, & Jeffrey M. Drazen, *Letting the Genome out of the Bottle—Will We Get Our Wish?*, 358 *NEW ENG. J. MED.* 105, 105 (2008).

31. "Phenotype" refers to any observable characteristic or trait of an organism: such as its morphology, development, biochemical or physiological properties, or behavior.

pany, and provide the consumer with updated readouts of his or her level of risk for specific conditions, often as a subscription service.<sup>32</sup> In many instances, DTC companies do not conduct the tests themselves, but instead outsource the testing services and act as the consumer-friendly intermediary.<sup>33</sup> Because operational costs required for performing these testing services often remain a trade secret, it is unclear if selling genetic tests is the main source of revenue for many companies.

The second business model is based on the value of the aggregate genetic data collected from customers. As described in Section I.B, gene-trait association studies are an important tool in understanding how genetic factors impact disease and behavior.<sup>34</sup> By collecting genetic information from hundreds of thousands of individuals, DTC firms hope to develop a valuable resource for drug development, discovery of rare genetic markers and rare carriers, and identification of target populations for clinical trials.<sup>35</sup> While specific dollar prices would be determined by the size and scope of the databases, this collection of genetic profiles—and customer contact information—holds significant financial value.<sup>36</sup>

DTC companies also look to a third model for developing revenue—using genetic testing as a form of targeted marketing. Using this approach, DTC companies would combine their testing with the sale of products or treatments. For example, the sale of a vitamin regimen “tailored” to an individual consumer based on their genetic tests.<sup>37</sup> 23andMe, a Google-backed leader in DTC genetic testing reserves the right to enter into commercial arrangements to provide products and services to individual users and to collect fees from these referrals.<sup>38</sup> The financial value of such ser-

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“Phenotype databases” utilize a standardized lexicon of phenotypes, phenotypic descriptions and known causes.

32. *Id.*

33. *See* 23andMe, *supra* note 28.

34. David Gurwitz, Jeantine E. Lunshof, & Russ B. Altman, *A Call for the Creation of Personalized Medicine Databases*, 5 *NATURE REV. DRUG DISCOVERY* 23, 24 (2006).

35. *Id.*

36. *Id.*

37. *At Home DNA Tests: Marketing Scam or Medical Breakthrough?: Hearing Before the S. Spec. Comm. on Aging*, 109th Cong. 13-25 (2006) [hereinafter *Hearing*] (testimony of Gregory Kutz, U.S. Gov. Accountability Office [GAO]).

38. 23andMe Privacy Statement, <https://www.23andme.com/about/privacy> (last modified Nov. 11, 2007).

vices is unclear, and the benefit to customers has been openly questioned.<sup>39</sup>

## II. THE INDUSTRY-WIDE DEBATE ON MARKETING GENETIC TESTS DIRECTLY TO CONSUMERS

As genetics has become more prominent in healthcare, issues surrounding privacy, consumer-patient protection, and discrimination have come to the forefront of the healthcare debate. The recently passed Genetic Information Nondiscrimination Act of 2008 (GINA),<sup>40</sup> highlighted the urgency in developing proper federal regulations surrounding genetic testing, stating:

The early science of genetics became the basis of State laws that provided for the sterilization of persons having presumed genetic “defects” such as mental retardation, mental disease, epilepsy, blindness, and hearing loss, among other conditions. The first sterilization law was enacted in the State of Indiana in 1907. By 1981, a majority of States adopted sterilization laws to “correct” apparent genetic traits or tendencies. Many of these State laws have since been repealed, and many have been modified to include essential constitutional requirements of due process and equal protection. However, the current explosion in the science of genetics, and the history of sterilization laws by the States based on early genetic science, compels Congressional action in this area.<sup>41</sup>

The emergence of direct to consumer genetic testing in particular has drawn further attention from a broad range of industry and nongovernmental stakeholders as well as a number of government regulatory agencies. In November 2004, the Secretary’s Advisory Committee for Genetics Health and Society (SACGHS), an advisory panel established to broadly consider and advise the Secretary of Health and Human Services on the impact of genetic technologies, formally urged then-Secretary Tommy Thompson to conduct an analysis of the public health impact of DTC advertising and access to genetic tests.<sup>42</sup> In a 2007 draft report titled “U.S. System of

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39. *Hearing, supra* note 37 (testimony of Gregory Kutz, GAO).

40. Genetic Information Nondiscrimination Act of 2008, Pub. L. No. 110-233, 122 Stat. 881 (to be codified in scattered sections of 29 and 42 U.S.C.).

41. *Id.* § 2(2).

42. Letter from Reed V. Tuckson, Chair of Sec’y’s Advisory Comm. on Genetics, Health & Soc’y, Nat’l Inst. of Health, to Tommy G. Thompson, Sec’y of Health & Hu-

Oversight of Genetic Testing: A Response to the Charge of the Secretary of HHS,” SACGHS identified a number of concerns, such as: the risk that tests used by DTC companies which have no clinical relevance would have a negative impact on public perception of genetics in medicine generally, the increasing burden on patients who must interpret complicated genetic data without the guidance of a medical professional, and the increasing strain on physicians who are not trained in genetics.<sup>43</sup> To further examine the possible negative consequences of DTC genetic testing, SACGHS recommended that HHS should increase efforts through collaborations among relevant federal agencies,<sup>44</sup> states, and consumer groups to assess the implications of DTC advertising and consumer-initiated genetic testing and, as necessary, propose strategies to protect consumers from potential harm.<sup>45</sup>

While the consequences of DTC genetic testing are still being assessed, most of the issues have been identified. Critics of the industry focus on the lack of comprehensive regulatory oversight of genetic tests, the need for trained physicians to properly understanding the results, and the need for consumer protection to ensure that individuals are properly informed as to the significance of their genetic results.<sup>46</sup> Proponents of the industry, however, claim that DTC genetic testing allows consumers to take control of their own health and that, on a fundamental level, consumers have the right to know about their own genetic makeup.<sup>47</sup>

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man Serv. (Dec. 8, 2004), available at <http://oba.od.nih.gov/oba/sacghs/reports/DTCletter.pdf>.

43. See SEC’Y’S ADVISORY COMM. ON GENETICS, HEALTH & SOC’Y [SACGHS], U.S. SYSTEM OF OVERSIGHT OF GENETIC TESTING: A RESPONSE TO THE CHARGE OF THE SECRETARY OF HEALTH & HUMAN SERVICES (2008), [http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS\\_oversight\\_report.pdf](http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS_oversight_report.pdf).

44. Relevant federal agencies include the Food and Drug Administration (FDA), the National Institutes of Health (NIH), the Centers for Disease Control and Prevention (CDC), and the Federal Trade Commission (FTC).

45. SACGHS, *supra* note 43, at 9.

46. Gail H. Javitt & Kathy Hudson, *Federal Neglect: Regulation of Genetic Testing*, 22 ISSUES SCI. & TECH. 59, 63 (2006) (discussing the potential harms of direct-to-consumer genetic testing).

47. 23andMe Policy Forum: Direct to Consumer Genetic Testing, <https://www.23andme.com/about/policy/> (last visited Dec. 10, 2008).

## A. Direct-to-Consumer Genetic Testing Raises Three Central Criticisms

The rise of the DTC genetic testing industry has brought with it many benefits, concerns, and much debate. Three central criticisms comprise the bulk of the debate. First, federal regulation of genetic tests is not clearly defined, regardless of whether the tests are used in healthcare or through DTC genetic testing services. The inadequacy of regulation has resulted in no oversight or assurances as to the validity of genetic tests. Second, concern exists over administration of the tests without the counsel of trained healthcare professionals, such as doctors, nurses, or genetic specialists. This concern stems from the varying complexity of genetic tests, many of which require proper interpretation from a licensed professional. Finally, little active regulation exists to prevent false or misleading advertisement by DTC companies, such as making exaggerated or misleading claims as to the efficacy of particular genetic analyses. This concern is complicated by the lack of approved methods for determining the utility and accuracy of genetic tests coupled with the evolving scientific understanding of gene-trait associations.

### 1. Federal Regulations of Genetic Tests Are Poorly Defined

At present, regulatory oversight of genetic testing, both within the DTC and healthcare context, is shared between three authorities: the Centers for Medicaid and Medicare Services (CMS) under the Clinical Laboratory Improvement Amendments of 1988 (CLIA),<sup>48</sup> the Food and Drug Administration (FDA), and state health agencies.<sup>49</sup> However, no single body has clear authority over the accuracy, design and application of many genetic tests being used today.<sup>50</sup>

The authority that each agency has over genetic testing depends on how the genetic test in question is marketed, produced, performed, and interpreted.<sup>51</sup> For example, Laboratory Developed Tests (LDTs)—genetic tests that are designed and produced within a clinical laboratory—must be compliant with CMS regulations.<sup>52</sup> In contrast, test kits—genetic tests

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48. Clinical Laboratory Improvement Amendments of 1988, 42 U.S.C. § 263a (2006).

49. Javitt & Hudson, *supra* note 46, at 59-62 (discussing the various forms of regulatory oversight).

50. *Id.* at 61.

51. *Id.*

52. *Id.* at 60.

which are produced, marketed, and sold by a manufacturer—fall under FDA regulation.<sup>53</sup> However, in some instances FDA authority extends to LDTs, while some test kits can only be conducted in a CLIA compliant laboratory.<sup>54</sup> Due to this patchwork of regulations, a number of regulatory gaps exist that have allowed questionable tests to be directly marketed to consumers without physician involvement.

a) Regulations of Genetic Testing Under CLIA

Laboratories that perform genetic testing for health-related purposes must be certified by the CMS under the Clinical Laboratories Improvements Amendments of 1988 (CLIA).<sup>55</sup> Congress enacted CLIA to ensure that medical testing within clinical laboratories is consistently executed in a valid and reliable manner;<sup>56</sup> the validity of the tests themselves, however, is not under CLIA authority. The stringency of CMS oversight under CLIA depends on the complexity of the test. “High complexity” tests are generally grouped according to “specialty areas” and are subject to additional requirements to ensure safety.<sup>57</sup> In particular, they are subject to specified “proficiency testing” standards, where they must show their ability to accurately perform their tests.<sup>58</sup> There is at present no CLIA specialty area for genetic testing, and in 2007, over the protests of industry and public policy stakeholders, the CMS announced its intent to not create such a specialty.<sup>59</sup> The result is that, although clinical laboratories must meet specific requirements regarding the accuracy of genetic tests, there is no requirement under CLIA for genetic tests to meet standards for validity or utility regarding the interpretation of the genetic test result.<sup>60</sup>

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53. *Id.* at 61.

54. *Id.*

55. 42 U.S.C. § 263a.

56. *See id.*

57. *See* 42 C.F.R. § 493 (describing requirements for “high complexity” tests as well as specialty areas).

58. *See, e.g.*, Testimony of Judy Yost, Sec’y’s Advisory Comm. on Genetics, Health & Soc’y, Nov. 13, 2006, [http://www4.od.nih.gov/oba/SACGHS/meetings/Nov2006/transcripts/Genetic\\_Tech-Hamilton-Yost.pdf](http://www4.od.nih.gov/oba/SACGHS/meetings/Nov2006/transcripts/Genetic_Tech-Hamilton-Yost.pdf).

59. *See* Petition for Rulemaking from Kathy Hudson et al., Genetics & Pub. Policy Ctr., to Mark McClellan, Ctr. for Medicare & Medicaid Serv. (Sept. 26, 2006), *available at* [http://www.dnapolicy.org/resources/Petition\\_For\\_Rulemaking\\_September\\_2006.pdf](http://www.dnapolicy.org/resources/Petition_For_Rulemaking_September_2006.pdf); Reply from Dennis Smith, Ctr. for Medicare & Medicaid Serv., to Kathy Hudson, Genetics & Pub. Policy Ctr., *available at* [http://www.dnapolicy.org/resources/CMSresponse\\_8.15.07.pdf](http://www.dnapolicy.org/resources/CMSresponse_8.15.07.pdf).

60. *See* GENETICS & PUB. POLICY CTR., WHO REGULATES GENETIC TESTS?, (2006), [http://www.dnapolicy.org/images/issuebriefpdfs/Who\\_Regulates\\_Genetic\\_Tests\\_Issue\\_Brief.pdf](http://www.dnapolicy.org/images/issuebriefpdfs/Who_Regulates_Genetic_Tests_Issue_Brief.pdf).

## b) Regulation of Genetic Testing Under the FDA

While CLIA regulations cover proficiency standards of genetic-testing laboratories, the Medical Device Amendment of the Federal Food, Drug, and Cosmetic Act (FDCA) grants the FDA authority to regulate the genetic tests themselves.<sup>61</sup> FDA regulation categorizes genetic tests as “medical devices,” which include an “article” that is “intended for use in the diagnosis of disease or other conditions, or in the cure, mitigation, treatment, or prevention of disease.”<sup>62</sup> Medical devices that are used in laboratory analysis of human specimens are termed “in vitro diagnostic devices” (IVDs).<sup>63</sup> At present, the FDA regulates genetic tests as IVDs (also called “test kits”) if the components of a genetic test are bundled, labeled and sold to a laboratory as a unit.<sup>64</sup> Under FDA regulations, IVDs must undergo successful premarket review of safety, accuracy, and utility before they may be distributed commercially.<sup>65</sup>

But most of the 1,400 genetic tests available today are not available as test kits. Instead, they are derived or assembled within the clinical laboratories themselves. Known as “laboratory developed tests” (LDTs), these genetic tests are developed in-house using either commercial or custom components or components laboratories create themselves. With one exception,<sup>66</sup> the FDA exercises “enforcement discretion” with respect to

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61. 21 U.S.C. § 360k (2006).

62. 21 U.S.C. §§ 201(h), 321(h) (2006).

63. See GENETICS & PUB. POLICY CTR, FDA REGULATION OF GENETIC TESTS (2006), [http://www.dnapolicy.org/images/issuebriefpdfs/FDA\\_Regulation\\_of\\_Genetic\\_Test\\_Issue\\_Brief.pdf](http://www.dnapolicy.org/images/issuebriefpdfs/FDA_Regulation_of_Genetic_Test_Issue_Brief.pdf) (discussing how IVDs are defined as “reagents, instruments, and systems intended for use in the diagnosis of disease or other conditions, including a determination of the state of health, in order to cure, mitigate, treat, or prevent disease or its sequelae”).

64. *Id.*

65. *Id.*

66. In September 2006, the FDA released a draft guidance document addressing a subset of LDTs that the agency termed in vitro diagnostic multivariate index assays (IVDMIAAs). These tests use laboratory data and an algorithm (analytical tool) to generate a result for the purpose of diagnosing, treating, or preventing disease. Examples of IVDMIAA tests include those used to diagnose and guide treatment decisions for breast cancer, prostate cancer recurrence, cardiovascular disease, and Alzheimer disease. The draft guidance stated that the FDA considered IVDMIAAs to be medical devices and that the FDA would require them to undergo premarket review before being marketed. The FDA issued a revised draft guidance document in July 2007. See FDA, DRAFT GUIDANCE FOR INDUSTRY, CLINICAL LABORATORIES, AND FDA STAFF: IN VITRO DIAGNOSTIC MULTIVARIATE INDEX ASSAYS (2007), available at <http://www.fda.gov/cdrh/oivd/guidance/1610.pdf>.

LDTs; as a result, many LDTs do not have to undergo any prior review to assess their clinical validity before clinical use. It is left to the discretion of the manufacturer or laboratory to determine whether a test will be developed as a test kit or as an LDT. As a result, about twelve of the nearly 1,400 available genetic tests have undergone FDA review as of 2008.<sup>67</sup> Further, the FDA has not yet officially substantiated the claimed accuracy of the majority of DTC genetic tests.<sup>68</sup> The absence of a cohesive regulatory system for genetic tests has left consumers vulnerable to genetic tests that have unproven medical value.

c) State Regulation of Genetic Testing

CLIA licensure was delegated by the CMS to state authorities, although some states, namely New York and Washington, have adopted standards more stringent than those required for CLIA compliance and have therefore received waivers from CLIA.<sup>69</sup> States also have the authority to regulate who may order genetic tests from laboratories and who may receive the results of those tests, which is particularly relevant to DTC genetic testing.<sup>70</sup> CLIA regulations stipulate that tests can only be ordered by and results reported to an “authorized person,” though states can define for themselves who is an “authorized person.”<sup>71</sup> Often, consumers themselves are considered “authorized persons.”<sup>72</sup>

As of 2007, twenty-five states and the District of Columbia permit DTC testing without restriction, meaning a consumer can order a genetic test and receive his or her results directly, while thirteen states specifically prohibit it.<sup>73</sup> Other states are silent on the issue, effectively allowing DTC testing.<sup>74</sup> In states requiring a physician to order a test and receive the results, companies marketing DTC genetic testing commonly have an ar-

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67. *See id.* For a current list of clinically used genetic tests, see GeneTests Home Page, <http://www.genetests.org> (last visited Feb. 2, 2009).

68. *Id.*

69. Gail H. Javitt, et al., *Direct-to-Consumer Genetic Tests, Government Oversight, and the First Amendment: What the Government Can (and Can't) Do to Protect the Public's Health*, 57 OKLA. L. REV. 251, 274 (2004).

70. *Id.*

71. Aruna Prabhala, *Center Releases Analysis of State DTC Laws*, GENETICS & PUB. POL'Y CENTER NEWSL., July 2007, [http://www.dnapolicy.org/news.eneews.article.nocategory.php?action=detail&newsletter\\_id=24&article\\_id=100](http://www.dnapolicy.org/news.eneews.article.nocategory.php?action=detail&newsletter_id=24&article_id=100).

72. *Id.*

73. *Id.*

74. GENETICS & PUB. POLICY CTR., *DIRECT-TO-CONSUMER GENETIC TESTING: EMPOWERING OR ENDANGERING THE PUBLIC?* (2006), [http://www.dnapolicy.org/images/issuebriefpdfs/2006\\_DTC\\_Issue\\_Brief.pdf](http://www.dnapolicy.org/images/issuebriefpdfs/2006_DTC_Issue_Brief.pdf).

rangement with a physician whose name is used to order tests and report results to the consumer.<sup>75</sup> This practice is also widespread among DTC companies selling tests over the Internet so that requirements under state regulations are met.<sup>76</sup> However, an affiliation with a physician does not necessarily mean that the patient receives any counseling from that physician about the meaning of the results and about future health plans.

At least two state health agencies are trying to regulate DTC genetic testing by asserting that consumers in their state can only take a genetic test with the advice of a doctor.<sup>77</sup> In June 2008, thirteen genetic-testing companies received warning letters from the California Department of Public Health (CDPH) ordering them to stop marketing their genetic tests to California residents without a state license or the involvement of a state-licensed physician.<sup>78</sup> The CDPH's letters followed similar cease-and-desist orders that the New York State Department of Health sent to thirty-one DTC genomics-services and genetic-testing companies in April.<sup>79</sup>

## 2. *Critics Argue for Physician Involvement*

The variation in state regulations of genetic testing highlights a concern that consumers are not able to interpret the results of genetic tests without the involvement of trained counselors or physicians. A 2002 report published in the *Journal of American Medical Association (JAMA)* argued that the complexity of the information involved in genetic tests, the lack of consensus about the clinical utility of some tests, and the complicated social context surrounding genetics all affect the appropriateness of DTC genetic testing.<sup>80</sup> An underlying element of these concerns is that consumers lack the requisite knowledge to make appropriate decisions on whether a test is necessary or how to interpret test results. Even clinically available tests that provide legitimate information as to an individual's ge-

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75. Adam J. Wolfberg, *Genes on the Web—Direct-to-Consumer Marketing of Genetic Testing*, 355 *NEW ENG. J. MED.* 543, 545 (2006).

76. *Id.*

77. See Pollack, *supra* note 5.

78. *Id.*

79. *Id.*

80. Sarah E. Gollust, Sara Chandros Hull & Benjamin Wilfond, *Limitations of Direct-to-Consumer Advertising for Clinical Genetic Testing*, 288 *JAMA* 1762, 1763 (2002).

netic predisposition can be difficult to interpret without the assistance of trained genetics specialists.<sup>81</sup>

The American Medical Association (AMA) voiced this concern in its 2008 position against DTC genetic testing without the personal involvement of a physician.<sup>82</sup> In its resolution, the AMA claimed that the complexity of many of these tests warrants proper interpretation by medical professionals.<sup>83</sup> Unlike a simple pregnancy test that yields a “positive” or “negative” result, or even a more complicated cholesterol test that references a “normal range” but requires interpretation in the context of other factors, results of a genetic test can be much more challenging to interpret.<sup>84</sup> For example, a “positive” result of a genetic test may not necessarily indicate a clinical diagnosis, but instead only reflect an increased risk for developing a disease or condition. The AMA found that DTC companies have the potential to mislead patients into thinking that the results hold significant meaning for their health.<sup>85</sup> Without the proper pre- and post-genetic-test counseling from a health care provider, many of these tests are at best a waste of consumers’ money and at worst could lead consumers into making ill-informed health decisions.

Similar recommendations for the restriction of DTC genetic testing have been made by other groups and professional organizations. In 2007, the American College of Medical Genetics (ACMG) reaffirmed a 2004 policy that “genetic testing should be provided to the public only through the services of an appropriately qualified health care professional,” and warned that “due to the complexities of genetic testing and counseling, the self-ordering of genetic tests by patients over the telephone or the Internet, and their use of genetic ‘home testing’ kits, is potentially harmful.”<sup>86</sup> Even the DTC companies themselves recommend that customers consult licensed practitioners, although their recommendations often fall short of making such a consultation a prerequisite for testing.

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81. Nat’l Human Genome Research Inst., Direct to Consumer Marketing of Genetic Tests, Mar. 23, 2004, <http://www.genome.gov/12010659>.

82. EDWARD LANGSTON, CHAIR, AM. MED. ASS’N, REPORT OF THE BOARD OF TRUSTEES: DIRECT-TO-CONSUMER ADVERTISING AND PROVISION OF GENETIC TESTING (2008), [www.ama-assn.org/ama1/pub/upload/mm/471/bot7.doc](http://www.ama-assn.org/ama1/pub/upload/mm/471/bot7.doc).

83. *Id.*

84. *Hearing, supra* note 37 (statement of Dr. Kathy Hudson, Dir. Genetics & Pub. Policy Ctr.).

85. *See* LANGSTON, *supra* note 82.

86. Am Coll. of Med. Genetics Bd. of Dir., *ACMG Statement on Direct-to-Consumer Genetic Testing*, 6 GENETICS MED. 60 (2004).

3. *Consumer Protections: Ensuring Accurate Marketing of DTC Genetic Tests*

Concern over accuracy in advertising focuses on claims surrounding the interpretation of genetic results, rather than the results themselves. This concern arises from a broad range of underlying causes, starting with poorly understood or exaggerated scientific findings and ranging all the way to consumer fraud. The complexity of gene-trait associations and the lack of concise federal regulations add to this issue, as there is no industry standard to determine when a genetic test is considered “valid.”<sup>87</sup>

In July 2006, the Senate Special Committee on Aging held a hearing based on a year-long Government Accountability Office (GAO) investigation of DTC marketing of genetic tests, part of which involved submitting profiles to various DTC companies for diagnosis.<sup>88</sup> While researching four separate DTC companies, investigators posed as fourteen individual consumers, but used DNA from only two people, a forty-eight-year-old man and a nine-month-old girl.<sup>89</sup> Despite this, investigators received test results that were contradictory and warned of risks for various conditions for diseases such as cancer, heart disease, and “brain aging.”<sup>90</sup> One DTC company further recommended consumers purchase “personalized” nutritional supplements at \$1,200 per year, although the ingredients of such supplements were valued at only \$35 per year.<sup>91</sup> Alan Guttmacher, deputy director of the U.S. National Human Genome Research Institute in Bethesda, Maryland, stated that “it’s clear [the DTC companies] went way out ahead of the science.”<sup>92</sup>

Issues regarding misleading or fraudulent advertising fall under the jurisdiction of the Federal Trade Commission (FTC).<sup>93</sup> The FTC is charged with protecting consumers against unfair or deceptive trade practices, such as false or misleading advertising claims, as authorized by the Federal

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87. Gollust, *supra* note 80.

88. *Hearing, supra* note 37 (testimony of Gregory Kutz, GAO).

89. *Id.*

90. *Id.*

91. *Id.*

92. Gene Russo, *Home Health Tests Are ‘Genetic Horoscopes’*, 443 NATURE 497 (2006).

93. 15 U.S.C. §§ 45(a) 52 (2006); *see also* FTC Policy Statement on Deception, *appended to* In re Cliffdale Associates, Inc., 103 F.T.C. 110, 174 (1984), *available at* <http://www.ftc.gov/bcp/policystmt/ad-decept.htm>.

Trade Commission Act of 1914.<sup>94</sup> This charge has a specific relevance in DTC genetic testing, where companies advertise their products directly to consumers without a health-care intermediary.<sup>95</sup> Under such authority, the FTC is in the position to check the accuracy and scientific support of DTC companies' advertising claims.<sup>96</sup>

The FTC has asserted jurisdiction against manufacturers of a variety of purported health products available without a prescription, including companies claiming to sell products that can result in hair re-growth, cure cancer, or cause weight loss.<sup>97</sup> As many DTC genetic testing services are sold over the Internet, the FTC has asserted its jurisdiction to take action against genetic test advertising that is false or misleading. The FTC conducts periodic sweeps of the Internet to detect fraudulent health claims and sends warnings to those companies in violation.<sup>98</sup> The FTC also initiated a joint effort with the FDA and NIH to identify appropriate targets for legal action.<sup>99</sup> Finally, in 2006, the FTC released a consumer alert titled "At Home Genetic Tests: A Healthy Dose of Skepticism May Be the Best Prescription."<sup>100</sup> This alert warned consumers about companies that claim they can measure the risk for specific diseases, offer "customized" dietary or health recommendations, suggest that certain consumers may be able to withstand certain risks such as smoking or diet, or give information about how a patient may respond to certain prescription drugs.<sup>101</sup>

The FTC's ability to enforce the Federal Trade Commission Act is limited by a few factors. First, the relatively small size of the agency and its scarce resources have forced the FTC to use enforcement discretion in identifying which advertising claims to pursue.<sup>102</sup> The agency has restricted its pursuits to blatantly false claims and health products whose false advertising would cause concrete harm to a large number of people. Operation Cure.all, an FTC enforcement and consumer education campaign, conducted periodic sweeps of the Internet to detect fraudulent

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94. Federal Trade Commission Act of 1914, 15 U.S.C. §§ 41-58 (2006).

95. GENETICS & PUB. POLICY CTR., *supra* note 71.

96. *Id.*

97. Javitt & Hudson, *supra* note 46 at 65.

98. *Id.*

99. *Id.*

100. FTC, AT-HOME GENETIC TESTS: A HEALTHY DOSE OF SKEPTICISM MAY BE THE BEST PRESCRIPTION (2006), available at <http://www.ftc.gov/bcp/edu/pubs/consumer/health/hea02.pdf>.

101. *Id.* at 3.

102. Javitt & Hudson, *supra* note 46 at 65.

health claims and sent warnings to companies in violation.<sup>103</sup> Until now, however, the FTC appears to have taken no legal action against any genetic test advertisements, even those that would appear clearly false and misleading on their face.

In addition, the FTC's ability to enforce the Federal Trade Commission Act is limited since the boundary between what is truthful and what is misleading is not clear. The First Amendment provides broad protection for commercial speech,<sup>104</sup> and the government bears a high burden in proving that speech is harmful and that restrictions are needed to mitigate or prevent such harms.<sup>105</sup> First Amendment protection is afforded only to truthful commercial speech about a lawful activity, but the Supreme Court has provided little guidance in determining what constitutes misleading commercial speech.<sup>106</sup> Even in circumstances in which the Court has identified commercial speech as potentially misleading, the remedy is usually additional disclosure, such as warning labels, instead of an entire ban on the speech.<sup>107</sup>

## B. The DTC Companies' Responses

Criticisms discussed in this Note—lack of clear regulatory authority, importance of physician involvement, and accuracy in advertising claims—have been met with a broad range of responses from DTC genetic testing companies. In the absence of clear federal regulations, many DTC companies have made efforts to comply with state CLIA certification requirements and some have instituted a grading system to reflect the scientific confidence in genetic tests.<sup>108</sup> Further, many DTC companies have employed a trained genetic counselor to either assist with the interpretation of test results or to help guide consumers in understanding the significance of their outcomes.<sup>109</sup> Finally, many companies take a caveat emp-

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103. Press Release, FTC, "Operation Cure.all" Targets Internet Health Fraud (June 24, 1999), available at <http://www.ftc.gov/opa/1999/06/opcureall.shtm>.

104. See 44 Liquormart, Inc. v. Rhode Island, 517 U.S. 484 (1996); Florida Bar v. Went For It, Inc., 515 U.S. 618 (1995); Rubin v. Coors Brewing Co., 514 U.S. 476 (1995).

105. Javitt et al., *supra* note 69 at 287.

106. *Id.* at 254.

107. *Id.* at 298.

108. See discussion *infra* Sections II.B.1-2.

109. *Id.*

tor stance, stating that they have fulfilled their duty to disclose, and that consumers actually have a right to know their genetic information.<sup>110</sup>

1. *Many DTC Companies Institute Their Own Genetic Test Rating System*

While many DTC genetic testing services operating today are CLIA compliant, CLIA itself does not explain how to determine which genetic tests are scientifically valid.<sup>111</sup> DTC services have met this concern in varying manners. Some services, such as 23andME, take an all-inclusive, transparent approach by disclosing the limits of genetic testing, accompanied by a reliability rating or a “research confidence” index, based upon the number and size of the cohort studies.<sup>112</sup> The highest possible rating is “4-stars” and requires multiple cohort studies of 1000 individuals or more.<sup>113</sup> Other DTC services take a “black-box” strategy, exemplified by deCODEme’s enigmatic statement in their FAQ section, that “[t]he information provided by the deCODEme website is as reliable as the statistics of the scientific studies that our calculations are based on.”<sup>114</sup> But deCODEme augments this black-box approach by employing a higher level of selectivity in what it reports to the consumer, stating that it “only reports risk based on well-validated genetic variants” and that “[t]o include risk estimates based on unverified variants, that have only marginal evidence behind them, is unjustified and scientifically unsound.”<sup>115</sup>

The measures taken by these DTC companies do not address the underlying lack of an industry-wide threshold for what constitutes a “scientifically sound” gene-trait association. Issues such as multi-gene factors, rarity of alleles, and population sampling<sup>116</sup> can make it hard to understand gene-trait associations.<sup>117</sup> In 2007, for example, an independent advisory board to the CDC studied the interaction between CYP450 and the metabolism of a class of antidepressant drugs, the selective serotonin reuptake inhibitors (SSRIs). As there was an established interaction between CYP450 and SSRIs, medical professionals understood that genetic testing of the CYP450 genes should help determine the effectiveness and dosage

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110. *Id.*

111. GENETICS & PUB. POLICY CTR., *supra* note 60.

112. 23andMe Home Page, *supra* note 28.

113. *Id.*

114. See deCODEme Frequently Asked Questions, <https://www.decodeme.com/faq> (last visited Feb. 22, 2009).

115. *Id.*

116. For example, all members of a cohort might be eastern European.

117. Khoury, *supra* note 6, at 803.

requirements of certain prescriptions.<sup>118</sup> But the advisory committee, going against conventional understanding, found no evidence “showing that the results of CYP450 testing influenced SSRI choice or dose and improved patient outcomes”.<sup>119</sup> At present, at least fifteen businesses continue to offer CYP450 genotyping services, with four companies making specific claims about the benefit of such testing for SSRI prescribing or dosing.<sup>120</sup> In the absence of an agreed upon and scientifically derived standard for describing the value of a particular genetic test, consumers will not be able to appreciate the uncertainty surrounding many gene-trait associations.

## 2. *Some DTC Companies Involve a Physician or Trained Genetics Expert*

DTC genetic testing services also differ in how they involve a licensed physician. Navigenics and DNA Direct, two industry leaders in the DTC genetic testing field, include a live genetic counselor to discuss results with customers.<sup>121</sup> They market it as a service to help consumers understand the results of their genetic tests. It is unclear, though, whether this is sufficient to inform consumers.<sup>122</sup>

In contrast, many companies, following state law, put the responsibility on the consumer to seek proper consultation from third-party medical professionals. DeCODEme complies with state law so that “unless the Genetic Scan is ordered under the supervision of a physician who provides appropriate counseling, the deCODEme service may omit certain genetic risk information to residents of states where providing such information is restricted.”<sup>123</sup> When there is no state law, deCODEme informs its customers that they “must seek the advice of [their] physician or other qualified

118. S. H. Katsanis et al., *A Case Study of Personalized Medicine*, 320 *SCIENCE* 53, 53-54 (2008).

119. *Id.*

120. *Id.* Inconsistencies regarding which specific CYP450 genes should actually be genotyped for each of the five SSRIs were also observed. This finding demonstrates the lack of consensus within the genetic testing community as to what genes are actually relevant to test for each SSRI. *Id.*

121. DNA Direct Home Page, <http://www.dnadirect.com>; Navigenics Home Page, <http://www.navigenics.com>. (last visited Dec. 10, 2008).

122. See M. K. Cho, M. Arruda & N. A. Holtzman, *Educational Material About Genetic Tests: Does It Provide Key Information for Patients and Practitioners?*, 73 *AM. J. MED. GENETICS* 314 (1997).

123. deCODEme Service Agreement, [http://www.decodeme.com/information/service\\_agreement](http://www.decodeme.com/information/service_agreement) (last visited Dec. 10, 2008).

health provider with any questions [regarding medical matters and] must not disregard professional medical advice or delay in seeking it because of the results of [the consumer's] Genetic Scan or anything [the consumer has] read on the deCODEme Site."<sup>124</sup>

Encouraging customers to seek a physician's advice may help DTC genetic testing companies avoid liability through the "learned intermediary" tort law defense.<sup>125</sup> Used primarily with prescription drugs and medical devices, the duty of care falls on the physicians, or "learned intermediaries," who are in the position to apply their prescribing power to determine whether a drug, treatment, or procedure is appropriate for a patient.<sup>126</sup> The Fifth Circuit stated the policy behind the doctrine as follows:

Prescription drugs are likely to be complex medicines, esoteric in formula and varied in effect. As a medical expert, the prescribing physician can take into account the propensities of the drug, as well as the susceptibilities of his patient. . . . The choice he makes is an informed one. . . . Pharmaceutical companies then, who must warn ultimate purchasers of dangers inherent in patent drugs sold over the counter, in selling prescription drugs are required to warn only the prescribing physician, who acts as a "learned intermediary" between manufacturer and consumer.<sup>127</sup>

Manufacturers of prescription drugs and medical devices similarly meet their duty of care to patients by providing warnings to the prescribing physicians.<sup>128</sup>

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124. *Id.*

125. The learned intermediary doctrine is included within RESTATEMENT (THIRD) OF TORTS § 6(d) (1997). The learned intermediary doctrine provides that:

A prescription drug or medical device is not reasonably safe due to inadequate instructions or warnings if reasonable instructions or warnings regarding foreseeable risks of harm are not provided to:

- (1) prescribing and other health-care providers who are in a position to reduce the risks of harm in accordance with the instructions or warnings; or
- (2) the patient when the manufacturer knows or has reason to know that health-care providers will not be in a position to reduce the risks of harm in accordance with the instructions or warnings.

*Id.*

126. Pilar N. Ossario, *Product Liability for Predictive Genetic Tests*, 41 JURIMETRICS J. 239, 255 (2001).

127. *Reyes v. Wyeth Labs.*, 498 F.2d 1264, 1276 (5th Cir. 1974).

128. *Id.*

Since the increase of DTC advertising of pharmaceuticals in the 1990s, the learned intermediary defense has been weakened by the courts as the duty to warn has been partially shifted back to drug manufacturers. In *Perez v. Wyeth Laboratories, Inc.*, a group of women sued Wyeth for injuries suffered while using the contraceptive Norplant.<sup>129</sup> They contended that the manufacturer failed to adequately warn them of the drug's side effects.<sup>130</sup> The trial court dismissed their claim under the learned intermediary doctrine, but the New Jersey Supreme Court reversed.<sup>131</sup> The state supreme court held that Wyeth engaged in a nationwide DTC advertising campaign targeting women, not physicians.<sup>132</sup> Because Wyeth had marketed their drug directly to consumers, they had a duty to properly warn consumers of the drug's adverse effects.<sup>133</sup>

Though still an unsettled question, a duty to inform—as well as the learned intermediary defense—may well apply to DTC genetic testing. While many DTC genetic testing services are available without a prescription, these tests are still highly complex, individualized by nature, and their results can significantly influence the health and lifestyle choices of a consumer. Many of the more severe concerns discussed by medical ethicists, such as undergoing an abortion based on prenatal genetic testing,<sup>134</sup> typically require a physician to administer the procedure. But lesser harms, such as consumers using genetic test results to alter their own drug regimen, can be conducted independent of a doctor's orders. Further, most practicing physicians are poorly trained in medical genetics and may be unable to effectively determine the appropriateness of a genetic test for a particular patient.<sup>135</sup> By marketing directly to consumers without the in-

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129. 734 A.2d 1245 (N.J. 1999).

130. *Id.* at 1248.

131. *Perez*, 734 A.2d 1245.

132. *Id.* at 1262.

133. *Id.*

134. Victoria Colliver, *Home DNA Tests: When You Just Have to Know*, S.F. CHRON., Aug. 21, 2007, at C1.

135. See Louise Wilkins-Haug et al., *Gynecologists' Training, Knowledge and Experiences in Genetics: A Survey*, 95 OBSTETRICS & GYNECOLOGY 421, 424 (2000) (reporting that 65% of gynecologists responded that they had received no formal training in the use of molecular genetic tests); see also A. Hunter et al., *Physician Knowledge and Attitudes Towards Molecular Genetic (DNA) Testing of Their Patients*, 53 CLINICAL GENETICS 447, 450 (1998) (finding that 57% of physicians were neutral or uncertain about their ability to counsel patients about cystic fibrosis risks based on genetic test results, and that over half of physicians surveyed were neutral or uncertain about their abilities to counsel patients about risks for common genetic disorders).

volvement of a physician or trained counselor, the duty to warn of the risks associated with genetic testing could fall to the DTC companies themselves.

At least one industry leader has taken a publicly defiant approach regarding the ability of consumers to interpret their own genetic test results. 23andMe, a leader in the DTC industry, has argued that individual autonomy has made knowledge of one's own genes a right.<sup>136</sup> In their public policy statement, 23andMe states:

Genetic information is a fundamental element of a person's body, identity and individuality. As such, the rights that people enjoy with regard to financial, medical and other forms of personal information should apply to genetic information as well. . . . We believe our customers are capable of understanding the context of the information we provide them. We also think the benefits our customers accrue in accessing their genetic information outweigh potential risks.<sup>137</sup>

This argument, however, is only partially valid in the given context. As discussed above in Section I.I.C, DTC companies not only report on a customer's genes but also interpret the results for them. Customers receive not only their specific genetic makeup but also receive indices describing their risk for heart attack, cancer, and a host of other genetically-linked diseases.<sup>138</sup> This service goes beyond providing information for consumers and ventures into the realm of medical advice.

### III. DISCUSSION

This Note has so far discussed three core concerns around DTC genetic testing. First, regulatory gaps offer little control over scientific standards on how to determine which genetic tests should and should not be marketed to individuals.<sup>139</sup> Second, administering test results without proper guidance from a trained health-care practitioner could lead to misapplication of the results, increasing the risk to the consumer.<sup>140</sup> Third, consumer protections are ineffective against false or misleading advertising and are ineffective at ensuring consumers understand the limits to genetic tests.<sup>141</sup>

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136. 23andMe Policy Forum, *supra* note 47.

137. *Id.*

138. *See* 23andMe, *supra* note 28.

139. *See supra* Section II.A.1.

140. *See supra* Section II.A.2.

141. *See supra* Section II.A.3.

These issues not only pose risks to individuals, but also create an environment that could threaten the growth of an industry with powerful health and scientific potential. DTC testing companies have taken steps toward meeting the concerns of critics, but these efforts have been incomplete and ad hoc.

There are at least two approaches for safeguarding against these risks. The first is through improving government oversight of both medical genetic testing and DTC genetic testing. A fundamental element of this strategy is developing a transparent method for assessing and communicating the scientific validity of available genetic tests. Adopting validity standards would facilitate the development of comprehensive regulations, whether through existing authorities or new legislation. The second approach is through industry self-regulation. In this approach, industry must adopt scientific standards for genetic tests and improve communication with consumers. These two approaches are not mutually exclusive and can be used in conjunction with one another. Thus, it may be in the best interest of the DTC industry itself to impose reasonable regulation in the near future in order to limit its tort liability.

#### **A. Improving Government Oversight of DTC Genetic Testing**

The role of relevant government authorities—CMS, FDA and FTC—in the regulation of genetic testing is poorly defined. A number of stakeholder groups and the authors of pending legislation have all expressed the need for a transparent and robust approval process for genetic tests. But no regulatory authority can effectively develop an approval process without a uniform mechanism for assessing the accuracy, quality, and utility of genetic tests.

##### *1. Developing Standards in Genetic Testing: the ACCE Framework*

At present, the scientific support for many gene-trait associations is weak due to limited data.<sup>142</sup> Because of the observational nature of gene-trait association studies, the association between a single gene variant and disease state can be confounded by a number of factors, including variation in other genes, environmental exposures, population stratification, and

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142. Joel N. Hirschhorn et. al., *A Comprehensive Review of Genetic Association Studies*, 4 GENETICS MED. 45, 60 (2002).

other individual differences between cases and controls.<sup>143</sup> Even if replicated, association studies do not necessarily imply causality.<sup>144</sup>

In 2004, the Centers for Disease Control and Prevention (CDC) initiated a pilot project titled “Evaluation of Genomic Applications in Practice and Prevention” (EGAPP), an independent expert panel, to expand and contextualize the term “clinical utility.”<sup>145</sup> The project’s goal is “to establish and evaluate a systematic, evidence-based process for assessing genetic tests and other applications of genomic technology in transition from research to clinical and public health practice.”<sup>146</sup> Out of this mission, EGAPP developed a framework to evaluate the overall benefit of genetic tests based upon four criteria: (1) “the analytic utility”—the accuracy of test identifying the biomarker, (2) “the clinical validity”—the relationship between the biomarker and clinical status, (3) “the clinical utility”—the likelihood that test will lead to an improved health outcome, and (4) “the ethical, legal and social implications.” This four-factor test is known as the “ACCE” framework.<sup>147</sup> Each of the four components contained a series of specific criteria, from which the overall benefit of the genetic tests could be determined.

Adopting the ACCE model or a similar framework to communicate the quality and validity of a genetic test is an important first step. The current oversight system does not ensure the analytic validity, clinical validity, or the clinical utility of genetic tests.<sup>148</sup> The development of such standard criteria would increase consumer understanding of test quality, protect consumers from erroneous results, and assist in properly conveying the nature of understanding behind a gene-trait association. Further, the adoption of such standards is likely a prerequisite for the government or industry to be able to regulate the industry.

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143. John D. Potter, *At the Interfaces of Epidemiology, Genetics and Genomics*, 2 NATURE REV. GENETICS 142-147 (2001).

144. CDC, Genetic Testing, <http://www.cdc.gov/genomics/gtesting.htm> (last visited Dec. 10, 2008).

145. CDC, Evaluation of Genomic Applications in Practice and Prevention Home Page, <http://www.cdc.gov/genomics/gtesting/EGAPP/about.htm> (last visited Dec. 10, 2008).

146. *Id.*

147. CDC, ACCE Framework, <http://www.cdc.gov/genomics/gtesting/ACCE.htm> (last visited Dec. 10, 2008).

148. GENETICS & PUB. POLICY CTR., *supra* note 60.

## B. Enforcing Accuracy Standards in Genetic Testing Through CLIA

Once a standard for assessing test validity has been established, the development of effective federal regulation would require two measures. First, CMS must develop a CLIA specialty area for genetic testing to ensure proficiency testing of genetic tests, appropriate quality control standards, and to maintain accuracy in testing.<sup>149</sup> The failure of CMS to create a genetic testing specialty has resulted in inadequate federal oversight of the laboratories conducting genetic tests.<sup>150</sup> As the number of new genetic testing technologies continues to increase, the need to ensure that laboratories properly use these advances grows.<sup>151</sup> By creating a CMS genetic test specialty focused on key quality requirements<sup>152</sup> and proficiency standards for laboratory personnel, CMS would help ensure that both medical and DTC genetic testing are conducted accurately and consistently.

This approach is not without support. In May 2000, CMS published a Notice of Intent in the Federal Register regarding the development of a genetic specialty area under CLIA.<sup>153</sup> Of the fifty-seven responses received, 93% supported the recommendation.<sup>154</sup> In September 2006, three nonprofit centers, Johns Hopkins University's Genetics & Public Policy Center, Public Citizen, and Genetic Alliance, filed a Citizen Petition asking CMS to strengthen its oversight of genetic tests by creating a genetic testing specialty, concluding:

Making sure that laboratories can accurately and reliably perform genetic tests is a fundamental requirement for the success of genetic medicine, and a fundamental obligation of CMS under CLIA . . . [B]ecause of CMS' inattention regarding laboratories performing genetic tests, neither health care providers nor consumers can be confident in the oversight mechanisms in place to

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149. JULI MURPHY, GAIL JAVITT & KATHY HUDSON, CREATING A GENETIC TESTING SPECIALTY UNDER CLIA: WHAT ARE WE WAITING FOR? (Genetics & Pub. Policy Ctr. 2006), <http://www.dnapolicy.org/resources/McClellanpaper.pdf>.

150. *Id.* at 3.

151. *Id.*

152. For example, test accuracy and clinical validity criteria would be similar to the criteria described under the ACCE framework.

153. Notice of Intent, 65 Fed. Reg. 25928 (May 4, 2000).

154. *Id.*

ensure that laboratories performing genetic tests provide accurate and reliable test results.<sup>155</sup>

Despite the community support, however, CMS stated it no longer intends to create such a specialty. In its response to the citizen petition, issued August 15, 2007, CMS cited both technical challenges and cost-effectiveness as justifications for not developing standards for genetics proficiency testing.<sup>156</sup>

### C. Enforcing Validity and Utility Standards of Genetic Testing Through the FDA

In addition to creating a CMS specialty area for genetic testing, effective federal regulation must involve the FDA. FDA oversight should extend to LDTs as medical devices, including both pre- and post-market evidence made available to the public, but should set separate standards for tests with medical purposes and those with nonmedical purposes.<sup>157</sup> The FDA could classify a submission as deficient if it relies on unverified science or does not adequately summarize the peer-reviewed literature. Although very few genetic tests on the market or currently under development have been regulated as medical devices,<sup>158</sup> the FDA's current—and still evolving—policy shows that previously unregulated diagnostics could require FDA approval before marketing.<sup>159</sup> This requirement would address the uncertain scientific evidence behind most genetic tests, dealing with both the clinical validity and clinical utility considerations under the ACCE framework.

FDA standards should consider the complexity and the intended use of the results of any given test. In particular, tests that provide medical information should be held to a higher standard than those that provide nonmedical information. Standards should be determined with the input of both industry experts and professional organizations, and should aim to minimize both the time and cost of review. To avoid overly cautious regulatory burdens, higher review standards should only apply to those tests which may convey a medical outcome, but not for those for which the ap-

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155. Petition for Rulemaking from Kathy Hudson, *supra* note 59, at 17.

156. Reply from Dennis Smith, *supra* note 59.

157. "Nonmedical" purposes in this context refers to genetic testing that convey no direct health risks or implications, such as ancestral DNA testing, paternity testing, and genotyping for certain personality or physical characteristics such as eye color.

158. See e.g., GeneTests Home Page, <http://genetests.org> (last visited Dec. 10, 2008).

159. See FDA, DRUG-DIAGNOSTIC CO-DEVELOPMENT CONCEPT PAPER (2005), available at <http://www.fda.gov/Cder/genomics/pharmacoconceptfn.pdf>.

plication is education, nonmedical, or entertainment.<sup>160</sup> For example, DTC genetic tests which describe predisposition for hypertension should meet a higher standard of validity and utility, while a genetic test that conveys information such as ancestry may be required to meet a more relaxed standard.

The adoption of this tiered approach to genetic testing standards would assist the FTC in pursuing claims of false or misleading advertising in genetic testing. The FTC's limited personnel poses a practical concern, however: the FTC does not have the in-house expertise able to properly understand the nuances of technical claims. This challenges its ability to prioritize possible transgressors and pursue them.<sup>161</sup> By developing targeted scientific standards and clear-cut regulation requirements through the FDA and CMS/CLIA, the FTC will more easily be able to identify DTC companies that are making claims unsupported by scientific understanding.

#### **D. Proposed Legislation for Further Regulation of DTC Genetic Testing**

In instances where regulatory agencies are unable or unwilling to act, legislation may be necessary. Two bills currently proposed in Congress could significantly mitigate the concerns regarding genetic testing. The Genomics and Personalized Medicine Act of 2007,<sup>162</sup> introduced by then-Senator Obama, and the Laboratory Test Improvement Act of 2007,<sup>163</sup> introduced by Senator Kennedy, both call upon CMS to create a CLIA specialty category for genetic testing. This would mark an important change in current regulations, as current CLIA requirements only provide oversight to the laboratories themselves but do not require specific proficiency requirements for genetic tests. Creating a specialty category for genetic tests within CLIA would ensure that genetic testing is carried out in a reli-

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160. Karl V. Voelkerding, *Finding a Workable Balance: Regulation of Genetic Testing in the Human Genome Era*, 2 J. MOLECULAR DIAGNOSTICS 57 (2000)

161. Nat'l Human Genome Inst., Workshop Summary: Direct to Consumer (DTC) Advertising of Genetic Tests, Welcome and Introductions (Mar. 23, 2004), <http://www.genome.gov/12010660>.

162. S. 976, 110th Cong. (2007) (proposing the creation of an interagency working group, the development of a national biobank for the collection of genomic data, and an increase in genetics and genomics training).

163. S. 736, 110th Cong. (2007) (deeming laboratory tests to be devices for the purposes of regulation, requiring public disclosure concerning the tests, and requiring reporting of direct-to-consumer genetic tests under 21 U.S.C. § 360k).

able and consistent manner, thereby enhancing the significance of the test results.

Outside of alterations to CLIA, the two bills take markedly different approaches towards genetic testing oversight. The first and more comprehensive one is the Kennedy Laboratory Improvement Act, which would require laboratories performing LDTs to be registered with the FDA and to submit data demonstrating the analytical and clinical validity of the tests.<sup>164</sup> Some tests under the bill would only be obtainable through a licensed physician, posing a strict regulation on many DTC firms.<sup>165</sup> In addition, the impact of the bill would be much broader than current regulations, affecting *any* test performed by a laboratory, including LDTs.<sup>166</sup> To date, the Senate has not taken action on this measure.

Taking a more reserved approach, the Genomics and Personalized Medicine Act would create a formalized working group to first solicit outside expert advice before finalizing any specific regulation.<sup>167</sup> The bill authorizes a new interagency working group and the Institute of Medicine to study issues including analytic validity, clinical validity, and clinical utility. They would then make recommendations to Congress on these key issues.<sup>168</sup> Once the report is submitted, the Secretary is to develop and propose a decision matrix to help labs and other test makers know which types of tests require which level of review and who is responsible for the review—CMS, the FDA, or both.<sup>169</sup> The bill also requests a study by the National Academies of Sciences on incentives to stimulate advances in designing and developing new genetic testing technologies.<sup>170</sup> This measure could take great strides in developing a framework to assess the scientific and clinical value of genetic tests, an area not fully covered by CMS or FDA regulation.

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164. *Id.*

165. *Id.*

166. *Senator Kennedy introduces the Laboratory Test Improvement Act*, GENETICS & PUB. POL'Y CENTER NEWSL., Mar. 2007, available at [http://www.dnapolicy.org/news.eneews.article.nocategory.php?action=detail&newsletter\\_id=20&article\\_id=78](http://www.dnapolicy.org/news.eneews.article.nocategory.php?action=detail&newsletter_id=20&article_id=78).

167. S. 976, 110th Cong. (2007).

168. *Id.*

169. *Id.*

170. *Id.*

### E. Instituting Industry Self Regulation to Diminish Consumer Risk

Consumer risk can also be significantly decreased through appropriate industry self regulation. Such regulation would first need an agreed-upon standard for communicating the quality and risks of genetic tests. As discussed above in Section II.II.B.1, current DTC testing services employ varying methods to communicate the scientific confidence behind genetic tests, ranging from a community-based review to a “black box” standard. DTC companies should inform consumers in an understandable fashion as to the sensitivity, specificity, and predictive value of a genetic test, including the scientific evidence on which any claims of benefit are based.

Coordinated efforts at self-regulation in this area may convince legislators that overt regulation is unnecessary.<sup>171</sup> But more formal regulation would eventually be required to resolve regulatory confusion of LDTs and test kits—particularly in the area of DTC genetic testing.<sup>172</sup> Industry self-regulation only provides preliminary protection of consumer privacy of personal information; it does not foster standards that would ensure the accuracy of marketing claims and the efficacy of tests. Proper policing is best addressed through government agency regulation.

The argument for self regulation is, in part, one of industry self protection as well. As discussed above in Section II.II.B.2, DTC companies have not acknowledged a duty to warn customers. For two reasons, the duty to warn may well fall on the shoulders of the DTC genetic testing companies. First, the learned-intermediary rule contains an exception in the case of over-the-counter drugs. Under this exception, the duty to warn the consumer of any hazards or recommended precautions associated with over-the-counter drugs rests with the manufacturer.<sup>173</sup> Similarly, as DTC genetic tests are made available directly to the consumers without a trained intermediary, the duty to warn customers of potential hazards may rest with the DTC genetic testing service.

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171. Bryn William-Jones, *Where There's a Web, There's a Way: Commercial Genetic Testing and the Internet*, 6 COMMUNITY GENETICS 46, 53 (2003).

172. *Id.*

173. *E.g.*, *Plummer v. Lederle Laboratories*, 819 F.2d 349, 355-56 (2d Cir. 1987) (applying California law); *In re Rezulin Prod. Liab. Litig.*, 133 F. Supp. 2d 272, 282 (S.D.N.Y. 2001) (applying Mississippi law); *Stone v. Smith, Kline & French Lab.*, 447 So.2d 1301, 1305 (Ala. 1984); *Torsiello v. Whitehall Lab.*, 398 A.2d 132, 138 (N.J. Super. App. Div. 1979).

Second, internet-based DTC genetic testing undermines the health professional's role as gatekeeper and mediator of complex health technologies. Because there is little or no opportunity for the type of personalized support typically provided by physicians or genetic counselors, consumers may be left psychologically unprepared to deal with potentially traumatizing genetic test results.

While the physical risks associated with genetic testing are small, there are other risks, such as privacy, confidentiality, and security threats, and these could lead to emotional, social, and financial consequences. For example, while recently passed legislation prohibits the use of genetic information for discriminatory purposes by health insurers and employers,<sup>174</sup> no regulations currently prevent the use of genetic information in life insurance policies or for the purposes of law enforcement. Further, while DTC companies who compile genetic databases claim to "de-identify" genetic information before disclosing that information to a third party for research purposes,<sup>175</sup> whether that is in fact possible is unclear.<sup>176</sup> Finally, the physical security of the genetic information is also at risk of theft, just like social security numbers or credit card information.<sup>177</sup>

For its part, one leader in the DTC genetic testing industry has taken a proactive stance in developing industry-wide standards.<sup>178</sup> On April 8, 2008, Navigenics announced that it will develop a set of industry standards for consumer genomic testing services, and that it will consult with the many stakeholders involved.<sup>179</sup> Among the ten standards for performance criteria were validity, accuracy and quality, clinical relevance, actionability, and access to genetic counseling.<sup>180</sup> These criteria address

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174. Genetic Information Nondiscrimination Act of 2008, Pub. L. No. 110-233, 122 Stat. 881 (to be codified in scattered sections of 29 and 42 U.S.C.).

175. See, e.g., 23andMe Privacy Statement, *supra* note 38.

176. See Amy L. McGuire & Richard A. Gibbs, *Genetics: No-Longer De-Identified*, 312 SCIENCE 370-371 (2006) (discussing challenges in de-identifying genetic material); see also *DNA Databases Shut After Identities Compromised*, NATURE NEWS, Sept. 3, 2008, [http://www.nature.com/news/2008/080903/full/455013a.html?s=news\\_rss](http://www.nature.com/news/2008/080903/full/455013a.html?s=news_rss).

177. Victoria Lambert, *DNA Database: Will Your DNA Get Lost, Too?*, DAILY TELEGRAPH, Nov. 28, 2008, at 27; see also David Migoya, *Stolen State Database Puts 1.4 Million at ID-Theft Risk*, DENVER POST, Nov. 3, 2006, at 1.

178. *Navigenics Proposes Standards for Personal Genomics Services, Coupled With Prospective Outcomes Studies, to Safeguard Consumers Company Acts to 'Ensure the Integrity of This Critical Step Toward Personalized Health Care,'* REUTERS, Apr. 8, 2008, <http://www.reuters.com/article/pressRelease/idUS100153+08-Apr-2008+PRN20080408>.

179. *Id.*

180. *Id.*

many concerns of DTC testing and could serve as the foundation for effectively reducing consumer risk.

#### **IV. CONCLUSION**

The science of genetic testing is advancing at a rapid pace, but the lack of regulation in this field could put individuals at risk. Marketing genetic tests directly to consumers allows companies to bypass what little oversight exists, avoid accountability for their advertising claims, and provide consumers with genetic information without counseling from doctors. The first step in reducing this risk is to develop scientific standards that communicate the quality and value of particular genetic tests. Tighter government oversight requires clear regulatory authority by CLIA or the FDA. In addition, industry self-regulation could address these concerns and protect the DTC companies from possible liability. Responsible regulations will help ensure that the DTC industry does not move at a faster pace than the science can support and will promote the growth of this exciting field.