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Molecular genetic research has expanded rapidly since the first description of DNA's double helix structure was published in 1953.[1] The modern "Genomic Era" began in 2003 when the Human Genome Project announced the completed sequencing of the human genome.[2] Since the completion of the Human Genome Project, advances in genetic testing have increased dramatically.[3] Although there are currently genetic tests available for over 2,000 diseases,[4] less than ten percent of the tests available for clinical use have been reviewed for clinical validity and utility.[5] Furthermore, the United States Food and Drug Administration (FDA) has approved less than a dozen of the commercially available genetic tests.[6]

The substantial proliferation of genetic testing has led to many debates about the safety and appropriate use of this technology.[7] Discussion regarding the necessary and appropriate amount of regulation for genetic testing has continued for over fifteen years.[8] Despite this prolonged

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1. Alan E. Guttmacher & Francis S. Collins, *Welcome to the Genomic Era*, 349 NEW ENG. J. MED. 996, 996 (2003); *see generally* James D. Watson & Francis H.C. Crick, *A Structure for Deoxyribose Nucleic Acid*, 171 NATURE 737 (1953).) (theorizing on DNA's double helix structure).

2. Guttmacher & Collins, *supra* note 1, at 996.

3. *See generally GeneTests: Growth of Laboratory Directory*, NAT'L CTR. FOR BIOTECHNOLOGY INFO., http://www.ncbi.nlm.nih.gov/projects/GeneTests/static/whatsnew/labdirgrowth.shtml. (last visited Sept. 29, 2011) (charting the increasing number of diseases for which genetic testing is available).

4. *Id.*

5. Amy L. McGuire et al., *Regulating Direct-to-Consumer Personal Genome Testing*, 330 SCIENCE 181, 181 (2010).

6. Joan L. McGregor, *Why John Stuart Mill Would Support Restriction on DTC Marketing of Genetic Tests*, 8 AM. J. BIOETHICS 9, 9 (2008).

7. Stuart Hogarth et al., *Closing the Gaps—Enhancing the Regulation of Genetic Tests Using Responsive Regulation*, 62 FOOD & DRUG L.J. 831, 831 (2007).

8. Stuart Hogarth, *Myths, Misconceptions and Myopia: Searching for Clarity in the*

discussion, "no new issues are [currently] on the table."[9] The debate about genetic test regulation continues to focus on concerns regarding the predictive quality and proper interpretation of genetic test results, the potential misuse of genetic information, the appropriate uses of direct-to-consumer (DTC) genetic test marketing, and the ability to distinguish clinically-valid tests from flawed pseudoscience.[10]

Many of the concerns about genetic testing may be traced to the special nature of genetic information and the fact that genetic testing differs from other diagnostic tests and medical treatments.[11] The debate about the necessary and appropriate regulation of genetic testing may be based on the perception that genetic information is more central to our individuality than other biological and medical information.[12] Genetic information is unique to the individual, but is also inherently familial because the test results can reveal information about that individual's genetic relatives.[13] Genetic testing differs from other medical tests because the tests may be used to determine the probability of disease development and to identify risks in individuals without symptoms.[14] Such predictive genetic tests may provide information about diseases, conditions, and disorders for which there are no available treatments or preventive measures.[15] Furthermore, even with a positive genetic test result, the probability that an individual will develop the disease, condition, or disorder is uncertain.[16] A negative test result merely indicates the absence of a particular gene sequence that is associated with a particular condition and does not completely rule out the possibility

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Debate about the Regulation of Consumer Genetics, 13 PUB. HEALTH GENOMICS 322, 324 (2010).

9. *Id.*

10. *See* Guttmacher & Collins, *supra* note 1, at 997–98 (outlining the concerns already evident at the beginning of the genomic era).

11. Gail H. Javitt, Erica Stanley & Kathy Hudson, *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, 260 (2004).

12. Michael J. Green & Jeffrey R. Botkin, *"Genetic Exceptionalism" in Medicine: Clarifying the Differences between Genetic and Nongenetic Tests*, 138 ANNALS INTERNAL MED. 571, 572 (2003).

13. Patricia Roche, *Caveat Venditor: Protecting Privacy and Ownership Interests in DNA*, HUMAN DNA: LAW AND POLICY INTERNATIONAL AND COMPARATIVE PERSPECTIVES 33, 34 (Bartha M. Knoppers ed., 1997).

14. Green & Botkin, *supra* note 12, at 571.

15. SEC'Y'S ADVISORY COMM. ON GENETICS, HEALTH, AND SOC'Y, U.S. DEP'T OF HEALTH & HUMAN SERVS., U.S. SYSTEM OF OVERSIGHT OF GENETIC TESTING: A RESPONSE TO THE CHARGE OF THE SECRETARY OF HEALTH AND HUMAN SERVICES 18 (April 2008), *available at* http://oba.od.nih.gov/oba /SACGHS/reports/SACGHS_oversight_report.pdf.

16. PROMOTING SAFE AND EFFECTIVE GENETIC TESTING IN THE UNITED STATES: FINAL REPORT OF THE TASK FORCE ON GENETIC TESTING 3 (Neil A. Holtzman & Michael S. Watson eds., 1998) [hereinafter FINAL REPORT], *available at* http://www.genome.gov/10001733.

of future disease development.[17] Similarly, a positive test result merely indicates the presence of a particular gene sequence; a positive result does not mean that the individual will definitely develop that particular disease.[18] Even if the presence of a particular genetic sequence is directly correlated to the future development of a particular condition, the positive test result does not indicate when the condition will develop or the severity of the condition.[19] Complex interactions between an individual's genes and environmental factors influence and determine if and how a condition develops.[20]

Recent developments in health care reform and new proposals by genetic testing companies have once again sparked the debate about the regulation of genetic testing. Two distinct approaches have been proposed by two different agencies within the Department of Health and Human Services (DHHS) to address some of the concerns about the accuracy, validity, and ability of this technology. Part I of this article will discuss the current federal regulatory framework for genetic testing and genetic information.[21] Part II will explore the recent developments that have prompted the agencies to propose changes to the current regulatory framework.[22] Part III will address some of the concerns and challenges facing the implementation of these proposed changes.[23] Finally, Part IV will evaluate the two proposals and the competing interests in order to suggest how genetic testing may best be regulated to meet the needs of the industry, clinicians, researchers, patients, and consumers.[24]

I. THE CURRENT FEDERAL REGULATORY FRAMEWORK

Genetic testing is currently covered by three different regulatory schemes. The FDA, under the Federal Food, Drug, and Cosmetic Act (FDCA), may regulate genetic tests as medical devices.[25] The Clinical Laboratory Improvement Amendments of 1988 (CLIA) give the Centers for Medicare and Medicaid Services (CMS) the authority to ensure the validity and reliability of clinical laboratory testing.[26] The Genetic Information Nondiscrimination Act of 2008 (GINA) protects the use and dissemination

17. *Id.*
18. *Id.*
19. Javitt, Stanley & Hudson, *supra* note 11, at 260–61.
20. Guttmacher & Collins, *supra* note 1, at 997.
21. *See infra* Part I.
22. *See infra* Part II.
23. *See infra* Part III.
24. *See infra* Part IV.
25. 21 U.S.C. §§ 301–399 (2006).
26. 42 U.S.C. § 263a (2006).

of genetic information.[27]

A. *Medical Device Regulation*

The FDCA[28] and the Medical Device Amendments of 1976[29] give authority to the FDA to regulate medical devices.[30] A medical device is defined as "an instrument, apparatus, implement, machine, contrivance, implant, in vitro reagent, or other similar or related article, including any component, part, or accessory, which is . . . intended for use in the diagnosis of disease or other conditions, or in the cure, mitigation, treatment, or prevention of disease[.]"[31]

The FDA uses three categories of medical devices to determine how much review is necessary to ensure the safety and effective use of each device.[32] Class I devices are subject to the least FDA regulation oversight and may be introduced directly into U.S. commerce.[33] Class II devices have an increased safety risk and are subject to greater FDA controls including post-marketing surveillance and device performance standards to ensure safety and effectiveness.[34] Class III devices are defined as those: 1) for which insufficient information exists to determine a reasonable assurance of safety and effectiveness through general or special controls; and 2) which are either for (a) supporting, sustaining, or preventing impairment of human health, or (b) present a potential unreasonable risk of illness of injury.[35] This final class is subject to the greatest FDA oversight.[36]

Although the definition of a medical device provides the FDA broad authority to regulate genetic tests, the FDA has previously chosen to limit its regulation of genetic tests.[37] This limited regulation of genetic tests may be attributed to the FDA regulatory distinction between genetic "home brew" or "laboratory developed" tests (LDTs), which are developed and analyzed completely within one laboratory, and genetic tests that contain

27. Genetic Information Nondiscrimination Act, Pub. L. No. 110-233, 122 Stat. 881 (2008) (to be codified in scattered sections of 26, 29 & 42 U.S.C.).

28. 21 U.S.C. §§ 301–399 (2006).

29. Pub. L. No. 94-295, 90 Stat. 539 (1976) (codified in scattered sections of 21 U.S.C.).

30. 21 U.S.C. § 321(h) (2006).

31. *Id.*

32. Neil A. Holtzman, *FDA and the Regulation of Genetic Tests*, 41 JURIMETRICS 53, 59 (2000).

33. 21 U.S.C. § 360c(a)(1)(A) (2006).

34. 21 U.S.C. § 360c(a)(1)(B).

35. 21 U.S.C. § 360c(a)(1)(C).

36. *Id.*

37. *See* Andrew S. Robertson, *Taking Responsibility: Regulations and Protections in Direct-to-Consumer Genetic Testing*, 24 BERKELEY TECH. L.J. 213, 223–24 (2009) (explaining that the FDA has exercised "enforcement discretion" in the regulation of certain genetic tests).

components that travel in interstate commerce.[38] The FDA has previously chosen to regulate only the individual components and reagents used in genetic tests and has declined to regulate the LDTs or the laboratories that develop and analyze them.[39] Consequently, these LDTs are classified as Class I or II medical devices that are subject to less regulation than other genetic tests.[40] Practically, this allows certain genetic tests to reach the consumer market without any FDA evaluation for safety, effectiveness, or accuracy.[41] Consumers who purchase these DTC genetic tests "are likely unaware that the FDA is not involved in [any] quality manufacturing control or efficacy testing" of the products.[42]

In addition to these self-imposed regulatory distinctions, the FDA has previously stated that its regulatory power over medical devices cannot be extended to regulate genetic tests marketed as services.[43] This allows companies that market their LDTs as services to circumvent the entire FDA pre-market approval process.[44]

The limited regulation of genetic tests may also be due to the availability of the FDA's pre-market notification process.[45] The pre-market notification process, also known as 510(k) abbreviated marketing clearance, requires that developers "demonstrate that their new test is substantially equivalent to a medical device currently on the market."[46] If the genetic test developer can produce data that demonstrates that a new test is substantially equivalent to a test already available on the market, the new test can be marketed in the same class as that test, and the developer does not need to prove the new test's safety and effectiveness in order to gain pre-market approval.[47]

In addition to the market approval regulations, the FDA has regulations

38. Rebecca Antar Novick, Note, *One Step at a Time: Ethical Barriers to Home Genetic Testing and Why the U.S. Health Care System is Not Ready*, 11 N.Y.U. J. LEGIS. & PUB. POL'Y 621, 629 (2008).

39. Bruce Patsner, *New "Home Brew" Predictive Genetic Tests Present Significant Regulatory Problems*, 9 HOUS. J. HEALTH L. & POL'Y 237, 251–52 (2009). *See also* Medical Devices; Classification/Reclassification; Restricted Devices; Analyte Specific Reagents, 62 Fed. Reg. 62,243, 62,245 (Nov. 21, 1997) (codified at 20 C.F.R. §§ 809, 864) (discussing the FDA's intention not to regulate the reagents used in genetic tests differently from other Class I medical devices).

40. Patsner, *supra* note 39, at 249.

41. *Id.* at 254.

42. *Id.* at 249–50.

43. Holtzman, *supra* note 32, at 61.

44. Patsner, *supra* note 39, at 254.

45. *See* Lauren Solberg, Note, *Over the Counter but Under the Radar: Direct-to-Consumer Genetics Tests and FDA Regulation of Medical Devices*, 11 VAND. J. ENT. & TECH. L. 711, 730 (2009) (introducing the pre-market notification process).

46. *Id.*

47. Holtzman, *supra* note 32, at 60.

regarding medical device labeling that may be applied to genetic tests.[48] Genetic tests that include FDA regulated components must comply with the promulgated regulations regarding general medical device labeling,[49] as well as with regulations for *in vitro* diagnostic products.[50] Pursuant to this authority, the FDA requires that the advertising and promotional materials for some genetic tests "state that their '[a]nalytic and performance characteristics are not established' . . . because they are 'not clinically validated.'"[51] Result reports generated from these tests are also required to include a notification that the test is not cleared or approved by the FDA.[52] However, these provisions have not been uniformly enforced. Furthermore, genetic tests that are classified as Class I or II medical devices are not considered restricted medical devices, and thus are not subject to the additional advertising regulations.[53] DTC genetic tests and LDT kits are generally not subject to these regulations, so the labels and materials included with these tests may include information that has not been substantiated.[54]

B. Clinical Laboratory Regulation

CMS has authority to regulate medical testing within clinical laboratories under CLIA.[55] CLIA establishes regulations to ensure that every laboratory determines the analytical validity of its laboratory tests before offering them for clinical purposes, such as use for disease diagnosis, prevention, or treatment.[56] Although CLIA was enacted to certify valid and reliable medical testing, it does not authorize or certify the validity of the individual

48. Solberg, *supra* note 45, at 728–29.

49. 21 C.F.R. § 801.1 (2010).

50. 21 C.F.R. § 809.10 (2010).

51. Jennifer E. Spreng, *The Food and Drug Administration and the Pharmacy Profession: Partners to Ensure the Safety and Efficacy of the Pharmacogenomic Therapy*, 13 J. HEALTH CARE L. & POL'Y 77, 91–92 (2010).

52. *Id.*

53. *See Marketing or Medicine: Are Direct-to-Consumer Device Ads Playing Doctor?: Hearing Before the S. Special Comm. on Aging*, 110th Cong. 67 (2008) (statement of Daniel Schultz, Dir., Ctr. for Devices and Radiological Health, Food & Drug Administration), *available at* http://www.fda.gov/NewsEvents/Testimony/ucm096272.htm (explaining that "few Class I and Class II devices are restricted by regulation").

54. Solberg, *supra* note 45, at 729.

55. 42 U.S.C. § 263a (2006); *see also Clinical Laboratory Improvement Amendments (CLIA) Overview*, CTRS. FOR MEDICARE & MEDICAID SERVS., http://www.cms.gov/CLIA/ (last visited Oct. 1, 2011).

56. BIN CHEN ET AL., CTRS. FOR DISEASE CONTROL & PREVENTION, 58 MORBIDITY AND MORTALITY WEEKLY REPORT: GOOD LABORATORY PRACTICES FOR MOLECULAR GENETIC TESTING FOR HERITABLE DISEASES AND CONDITIONS 3 (2009); Holtzman, *supra* note 32, at 57.

tests.[57]

CLIA imposes basic requirements for laboratory methodology and documentation, as well as standards for personnel qualification, in order for a laboratory to receive certification.[58] CLIA certification requires periodic facility inspections and the examination of proficiency sample tests for high-complexity tests.[59] Although genetic tests are highly complex and require precise skill to perform and interpret, CMS does not require laboratories that perform genetic testing to undergo specific proficiency examinations for the genetic tests.[60] Under CLIA, "there are no specified quality control, personnel, or proficiency testing requirements mandated . . . for most genetic tests."[61] Although laboratories that perform genetic tests must meet some personnel requirements to fulfill the certification requirements for high-complexity testing, the actual laboratories are only subject to the general CLIA methodology and documentation requirements.[62] Furthermore, CLIA "does not require [that] laboratories address the *clinical* validity or utility of tests [performed]."[63] This is particularly problematic for "home brew" genetic tests because CLIA does not include explicit authorization or a process for evaluating test accuracy.[64]

Although CLIA requires that clinical laboratories report any changes regarding offered examinations, tests, and procedures to the Secretary of DHHS,[65] CLIA does not authorize CMS to limit or restrict the offering of a particular test.[66] The decision to offer new genetic tests is left to the discretion of each individual laboratory.[67]

C. Genetic Information Regulation

Prior to the passage of GINA in May 2008, the protection of genetic information in the United States was governed by an assortment of federal

57. Robertson, *supra* note 37, at 222.

58. 42 U.S.C. §§ 263a(d)(1), (e)(2)(D), (f)(1) (2006).

59. 42 U.S.C. §§ 263a(d)(3), (f)(3)(A); Gail H. Javitt & Kathy Hudson, *Federal Neglect: Regulation of Genetic Testing*, 22 ISSUES IN SCI. & TECH. 59, 59 (2006), *available at* http://www.dnapolicy.org/resources/Issues_in_Science_and_Technology.pdf.

60. Javitt & Hudson, *supra* note 59, at 59–61.

61. *At Home DNA Tests: Marketing Scam or Medical Breakthrough: Hearing Before the S. Special Comm. on Aging*, 109th Cong. 35–36 (2006) (statement of Kathy Hudson, Dir., Genetics & Pub. Policy Ctr., and Assoc. Professor, Berman Bioethics Inst., Inst. of Genetic Med. and Dep't of Pediatrics, Johns Hopkins Univ.).

62. 42 C.F.R. § 493.1495 (2010); *see* CHEN, *supra* note 56, at 3.

63. Holtzman, *supra* note 32, at 57.

64. Javitt & Hudson, *supra* note 59, at 61.

65. 42 U.S.C. § 263a(d)(2)(B) (2006).

66. Javitt & Hudson, *supra* note 59, at 61.

67. *Id.*

and state statutes and regulations.[68] The states' earliest efforts began in the 1970s and were designed as anti-discrimination responses to specific genetic conditions.[69] Following the establishment of the Human Genome Project, many more states began to develop regulations and restrictions governing the use of genetic information. Since each state created its own policies, the laws varied widely in their approach, classification, application, and level of protection.[70] Some states relied on privacy grounds to justify the protection of genetic information, while other states relied on property rights.[71] State laws were separated into three categories of protection: "(1) trait protection; (2) prohibition of discrimination from the results of genetic testing; and (3) forbidding discrimination in regards to genetic information."[72] The wide variety of approaches and protections led to compliance complications for employers, healthcare providers, and individuals.[73]

Prior to GINA, a number of federal statutes also governed the protection of genetic information, including: Title VII of the Civil Rights Act of 1964; the Rehabilitation Act of 1973; the Employee Retirement Income Security Act of 1974; Title XVIII of the Social Security Act; the Americans with Disabilities Act; the Family and Medical Leave Act; the Occupational Safety and Health Act; the Public Health Service Act; Executive Order 13145;[74] and the Health Insurance Portability and Accountability Act of 1996.[75] In addition, provisions in the Fourth, Fifth, and Fourteenth Amendments concerning unreasonable searches and due process were interpreted to provide another basis for the preservation of genetic information and privacy.[76] Despite the large number and wide variety of federal statutes and Constitutional provisions that potentially applied to

68. *See* Karen H. Rothenberg, *Social Implications of Genetic Testing*, 3 N. AM. ACTUARIAL J. 133, 134 (1999) (discussing the evolution of genetic testing in the 1970s and 1980s); Daniel Schlein, *New Frontiers for Genetic Privacy Law: The Genetic Information Nondiscrimination Act of 2008*, 19 GEO. MASON U. C.R. L.J. 311, 347 (2009).

69. Rothenberg, *supra* note 68, at 134; Schlein, *supra* note 68, at 347.

70. Schlein, *supra* note 68, at 347.

71. Morse Hyun-Myung Tan, *Advancing Civil Rights, The Next Generation: The Genetic Information Nondiscrimination Act of 2008 and Beyond*, 19 HEALTH MATRIX 63, 88 (2009).

72. *Id.* at 89.

73. *See id.* at 92 (explaining that individuals and genetic information may easily cross state lines, which "present[ed] challenges for both individuals seeking protection and governments enforcing [the] provisions.").

74. Patricia Nemeth & Terry W. Bonnette, *Genetic Discrimination in Employment*, 88 MICH. B.J. 42, 44 (2009) ("Since February 10, 2000, federal employees have been protected under Executive Order 13145 against discharge or other restrictions in their employment or employment benefits on the basis of genetic information.").

75. Tan, *supra* note 71, at 93–94; Schlein, *supra* note 68, at 318.

76. Schlein, *supra* note 68, at 345.

protect genetic privacy, substantial loopholes and irregularities in the regulations did not adequately protect against the possibility of genetic privacy breaches. After many years of political debate, President George W. Bush signed GINA on May 21, 2008.[77]

GINA prohibits discrimination on the basis of genetic information for health insurance and employment purposes.[78] Genetic information is defined as information about an individual's genetic tests, the genetic tests of family members of such individual, and the manifestation of a disease or disorder in family members of such individual.[79] Genetic information does not include information about the sex or age of an individual.

Title I of GINA prohibits group health plans from adjusting premium or contribution amounts on the basis of genetic information. To protect individuals from discrimination by group health insurance providers, the insurance issuer may not:

> request or require an individual or family member of such individual to undergo a genetic test [. . . , may] not request, require, or purchase genetic information for underwriting purposes [. . . , and may] not request, require, or purchase genetic information with respect to any individual prior to such individual's enrollment under the plan or coverage in connection with such enrollment.[80]

Violation of these prohibitions results in financial penalties unless the insurer can demonstrate that reasonable diligence was used to avoid noncompliance.[81]

Title II of GINA prohibits employers from using genetic information to make decisions regarding hiring, firing, job placement and promotions in addition to regulating how employers may acquire an employee's genetic information. Title II makes it "an unlawful employment practice for an employer to fail or refuse to hire, or to discharge, any employee, or otherwise to discriminate against any employee with respect to the compensation, terms, conditions, or privileges of employment of the employee, because of genetic information."[82] It is also unlawful "to limit, segregate, classify [. . .] or otherwise adversely affect the status of the employee [. . .]" because of genetic information.[83] Employers are also

77. Press Release, White House, President Bush Signs H.R. 493, the Genetic Information Nondiscrimination Act of 2008 (May 21, 2008).

78. Genetic Information Nondiscrimination Act of 2008, Pub. L. No. 110-233, 122 Stat. 881 (to be codified in scattered sections of 26, 29, and 42 U.S.C.).

79. § 101(d), 122 Stat. at 885.

80. § 101(b), 122 Stat. at 883–84.

81. § 101(e), 122 Stat. at 886–87.

82. § 202(a)(1), 122 Stat. at 907.

83. § 202(a)(2), 122 Stat. at 907.

prohibited from requesting, requiring, or purchasing the genetic information of an employee or family member of the employee, with six exceptions for inadvertent discovery, company offered health services, and workplace genetic monitoring.[84]

II. RECENT DEVELOPMENTS AND PROPOSED CHANGES

A. FDA and Congressional Actions

In May 2010, Pathway Genomics (Pathway) announced a plan to sell one of its genetic tests directly to consumers at national retail pharmacies throughout the United States.[85] In response, the FDA sent a letter to Pathway stating that the home-use genetic testing kit was a medical device as defined in the FDCA.[86] The FDA informed Pathway that the agency had not cleared or approved their test and asked the company to either provide documentation of the clearance, or approval, or the basis for their determination that the company was not required to obtain FDA clearance for the device.[87] Pathway's attempt to sell genetic tests in retail pharmacies also prompted the House Committee on Energy and Commerce (House Committee) to begin a broad investigation of DTC genomic companies.[88] The House Committee sent letters to three companies requesting information about the conditions and diseases the companies test for, guidance materials related to genetic counseling and physician consultations, and documentation about test accuracy, processing policies, and FDA compliance.[89] Although Pathway stated that their test fell within

84. § 202(b), 122 Stat. at 907–08; *see also* Schlein, *supra* note 68, at 357–59 (explaining the distinctions between the six exceptions to GINA's "presumption of unlawfulness when an employer asks, requires, or purchases genetic information of an employee or an employee's family member.").

85. Rob Stein, *Company Plans to Sell Genetic Testing Kit at Drugstores*, WASH. POST, May 11, 2010, http://www.washingtonpost.com/wp-dyn/content/article/2010/05/10/AR2010051004904.html.

86. Letter from James Woods, Deputy Dir., Patient Safety and Product Quality, Office of *In Vitro* Diagnostic Device Evaluations and Safety, Ctr. for Devices and Radiological Health, to James Plante, Founder & CEO, Pathway Genomics Corp. (May 10, 2010), *available at* http://www.fda.gov/downloads/MedicalDevices/ResourcesforYou/Industry/UCM211875.pdf.

87. *Id.*

88. Turna Ray, *As Congress Investigates DTC Genomics Market, Firms Vow Cooperation*, PHARMACOGENOMICS REP. (May 26, 2010), http://www.genomeweb.com/dxpgx/congress-investigates-dtc-genomics-market-firms-vow-cooperation.

89. Letter from Henry A. Waxman, Chairman, H. Comm. on Energy and Commerce, et al., to James Plante, Chief Exec. Officer, Pathway Genomics Corp. (May 19, 2010), *available at* http://democrats.energycommerce.house.gov/documents/20100519/PlantePathwayGenomics.2010.5.19.pdf; Letter from Henry A. Waxman, Chairman, H. Comm. on Energy and Commerce, et al., to Vance Vanier, Pres. & CEO, Navigenics (May 19, 2010),

the current regulatory guidelines, the FDA's letter and the House Committee's investigation prompted the national retail pharmacies to halt retail genetic test sales until the regulatory concerns were resolved.[90]

In June 2010, less than one month after the FDA and House Committee began their investigations, 23andMe, a DTC genetic test company, determined that a number of customer samples were incorrectly processed.[91] This mix-up resulted in a number of customers receiving and viewing genetic data and test results that were not their own.[92] Following this mix-up, the House Committee asked 23andMe for additional information about the company's policies and communications related to the collection, processing, and analysis of customer samples.[93] The FDA sent 23andMe and four other genetic testing companies letters, similar to the letter Pathway received, stating that the companies' tests were medical devices subject to regulation under the FDCA.[94] However, in these letters, the FDA specified that the companies' services were not considered LDTs

available at http://democrats.energycommerce.house.gov/documents/20100519/Vanier.Navigencis.2010.5.19.pdf; Letter from Henry A.Waxman, Chairman, H. Comm. on Energy and Commerce, et al., to Anne Wojcicki, Pres., 23andMe, Inc. (May 19, 2010), *available at* http://democrats.energycommerce.house.gov/documents/20100519/Wojcicki.23andME.2010.5.19.pdf.

90. Ray, *supra* note 88.

91. 23andMe, *Update from 23andMe*, THE SPITTOON (June 8, 2010, 2:56 PM), http://spittoon23andme.com/2010/06/08/update-from-23andme/.

92. Turna Ray, *UPDATE: 23andMe Says LabCorp Incorrectly Processed 96 Samples, Mixing Up Customer Data*, PHARMACOGENOMICS REP. (June 9, 2010), http://www.genomeweb.com/dxpgx/update-23andme-says-labcorp-incorrectly-processed-96-samples-mixing-customer-dat.

93. *Congress Seeks More Information on 23andMe Data Mix-up*, GENOMEWEB DAILY NEWS (June 14, 2010), http://www.genomeweb.com/dxpgx/congress-seeks-moreinformation-23andme-data-mix.

94. Letter from Alberto Gutierrez, Office of *In Vitro* Diagnostic Device Evaluation and Safety, Ctr. for Device and Radiological Health, to Earl M. Collier, Jr., CEO, deCODE Genetics, (June 10, 2010), *available at* http://www.fda.gov/downloads/MedicalDevices/ResourcesforYou/Industry/UCM215241.pdf; Letter from Alberto Gutierrez, Office of *In Vitro* Diagnostic Device Evaluation and Safety, Ctr. for Device and Radiological Health, to Jorge Conde, Co-Founder & CEO, Knome, Inc. (June 10, 2010), *available at* http://www.fda.gov/downloads/MedicalDevices/ResourcesforYou/Industry/UCM215239.pdf ; Letter from Alberto Gutierrez, Office of *In Vitro* Diagnostic Device Evaluation and Safety, Ctr. for Device and Radiological Health, to Jay T. Flatley, President & CEO, Illumina, Inc. (June 10, 2010), *available at* http://www.fda.gov/downloads/MedicalDevices/ResourcesforYou/Industry/UCM215242.pdf; Letter from Alberto Gutierrez, Office of *In Vitro* Diagnostic Device Evaluation and Safety, Ctr. for Device and Radiological Health, to Vance Vanier, President & CEO, Navigenics (June 10, 2010), *available at* http://www.fda.gov/downloads/MedicalDevices/ResourcesforYou/Industry/UCM215243.pdf; Letter from Alberto Gutierrez, Office of *In Vitro* Diagnostic Device Evaluation and Safety, Ctr. for Device and Radiological Health, to Anne Wojcicki, President & Co-Founder, 23andMe, Inc. (June 10, 2010), *available at* http://www.fda.gov/downloads/MedicalDevices/ResourcesforYou/Industry/UCM215240.pdf.

because the tests were not developed by and used in a single laboratory.[95] In July 2010, the FDA sent letters to fourteen additional companies that manufacture and market genetic tests.[96] As in the previous letters, the FDA explained that the companies' genetic tests were improperly being marketed without FDA clearance or approval.[97]

During the summer of 2010, the FDA also announced a public meeting and requested comments regarding the oversight of LDTs.[98] The FDA reached out to "interested stakeholders regarding reasonable and effective regulation of LDTs."[99] In the Federal Register notice, the FDA acknowledged that the agency had previously "exercised enforcement discretion and [had] not enforced applicable regulations with respect to LDTs[.]"[100] The agency explained that this limited regulation was acceptable when the initial LDTs, simple pathology tests, were developed and used by clinicians working in the laboratories that developed and interpreted the patients' results.[101] However, the FDA noted that today's LDTs are increasingly complex, use components that are not individually regulated by the FDA, and assess high-risk conditions and diseases, often in a remote laboratory that is removed from the test development laboratory and the patient care offices.[102] To address growing concerns about the corporate development of LDTs and the increased use of these tests in clinical disease management, the FDA suggested that a "risk-based application of oversight to LDTs is the appropriate approach" to regulation.[103] The FDA held a public meeting over two days in July 2010

95. Letter from Gutierrez to Conde, *supra* note 94; Letter from Gutierrez to Wojcicki, *supra* note 94; Turna Ray, *FDA Warns DTC Genomics Firms that Genetic Tests are Not LDTs, May Need Clearance or Approval*, PHARMACOGENOMICS REP. (June 15, 2010), http://www.genomeweb.com/dxpgx/fda-warns-dtc-genomics-firms-genetic-tests-are-not-ldts-may-need-clearance-or-ap (reporting on the letters sent to five genetic test companies).

96. *See, e.g.*, Letter from James Woods, Deputy Dir., Patient Safety and Product Quality, Office of *In Vitro* Diagnostic Device Evaluations and Safety, Ctr. for Devices and Radiological Health, to Harry F. Hixson, Jr., Chairman & CEO, Sequenom, Inc. (July 19, 2010), *available at* http://www.fda.gov/downloads/MedicalDevices/ProductsandMedicalProcedures/InVitroDiagnostics/UCM219595.pdf. Links to the other thirteen letters are available at http://www.fda.gov/MedicalDevices/ProductsandMedicalProcedures/InVitroDiagnostics/ucm219582.htm (last visited Oct. 23, 2011). Three additional companies that market genetic tests received letters from the FDA in May 2011. *FDA Warns More DTC Genetics Firms*, GENOMEWEB DAILY NEWS (May 17, 2011), http://www.genomeweb.com/dxpgx/fda-warns-more-dtc-genetics-firms.

97. Letter from Woods to Hixson, Jr., *supra* note 96.

98. Oversight of Laboratory Developed Tests; Public Meeting; Request for Comments, 75 Fed. Reg. 34,463 (June 17, 2010).

99. *Id.*

100. *Id.*

101. *Id.*

102. *Id.*

103. *Id.* at 34,464.

and addressed four topics: 1) patient considerations, 2) challenges facing laboratories, 3) DTC marketing of genetic testing, and 4) education and outreach.[104] At the beginning of the meeting, Dr. Jeffrey Shuren, the FDA's Director of the Center for Devices and Radiological Health, stressed that although the agency had formally decided to regulate LDTs, it had not yet decided how the agency would exercise that authority.[105]

In the same week as the FDA's public meeting, the House Committee also held a hearing about the marketing and regulation of genetic tests and the effects of these tests on public health.[106] At the House Committee hearing, the Government Accountability Office (GAO) disclosed an undercover report stating that the services of four DTC genetic test companies were deceptive, fraudulent, and nearly useless.[107] The GAO purchased tests from four different companies in order to investigate the companies' marketing practices, testing data, and customer support.[108] This investigation was conducted to reassess the claims made by DTC genetic testing companies following a prior investigation into claims about personalized nutrition and lifestyle recommendations made by four DTC genetic testing companies in 2006.[109] Following the first undercover investigation, Gregory Kutz, the Managing Director of Forensic Audits and Special Investigations for the GAO, testified before the Senate Special Committee on Aging that the sampled genetic tests at best provided little or no value to consumers, and at worst "could frighten a consumer into

104. *Id.*; *see also* U.S. Dep't of Health & Human Servs., Food & Drug Admin., *Public Meeting on Oversight of Laboratory Developed Tests Before the Food & Drug Admin.*, (July 19, 2010) [hereinafter *Public Meeting I*], *available at* http://www.regulations.gov/Search/Regs/home.html#documentDetail?R=0900006480b65834; U.S. Dep't of Health & Human Servs., Food & Drug Admin., *Public Meeting on Oversight of Laboratory Developed Tests* (July 20, 2010), *available at* http://www.regulations.gov/search/Regs/home.html#documentDetail?R=0900006480b6591.

105. *Public Meeting I*, *supra* note 104, at 6–7.

106. Memorandum from the Comm. on Energy & Commerce Staff to the Subcomm. on Oversight & Investigations Members & Staff (July 20, 2010), *available at* http://democrats.energycommerce.house.gov/documents/20100720/Briefing.Memo.oi.2010.7.20.pdf.

107. Matt Jones, *DTC Genetics Services Hit Hard on the Hill as FDA Mulls Options*, GENOMEWEB DAILY NEWS (July 23, 2010), http://www.genomeweb.com/dxpgx/dtc-genetics-services-hit-hard-hill-fda-mulls-options.

108. *GAO's Stealth Report Attacks "Deceptive" DTC Genetics*, GENOMEWEB DAILY NEWS (July 22, 2010), http://www.genomeweb.com/dxpgx/gaos-stealth-report-attacks-deceptive-dtc-genetics.

109. U.S. GOV'T ACCOUNTABILITY OFFICE, GAO-10-847T, DIRECT-TO-CONSUMER GENETIC TESTS: MISLEADING TEST RESULTS ARE FURTHER COMPLICATED BY DECEPTIVE MARKETING AND OTHER QUESTIONABLE PRACTICES 1–2 (2010) [hereinafter MISLEADING TEST RESULTS]; *see also* U.S. GOV'T ACCOUNTABILITY OFFICE, GAO-06-977T, NUTRIGENETIC TESTING: TESTS PURCHASED FROM FOUR WEB SITES MISLEAD CONSUMERS 2 (2006).

thinking that they will develop cancer, osteoporosis, heart disease, or brain aging."[110] After investigating the claims of four new DTC genetic testing companies, in July 2010 Mr. Kutz testified before the House Committee that the new test results the GAO received were just as "misleading and of little or no practical use to consumers."[111] After informing the FDA, National Institutes of Health (NIH), and Federal Trade Commission (FTC) of the findings, the GAO referred all of the companies investigated to the FDA and FTC for appropriate action related to their claims.[112]

Even though discussions about the need for improved regulation of LDTs and DTC genetic tests began in the spring of 2010, as of September 2011 the FDA was still in the drafting phase and had issued limited guidance about how the regulations will develop.[113] This may be related to the large number of public submissions that the FDA received in response to the notice for public comment and at the public meetings.[114] The House Committee has also not taken any significant action since the hearings in July 2010. One of the greatest steps towards improved regulation occurred in June 2010, when CMS and the FDA signed a Memorandum of Understanding (MOU) that acknowledged the need for greater collaboration between the two agencies.[115] The agencies "agree[d] to work together to promote initiatives related to the review and use of FDA-regulated . . . medical devices[.]"[116] Throughout the discussions of increased FDA regulation, the FDA has mentioned using a test registry to monitor available genetic tests.

110. *At Home DNA Tests: Marketing Scam or Medical Breakthrough: Hearing Before the S. Special Comm. on Aging*, 109th Cong. 3–4 (2006) (statement of Gregory Kutz, Managing Director of Forensic Audits and Special Investigations, United States Government Accountability Office).

111. MISLEADING TEST RESULTS, *supra* note 109, at 4.

112. *Id.* at 19.

113. Molika Ashford, *At Dx Conference, FDA's Gutierrez Sheds Further Light on Plans for Risk-Based LDT Regulation*, PHARMACOGENOMICS REP. (Sept. 7, 2011), http://www.genomeweb.com/mdx/dx-conference-fdas-gutierrez-sheds-further-light-plans-risk-based-ldt-regulation. *See also FDA in Drafting Phase for LDT Oversight*, GENOMEWEB DAILY NEWS (Oct. 5, 2010), http://www.genomeweb.com/Dxpgx/fda-drafting-phase-ldt-oversight.

114. Over one hundred public presentations and comments regarding the regulation of LDTs are available at Regulations.gov under Docket ID. FDA-2010-N-0274.

115. CTRS. FOR MEDICARE & MEDICAID SERVS. & U.S. FOOD & DRUG ADMIN., MOU 225-10—0010, MEMORANDUM OF UNDERSTANDING BETWEEN UNITED STATES FOOD AND DRUG ADMINISTRATION AND CENTERS FOR MEDICARE & MEDICAID SERVICES (2010), *available at* http://www.fda.gov/aboutfda/partnershipscollaborations/Memorandaofunderstandingmous/domesticmous/ucm217585.htm.

116. *Id.*

B. NIH Genetic Test Registry

In March 2010, the NIH announced the creation of the Genetic Testing Registry (GTR), a public database that will provide increased access to information about the availability, validity, and usefulness of genetic tests to researchers, consumers, health care providers, and the public.[117] The GTR is intended to be a comprehensive resource that provides detailed information about genetic tests currently available to patients.[118] Genetic test companies will voluntarily provide the information included in the GTR.[119]

The NIH published a detailed notice and request for comments about the development of the GTR in June 2010.[120] In addition to stressing that the GTR will provide a centralized public resource with information about the intended use, validity, and utility of genetic tests, the Federal Register notice also indicated that the GTR will be used to facilitate the exchange of Health Information Technology.[121] Although the NIH clearly expressed that certain information is expected to be incorporated in the GTR, including information about the molecular basis and methods used for testing, the NIH specifically requested comments regarding the types of genetic tests and data elements that should be included in the GTR.[122] The NIH also asked to receive comments about the potential uses, benefits, and risks of the GTR.[123] In November 2010, the NIH held a public meeting to discuss the development of the GTR.[124] Although the GTR was expected to be available for public use in 2011,[125] the NIH has not yet issued any specific statements regarding further development of the GTR.[126]

II. CHALLENGES FACING THE PROPOSED CHANGES

Although there are already three different pieces to the genetic testing regulatory puzzle, two separate agencies in the DHHS have introduced

117. Press Release, Nat'l Insts. of Health, NIH Announces Genetic Testing Registry (March 18, 2010), *available at* http://www.nih.gov/news/health/mar2010/od-18.htm.

118. *Id.*

119. *Id.*

120. Request for Information (RFI) on the National Institutes of Health Plan to Develop the Genetic Testing Registry, 75 Fed. Reg. 33,317 (June 11, 2010).

121. *Id.*

122. *Id.* at 33,318.

123. *Id.* at 33,318–19.

124. Plan to Develop a Genetic Testing Registry at the National Institutes of Health; Public Meeting; Request for Comments, 75 Fed. Reg. 62,406 (Oct. 8, 2010).

125. *Id.* at 62,407.

126. The GTR is now expected to launch in early 2012. *Genetic Testing Registry*, NAT'L CTR. FOR BIOTECHNOLOGY INFO., http://www.ncbi.nlm.nih.gov/gtr/ (last visited Oct. 23, 2011).

proposals to address lingering concerns about genetic testing.[127] These proposals are intended to fill in some of the holes that exist in the current genetic testing regulatory framework. However, the FDA's decision to revamp the regulation of LDTs, and the NIH's decision to create a GTR, raise concerns about the implementation of these changes and the use of agency and industry resources.

A. Revamped FDA Regulations

The public submissions responding to the FDA's notice for comments and meeting highlighted a number of concerns regarding the development and implementation of revamped LDT regulations. Many of the comments stressed the need for clarity and consistency in the FDA regulation of LDTs in order to ensure that consumers and researchers have access to clinically valid and useful tests.[128] Some of the laboratories and researchers raised concerns that LDTs are currently used to respond to and diagnose emerging health issues, and that overly burdensome regulations would limit the availability of these tests in public health situations.[129]

These comments stressed the need to ensure that any new regulatory approach is flexible enough to accommodate rapid access to LDTs for rare diseases or in public health emergencies.

The comments also expressed distinct views concerning the level of regulation to which LDTs are currently subject. Some comments focused on the fact that LDTs are already subject to regulation under CLIA and that the reagents used in LDTs are already subject to FDA regulation.[130] Other comments focused on the increasing complexity of LDTs and expressed concern that certain tests enter the market without an adequate assessment of analytical and clinical performance.[131] Underlying both sides of this debate are concerns that new regulations will force manufacturers of LDTs

127. *See supra* Part II.A–B.

128. Turna Ray, *DTC Genomics Gets Time Before FDA as Agency Considers Regulatory Strategy*, PHARMACOGENOMICS REP. (July 21, 2010), http://www.genomeweb.com/dxpgx/dtc-genomics-gets-time-fda-agency-considers-regulatory-strategy.

129. *See, e.g.*, Letter from Patrick Luedtke, President, & Scott J. Becker, Exec. Dir., Ass'n of Pub. Health Labs., to Food & Drug Admin. (Aug. 4, 2010) (explaining that LDTs are used when there is no equivalent FDA cleared test or when an outbreak occurs and the community health leaders require timely disease confirmation).

130. *See, e.g.*, American Society of Microbiology, Comments to FDA/CDRH at Public Meeting on Oversight of Laboratory Developed Tests: Clinical Lab Challenges (July 20, 2010), http://www.asm.org/index.php/policy/ldtcomments7-20-10.html (discussing the on-going validation of LDTs performance characteristics).

131. *See, e.g.*, Letter from Justine Handelman, Exec. Dir., Office of Legislative and Regulatory Policy, & Allan Korn, Senior Vice Presidet & Chief Med. Officer, Office of Clinical Affairs, BlueCross BlueShield Ass'n, to Leslie Kux, Acting Assistant Comm'r for Policy, Food & Drug Admin. (Aug. 2010) (stating that LDTs prepared without clinical performance tests should be categorized as investigational rather than clinical tests).

to meet divergent quality system requirements for the FDA and CLIA. The duplication of verification and regulation would likely increase the costs of these tests for consumers.[132] However, as Dr. Shuren testified, the FDA and CLIA regulations are supposed to complement one another; CLIA regulations focus on the quality of the testing process, while FDA regulations focus on the safety, effectiveness, quality, and manufacture of diagnostic tests.[133] Since the FDA and CMS signed a MOU regarding this issue in June 2010, it is likely that any new regulations for LDTs will adequately balance the FDA and CLIA requirements.

One underlying problem is the concern that the FDA does not have an adequate clearance pathway for genetic tests because the current clearance pathway for medical devices is based on a demonstration of effectiveness and safety.[134] The FDA has indicated that it intends to develop new LDT regulations based on the level of risk each test presents, however, it is difficult to determine how much risk a predictive genetic test presents. The pre-market approval process does not really work for this kind of technology because there is no definitive measure of "effectiveness."

Despite the debate about the necessary level of regulation the FDA should have over LDTs, it makes sense for the FDA to exercise greater regulation over these kinds of genetic tests. The initial distinctions drawn between LDTs and genetic tests that include components that travel in interstate commerce were tenuous at best. A large number of these tests are now performed in laboratories separate from the developmental laboratories and the patient care offices;[135] the tests are no longer completed entirely in one laboratory setting. In addition, in order to ensure that consumers and researchers have access to accurate and valid tests, it makes more sense for all tests to be subject to the same levels of regulation. However, there is lingering concern because not all LDTs are genetic tests. The FDA's decision to increase regulation of LDTs had already raised some questions about the retail sale of other diagnostic tests to consumers. For example, in November 2010, the FDA sent a letter to Identigene saying that the company's home-use urine collection test for chlamydia and gonorrhea had not been approved by the agency.[136] As some of the comments suggested, it

132. American Society of Microbiology, *supra* note130.

133. *Direct-to-Consumer Genetic Testing and the Consequences to the Public Health: Hearing Before the Subcomm. on Oversight and Investigations of the H. Comm. on Energy and Commerce*, 110th Cong. 5 (2010) (statement of Dr. Jeffrey Shuren, Dir., Ctr. for Devices and Radiological Health, U.S. Food & Drug Administration), *available at* http://democrats.energycommerce.house.gov/documents /20100722/Shuren.Testimony.07.22.2010.pdf.

134. *See* 21 U.S.C. § 360c (2006) (indicating that the classification of medical devices intended for human use is based on the safety and effectiveness of the particular device).

135. Oversight of Laboratory Developed Tests, 75 Fed. Reg. 34,463 (June 17, 2010).

136. Letter from James Woods, Deputy Dir., Patient Safety and Product Quality, Office

may be necessary for the FDA to phase in any new regulations to ensure continued patient access to LDTs currently being used.[137] Phased-in regulations may also allow the FDA to develop the staffing and resources necessary to implement these changes.[138]

B. NIH Registry Concerns

The NIH also received a large number of responses to its request for information regarding the development of the GTR.[139] Although the proposed GTR is voluntary, the initial recommendation of the Secretary's Advisory Committee on Genetics, Health, and Society was to develop a mandatory registry.[140] However, a number of genetic test companies stated that they would discontinue their genetic test offerings if the GTR became mandatory.[141] There is still some lingering uncertainty as to whether genetic test companies will participate and provide meaningful or useful information about their tests in a voluntary database.[142] The disclosure of some information may be hampered by the NIH's expectation that the GTR will be used to "[f]acilitate genetic and genomic data-sharing for research and new scientific discoveries."[143] Diagnostic firms and laboratories also contend that posting clinical validity or utility information while that data is still developing and emerging will not be a helpful measurement of the tests' use.[144] In addition, some test manufacturers have suggested keeping

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of *In Vitro* Diagnostic Device Evaluations and Safety, Ctr. for Devices and Radiological Health, to Steve Smith, Exec. Dir., IDENTIGENE (Nov. 4, 2010), *available at* http://www.fda.gov/MedicalDevices/Productsand MedicalProcedures/InVitroDiagnostics/ucm232746.htm *See also* Andrew Pollack, *F.D.A. Questions Drug Store Tests for Sexually Transmitted Diseases*, N.Y. TIMES (Nov. 5, 2010, 8:41 PM), http://prescriptions.blogs.nytimes.com/2010/11/05/f-d-a-questions-drug-store-tests-for-sexually-transmitted-diseases/.

137. *See, e.g.*, Letter from Am. Soc'y for Clinical Pathology & The Joint Comm'n, to Food & Drug Admin. (Aug. 13, 2010), *available at* http://www.regulations.gov/#!document Detail;D=FDA-2010-N-0274-0078 (outlining a process for applying a new regulatory scheme to existing LDTs).

138. Kirell Lakhman, *FDA's Effort to Update 'Consumers' on New MDx Developments Are Not Up to Snuff*, THE SAMPLE (Nov. 19, 2010), http://www.genomeweb.com/blog/fdas-effort-update-consumers-new-mdx-developments-are-not-snuff.

139. *See Request For Information Comments*, OFFICE OF BIOTECH. ACTIVITIES., http://oba.od.nih.gov/GTR/gtr_rfi_comments.html (last visited October 25, 2011).

140. SEC'Y'S ADVISORY COMM. ON GENETICS, HEALTH, AND SOC'Y, *supra* note 15, at 112–13.

141. Kirell Lakhman, *Some AMP Members Would Discontinue Tests if NIH's Genetic Test Registry Becomes Mandatory*, THE SAMPLE (July 29, 2010), http://www.genomeweb .com/blog/some-amp-members-would-discontinue-tests-if-nihs-genetic-test-registry-becomes-m.

142. Nat'l Inst. of Health, *supra* note 117.

143. *Id.*

144. Turna Ray, *Comments on Genetic Testing Registry Highlight Wide Range of*

information about cost and reimbursement off the registry to avoid the possibility that health insurance companies will use this information to determine coverage.[145]

Another concern regarding the development of the GTR is that this project seems to duplicate two already existent registries. The National Center for Biotechnology Information (NCBI) already runs GeneTests, a publicly funded medical information resource,[146] and GeneReviews, a database of "expert-authored, peer-reviewed, current disease descriptions that apply genetic testing to the diagnosis, management and genetic counseling of patients and families with specific inherited conditions."[147] The NIH has indicated that GeneTests will continue to receive funding after the GTR is developed,[148] and that GeneReviews will not be replaced.[149] Since the GTR will be developed by the NCBI,[150] the same program that already runs the GeneTests and GeneReviews databases, individuals in the industry and working for the government have expressed concern that the new NIH GTR will duplicate the two existing databases.[151] During the annual meeting of the Association for Molecular Pathology in November 2010, a highly respected lab director from the Weill Cornell Medical Center in New York, specifically asked NIH officials why the GTR was being created.[152] In response to concerns about duplicative efforts, the Office of Biotechnology Activity has explained that GeneTests was not built to support the technologies that the GTR is intended to cover, so it makes more sense technically and financially to develop a new database.[153] However, in order for the GTR to be a useful resource, the NIH needs to

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Stakeholder Concerns, PHARMACOGENOMICS REP. (Oct. 20, 2010), http://www.genomeweb.com/dxpgx/comments-genetic-testing-registry-highlight-wide-range-stakeholder-concerns.

145. *Id.*

146. *GeneTests*, NAT'L CTR. FOR BIOTECHNOLOGY INFO., http://www.ncbi.nlm.nih.gov/sites/GeneTests/?db=GeneTests (last visited Oct. 25, 2011).

147. *GeneReviews*, NAT'L CTR. FOR BIOTECHNOLOGY INFO.,http://www.ncbi.nlm.nih.gov/projects/GeneTests/static/about/content/reviews.shtml (last visited Oct. 25, 2011).

148. Kirell Lakhman, *Behind ACLA's 'Vapid,' 'Confusing,' 'Unhelpful,' and 'Ridiculous' LDT-IVD Meeting*, THE SAMPLE (Dec. 6, 2010), http://www.genomeweb.com//node/957125?hq_e=el&hq_m=889576&hq_l=7&hq_v=2a000b285a.

149. Matt Jones, *NIH Meets Public on Genetic Test Registry*, GENOMEWEB DAILY NEWS (Nov. 3, 2010), http://www.genomeweb.com/dxpgx/nih-meets-public-genetic-test-registry.

150. Plan to Develop a Genetic Testing Registry at the National Institutes of Health, 75 Fed. Reg. 62,406, 62,407 (Oct. 8, 2010).

151. Lakhman, *supra* note 148.

152. Kirell Lakhman, *Lab Director Sparks Controversy at AMP Conference by Questioning 'Credibility' of NIH Genetic Testing Registry*, THE SAMPLE (Nov. 20, 2010), http://www.genomeweb.com/blog/lab-director-sparks-controversy-amp-conference-questioning-credibility-nih-genet.

153. *Frequently Asked Questions*, OFFICE OF BIOTECHNOLOGY ACTIVITY, http://oba.od.nih.gov/GTR/gtr_faqs.html#GTR_FAQ023 (last visited Dec. 15, 2010).

avoid burdening the limited resources of laboratories with an additional responsibility to update the same or similar information in multiple places.[154]

Perhaps the greatest concern raised regarding the development and implementation of the GTR relates to practical concerns about how to best organize the GTR so that the information is useful and can be easily accessed and understood by all interested parties. Although some consumer groups worry that the genetic test companies will not include enough useful information in the registry, the corollary concern is that too much information will be included in the registry, which will render it useless for certain populations. Depending on what information is disclosed by the genetic test companies, the information included in the registry may be too technical for consumers, too simple for industry users, or too much for anyone to sufficiently understand.

C. Another Mixed Message Regarding the Genetic Testing Industry

An additional source of concern regarding the proposed changes to genetic testing regulation is the recent decision by the Secretary of DHHS to let the charter for the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) expire.[155] The SACGHS was established in 2002 to:

> (1) [p]rovide a forum for expert discussion and deliberation and the formulation of advice and recommendations on the range of complex and sensitive medical, ethical, legal and social issues raised by new technological developments in human genetics; (2) assist the Department of Health and Human Services and... other Federal agencies in exploring issues raised by the development and application of genetic technologies; and, (3) make recommendations to the Secretary of Health and Human Services concerning how such issues should be addressed.[156]

A scheduled review determined that the SACGHS had fulfilled its mandate by successfully addressing the major topics related to genetic technologies through comprehensive reports and recommendations.[157]

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154. Lakhman, *supra* note 141.

155. Turna Ray, *HHS Will Not Renew SACGHS Charter, Ending Committee's Decade-Long Tenure in October*, PHARMACOGENOMICS REP. (Sept. 29, 2010),http://www.genomeweb.com/dxpgx/hhs-will-not-renew-sacghs-charter-ending-committees-decade-long-tenure-october.

156. Establishment of the Secretary's Advisory Committee on Genetics, Health, and Society, 67 Fed. Reg. 65,126, 65,126 (Oct. 23, 2002).

157. Matt Jones, *UPDATE: HHS to Let Secretary's Genetics Committee Expire*, GENOMEWEB DAILY NEWS (Sept. 28, 2010), http://www.genomeweb.com/hhs-let-secretarys-genetics-committee-expire.

However, as evidenced by the recent FDA, Congressional, and NIH actions, and as stated by an official from the NIH, "genetics-related issues are not going away."[158] Although other DHHS committees will assume some of the SACGHS's duties, the number of recommendations set forth at the SACGHS's last meeting, suggest that the Committee's work will not be easily passed on.[159]

The termination of the SACGHS also sends a mixed message to the industry and consumers regarding the status of genetic testing. The SACGHS was first to suggest the development of a genetic test registry as a way to provide more information about specific genetic tests and the laboratories that perform them.[160] The SACGHS was also first to suggest that the FDA extend its regulatory oversight to all genetic tests.[161] Now that these recommendations are being considered and implemented, it seems odd to dissolve the committee that first determined the need and basis for change. Widespread, clinically-useful genetic testing still faces multiple challenges and it is unclear whether the FDA, the NIH, and the DHHS are fully prepared to respond to the competing interests and concerns that remain.

IV. CONCLUSION

Genetic testing is currently regulated by three separate schemes and the DHHS has proposed adding two significant pieces to the regulatory puzzle. While there is certainly a need to complete the regulatory puzzle, the FDA and NIH must ensure that any new pieces will help complete the picture without stifling industry innovation.

One of the underlying problems with regulating genetic testing is that the government, industry, clinicians, and consumers alike are still not sure how this technology can best be used and what consequences will result from using it. There is concern that the commercial viability of consumer genetics is unproven,[162] and neither of the current proposals address concerns that individuals who receive genetic test results may not be able to fully understand the results they receive.[163] Although there is some debate

158. *Id.*

159. *SACGHS Wraps Up with Recommendations on Sequencing, Comparative Effectiveness*, GENOMEWEB DAILY NEWS (Oct. 7, 2010), http://www.genomeweb.com/sacghs-wraps-recommendations-sequencing-comparative-effectiveness.

160. *See* SEC'Y'S ADVISORY COMM. ON GENETICS, HEALTH, AND SOC'Y, *supra* note 15, at 112–13.

161. *Id.* at 112.

162. Hogarth, *supra* note 8, at 325.

163. *See* Guttmacher & Collins, *supra* note 1, at 997 (explaining that the "[p]roper interpretation of screening results demands an understanding of the clinical implications of specific genotypes.").

about whether the concern that individuals will misunderstand their genetic test results is exaggerated,[164] these concerns are based on the notion that genetic information is unique.

Previous legislation and regulations have reaffirmed that genetic information is different than other medical information and therefore needs special protection.[165] Following this precedent, it makes sense to develop specific regulations and policies that address the issues related to consumer understanding of genetic test results. For example, the Center for Genetic Research Ethics and Law, a recent venture developed through a partnership between the Cleveland Clinic and Case Western Reserve University, is trying to diffuse the misperceptions that many have about genetic testing and research.[166] Similarly, a requirement that a board-certified practitioner complete the genetic test result interpretation would address concerns that individuals do not receive enough sound medical information with their test results.[167] Another possible solution would create a system where certain high-risk genetic tests are only available through a physician and other tests are available through the consumer market.[168]

Finally, Senator Orrin Hatch has suggested creating a new division within the FDA, the Center for Advanced Diagnostics Evaluation and Research, dedicated to the evaluation of diagnostic tests.[169] This new division would be responsible for ensuring the safety and efficacy of "advanced personalized diagnostics," a new category of tests that includes genetic tests and other LDTs.[170] Senator Hatch's proposal defines an

164. Ciara Curtin, *Researchers Report on Users' Understanding of Direct-to-Consumer Genetic Test Results*, GENOME TECH. (Nov. 30, 2010), http://www.genomeweb.com/dxpgx/researchers-report-users-understanding-direct-consumer-genetic-test-results (explaining that two recent studies found that early users of DTC genetic tests had a "moderately good understanding of their risks."); *cf.* Turna Ray, *ASHG Studies Shed Light on Consumers' Understanding of Gene Scan Reports, Response to Results*, PHARMACOGENOMICS REP. (Nov. 10, 2010), http://www.genomeweb.com/dxpgx/ashg-studies-shed-light-consumers understanding-gene-scan-reports-response-resul (finding that DTC genetic test consumers tended to feel that they understood more about their genetic test results than they actually did).

165. *See* discussion of GINA *infra* Part I.C.

166. *Center to Help Improve Patient Understanding of Genetic Testing*, BIOETHICS REFLECTIONS (Cleveland Clinic, Cleveland, Ohio), Fall 2010, at 6, *available at* http://my.clevelandclinic.org/Documents/Bioethics/reflections/fall10.pdf.

167. *Genetics Experts Consider Tighter Regulation, Potential Ban on DTC Genetic Testing*, GENOMEWEB DAILY NEWS (Aug. 11, 2010), http://www.genomeweb.com/dxpgx/genetics-experts-consider-tighter-regulation-potential-ban-dtc-genetic-testing.

168. Hogarth, *supra* note 8, at 324.

169. Turna Ray, *Draft Bill Proposes FDA Create New Division to Review 'Advanced Personalized Diagnostics'*, PHARMACOGENOMICS REP. (June 23, 2010), http://www.genome web.com/dxpgx/draft-bill-proposes-fda-create-new-division-review-advanced-personalized-diagnos.

170. *Id.*

advanced personalized diagnostic as a medical product "distinct from a device" and suggests "establish[ing] regulatory criteria that address the unique characteristics of such products."[171] Although Senator Hatch has not yet introduced a bill with his proposal,[172] this new legislation could significantly impact any choices the FDA makes regarding the regulation of LDTs.

Despite the flurry of activity, investigations, and comments collected throughout 2010, the FDA, the NIH, and the DHHS are still trying to sort out the pieces of the genetic testing regulatory puzzle. In the meantime, DTC genetic testing companies continue to find new avenues into the consumer marketplace. For example, in addition to expanded genetic counseling offerings,[173] 23andMe recently announced a new annual subscription-based plan that will enable the company to update its tests and its customers' test results with an average of two to five new genetic discoveries per month.[174] This is just one example of the wide range of rapid changes that occur throughout the genetic testing industry, and demonstrates why the DHHS needs to ensure that all pieces of the regulatory puzzle are put into place without limiting industry innovation. Better FDA regulation of LDTs and an organized, accessible, and useful GTR may help create a complete picture of genetic test regulation.

171. Turna Ray, *Latest Draft of Hatch IVD Bill Contains New Regulatory Proposals; Pricing Reforms Under Discussion*, PHARMACOGENOMICS REP. (July 6, 2011), http://www.genomeweb.com/node/973340/?hq_e=el_m=1046128_l=8_v=068ee2d6b0.

172. *Id.*

173. *23andMe Partner to Provide Genetic Counseling*, GENOMEWEB DAILY NEWS (June 7, 2010), http://www.genomeweb.com/dxpgx/23andme-partner-provide-genetic-counseling.

174. *23andMe Launches New Pricing Model, Charges Monthly Fee*, GENOMEWEB DAILY NEWS (Nov. 24, 2010), http://www.genomeweb.com/dxpgx/23andme-launches-new-pricing-model-charges-monthly-fee.