THE NEED FOR INCREASED OVERSIGHT OF GENETIC TESTING:
A DETAILED LOOK AT THE GENETIC TESTING PROCESS

David C. Bonnin*

INTRODUCTION

With every great blessing comes responsibility. The medical genomics revolution is no exception. The sequencing of the Human Genome and the ever increasing understanding of the molecular biology of disease have enabled the development of gene therapies, opened the possibilities of therapeutic cloning techniques, and given rise to many other avenues of discovery for preventing and curing disease. One such developing technique is predictive genetic testing.¹ Physicians can now take a sample of blood or tissue from a patient and send it to a testing lab that will run comparative genetic tests yielding data on the patient’s predisposition to various forms of genetic disease.² Using this information, patients and physicians can determine a course of lifestyle, diet, and sometimes medication that can work to counteract the predisposition and mitigate the ef-

---

* Candidate for Doctorate of Jurisprudence, University of Houston Law Center: May, 2004. The author would like to thank his former colleagues at the Human Genome Sequencing Center at Baylor College of Medicine, the faculty of the University of Houston Health Law & Policy Institute, most notably Professors Rich Saver and Mary Anne Bebitchki for their guidance early on, and his colleagues and mentors at the Houston Journal of Health Law & Policy for their patience and tireless dedication.

¹ Genetic testing is defined as “the analysis of chromosomes, genes, and/or gene products to determine whether a mutation is present that is causing or will cause a certain disease or condition.” A Public Consultation on Oversight of Genetic Tests, Secretary’s Advisory Committee on Genetic Testing, December 1, 1999 – January 31, 2000, available at http://www4.od.nih.gov/oba/sacgt/reports/Public_Consultation_document.htm (last visited Sept. 13, 2003) [hereinafter SACGT Public Consultation].

fects of the patient's genetic shortcomings. This technique has the potential to prolong many lives and save countless people from the pain and suffering that can accompany genetic diseases.

However, genetic testing in the current environment has raised a number of very real concerns for patients, physicians, marketers of tests and testing service providers. Questions remain concerning problems such as quality assurance of laboratories and laboratory staff offering such testing, clinical validity of the tests offered, analytical validity, accuracy, physician education about the significance of test results, physician competency in pre- and post-testing counseling, and maintaining patient privacy with respect to the results of predictive tests. Social concerns regarding the creation of a genetic underclass and genetic discrimination in the workplace and in health insurance availability are also of paramount importance.

The difficulty of these issues brought about early attempts to shape national policy in the face of blossoming genetic discovery in the 1990s. In response to calls by two of these early advisory groups, The Task Force on Genetic Testing (TFGT) and the Joint NIH/DOE Committee to Evaluate the Ethical, Legal, and Social Im-

---

1 SAGCT Public Consultation, supra note 1. Genetic testing has many purposes, including "prenatal diagnosis, newborn screening, carrier testing, diagnosis/prognosis, presymptomatic testing and predictive testing." Id.

2 Id.


4 See generally id.


6 SAGCT Public Consultation, supra note 1.

lications Program of the Human Genome Project, then-Secretary of Health and Human Services Donna Shalala chartered the Secretary's Advisory Committee on Genetic Testing (SACGT) in June, 1999. Following the lead of the TFGT recommendations, and with an eye to the contributions of the SACGT, this article will attempt to crystallize the current calls for increased regulatory oversight of genetic testing in three areas: the regulation of new genetic tests and testing services, the quality assurance function of federal regulation of laboratories performing genetic tests, and the improvement of physician care with respect to genetic testing.

STATE OF THE ART: Genetic testing has been defined as "a test for determining the presence or absence of an inherited genetic characteristic in an individual, including tests of nucleic acids such as DNA, RNA and mitochondrial DNA, chromosomes or proteins in order to identify a predisposing genetic characteristic." Genetic testing can be either predictive or diagnostic in nature, and can have many different purposes. Currently, genetic testing is being used for pre-implantation diagnosis following in vitro fertilization, prenatal diagnosis, newborn screening, carrier screening, diagnostic or confirmatory testing, and predictive testing. Predictive testing is largely recommended to those who have a family history of a given disease and can be used to assess a predisposition to genetic disease.

10 See Press Release, Health and Human Services, HHS Forms Genetic Testing Advisory Board (Aug. 7, 1998) available at http://www4.od.nih.gov/oba/SACGT/press_releases/PR89797.htm (quoting Secretary Shalala as saying "we need to ensure that test results are accurate and medically valid and that this information remains confidential") (on file with author).

11 See generally TFGT Final Report, supra note 5. The report also contains recommendations for dealing with the emerging problem of encouraging a marketplace for and increased regulation of genetic testing of rare genetic diseases in particular, a topic not addressed in this essay. Id.

12 Michael Malinowski, Separating Predictive Genetic Testing From Stake Oil: Regulation, Liabilities, and Lost Opportunities, 41 JURIMETRICS J. 23, 31 (citing the New Jersey Definition, codified at N.J.STAT. ANN. § 17B:30-12 (West 1998)). Another more technical definition is found in the Recommendations of the SACGT: "A genetic test is an analysis performed on human DNA, RNA, genes and/or chromosomes to detect heritable or acquired genotypes, mutations, phenotypes, or karyotypes that cause or are likely to cause a specific disease or condition. A genetic test also is the analysis of human tissues and certain metabolites, which are predominantly used to detect heritable or acquired genotypes, mutations, or phenotypes." Secretary's Advisory Committee on Genetic Testing, National Institutes of Health, Enhancing the Oversight of Genetic Tests: Recommendations of the SACGT 1 (July 2000) [hereinafter SACGT Recommendations].

13 SACGT Public Consultation, supra note 1.

14 SACGT Recommendations, supra note 12, at 2.
when used in combination with family history information. The predictive power of genetic testing with respect to the onset of symptoms can be wildly variable and is dependent on a number of factors, including the accuracy of the test and variances in gene expression. Despite these difficulties, the technology is advancing rapidly. Combined with the exponential development of bioinformatics systems, emerging scientific breakthroughs will likely expand genetic testing capabilities in a breathtaking fashion. For example, Affymetrix, a private biotech company, is developing biochips the size of a quarter that could potentially test for 400,000 DNA arrays at a time. However, despite these quantum leaps in technical proficiency, in most cases current understanding of the association of genetic mutations and disease is fledgling at best. Furthermore, a test might not always detect all the mutations of a gene, not all mutations have the same effects, and diseases can be caused by complex interactions between genetic and environmental factors that are not always clearly understood.

STATE OF THE INDUSTRY: The high profile success of the Human Genome Project and its private counterpart, Celera Genomics, has provided a significant flow of investment dollars for big biotech companies and small startups alike. Development of testing for high penetrance genetic diseases that show simple inheritance pat-
terns has come to a virtual halt. Nonetheless, the marketplace for genetic testing is increasing at a rate of thirty percent annually. Currently there are 1086 genetic medicine clinics and 579 laboratories offering testing for 990 diseases. This is more than three times the number of diseases detectible by genetic testing just two-and-a-half years ago. Turn around times vary and costs can be as low as $100 to as high as $2000 based on factors such as contractual agreements for volume discounts, sample handling requirements, volume of the particular type of testing performed, testing methodology, etc. Estimates are that in 2000 the U.S. genetic testing industry generated $319.9 million in revenue, a number projected to grow to $1.17 billion in 2007. New genetic screening technology using tandem mass spectroscopy has amplified the number of tests that can be performed on a single sample. Indeed, Germany is on the verge of implementing a mandatory mass spec screening program for all

25 Henry T. Greely, Lecture Series on Genetics in the Law, University of Houston Law Center (Jan. 2003) (on file with author). Perhaps the reason for this is that all of the obvious, high penetrance, well correlated disease genes have been found and identified. From an evolutionary biological perspective, it seems reasonable that there would be fewer of these types of genetic diseases, because they would be selected against most strongly over time. Id.; see also Ann Y. Huang, FDA Regulations of Genetic Testing: Institutional Reluctance and Public Guardianship, 53 FOOD & DRUG L.J. 555, 559 n.36 (1998) (stating that "most single-gene disorders are quite rare and have less market potential, so to earn high profits the tests must involve more common disorders, which often turn out to be more genetically complex.");


27 These statistics are updated daily by Genetests-Geneclinic, "a publicly funded medical genetics information resource developed for physicians, other healthcare providers, and researchers, available at no cost to all interested persons." available at http://www.genetests.org/ (last visited Sept. 29, 2003) [hereinafter Genetest].

28 SACGT Recommendations, supra note 12 at 3 (citing Genetest in July 2000, noting that 300 diseases were detectable by genetic testing).


31 Eliot Marshall, Fast Technology Drives New World of Newborn Screening, 294 Sci. 2272 (2001). The article touches on the interesting and very important debate currently raging over whether or not to introduce mandatory newborn screening for several diseases. See generally id.
newborns, a move that, if instituted here in the United States would possibly force wholesale changes in the regulatory framework. In light of this rapid growth and development in the genetic testing industry, commentators agree that a successful regulatory framework must be flexible enough to withstand dynamic market shifts and large jumps in market production.

**STRUCTURE OF PAPER:** This essay began with a brief introductory statement, proceeded to describe the current state of the art in predictive genetic testing and also included an introduction to the state of the genetic testing industry. The rest of the paper will walk through a generalized genetic testing procedure and examine the current regulatory situation governing each step. In some selected steps, it will discuss the need for increased or modified regulation and present a summary of the current thinking and recent steps toward a new regulatory framework.

In order for there to be genetic testing, beneficial genetic tests have to be developed. Thus the logical starting point for regulatory analysis is development of testing services or products. The first issue deals with what (if any) restrictions should be placed on the development of new tests or services. This section includes a discussion of possible criteria for deciding whether a test is beneficial and should be developed, who should develop the test, and how these standards should be enforced.

The next step is to prepare the delivery system for these new products and services. This section will deal largely with determining who should act as gatekeeper to these products and services and how to prepare the gatekeeper(s) to deliver genetic testing in an efficient, cost-effective manner that is beneficial to the patient.

Next we discuss the provider—patient interaction. This section will cover setting and enforcing standards for patient education, informed consent, genetic counseling, and will touch on the sensitivity of the privacy issues involved.

---

32 Hanna Cleaver, Germany May Screen All Newborns for 20 Disorders, REUTERS LIMITED, February 13, 2002 (quoting Professor Adelbert Roscher at the University Children’s Clinic in Munich: “We are far ahead of the USA and will be the first country in the world to do this. It could be possible to have all newborns in Germany tested as routine by the end of this year.”).

33 See Marshall, supra note 31.

The logical next step in the discussion will be to dissect the need for standards in gatekeeper prescription or recommendations for testing. Following the framework established above, this essay will discuss the development of criteria for deciding when a particular genetic test is appropriate for a particular patient, who will develop the criteria, and how these criteria should be enforced. In this context, the role of the tort system for deterring uninformed or careless decisions, as well as potential roles for physician licensing and accreditation organizations, professional organizations, and administrative oversight, will be discussed.

The essay continues with brief discussions of sample collection and transmission before moving on to the regulation of the testing itself. Here the paper will outline the current roles of regulatory tools like the Food and Drug Administration (FDA), Clinical Laboratories Improvements Amendments (CLIA) and state licensure as well as considering potential expansions of the impact of these tools in this area.

Finally, the essay concludes with a section on the last step in the genetic testing process—dissemination of results and data. Discussions of counseling and follow up will both surface here.

I. DEVELOPMENT OF TESTING PRODUCTS AND SERVICES

The issue of whether or not the development of genetic testing should be restricted at all is a central question. As discussed more fully below, under the current regulatory system, the FDA has thus far chosen not to restrict the development of genetic testing services. Furthermore, downstream regulation of the actual testing procedures does not place any restriction on the development of new tests so long as the methods used in the testing are analytically valid. This basically means that companies are free to experiment with and develop any genetic testing technology they like, only facing restrictions on testing using human subjects. While it is generally agreed that genetic testing has great potential to enhance human longevity and quality of life, it is also agreed that genetic

---

35 See Koenig, et al. infra note 214, and accompanying discussion.
36 Nell A. Holtzman, FDA and the Regulation of Genetic Tests, 41 JURIMETRICS J. 53, 57 n.17. Analytical validity is defined by Nell Holtzman as “the probability that (1) the test will be positive when the analyte it is intended to measure is present and (2) the test will be negative when the analyte it is intended to measure is absent.” Id.
37 See Anita Silvers & Michael Ashley Stein, An Equality Paradigm for Preventing Genetic Discrimination, 35 VAND. L. REV. 1341, 1348-49 (2002) (citing numerous potential benefits in-
testing, when done poorly, has the potential for great human harm.  

There are two ways to prevent tests which have a greater potential for harm than good from reaching consumers. Either allow the test to be developed, but restrict access to it, or develop a set of criteria by which to judge whether a test has a greater potential to help than harm. This second solution is advantageous from an economic efficiency standpoint because it enables marketability determinations before allowing expenditure of limited research resources.

So how should these criteria be developed and what should they be? One of the SACGT’s missions was to come up with a methodology for classification of genetic tests. Ostensibly, this would allow whatever regulatory agency in charge to set appropriate levels of scrutiny for developing genetic testing technologies or services.

In order to carry out this mission, the SACGT convened a working group in August of 2000. After just one day of discussion, the working group created a set of four criteria that would allow classification of genetic tests into one of two scrutiny levels for regulatory review. The four criteria included:

- test volume,
- usage of the test for population-based study,

including enabling individuals to prepare for disease onset, act prophylactically to delay or eliminate onset, and engage in life planning regarding offspring and career).


40 This is a bad solution for a number of reasons, not the least of which is that desperate people do desperate things. One can imagine a genetic testing black market that preys upon people with significant family history or other indications of possible disease propensity, marketing and selling genetic tests that have little or no value to the patient. Henry T. Greely, Lecture Series on Genetics in the Law, University of Houston Law Center (Jan. 2003) (on file with author).


42 Id. (stating "SACGT initially considered a classification scheme to be an essential initial step in the process of test evaluation.").

43 Id. (stating "the Working Group was chaired by SACGT member Dr. Wylie Burke and composed of SACGT members, SACGT ex officio members, and ad hoc experts representing relevant professional and private sector organizations.").

44 Id.
diagnostic or predictive nature of the test, and
a trio of questions related to:
the availability of a treatment,
the predictive value of the test, and
the potential for social or medical harms.44

After an initial acceptance by the SACGT proper, problems
arose regarding the proposed classification methodology when the
Centers for Disease Control and Prevention’s (CDC) Genetic Labo-
ruminary Forum conducted pilot-tests of the methodology.45 The
SACGT went back to the drawing board in an attempt to modify its
proposed methodology to eliminate test volume, add analytical va-
lidity and further define population screening and disease fre-
quency.46 Submission of this new classification system for public
comment yielded numerous and varied responses from a broad
cross section of commentators.47 Unfortunately, in light of “irresolv-
able questions” raised by the commentators about the proposed
classification methodology, the SACGT decided to table discussion
in this area.48 However, SACGT did make note of the fact that the
work they had done could be useful in future determinations.49

The biggest problem with the SACGT methodology was that
they attempted to set up the classification scheme based on a “lim-
ited set of elements in a simple, linear fashion.”50 Thus, when they
opened up for public comment, they were confronted with a large
number of elements and factors that the methodology did not con-
sider.51 Among those listed were:
test sensitivity and specificity,
genetic heterogeneity,
penetrance,
low predictive value,
potential for social stigma,

44 Id.
45 Classification Method, supra note 40. These problems largely revolved around the test
volume criterion, which the CDC found to be unreliable in light of the dynamic potential
value over time. Id. Furthermore, definitional questions with regard to population
screening and the risks of off-label use contributed to the downfall of this first proposed
methodology. Id.
46 Id.
47 Id.
48 Id.
49 Id.
50 Classification Method, supra note 40.
51 See id.
predictive tests,
tests for behavioral disorders,
pharmacogenetic testing,
complexity of test,
difficulty of test interpretation,
burden of disease,
pattern of inheritance,
late onset disorders,
availability of proven treatments or prevention,
clinical utility,
prenatal testing,
disease incidence or progression,
availability and strength of confirmatory procedures and
the reliability of clinical corroboration.\textsuperscript{32}

It is easy to understand how such an influx of factors to consider would be overwhelming to SACGT. It is also easy to conceive that perhaps the problem suggests its own solution. Because the development of a linear system could not possibly work with so many factors, all or any of which may have bearing on the impact of the next, it seems that a balancing of these factors would be the next best option. However, it is more difficult to see how each of these factors might cut in a balancing test. For instance, should pharmacogenetic testing be subject to higher or lower scrutiny? What is needed, then, if such a classification methodology is going to work, is a thorough evaluation of each factor. This is likely to be a very daunting task, perhaps more daunting than developing a linear model using a limited number of factors, for a number of reasons. Because of these difficulties, the SACGT chose to table further discussion of classification methodologies.\textsuperscript{33}

In the meantime, several alternative approaches to regulating test development have arisen. Of course, the FDA could apply its traditional regulatory framework to genetic tests without classification schemes to determine levels of scrutiny.\textsuperscript{34} Indeed, the SACGT seemingly convinced the FDA to take a bigger bite of the regulatory apple. In the early days of the most recent Bush administration, the FDA was working on a novel regulatory “template” to tackle this

\textsuperscript{32} Id.

\textsuperscript{33} Id.

\textsuperscript{34} See generally Barbara Koenig, et al., Genetic Testing for BRCA1 and BRCA2: Recommendations of the Stanford Program in Genomics, Ethics, and Society, 7 J. Women's Health 531, 540 (1998).
issue. A discussion of these alternatives is the next major topic of this paper. As SACGT points out, in the existing regulatory framework, there is no official distinction between genetic and nongenetic medical testing. The following section discusses the current regulatory scheme for nongenetic testing, which is applicable to genetic testing by default.

The FDA has oversight of all laboratory tests and their components under the Federal Food, Drug and Cosmetic Act (FDCA), as amended by the Medical Devices Act (MDA) and the Safe Medical Devices Act of 1990. Under the act, laboratory tests are considered to be diagnostic devices and tests packaged and marketed as kits to multiple laboratories require pre-market approval by the FDA. The FDA classifies devices according to the risks involved in their


56 SACGT Recommendations, supra note 12, at 8.

57 The wisdom of this non-distinction touches on a very important debate regarding what is called the theory of genetics exceptionalism. While it does not directly impact the subject of this paper, this interesting and important subject tangentially informs the discussion. Several articles on the debate have been written. The basic argument is over the degree of special treatment that should be given to our genes, genetic information, and the science of genetics in general. There is a popular belief among the semi-informed public that our genes are "magical" in that they determine who and what we are on the most basic levels. On the other hand, many academics are now beginning to argue that this view is unfounded, unsafe, or just plain incorrect. See generally Thomas H. Murray, Genetic Exceptionalism and "Future Diaries": Is Genetic Information Different from Other Medical Information?, in GENETIC SECRETS: PROTECTING PRIVACY AND CONFIDENTIALITY IN THE GENETIC ERA 60, 61 (Mark A. Rothstein ed., 1997); DOROTHY NELKIN & M. SUSAN LINDEE, THE DNA MYSTIQUE: THE GENIE AS A CULTURAL ICON 166-68 (1995); LAWRENCE O. GOSTIN & James G. Hodge, Jr., Genetic Privacy and the Law: An End to Genetics Exceptionalism, 40 JURIMETRICS J. 21, 33-34 (1999); Henry T. Greely, Point/Counterpoint: Genetic Discrimination: The Complex Case for Some Legislative Protection, 149 U. PA. L. REV. 1483 (2001); Ronald M. Greem & A. Mathew Thomas, DNA: Five Distinguishing Features for Policy Analysis, 11 HARV. J.L. & TECH. 571 (1998); Pauline T. Kim, Genetic Discrimination, Genetic Privacy: Rethinking Employee Protections for a Brave New Workplace, 96 N.W. U. L. REV. 1497, 1543-46 (2002); Glenn McGee, Forward: Genetic Exceptionalism, 11 HARV. J. L. & TECH. 565 (1998); Mark A. Rothstein, Why Treating Genetic Information Separately Is a Bad Idea, 4 TEX. REV. L. & POL. 33 (1999); Sonia M. Suter, The Allure and Peril of Genetics Exceptionalism: Do We Need Special Genetics Legislation?, 79 WASH. U. L. Q. 669 (2001).

use and imposes various levels of restriction and review for each classification level.\textsuperscript{60} Due to the complex nature of the tests themselves, most genetic testing kits are considered Class III devices,\textsuperscript{61} which do require pre-market approval.\textsuperscript{62} To get this approval, kit manufacturers are required to submit an application.\textsuperscript{63} The application is subject to review by an FDA examiner, one of whom the FDA has designated to specialize in genetic tests.\textsuperscript{64}

Although the FDA regulates genetic tests marketed through interstate commerce as kits under the Medical Device Amendments to the FDCA, they do not regulate genetic testing when it is offered as a service.\textsuperscript{65} This distinction is important. While some are more inclined to support genetic testing as a do-it-yourself type of procedure for privacy and anti-discrimination reasons, concerns regarding the availability and nature of genetic counseling that will accompany such test kits are serious.\textsuperscript{66} Numerous academic articles and commission reports stress the importance of counseling.\textsuperscript{67} One can reasonably assume that a patient would get greater access to such counseling when the test is administered through a physician-referral clinic rather than a take-at-home test. It is unclear however, which type of testing, a service or a kit, is likely to yield more accurate results. While it may seem that a testing service would have in place more sophisticated quality control and assurance mechanisms, the fact that testing kits have to pass FDA muster may indicate that

\textsuperscript{60} See Huang, supra note 25, at 588 (citing 21 U.S.C. § 360c (FDCA § 513)).


\textsuperscript{63} Id.

\textsuperscript{64} Id. at n.231 (citing telephone interview with Dr. Steven Gotman, Director of Division of Clinical Laboratory Devices, Food and Drug Administration (May 1, 1998); Food and Drug Admin., Center for Devices and Radiological Health, Monitoring the Human Genome Project for Impact on Developments in Medical Devices [contents available at the FDA Freedom of Information Office, 5600 Fisher’s Lane # 12A16, Rockville, MD 20857]).

\textsuperscript{65} Holtzman, supra note 36, at 53-54 n.3.

\textsuperscript{66} Huang, supra note 25, at 589. Reportedly, the FDA at one time considered requiring companies who were developing genetic testing kits to provide for counseling. Id.

\textsuperscript{67} See generally, e.g., Laura M. McConnell et al., Genetic Testing and Alzheimer Disease: Recommendations of the Stanford Program in Genomics, Ethics, and Society, 3 GENETIC TESTING 3 (May 1999).
they have a higher accuracy. Furthermore, while a testing service might have in place skilled (or at least highly trained) technicians, a person is more likely to pay very close attention to any protocol if it is his own tissue he is testing. On the other hand, some commentators suggest that despite this practical reasoning, those genetic tests regulated by the FDA are generally of better quality. Regardless of how they stack up quality-wise, it remains clear that predictive genetic testing services are a growth business.

In addition, it should be noted that though the FDA does not currently regulate genetic tests offered through laboratory testing services, this is not because they lack the power to do so. The FDA has expressed publicly that it could expand its regulatory reach in this area. Despite these public announcements of authority, questions remain about the FDA’s legal jurisdiction over genetic testing services. The FDCA expressly rests upon federal commerce power, so by the statute, the FDA “must be able to point to a product used in such services that has moved or will move in interstate commerce” as a target for its regulation. Another issue is raised by the FDA’s longstanding position that it may not regulate the practice of medicine.

If jurisdiction can be established, many commentators have highlighted good reasons for choosing the FDA as the primary regulatory agency for genetic testing services. Professor Michael Malinowski summarized these reasons expertly in his article published

---

66 See Holtzman, supra note 36.
67 Other distinctions exist, including the utility of anonymous data gathered by testing services for disease surveillance efforts.
69 Fiscal Year 2001 FDA Budget Request, supra note 26 and accompanying text.
70 See TFGT Final Report, supra note 5, at 30 (citing Personal Communications from D. Bruce Burlington, M.D. Director, Center for Devices and Radiological Health, FDA, April 3, 1996).
72 Id.
73 Id. (citing 21 U.S.C. § 331 and Peter Barton Hutt & Richard A. Merrill, FOOD AND DRUG LAW 1066 (2d ed. 1991)).
in Fall 2000.\textsuperscript{77} Among the positive reasons for choosing the FDA were FDA familiarity with regulating medical devices and evidence of past success using the current device classification approach.\textsuperscript{78}

There are also potential drawbacks for FDA expansion into regulation of genetic testing services. For one, the FDA may not have sufficient resources to carry out this mission effectively.\textsuperscript{79} There are the matters (discussed above) concerning regulatory authority and potential regulation of the practice of medicine. Furthermore, it is not at all clear at this point that the FDA is willing to take on such a task. Until recently, the agency resisted suggestions to regulate sperm, \textit{in vitro} fertilization clinics, and egg banks.\textsuperscript{80} Finally, there are real concerns regarding FDA competence to address the complex social issues attached to genetic testing which go beyond mere product performance concerns — issues that “lie outside FDA’s statutory mandate and recognized expertise.”\textsuperscript{81}

Another important regulatory monitor of developing testing services could be the use of Institutional Review Boards (IRBs). Under the Medical Device Amendments to the FDCA, FDA mandates Institutional Review Board (IRB) approval for all protocols used to develop new medical devices.\textsuperscript{82} As mentioned above, the FDA has not enforced this requirement on laboratories that are developing genetic tests to be offered as services.\textsuperscript{83}

IRB review is also required for organizations that wish to use federal grant funds to develop genetic tests.\textsuperscript{84} However, due to the administrative difficulties in obtaining IRB approval and the speculative nature of the efficacy of many genetic tests in development,

\textsuperscript{77} Malinowski, supra note 12, at 43.

\textsuperscript{78} Id. Other positives listed: “FDA regulation would carry assurances of compliance with good manufacturing practice standards, and the agency could expedite clearance through its authority over labeling, advertising, marketing, and through post marketing controls.”

\textsuperscript{79} Id. FDA could self-fund this task to some degree by enacting user fees. See Merrill, supra note 73, at 64-66.

\textsuperscript{80} Merrill, supra note 73, at 65.


\textsuperscript{82} Merrill, supra note 73, at 64. Specifically, Professor Merrill refers to issues of test recipient autonomy, informed consent and privacy. Id.


\textsuperscript{84} Id.
many testing services are developing their products with private funds, and have not sought IRB approval of their protocols. According to the Task Force, IRBs were created to protect human subjects from the risks associated with participation in investigational research. In order to accomplish this mission, IRBs “must recognize the risks posed by genetic test development and determine that investigators have taken adequate steps to apprise subjects of these risks and reduce the chance of harm from those risks.” The Task Force for Promoting Safe and Effective Genetic Testing in the United States recommended expanding IRB requirements to the development of all genetic testing, whether to be marketed as a kit, a service, developed with federal or private funding.

A third tool in the regulatory arsenal is the Clinical Laboratories Improvements Act (CLIA), passed by Congress in 1988. As early as 1992, commentators recognized that the CLIA of 1988 could be extended to include federal oversight of DNA analyses.

As outlined in more detail later, CLIA functions to establish minimum quality levels in laboratory testing practices. The regulations establish three levels of test complexity, classify laboratories according to the complexity of the tests they perform and set forth standards for “quality control, quality assurance, patient test management, personnel, inspections and proficiency testing.”

Many have pointed out, however, that without serious overhauling, the statute is totally inadequate to serve as a complete regulation of genetic testing labs. As it stands now, CLIA does not

---

85 Id.
86 Id. at 31.
88 Id. at 34. “Protocols for the development of genetic tests that can be used predictively must receive the approval of an IRB when subject identifiers are retained and when the intention is to make the test readily available for clinical use….. IRB review should consider the adequacy of the protocol for: (a) the protection of human subjects involved in the study, and (b) the collection of data on analytic and clinical validity and data on the test’s utility for individuals who are tested.” Id. at 30.
90 See Robin Elizabeth Margolis, Genetic Testing: Should Clinical Laboratory Regs Cover DNA Analysis?, 9 No. 9 HEALTHSPAN 18, 18 (1992).
92 Huang, supra note 25, at 589 n. 239.
require an IRB review of research protocols used by the laboratories it governs. Additional criticisms of the CLIA provisions as regulatory tools for genetic testing include the lack of a specific category for genetic tests and the fact that CLIA requires laboratories to show analytical validity, but contains no requirement for clinical validity or utility. Furthermore, the Health Care Finance Administration (HCFA) admits that CLIA does not address issues relating to genetic counseling or informed consent. The HCFA and CDC are attempting to develop specific requirements for genetic testing, among them provisions for both pre- and post-analytical phases of the testing process.

A fourth set of regulatory tools used to keep genetic test development safe are Human Research Subject Protection Laws. These are administered by two different agencies, depending on the nature for the research. OHRP has oversight duties for research funded by the Department of Health and Human Services, while the FDA covers human subjects in investigational trials of devices, drugs and other biologics being developed for commercial testing. Both oversight organizations, however, enforce similar regulatory rules. Basically, these regulations require IRB review of a study with a focus on three elements: 1) safety of the subjects; 2) sufficiency of the informed consent process; and 3) balance of the risks.
and potential benefits of the study. There are loopholes in this scheme with respect to genetic testing services. First, the FDA regulations apply only to kits and not to services. Second, OHRP regulations only apply if the genetic test is used for patient care purposes. It seems that this is another area where governmental reluctance is allowing commercial genetic testing services to slip through regulatory cracks.

Given the regulatory structure outlined above, is there a need for more regulation? The SACGT thought so. Several other commentators agree. Most of those calling for increased regulation agree that the FDA is the most appropriate vehicle for the reasons discussed above. Some commentators are more tentative in vesting the FDA with full regulatory power.

Despite this near unanimity, the clamor for increased regulation of genetic testing services is receiving little attention. As recently as last year, published remarks by the FDA’s Acting Principal Deputy Commissioner of Food and Drugs contained only fleeting mention of genetic testing and no discussion of regulation. However, other departments have stepped up:

In response to the SACGT recommendations, the Office of Device Evaluation’s (ODE) Division of Clinical Laboratory Devices (DCLD) has under development a “Genetics Template” which will serve as an outline for collecting administrative, analytical, and clinical data on tests used to detect the presence of genetic diseases. DCLD has been developing this template in collaboration with professional laboratory and clinical organizations. Additional steps for

104 Id. at 10-11.
105 See id. at 10.
106 See SACGT Recommendations, supra note 12, at 11 ("meaning that results are provided to a subject, the subject’s family, or to the subject’s health care provider.");
107 Id. at ix. "Based on the rapidly evolving nature of genetic tests, their anticipated widespread use, and extensive concerns expressed by the public about their potential for misuse or misinterpretation, additional oversight is warranted for all genetic tests." Id.
109 See e.g., SACGT Recommendations, supra note 12, at 13; Holtzman, supra note 36, at 54; Malinowski, supra note 12, at 41; Huang, supra note 25, at 589-90.
110 Geetter, supra note 70, at 37 (stating "[FDA’s] track record suggests neither an eagerness nor a clear sense of how regulation should be structured"); Merrill, supra note 73, at 66 (stating "I do not oppose giving FDA a larger role"); Numnally, supra note 108, at 344-45.
111 See Bernard A. Schweitz, Remarks of the Acting Principal Deputy Commissioner of Food and Drugs, 56 Food & Drug L.J. 123 (2001).
the potential oversight of genetic disease testing are still in the planning stages but information collected in the templates could enable FDA to focus its attention to monitoring genetics testing activities.\textsuperscript{112}

Again, this statement seems to be directly limited to testing kits, as there is no mention of expanding FDA oversight to testing services.\textsuperscript{113} The SAGCT has been disbanded by Secretary of Health and Human Services, Tommy Thompson, which has been closely followed by a shift in FDA policy regarding the potential for an increased role in the regulation of genetic testing.\textsuperscript{114} Where the SAGCT had convinced the FDA to take on a larger role, there seems to have been an about-face and now the FDA is once again considering whether they have the authority to take any steps toward greater oversight.\textsuperscript{115}

II. PREPARING THE DELIVERY MECHANISM

Once a genetic test or testing service has been developed, the next step is preparation for its delivery into the commercial/clinical marketplace. In order for a test to be used properly, information about the test must be evaluated and disseminated to providers and to the public; providers must be trained to administer the tests, and delivery mechanisms must be established.

The Agency for Healthcare Research and Quality (AHRQ) is the lead federal agency in health care quality and plays an important role in the monitoring and dissemination of information pertaining to effectiveness of diagnostic and therapeutic medical interventions.\textsuperscript{116} As the "health services research arm of the U.S. Department of Health and Human Services" (HHS), the AHRQ complements the scientific research efforts of the National Institutes of Health.\textsuperscript{117} As a self-proclaimed science partner, the AHRQ works with the public and private sectors to investigate what works and

\begin{itemize}
\item \textsuperscript{113} Id.
\item \textsuperscript{115} Id. (quoting Neil Holtzman as saying, "this is a real turnaround. It's bad. It's terrible.").
\item \textsuperscript{116} SACGT Recommendations, supra note 12, at 11.
\item \textsuperscript{117} What is AHRQ?, http://www.ahrq.gov/about/whatis.htm (last visited Sept. 5, 2003).
\end{itemize}
does not work in health and health care, and translates this knowledge into practice and policymaking.\textsuperscript{118} AHRQ also works to benefit quality measurement and improvement in health services.\textsuperscript{119} To this end, they oversee the Patient Safety Task Force, which is a HHS effort to collect data about medical errors and reduce the number of injuries resulting from them.\textsuperscript{120}

The SACGT acknowledged that “AHRQ is expected to play a greater role” in the organization and dissemination of information about the effectiveness of genetic testing technologies.\textsuperscript{121} AHRQ is already participating by supporting U.S. Preventative Services Task Force evidence and effectiveness reviews of over 100 preventative medical interventions, including genetic testing for phenylketonuria (PKU) and Down’s Syndrome.\textsuperscript{122} After conducting these reviews, the Task Force forwards recommendations as to the appropriateness of the interventions for clinical usage.\textsuperscript{123} In addition, AHRQ is participating through its public information dissemination efforts, some of which focus on genetic testing issues.\textsuperscript{124}

Another way to regulate genetic testing is to set standards for the practice of administering the tests, or ensure that whoever delivers the testing to the public has been properly trained to do so.\textsuperscript{125}

The TFGT recommends a comprehensive strategy for improving physician understanding of the clinical value, and patient care implications of genetic testing.\textsuperscript{126} It is generally agreed that testing should be prescribed or recommended by health care service providers with some level of competency in medical genetics.\textsuperscript{127} However, the rate of genetic discovery that lead to the development of new tests and testing technologies is far outpacing the growth of

\textsuperscript{118} Id.
\textsuperscript{119} Id.
\textsuperscript{120} Id.
\textsuperscript{121} SACGT Recommendations, supra note 12, at 11.
\textsuperscript{122} Id.
\textsuperscript{123} Id.
\textsuperscript{124} See Three Factors Should be Considered When Deciding About the Use of Genetic Testing, http://www.ahcpr.gov/research/ocr02/1002RA11.htm#head2 (reporting conclusions of the CDC’s Human Genetic Epidemiology Workshop) (last visited Nov. 5, 2003); see also Wylie Burke et al., Genetic Test Evaluation: Information Needs of Clinicians, Policy Makers and the Public, 156 AM. J. EPIDEMIOLOGY 311, 318 (August 2002).
\textsuperscript{125} TFGT Final Report, supra note 5, at 60.
\textsuperscript{126} Id.
\textsuperscript{127} Koenig et al., supra note 54, at 536; see also TFGT Final Report, supra note 5.
available care providers trained to administer the tests to the public in a controlled and safe environment.  

To address this problem, TFGT recommends an increased "role for non-genetic health care professionals." This role would include, but not necessarily be limited to, eliciting risks of genetic disease in healthy patients through gathering of family history, past history, and offering of genetic testing services. In order for these services to be safely offered by primary care physicians and other non-genetic specialists, they would require knowledge of test validity, disease and mutation rates in various population groups, and myriad other information, including test sensitivity, positive predictive value, clinical utility, etc.

Currently, physician understanding of genetics and genetic testing is in many cases inadequate to allow them to safely act as a gateway to genetic testing services. There are many articles in the literature that document the level of physician understanding and awareness of genetic technologies. An issue that compounds the problem is the perception that many primary care physicians feel the decision is no longer theirs to make, given the increased public awareness of genetic testing and the strong persuasive forces present in the advertising of testing services directly to patients. Armed with this knowledge, many patients present their doctors with information they were not even aware of, putting the physician in the position of interfering with his patient's health care deci-

---

128 TFGT Final Report, supra note 5. The Final Report indicates that the number of entries in a database of unique gene loci grew at nearly ten times the rate of growth for the number of M.D.s and Ph.D.s certified by the American Board of Medical Genetics. Id.

129 Id.

130 Id.

131 Id. at 62.

132 See TFGT Final Report, supra note 5.

133 Id. (citing studies that show that provider competence in genetics was significantly correlated to year of graduation from medical school, having coursework in genetics, and being exposed to genetic problems in his or her practices).

134 See Huang, supra note 25, at 584 n.205 (citing K.J. Hoffman et al., Physicians’ Knowledge of Genetics and Genetic Tests, 68 ACAD. MED. 625 (1993)); see also id. at n.203 (citing AMA Survey: Americans Trust Physicians on Genetic Testing Information, U.S. Newswire, Mar. 31, 1996, available in Westlaw, Allnews Database ("Eighty-one percent of Americans say they're confident their primary care physician could tell them if they are at risk for developing an inherited disease; seventy-two percent believe their family practitioner could interpret the results of genetic tests.

135 Id. at 584.
sions. Given these challenges and the rapid development of novel technologies in the area, continuing medical education programs should become a priority for primary care physicians and genetic health care specialists alike, if for no other reason than to ensure that they keep up with the average knowledge of the patients they are treating. Physicians should be encouraged to participate in these programs by their hospital affiliates, insurance payors, professional organizations, and their patients.

Given that continuing education can also play an important role in maximizing test safety and efficacy, it should be pointed out that there are many potential sources for this type of education, including the testing industry itself, professional provider organizations, patients who may be self-educated and share their knowledge with practitioners, and the ARHQ. Another valuable resource for physician education is the CDC website. This site hosts HuGEnet, an acronym for Human Genetic Epidemiology network. As part of the network, care providers, researchers, legislators and patients are provided with several very important educational resources, including HuGE Fact sheets, technical review articles, and the Genome and Disease Prevention Database Tool. This tool includes extensive sources compiled in an easy to access database that is searchable by disease, gene, topics and other criteria. As with any large database, filtration and sorting of data can be difficult, particularly for those not trained in the field to know exactly what they are looking for.

136 Huang, supra note 25, at 384 (citing, e.g., Scott Gottlieb, Genetic Testing Outpaces Treatment, Times Union, Mar. 12, 1998, at A11 ("The problem is that doctors—entrusted with making such judgments have not stepped up to their anointed role. Faced with consumers increasingly aware of the latest discoveries in genetics, doctors are relenting to demands that they...order prescriptions for the test du jour.").

137 SACGT Recommendations, supra note 12, at vii; TFGT Final Report, supra note 5.

138 TFGT Final Report, supra note 5, at 10.

139 See id.


145 See id.
Another online educational resource is the Genetest—Geneclinics website, which offers detailed information about testing services from availability and contact information to specific sample types required by each facility and information on methods used.

One other issue deserves consideration here. As noted above, the pace of testing technology development and public awareness has translated into demand for testing services that outpaces the number of newly trained care providers who are qualified to administer this sort of care. This has led for some to call for investigation into alternative delivery frameworks. As an initial matter, the same educational resources listed above for physicians could just as easily be used for nurses, public health providers and social workers. To facilitate the understanding and beneficial application of such information, the Task Force recommends increased genetics training in schools of nursing, public health, and social work. In their Final Report, the TFGT has suggested that nurses and community and public health models could play a valuable role in the delivery of genetic testing services and the counseling that is so important before and following testing. We will discuss the role of nursing in more detail in the next section. While the Task Force recognizes that community screening has worked successfully in the past with Tay-Sachs screening, they recommend that additional training for nurses, public health personnel and social workers is needed before large-scale, community-based genetic testing services will be a viable alternative to the traditional primary care provider.

III. PATIENT PHYSICIAN INTERACTION

Within the traditional primary care physician delivery system, the typical genetic testing recommendation will likely encompass...
the taking of a family history, patient education, counseling, and informed consent procedures.

Regardless of who is acting as test gatekeeper, the taking of an effective genetic family history is a key starting point. There is not much regulation involved in this area, but the involvement of public health concerns indicate that the Federal Government may have a role to play, at least as far as aiding physicians in their task.152 The CDC has stepped up to the plate here and is considering working on a family history tool to help physicians carry out this function.153

Patient education is another important process. Numerous online resources exist for this purpose, including the Council for Responsible Genetics,154 Genetic Interest Group,155 and the Center for Medical Consumers.156 While these publicly available resources can go a long way toward educating potential recipients of genetic testing, most agree that there is no substitute for quality genetic counseling by a trained counselor or physician.157

In its Final Report, the TFGT also pointed out the need for expansion of informed consent requirements to cover genetic testing.158 As it stands now, informed consent is generally not required to collect of blood samples for analysis, with the exception of collection for purposes of Human Immunodeficiency Virus (HIV) testing.159 Many similarities have been drawn between the genetic testing and HIV testing with regard to informed consent.160 A stronger informed consent doctrine for genetic testing could lead to better counseling, amelioration of fears over the "racialization of genomic information", and allaying fears generally associated with genetic medicine by the public.161 To address this, at least one com-

153 Id.
157 See infra p. 64 (discussion on genetic counseling).
158 TFGT Final Report, supra note 5.
160 Id. at 349-50.
161 See generally id. at 385-88.
mentator has proposed a model statute for pre-testing counseling and informed consent.\textsuperscript{162} As a general matter, this proposal would require

\begin{quote}
\textit{a description of the test and a statement of its purpose; a description of the disease(s) or condition(s) for which a test will be conducted; an explanation of the risks of stigma and discrimination; and assurances that the patient's medical confidentiality will be protected, with any pertinent exceptions specifically stated.}\textsuperscript{163}
\end{quote}

IV.  \textbf{Physician Recommendation, Sample Collection and Sample Transmission}

The next few steps in the testing process would be difficult to regulate. Every genetic test is likely to be different for each patient.\textsuperscript{164} The decision to recommend a particular test requires analysis of a large number of factors, including test accuracy, and clinical utility, as well as factors likely to be unique to every patient such as family history and psychological state of the patient.\textsuperscript{165} Because of the number and variability of all of these factors, developing practice guidelines for genetic testing is not likely to be easy.\textsuperscript{166} That does not mean practice guidelines should not exist,\textsuperscript{167} but it does bring a need for close scrutiny of who should develop them and how they should be enforced.

Professional organizations could play a role here, and would seem to be a natural fit because their physician members have “front-line” knowledge of the intricacies of physician-patient interaction, and generally have at least a baseline level of competence.\textsuperscript{168} The tort system may also play a role since a physician’s duty to his

\begin{footnotesize}
\begin{enumerate}
\item\textsuperscript{162} Id. at 405.
\item\textsuperscript{163} Id. In her excellent article, Professor Cooper also outlines a model statute covering post—test counseling. Id.
\item\textsuperscript{165} See generally id.
\item\textsuperscript{166} Id. Professor Greely lists seven variables in the equation for cancer testing including “the form of inheritance of the genetic condition, the relationship between the gene and the disease, the type of genetic testing to be used, the nature of the disease, the patient’s age at testing, the costs of the tests and the tested patient.” Id. He further suggests that each of these issues has at least seven possible answers, leading to a matrix of over 250,000 possible scenarios. Id. at 172.
\item\textsuperscript{167} Id. at 178. (“Practice guidelines are never a panacea, and practice guidelines for genetic testing face major challenges . . . . They are, however, an important start”).
\item\textsuperscript{168} See generally Marshall B. Kapp, Physicians’ Legal Duties Regarding the Use of Genetic Tests to Proct and Diagnose Alzheimer Disease, 21 J. LEGAL MED. 445 (2000).
\end{enumerate}
\end{footnotesize}
patients with regard to genetic testing is evolving with the development of testing technology, and of course, providers could face legal liability if they inappropriately order a test.

Provider competence is another issue that arises in this context. The TFGT suggests that

Hospitals and managed care organizations, on advice from the relevant medical specialty departments, should require evidence of competence before permitting providers to order predictive genetic tests defined as needing stringent scrutiny or to counsel about them. Periodic, systematic medical record review, with feedback to providers, should also be used to ensure appropriate use of genetic tests.

They outline a three step process for implementing standards for provider competence. The first step is to classify tests that require a showing of provider competence. The second step should define competence and the third step is to enable providers to gain competence through "easily accessible educational modules.".

V. Testing of Samples

The next logical place for regulatory monitoring of the genetic testing process is in the testing itself, and the laboratories that conduct these tests. As one might expect, the Centers for Disease Control play a role in the regulation of medical testing services. The major legislative mandate for this regulatory authority is the Clinical Laboratory Improvements Amendments of 1988. CLIA seeks to assure the proficiency of the laboratories it regulates

---

169 See generally id.


171 TFGT Final Report, supra note 5, at 66.

172 Id.

173 Id.

174 See generally SACGT Recommendations, supra note 12.

through on site inspection and disclosure requirements. Labs seeking accreditation must apply, giving a description of the testing performed, including number, types, and methodologies of the tests. Participating laboratories must also disclose the qualifications of the scientists directing, supervising and performing the laboratory examinations. They must keep the Secretary up to date on changes made, permit inspections, make operational records available, and pledge to treat proficiency testing samples in the same manner as it treats normally collected samples. Generally, laboratories are also required to

“maintain a quality assurance and quality control program adequate and appropriate for the validity and reliability of the laboratory examinations and other procedures of the laboratory and to meet requirements relating to the proper collection, transportation, and storage of specimens and the reporting of results,” to maintain proper records, and to use only personnel meeting such qualifications as the Secretary may establish for the direction, supervision, and performance of examinations and procedures within the laboratory, which qualifications shall take into consideration competency, training, experience, job performance, and education and which qualifications shall, as appropriate, be different on the basis of the type of examinations and procedures being performed by the laboratory and the risks and consequences of erroneous results associated with such examinations and procedures.

The proficiency testing is straightforward. The HFCA sends controlled samples to the laboratory and the laboratory processes the samples as it would a normally collected sample. The results obtained by the laboratory are checked against the known value of the sample and if the laboratory’s test results fall within an acceptable range of similarity, the laboratory is deemed proficient. This sort of testing measures the analytical validity of the laboratory procedure. The specific standards imposed by CLIA regulation vary with the complexity of the testing performed. CLIA Regulation is administered by the CDC, acting in concert with the Health Care

176 § 263a(d)(1)(B)-(D).
179 § 263a(d)(1)(C)-(E); § 263a(d)(2)(B).
180 §263a(f)(1)(A)-(C).
181 Holtzman, supra note 36.
182 SACGT Recommendations, supra note 12, at 9. This complexity is determined by consideration of the following seven factors: the examination and procedures performed and the methodologies employed, the degree of independent judgment involved, the amount of interpretation involved, the difficulty of the calculations involved, the calibration and quality control requirements of the instruments used, the type of training required to oper-
Financing Administration’s Division of Laboratories and Acute Care Services. The SACGT Recommendations recognize the important role that private sector accreditation of laboratories and their staff can play in the oversight of genetic testing. They noted four organizations who help assure the quality of clinical laboratory practices beyond the governmental guidance provided by CLIA, including the College of American Pathologists, the NCCLS (formerly the National Committee on Clinical Laboratory Standards), the American College of Medical Genetics (ACMG), and COLA. They also recognized various other private sector academic societies and patient advocacy groups for their roles. While these groups should be applauded for their civic involvement, many times their zealous advocacy for a certain position can cloud or shorten their views of the big picture. That said, the tension created by their opposition to the administrative inertia of any regulatory system is likely to be beneficial.

The tort system can play a part in ensuring test quality, when the test is marketed as a kit under products liability doctrine or when the test is marketed as a service under negligence theories. Possible claims against test providers include breach of express or implied warranty, negligence, and products liability, which embraces manufacturing defects, design defects, and warning or instructional defects.

With regard to a negligence claim, “if a company fails to use due care in manufacturing, conducting a test, or reporting test results, and this failure causes harm to a test user, then the company may be liable.” This is a straightforward system with few novel

---

183 SACGT Recommendations, supra note 12, at 9.
184 Id. at 12.
185 Id.
187 Id. at 242 (citing William Hawklard, Relationship Between the Warranty of Merchantability and Strict Liability in Tort, 1 HAWKLAND UCC SERIES § 2-314:13 (1999)).
188 Id. (citing generally RESTATEMENT (THIRD) OF TORTS: PROD. LIAB. § 2 (1998)).
189 Id. at 243. As an illustration, Professor Ossario cites the case of Nancy Seiger who, due to misreporting of the results of her BRCA1 mutation test, had a prophylactic oophorectomy.
legal issues and thus does not require any new rule-making to
implement.\textsuperscript{191}

Problems arise however, in the context of products liability,
particularly with respect to design defects.\textsuperscript{192} A number of
jurisdictions have held that medical products are largely exempt from
design defect liability because these products are unavoidably
unsafe.\textsuperscript{193} While at least one commentator has made the
argument that genetic tests are not unavoidably unsafe, at least from a
physical standpoint,\textsuperscript{194} this author feels the suggestion that psychological
and social risks of genetic testing “should be addressed through
instructions, warnings and pre- and post-test counseling” is not a
sufficient protection.\textsuperscript{195} All of the warnings and counseling in
the world cannot protect a patient from these significant concerns, and
even if they could, they would do nothing to compensate a patient
who suffers in this way due to a poorly designed test. This leaves
us with a paradox: consumers cannot be protected from faulty
designs because of their very dangers. As a result, either special ex-
ceptions from the unavoidably unsafe rules must be instituted, or
products liability cannot be counted on to protect consumers.

Turning from tort protections to more direct regulatory mechani-
isms, it should be noted that while every state has in place
mandatory neonatal testing for such diseases as PKU\textsuperscript{196} and
galactosemia, and most states require testing for hypothyroidism,\textsuperscript{197}
the genetic testing licensure regulations in the states are far from uni-
form.\textsuperscript{198} The recently proposed Model Act for Genetic Privacy and

\begin{itemize}
\item An autosomal recessive genetic disease found in 1 in 15,000 live births, PKU is an illustra-
tive example of how many “genetic tests” are not genetic at all. The test for PKU is actu-
ally a test for a protein that affected cells fail to metabolize. It is not a complex procedure
at all, and costs only about $1.25 per test, but the necessity for accurate diagnosis elevates
the level of scrutiny it should receive. See Lynn B. Jordan, John C. Carey, Raymond L-
\item Kathryn E. Cox, et al., Model Act for Genetic Privacy and Control, 88 Iowa L. Rev. 121, 129-30
(2002).
\end{itemize}
Control establishes a regulatory framework for genetic testing that would operate largely through the operation of the State Departments of Health. State Departments of Health are already involved in the regulation of genetic testing through their deemed status for CLIA compliance determinations. Indeed, some states have tougher licensing requirements for lab personnel and quality assurance than those required by CLIA, including in some instances licensure requirements for genetic counselors. The California Health and Safety Code is an excellent example of very progressive state regulation of genetic testing. The California Code requires, among other things, “clinical testing procedures established for use [...] be accurate, provide maximum information” and “produce results that are subject to minimum misinterpretation”, post-test counseling for “all persons determined to be or who believe themselves to be at risk for a hereditary disorder as a result of screening programs”, informed consent for all testing other than the mandatory prenatal testing for PKU, and informed consent in compliance with federal protections of human subjects. The California regulatory scheme establishes a genetic disease unit within the state Department of Health that has a mandate to “promote a statewide program of information, testing and counseling services and shall have the responsibility of designating tests and regulations to be used in executing this program.” These state-sponsored and mandated newborn screening programs generally do not fall under HCFA-approved proficiency testing guidelines. However, that is not to say that they are not regulated. Indeed, states have relied upon the private accreditation of the National

---

199 Supra note 12, at 12.
200 Id. at 124.
201 SACGT Recommendations, supra note 12, at 12.
202 Id. (citing New York and California as among the tougher states).
203 Cox et al., supra note 198, at 126 (citing CAL. HEALTH & SAFETY CODE § 124981 (West Supp. 2002)). MAGPAC cites California as requiring licensure of genetic counselors, which, at present, CLIA does not do. Id. As noted supra, however, the CDC has proposed that CLIA be extended to regulate genetic counseling when used in conjunction with a genetic testing service. Id.
204 See CAL. HEALTH & SAFETY CODE §§ 124975, 124980, 125000 (West Supp. 2002).
205 CAL. HEALTH & SAFETY CODE § 124980 (d) (West Supp. 2002).
206 CAL. HEALTH & SAFETY CODE § 124980 (g) (West Supp. 2002).
207 CAL. HEALTH & SAFETY CODE § 125000 (a) (West Supp. 2002).
208 SACGT Recommendations, supra note 12, at 12.
Newborn Screening Quality Assurance Program\textsuperscript{206} for verifying analytical validity and meeting CLIA quality assurance standards.\textsuperscript{210} This sort of interaction between state regulation, national guidelines, and private sector accreditation is an encouraging example of the ability of the current patchwork system to function with some level of efficiency.

One of the fears with respect to state regulation of genetic testing is that there will be a large disconnect between the limited scientific understanding of the legislature and the rapid pace of technological development.\textsuperscript{211} As a result, some states, again notably California, have set up advisory boards to help bridge this gap.\textsuperscript{212}

\section*{VI. Dissemination of Results}

The last stage in the genetic testing process is dissemination of results. There is wide agreement that patients who receive results from genetic testing also must be offered counseling to help deal with the results of the test.\textsuperscript{213} Indeed, The Stanford Recommendations on Genetic Testing For BRCA1/2 bluntly stated that “genetic counseling is the linchpin of good care.”\textsuperscript{214} Releasing test results to uninformed or unprepared patients can cause serious psychological harm.\textsuperscript{215} Efforts must be made to increase the numbers of licensed and otherwise qualified genetic counselors available to consumers

\textsuperscript{206} Operated by the National Center For Environmental Health’s Division of Laboratory Services, the National Newborn Screening Quality Assurance Program “provides proficiency testing samples for laboratories performing newborn screening tests” and “is an essential resource for the laboratorians performing these tests; provides consultation and even on-site assistance in resolving difficult analytical problems.” Save Babies Through Screening— A Parent’s Resource On Newborn Screening, PHL Newborn Screening Fact Sheet: The Role of Laboratories in Newborn Screening at http://www.savebabies.org/ library/APHL\%20\%20NBS\%20Fact\%20Sheet.pdf (last visited Nov. 6, 2003).

\textsuperscript{210} SACGT Recommendations, supra note 12, at 12.

\textsuperscript{211} See Symposium, Genetic Testing and Individual Rights, 27 SUFFOLK U.L. REV. 1477, 1495-96 (Winter 1993). Dr. Mullinsky ridicules the ignorance of state legislators who don’t seek outside help with the science. \textit{Id.} He singled out the Georgia legislative House who “enacted statutes that required immunization against sickle cell.” \textit{Id.} at 1496.

\textsuperscript{212} CAL. HEALTH & SAFETY CODE § 125000(a) (West Supp. 2002).

\textsuperscript{213} See TEDT Final Report, supra note 5; Cooper, supra note 159, at 350; Nunnally, supra note 108, at 345.

\textsuperscript{214} Koenig, et al., supra note 54, at 538.

\textsuperscript{215} Nunnally, supra note 108, at 345.
and to make their presence known so that physicians can give ready referrals. 216

Another problem with state regulation (or any regulation at all) is the limited ability of smaller state budgets to deal appropriately with the implications of expanding genetic testing technologies. 217 For instance, suppose a state were to expand its mandatory neonatal testing regime to include twenty disorders. What public benefit would come of this testing if many of those found to be at risk could not be treated with state funds? If patients have knowledge that they are at high risk for a disease, but are unable to pay for treatment themselves and unable to obtain state health coverage for treatment, then testing would arguably be a grave disservice to the public and would raise serious ethical questions. Thus commentators have called for genetic testing to include “multidisciplinary follow up care.” 218

VII. Conclusion

This essay only touches on the broad range of issues facing those who would increase regulatory oversight of the genetic testing industry. While the work done thus far by various legal scholars, the Task Force On Genetic Testing and The Secretary’s Advisory Committee on Genetic Testing has been and continues to be extraordinarily valuable, 219 it is not complete. Implementation of the ideas thus far developed and further study of others is needed if we are to reap the rewards of all of their hard work.

Additional issues still must be addressed. This paper did not address privacy, ownership and control of genetic information, and regulation of the health insurance market for genetic testing, but nonetheless, these issues remain of paramount importance. While Health and Human Services Spokesman William Pierce has expressed HHS’s intention to form a new committee to deal with “a broader range of genetic technologies,” the department has yet to

216 See generally SACGT Recommendations, supra note 12.
217 Koenig, et al., supra note 54, at 542.
218 Id. at 539.
219 Ellen Wright Clayton, Review Essay: Genetic Testing Is Different, http://www.ahcpr.gov/clinic/fppl/clayton.htm (last visited Nov. 6, 2003). “The far-reaching process, as described in Post and Whitehouse, the Task Force, and the SACGT documents, that is evolving to govern the use of genetic testing should become a blueprint for evaluating the full array of medical practice.” Id.
name this committee or even publicize a mandate for it. This new committee could hold great promise, if given a broad enough mandate to work with, and if the proper cross-section of stakeholders is included.
