Center for American Progress

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Policy Considerations in the Age of Genetic Medicine

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April 2008

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Introduction and Summary

The world stands on the brink of a genome-based personalized-medicine revolution, with individual Americans poised to be the greatest beneficiaries. An international research consortium that includes our country's National Human Genome Research Institute recently announced its $50 million plan to sequence the genomes of at least 1,000 individuals from around the world. According to NHGHRI Director Francis Collins, this project will increase the sensitivity of disease discovery efforts across the human genome five-fold, and within gene regions (the portions of a chromosome on which a particular gene is located) at least 10-fold.[1]

What's more, Harvard University geneticist George Church has also embarked on the even more ambitious "Personal Genome Project." He aims to sequence 100,000 genomes at a cost of about $1 billion, and possibly expand the project until it reaches 1 million sequenced genomes.[2]

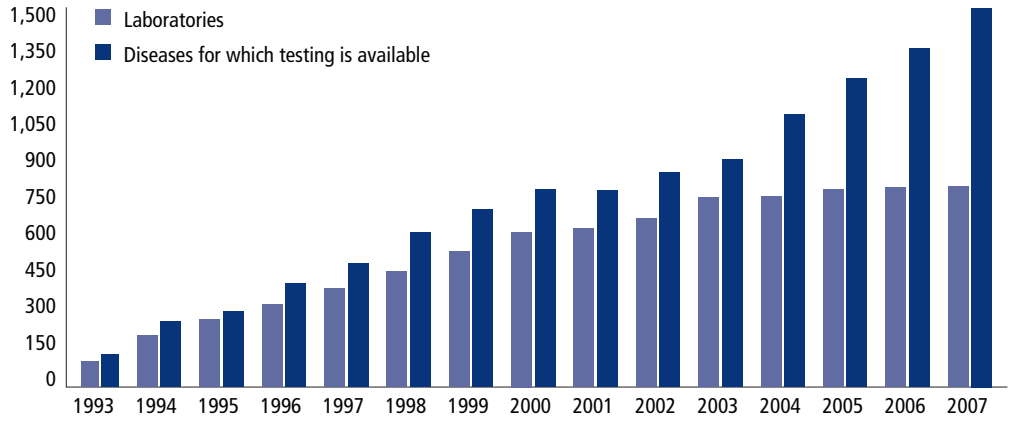
For individual Americans, this growth of genomic data means more accurate personal genetic information will become available to them, their physicians and, yes, their insurance companies, to make perhaps monumental health care decisions. This new information could well be a blessing or a curse, depending on how it is handled by patients, their doctors and their insurers. That's why legislation based on the best bioethical principles needs to be enacted by Congress this year. Genetic testing will become increasingly more accurate as more research is done.

But more importantly, in the meantime many genetic tests remain highly inaccurate. The results of genetic testing today are hardly reliable, yet they are already part of the medical marketplace. Congress's failure to enact federal genetic testing laws is one reason for this problem. A U.S. Department of Health and Human Services panel estimates that over the past decade over 1,100 genetic tests have become available. The website GeneTests reports that there are currently 1,546 diseases for which testing are available (see graph on page 2). However, some of these are only research tests; the number of clinical tests that are available is 1,258.[3] But until recently most tests were for very rare conditions. That percentage is beginning to rise, however, as genetic variants are being identified that increase risk for common complex diseases such as diabetes, cardiovascular disease and macular degeneration.

This sudden deluge of diagnostic information may cause great uncertainty, fear, and misunderstanding for average patients who, even now, do not know how to interpret and make use of their genetic information. Physicians also find themselves somewhat

GENE TESTING TAKES OFF

The number of gene testing laboratories and the number of diseases they test for are both on the rise



Source: GeneTests: Medical Genetics Information Resource (database online). Copyright, University of Washington, Seattle. 1993–2008. Available at http://www.genetests.org/servlet/access?id=8888891&key=RHSDvFjKqXHCP&fcn=y&fw=IJ5O&filename=/whatsnew/labdirgrowth.html. Accessed March 6, 2008."

perplexed as they now have to integrate genetic information into their evaluations and diagnoses of patients. This uncertainty arises because some genetic tests provide patients, physicians, and insurers with mathematical probabilities of disease that are seemingly much more precise than simple knowledge of family medical history.

Moreover, the preventative measures that may lead from genetic diagnoses will present new challenges to both patients and healthcare providers. Yet patients and healthcare providers will need to remain mindful of the fact that genes are *not* destiny; they will need to carefully weigh other clinical observations before taking drastic measures such as removing a body part that has a probability of one day becoming cancerous.

All these new medical dilemmas, however, are predicated on the assumption that patients will feel comfortable and safe having testing done and sharing it with their healthcare providers. The main reason patients would want to keep their genetic information private—or not even have the test in the first place—is fear of being discriminated against by employers or insurance companies. According to a 2007 survey by the Genetics and Public Policy Center, 93 percent of respondents thought that employers should not have access to their genetic test results, and the same percentage thought insurers should not have access.[4]

This fear of discrimination could lead to an under-utilization of genetic tests by patients, which would ultimately hurt employers and insurers, too, since patients will not be able to take preventative measures that could eliminate the need for expensive care, medical leaves, and sick days down the road. Although current state and federal laws do provide patients with some protection from genetic discrimination, they still remain inadequate and vague.

For instance, as of 1996, the Health Insurance Portability and Accountability Act, or HIPAA, prohibited group health insurers from using health-related information in making coverage decisions or setting premiums for an individual who is part of a group plan; and the Act specifically lists genetic information as such. Additionally, HIPAA states that genetic information in the absence of a diagnosis cannot be considered a preexisting condition.[5]

There are also some provisions in the Social Security Act, the Americans with Disabilities Act, and the Civil Rights Act that could apply to genetic information but do not mention genetic information explicitly. A patchwork of state laws regarding privacy, employment, and insurance practices also exist, with many state laws explicitly mentioning genetic information but all of them offering different levels and forms of protection.[6]

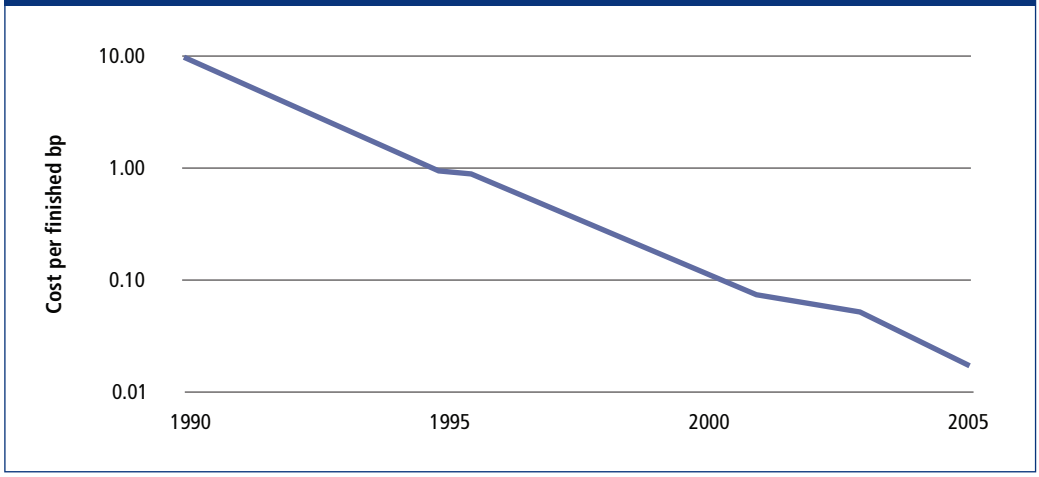
Federal policies that could be put in place by the proposed Genetic Information Nondiscrimination Act, or GINA (S. 358), now before Congress would further prevent discrimination by extending protections to holders of individual insurance plans, and would protect whole groups of insured people from being discriminated against based on the genetic information of some of the individuals in that group. The GINA legislation would also create a policy that protects individuals from discrimination by employers.

This legislation has broad support from legislators on both sides of the political aisle, but has yet to be enacted into law. Versions of GINA passed in the Senate in 2003 and 2005, but a version did not pass in the House until 2007. Now the Senate has yet to pass the 2007 version. This is because a group of Senators have placed a "hold" on the legislation. The possible objections that these Senators might have to the bill are most likely in line with the objections raised by groups such as the U.S. Chamber of Commerce, which feels that the existing state and federal protections are sufficient and that federal legislation would open insurance companies up to unnecessary lawsuits. Another objection might be similar to the one expressed in the Bush administration's statement that there is not a significant "firewall" between Title I of the bill, which deals with insurance, and Title II, which deals with employers.

Interestingly, the text of GINA was attached to the Paul Wellstone Mental Health and Addiction Equity Act (H.R.1424), which passed the House last month by a wide 268-to-148 margin. But the Senate version of the Mental Health bill (S. 558), which passed in September, does not contain the text of GINA. Unfortunately, the GINA language does not have a good chance of being included in the final version of the Mental Health bill that would emerge from a House-Senate conference committee.[7]

In fact, the Mental Health bill does not even have a good chance of going to conference to begin with because many Senators feel that the House version of the bill is too broad and would ruin the careful compromise worked out on the Senate version. These Senators include Sen. Edward Kennedy (D-MA) whose son, Rep. Patrick Kennedy (D-RI) authored the House version of the Mental Health bill. Therefore, the best option for getting GINA passed would be for Senate Majority Leader Harry Reid (D-NV) to override the hold by securing the 60 votes needed so that the stand-alone Senate version of the GINA bill (S.358) can pass.

GENE TESTING COSTS PLUMMET
Decrease in the cost of finished DNA sequencing



From Figure w/ caption "Free fall." Credit: Adapted from graph provided by Jeffrey Schloss/NHGRI from Service, SCIENCE 311: 1544 (2006). Courtesy: National Human Genom Research Institute. Please contact NHGRI for permission as well. Reprinted with permission from AAAS.

Opponents of GINA fear it will make employers vulnerable to frivolous lawsuits based on allegations of genetic discrimination, and that the new legislation will not be able keep up with further fast-moving scientific developments in the field of genetic testing that naturally outpace legislative action. Yet the influence of further scientific developments on possible genetic discrimination relies on the accuracy and significance of new genetic tests as they arise. Fear and uncertainty about the quality of genetic testing is bound to become more and more acute, especially as the cost of genetic sequencing comes down and low regulatory barriers to market entry encourages more new genetic-testing companies to jump into the marketplace (see graph above).

In fact, the genetic-test industry is growing at a breakneck pace. New entrant 23andMe, Inc. offers to decode 500,000 points on a customer's genome for $1,000. DNADirect, Inc., offers 17 condition-specific tests of varying cost directly to the consumer. Knome, Inc. will sequence a customer's entire genome for $350,000. And Sciona, Inc., sells "nutrigenetic tests" that claim to help consumers with diet and nutrition recommendations based on their genes. The proliferation of these new tests means that consumers will have direct access to much of their genetic information without their personal physician acting as a gatekeeper to review, interpret, or contextualize that information for them.

Once again, there are currently only a patchwork of state and federal regulations regarding the quality and parameters of genetic testing. These rules were enacted in response to the condition-specific genetic tests that have entered the market over the last decade. Tests are overseen by a variety of regulators. The Food and Drug Administration regulates the testing kits. The Federal Trade Commission regulates the labeling and advertising of genetic test kits. And the Centers for Medicare and Medicaid Services

regulates labs under the authority of the Clinical Laboratory Improvement Amendments Act of 1988.

Falling through the cracks in this system, however, are labs that develop their own genetic tests and offer testing as a service. In 2000, the Centers for Medicare and Medicaid Services announced that it would create a new set of protocols in order to better regulate genetic testing. CMS then did nothing for six years until it announced in April 2006 that the Department of Health and Human Services put the creation of new genetic testing rules on its regulatory agenda with the aim of having new regulations in place by November of that year—only to renege on that promise in July of 2006.

It was not until November 5, 2007 that the HHS Secretary's Advisory Committee on Genetics, Health, and Society released a draft of their report on the oversight of genetic testing. The SACGHS report was left open for comments until December 21, 2007 and will be released by the end of April 2008. One of the major recommendations that the SACGHS report will include is the creation of a registry for genetic tests. This registry could be housed at a federal agency or possibly at GeneTests, a National Institutes of Health-funded research center at the University of Washington-Seattle. Senate bill 736 also includes a similar provision.

Underpinning all of the concerns regarding the proper regulation of genetic tests and genetic privacy are key bioethical principles. For genetic testing, these principles protect the right of individuals to keep their genetic information private, underline the obligation of testing service-providers and physicians to actively prevent harm from befalling the patient through faulty tests or unsound recommendations based on tests, and ensure policies are in place so that individuals will not be discriminated against in employment or insurance. (See sidebar on page 10 for a detailed examination of the role of bioethics in genetic testing.)

GINA addresses many of these bioethical principles by specifying privacy protections and bans on discrimination. Our society needs to accommodate the rush of genetic information that is just over the horizon in a way that conforms to our firmly held moral values. If we fail to anticipate and plan for the implications of these developments, then the benefits will be denied to millions of Americans. GINA provides a coherent and timely public policy framework for the genetic medicine of the 21st century.

Presuming Congress passes this legislation—still a big "if" depending on whether the Senate is too busy to generate enough votes to override the hold—then regulators will be armed with many of the legal tools they need to address these issues. The next steps to take will be for members and regulators alike to delve into the complexities and the business hype of genetic tests. This paper addresses both of these issues in turn.

The Need for GINA

Cases of Genetic Discrimination Exist

Perhaps the easiest argument to dismiss against GINA is the suggestion that new policies to protect people from genetic discrimination are not needed. Trade groups representing employers and insurance companies contend that they do not use genetic information in making decisions, yet two high-profile cases and anecdotal evidence suggest otherwise.

In 1995, the Lawrence Berkeley National Laboratory was sued by employees who claimed that they had been subjected to medical screening without their consent. They alleged that among the tests performed was a genetic test for the sickle cell genetic trait. It is important to note that the genetic marker used at that time had an uncertain link to this condition. Testing for the sickle cell trait—even if efficacious—would indicate only carrier status for sickle cell disease.

In a 1998 ruling, the U.S. Court of Appeals for the Ninth Circuit sided with the workers and agreed that the practice violated the Civil Rights Act of 1964 and their right to privacy. The case was sent back to a lower court, where the employees settled with the laboratory in 2000.

Then, in 2001, the Equal Employment Opportunity Commission brought suit against the Burlington Northern Santa Fe Railway Corporation, alleging that the company was conducting unauthorized genetic tests on some of its employees. Those employees had filed workers compensation claims for carpal tunnel syndrome, and the company claimed that it was seeking to determine the cause of their condition. In this case the company agreed to pay the employees $2.2 million in an out-of-court settlement, and the courts therefore never ruled on the EEOC's application of the Americans with Disabilities Act to situations where employers violate employees' privacy by requiring genetic tests.

There are also many instances of genetic discrimination that never make it to the courtroom. At a 2004 meeting of the HHS Secretary's Advisory Committee on Genetics, Health and Society, for example, individuals testified about experiences in which they alleged that employers and health insurance companies used their genetic information in making decisions about them. These experiences included having to pay higher premiums and even not being able to obtain a health insurance policy in the first place because of the results of a genetic test.[8]

Cases of Genetic Discrimination

Lawrence Berkeley Laboratory (1999)

- Accused of conducting pre-employment screening for sensitive medical information, testing for genetic traits such as sickle cell trait, and for non-genetic factors such as syphilis and pregnancy
- Charges filed under: Title VII of the Civil Rights Act of 1964, and right to privacy as guaranteed by the U.S. and California Constitutions (also the Americans with Disabilities Act, but this was not affirmed by the courts)
- Company argument: sought to have case dismissed in summary judgment without a trial, claiming that the statute of limitations had run out
- Ruling: The U.S. Court of Appeals for the Ninth Circuit sided with the workers

Burlington Northern Santa Fe Railway Corporation (2002)

- Employees charged that those who had filed for workers compensation for carpel tunnel syndrome—a painful hand and wrist condition caused by repetitive motion—were tested for a genetic marker
  - Tests performed without their knowledge
  - Marker dubiously associated with carpel tunnel syndrome
- Charges filed under: Americans with Disabilities Act of 1990 by the Equal Employment Opportunity Commission
- Company argument: testing necessary to determine cause of injury for 36 employees who claimed to have job-related carpel tunnel syndrome
  - 20 employees were tested before program voluntarily suspended
- Settlement: Company agreed to halt testing and pay $2.2 million

Americans Are Fearful of Genetic Discrimination

The argument that genetic nondiscrimination legislation is not needed because cases of discrimination do not exist—aside from ignoring specific cases like the ones mentioned above—also discounts the danger that the fear of genetic discrimination poses. In the clinical setting, doctors report that patients are hesitant to be tested for genetic predispositions to develop disease based on the possibility that the results of such tests could end up in the hands of employers and insurance companies.

This means that patients are foregoing tests that could specify the elevated risk factors that they carry. As a result, they are not obtaining information that could inform their or their children's lifestyle and health care decisions and prevent disease. One key reason patients are doing this is the now standard practice among physicians and genetic counselors to warn patients of the ways in which test results could be used outside of the doctor's office and advise them of the safeguards that do or do not exist in their state.

A related problem occurs when people opt to pay for tests out-of-pocket in order to guarantee their confidentiality, and even choose not to share test results with their doctor in order to keep information out of their medical record. A February 2008 article in *The New York Times* reported on the lengths some patients will go in order to keep genetic information from insurance companies and doctors, sometimes putting their health at risk in the process.[9]

Similarly, laboratory researchers are concerned that people are opting not to participate in studies that involve genetic tests that could uncover correlations between genes and diseases. These potential subjects are concerned that

results from research tests could adversely affect them if the results somehow end up in the hands of third parties such as insurance companies or employers. Yet sometimes patients will ask doctors to put them in studies so that they can receive genetic tests that will not go on their medical record and probably be seen by insurance companies or employers.

In order to receive the highest standard of care, patients need to be able to undergo genetic tests with the assurance that the results will not be abused. They also need to be assured that they can share the results of such tests with their doctor without worrying about jeopardizing the confidentiality of the results and facing discrimination. And researchers need to be able to tell their subjects that their genetic information is private so that research can continue to help find new methods of disease identification and treatment.

Research conducted by the Genetics and Public Policy Center of Johns Hopkins University has focused on this issue, examining the extent to which Americans fear genetic discrimination. Their research has also examined whether there is public support for new legislation to prohibit discriminatory practices in the workplace and by insurers. In a 2007 survey, 93 percent of respondents expressed concern that the results of a genetic test indicating increased risk for a disease could be used in ways that were harmful to them. They found that less than one quarter of individuals would trust their employers or health insurers to have access to genetic information. More than three quarters support a law to prohibit them from using this information to discriminate.[10]

Current Federal Laws Do Not Adequately Protect Employees

The only federal policy that directly pertains to genetic discrimination by an employer is an executive order signed by President Clinton in February of 2000.[11] As a statement of federal policy it has a limited scope, protecting only those who work for the federal government. Executive Order 13145 prohibits departments and agencies within the executive branch from using "protected genetic informa-

The Fear of Genetic Discrimination

The Genetics and Public Policy Center, in its* U.S. Public Opinion on Uses of Genetic Information and Genetic Discrimination *(2007), conducted an online survey of 1,199 random adults in the United States about this issue. The results:

- 93 percent express concern that results of a genetic test indicating increased risk for a disease could be used in ways that are harmful to them
- 19 percent and 15 percent support use of genetic testing by employers (hiring and promotion) and health insurance companies (for qualification and pricing), respectively
- Three out of four Americans support a law forbidding genetic discrimination by health insurers and employers

tion" to discriminate against an applicant or employee. This policy is a move in the right direction, but protection from these practices is fundamental and needs to be expanded to workers in the private sector.

In 2005, the HHS Secretary's Advisory Committee on Genetics, Health, and Society commissioned a report examining the legal protections against genetic discrimination already in place. According to the report, there are two laws that apply to private sector employers and may protect their employees from genetic discrimination. Neither of these laws, however, makes specific reference to the use of genetic information as a basis of discriminatory practices, and neither has been tested by a court ruling.

The Americans with Disabilities Act, passed in 1990, protects individuals with a physical or metal impairment from discrimination. Since the conditions that qualify as falling under the purview of ADA are decided on a case-by-case basis, it has been argued that it protects those with a genetic predisposition to disease. The language of the statute sets up protection for three groups of people: those with a physical or mental impairment; those with a record of such impairment; or those who are regarded as having such impairment. This language makes it possible that ADA applies to genetic information under "prong one," where a genetic trait is considered a physical impairment, or "prong three," where those with a genetic trait are considered to be regarded by others as impaired.[12]

This law, however, applies only to employers meeting certain size criteria, and importantly, does not prevent those employers from collecting sensitive information in the first place. Moreover, this interpretation also remains untested. The only case to apply the ADA to an alleged instance of genetic discrimination—brought by the EEOC on behalf of Burlington Northern Santa Fe Railway Corporation workers—was settled out of court.

The Civil Rights Act of 1964 sets up another set of protections with a very limited scope. Title VII of the law specifically prohibits employment discrimination on the basis of race, color, religion, sex, or national origin. According to the SACGHS report, in those circumstances where genetic discrimination singles out one of these protected groups it could represent a violation of the Civil Rights Act.[13]

This leaves employers free to require genetic tests as long as the performance of the test and its results do not single out one of these groups. For example, a test would be allowed if it was administered to all employees and the results of the test did not segregate with any particular race, color, religion, sex, or national origin. Considering that most genetic disorders do not disproportionately affect a protected class, this prohibition does not offer significant protection.

Current Federal Laws Provide Inadequate Health Insurance Protection

A prominent argument against the passage of new laws prohibiting genetic discrimination by health insurance companies is that existing laws already protect consumers from such practices. Indeed there are two laws that apply to this industry, either by design or by extrapolation, but an examination of each reveals that they offer incomplete protections and leave much to be desired.

According to the SACGHS report, the Health Insurance Portability and Accountability Act of 1996 specifically

prohibits group health insurance plans from using genetic information to discriminate in certain ways.[14] Group plans, for example, cannot use genetic information to impose a preexisting-condition exclusion without an actual diagnosis, or establish eligibility requirements for an individual. Additionally, group plans cannot refuse to renew a policy based on genetic information.

Yet even though group insurers cannot use genetic information for the above practices, they are free to collect and retain that information and may even require that an individual submit to a genetic test as a condition of coverage. Furthermore, even in instances where genetic information cannot be used as a basis for denying coverage to or raising the premium of an individual, it can be used to justify raising the premium of an entire group. Finally, these limited protections of people in the group market have no bearing on the individual market, where insurers are not restricted by HIPAA.

The Americans with Disabilities Act also may protect individuals in cases of health insurance discrimination. Some U S. Court of Appeal Circuits have ruled that Title III of ADA, which guarantees equal access to places of public accommodation, applies to insurance policies.[15] This application is currently tenuous, however, as only two of seven circuits have expressed this interpretation. Even if insurance policies do qualify as places of public accommodation, it is still not clear that the law would apply to instances of genetic discrimination.

The States Were the Starting Point of Reform, but Regulations Still Remain Inadequate

The inadequacy of current federal rules governing genetic discrimination has in some ways been mitigated at the state level. This has certainly been the case with regard to genetic nondiscrimination policy. Beginning in the early 1990s,

Bioethical Principles

How genetic testing should be governed

Genetic testing carries with it some serious implications for personalized medicine. A quick analysis of risks and benefits of genetic testing allows us to consider the bioethical principles of **beneficence** and **non-maleficence**, or the medical injunction to both "do good" and "do no harm" to patients.

Perhaps the most disturbing feature of genetic discrimination is the asymptomatic nature of genetic information. Just because a person has a certain genotype (a set of genes for a characteristic), does not mean that they are manifesting the phenotype (the actual characteristic) associated with it. There is a difference between possessing a gene and expressing a gene. Thus, not only is genetic discrimination unjust because a gene is not something that a person acquires by choice, but often time this sort of discrimination is also often based on a faulty understanding of the way genetics works.

Moreover, genetic traits can be seen as more psychosocially stigmatizing than other medical conditions because people often identify genes as the foundation of their personal identity. Therefore, genetic tests could very well be considered harmful by default unless the tests lead to some sort of treatment that can improve the well-being of the patient.

Genetic information may tell a patient that they have a disease, a predisposing condition, or carrier status. The practical steps that could follow from acquiring such information might be a change in lifestyle, preventative medicine, removal of an organ or tissue, or certain decisions regarding reproduction. Yet in some cases the most practical step might be to do nothing or to simply wait and see without any definite prospect of benefit.

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Bioethical Principles *(continued)*

All of these options could create unnecessary feelings of uncertainty or anxiety in patients unless they are properly counseled, and the last option offers no benefit to offset the psychosocial consequences. This makes the ethics surrounding genetic testing particularly troublesome unless certain measures are taken to assuage psychosocial anxiety. These measures include better education of patients and physicians, and public policies that protect individuals and families from discrimination, faulty tests, and unreliable information.

What's worse, direct-to-consumer genetic tests might prove to be risky not only because DTC labs are not as reliable as doctor-administered or doctor-ordered tests, but also because patients might not even share the results with their physicians. Usually, the qualifications of a lab are certified by the Centers for Medicare and Medicaid Studies, or CMS, under the Clinical Laboratory Improvement Amendments, or CLIA. CLIA also assesses the analytical validity of 83 tests—none of which are genetic tests—where it demands proficiency testing. A private lab, however, does not need to be certified under CLIA if it defines itself as a research lab and not as a clinical lab. DNADirect and Navigenics, Inc. are two examples of private DTC labs that define themselves as clinical labs and are certified under CLIA because they return results to customers. 23andMe returns results to customers as well, but does not define itself as a clinical lab and is not certified under CLIA.

Even labs that do have CLIA certification are not required to be transparent about the accuracy of any of their tests unless those tests are one of the 83 tests specified in CLIA. However, even the data that CMS collects on the analytical validity of the 83 lab tests are difficult to obtain. Moreover, CMS is largely unable to interpret the data that they acquire.

Without making the data on the 83 lab tests publicly available, it is very difficult to hold labs accountable for these tests. Needless to say, this holds true for genetic tests, too, since they are not even included in the 83. When data about the margins of error for a test or the ratio of false positives to false negatives is neither obtained nor made available, patients are put at risk. For instance, a patient could obtain a direct-to-consumer genetic test that incorrectly comes up negative, and as a result would see no need to tell his physician about it. The patient might fail to seek out preventative screening due to a false sense of security and wait until it is too late.

Moreover, the Food and Drug Administration only regulates pre-packaged genetic testing "kits," which are manufactured and sold to labs. It does not regulate self-prepared "home brews," which are what most labs use.

A lack of reliability also arises when one company has a patent on the most reliable and medically useful form of a test, which means that any other company's test would most likely take a less reliable form. An example of this is breast cancer testing, which physicians consider most reliable when the entire sequence of a gene is analyzed. The only company that has a patent on this form of the test is Myriad Genetics, Inc. Most physicians would consider less thorough tests to be inconclusive and effectively useless.

This is because most of the other tests that are being marketed look only at single nucleotide polymorphisms, and do not sequence the entire gene. Moreover, physicians cannot be expected to read the results from every new form of genetic test that arises from every new company that sprouts up. This of course assumes that a physician actually gets to see and analyze the results and that the patient does not take the company's analysis at face value.

But assuming that the results of a genetic test are reliable and overseen by the patient's physician, they could prove to have considerable benefits for the patients. These benefits have, in fact, already been demonstrated in the medical context with genetic tests ordered by a physician. In some instances, such as the case of a predisposition to colon cancer, patients have been observed to be more likely to receive regular colonoscopies if they test positive for the gene or receive no test at all. In contrast, some patients have been known to adopt a fatalistic mindset when they find out that they test positive for a predisposing gene.

In reality, almost any definite conclusions from a genetic test are unwarranted. If a patient finds out that she has a gene that is known to be correlated with a certain disease, all she and her doctor in effect know is that there is higher numerical probability that she will develop that disease by a certain age than someone without the gene. What they do not know is the severity, exact age of onset, or the manner in which the symptoms will appear based on a simple numerical probability.

Moreover, these numerical probabilities are based on studies of large populations. Physicians cannot easily deduce an individual's unique susceptibility to a disease unless other factors are taken into account such as environment, lifestyle, diet, and family history. Genetic tests are usually most helpful when they are used by physicians in a diagnostic context. These kinds of tests occur when a physician suspects that a patient has a certain disease based on other factors and then tests the patient's genes in order to confirm the diagnosis and then recommend testing for the patient's family members.

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Bioethical Principles *(continued)*

Some might argue, however, that an individual has a right to know what is in their genes, regardless of whether a physician or genetic counselor oversees the test. This could be considered beneficial for individuals who are not in a particularly high-risk category and are just curious. Although this is an interesting theoretical consideration, it leaves consumers open to exploitation by tests with minimal gene-to-disease correlation data backing them up and which have not been proven to be reliable or useful.

This in turn leads to specious and often over-generalized claims such as those made by companies providing "nutrigenetic tests," which purport to provide nutritional advice based on genetic information, and which have recently been subjected to a scathing study by the Government Accountability Office. This is the problem with the application of the bioethical principle of **autonomy** to genetic tests. Patients certainly have a right to information about their bodies, health, and well-being; however, without proper discretion, that information could be useless at best or harmful at worst. Although true patient autonomy can only be exercised when the patient actually does have reliable information, DTC genetic testing companies can still co-opt the language of autonomy and "the right to know" to promote their tests while they downplay the issue of test reliability.

Finally, there is the principle of **justice**. As things currently stand, direct-to-consumer genetic tests are expensive, which limits access. And if testing companies do not offer counseling and patients take the results to their physicians, those physicians could become overburdened with the difficult and time-consuming task of interpreting these new genetic tests for their patients. This puts an extra strain on resources, especially when many physicians are not adequately trained in the specialty of medical genetics.

The good news, however, is that other specialists, such as oncologists and even some neurologists, have begun to incorporate the analysis of well-validated genetic tests into their clinical practice. Currently, the only federal regulation that keeps direct-to-consumer genetic testing companies honest about the claims they make is Section 5 of the FTC Act, which prevents deceptive commercial practices. This has led some genetic testing companies to admit in their fine print that "we cannot and do not diagnose diseases or medical conditions, provide medical advice or otherwise assess your health." Yet they still manage to make an authoritative impression on any consumer interested in pursuing biological fortunetelling.

For instance, in the fall of 2007, Myriad Genetics incited controversy with its advertising campaign for its genetic test for the breast cancer genes known as BRCA1 and BRCA2, both of which indicate a predisposition to breast and ovarian cancer. Although it is considered the most reliable and comprehensive genetic test for breast cancer—it sequences the entire gene—and can only be ordered for a patient by a physician, it is very expensive ($3,120).

There are also less expensive tests that simply test for the most common mutations on the gene and do not require a physician's order. DNADirect is one such company that does this; it also employs healthcare providers to assist and counsel to their customers. Nevertheless, this is still not the same as a doctor-patient relationship. Moreover, without a public database of genetic tests, there is no way for patients to objectively assess the claims of private testing companies and the healthcare providers who work for them.

Women with a mutation in the BRCA 1 or BRCA 2 gene have a 36 percent to 85 percent probability of developing breast cancer and a 16 to 60 percent probability of developing ovarian cancer depending on the population. Yet only 5 percent to 10 percent of women with breast or ovarian cancer have a hereditary form of cancer due to genetic mutations. In the general population, only 1 in 500 to 1 in 1000 individuals has BRCA 1 mutations. The prevalence of BRCA 2 mutation is unknown.

The upshot: Those with a family history of breast cancer may understandably grasp at any data that might inform their medical decision making, but these data on the very limited predictive power of breast cancer testing demonstrate how easy it is to be misinformed, attributing more significance to a genetic test than is warranted.

These caveats exist not only for breast and ovarian cancer, but for a whole host of diseases and conditions with genetic components. Furthermore, anticipation and awareness of these caveats, risks, and adverse consequences arises from a commitment to the universal bioethical principles of beneficence, non-maleficence, autonomy, and justice. Therefore, it is useful and prudent to employ similar discretion and caution—and even measured optimism—toward genetic testing as a whole.

many states crafted policies that provided individuals with some level of protection from discrimination either in the workplace, in healthcare, or in both. These state laws, however, have created a patchwork of protections that vary considerably by state and fail to provide the most essential guarantees, which is why federal legislation is still necessary.

Nevertheless, various state laws do provide some general guidelines as to how federal legislation should look. The National Conference of State Legislatures has compiled state genetic nondiscrimination laws and released statistics about the protections offered by each state. In November of 2007, NCSL found that, of the 50 states and Washington, DC, 34 had passed prohibitions outlawing the use of genetic information by employers.[22] Not surprisingly, protections offered in each state vary. Not all of the statutes extend the protection to inherited characteristics such as those of Arizona and Iowa, and only some define genetic information in broad enough terms to include the test results of family members, a family history, and information related to genetic tests.

For an individual American, this means that there are still loopholes in many states that employers could exploit to get at information about an employee's inherited characteristics regardless of whether those characteristics have any medical significance. Moreover, of the 34 states with established protections only 14 had put in place specific penalties to be levied when employers violate them.

NCSL found in the insurance arena that 47 states and Washington, DC had

State Laws Governing Genetic Discrimination

Compiled by the National Conference of State Legislatures in November, 2007

Genetic Privacy Laws

- The majority of state legislatures have taken steps to safeguard genetic information beyond the protections afforded to other types of health information
- 17 states require informed consent for a third party to either perform/require a genetic test or obtain genetic information or both
- 27 states require consent to disclose genetic information
- 5 states explicitly define genetic information as personal property
- 4 states mandate individual access to personal genetic information
- 18 states have established specific penalties (civil, criminal, or both) for violating genetic privacy laws

Employment Laws

- 34 states and the District of Columbia have genetic nondiscrimination in employment laws
  - ALL prohibit discrimination based on the result of genetic tests
  - MANY extend the protections to inherited characteristics
  - SOME include test results of family members, family history, and information about genetic testing
- Most also restrict employer access to genetic information
- Some also make exceptions to statutory requirements (for example, if genetic information may identify individuals who may be a safety risk in the workplace)
- 14 states have specific penalties for genetic discrimination in employment

passed some form of protection against genetic discrimination.[23] The details of these laws, however, greatly limit the number of policies that these laws cover as well as the protections that they afford policyholders. For example, NCSL counts only 24 states that prohibit insurance companies in the individual and group markets from requiring individuals to submit to genetic tests or provide genetic information. In addition, eight states have policies that apply to either the individual market for health insurance or the group market, but not both.

The fact that some state-level protections have been implemented has not put this issue to rest. A 2005 commentary in the magazine *Nature Genetics* explained "variation in language, coverage and effectiveness of state laws is part of the impetus to pass basic federal legislation enforcing genetic nondiscrimination."[24] Consideration of this issue by the federal government is necessary in order to create a comprehensive, national level of protection from genetic discrimination for all Americans.

Federal Action Now Required

The Genetic Information Nondiscrimination Act of 2007 (S. 358), which seeks to close many of the protection gaps in existing state and federal laws, is only the latest effort to ensure genetic privacy and safety. Legislation seeking similar goals has been introduced in every session of every Congress since 1995. In fact, versions of the current GINA bill were approved by the Senate in 2003 and 2005 but never made it through the House of Representatives. Last year, the House of Representatives passed GINA by a margin of 420 to 3 (H.R. 493), and on March 5 of this year it passed in the House, again, as an addition to the Paul Wellstone Mental Health and Addiction Equity Act (H.R.1424).

As for the stand-alone GINA bill, a small group of Senators have put a hold on the legislation, which under Senate rules prevents the Majority Leader of the Senate, Harry Reid (D-NV), from bringing the legislation to the floor of the Senate for a vote without first persuading 59 of his colleagues to agree to overrule the hold. Given the support of the legislation in the Senate, the House, and the general public, there is increasing practical necessity for the Senate to vote on S. 358 this session.

An Examination of GINA's Provisions

Understanding the misplaced opposition to GINA requires a careful examination of the legislation itself. GINA seeks to prohibit genetic discrimination in employment and health insurance by outlawing specific practices in each setting. Title I of the bill applies to health insurance policies, expanding the HIPAA protections for the group and individual markets and also restricting the acquisition, use, and disclosure of genetic information. It sets up a more robust set of protections for consumers by preventing group plans from accessing genetic information in the first place, or from requiring that people submit to genetic tests.

A Congressional Research Service Report concluded that GINA would prohibit group plans from "requesting, requiring, purchasing, using or disclosing genetic information for the purposes of underwriting, eligibility determination, premium rating, or the creation, renewal, or replacement of a health insurance plan or contract."[25] Addressing a specific gap in the HIPAA protections, it would prevent group plans from raising the premium of an entire group based on the genetic information of an individual within that group.

Provisions of the Genetic Information Non-discrimination Act

A Summary

Title I: Health Insurance

- Extends Health Insurance Portability and Accountability Act protections against discrimination by group health plans/issuers of insurance in group and individual markets, and restricts acquisition, use, and disclosure of genetic information
- Prohibits group plans and insurers from requesting or requiring that individuals or their family members undergo a genetic test (does not apply to health care professionals)
- Prohibits group plans and insurers from requesting, requiring, purchasing, using, or disclosing genetic information for purposes of underwriting, eligibility determination, premium rating, or the creation, renewal, or replacement of a health insurance plan or contract
- Prohibits plans and insurers in the group market from
  - Denying enrollment to an individual based on genetic information about that individual or their family (already addressed by HIPAA)
  - Adjusting a group's premium based on genetic information about an individual in the group or their family
- Prohibits insurers from denying enrollment or adjusting premiums based on genetic information about an individual or their family in the individual market
- Permits the Secretary to impose a penalty of $100 per day during a period of noncompliance with Title I. In cases of willful neglect minimum penalty of $2,500 or $15,000 for more severe/prolonged violations

Title II: Employment

- Would make it unlawful employment practice for an employer to discriminate against an employee on the basis of genetic information, and also would bar employers from acquiring genetic information except under certain specified circumstances
- Would cover employers and employees as defined in:
  - Civil Rights Act of 1964, Government Employee Rights Act of 1991, Congressional Accountability Act of 1995, Section 3 U.S.C. 411(c)
  - Also covers job applicants
- Prohibits employers, employment agencies, and labor organizations from requiring or requesting that an individual or family member undergo a genetic test
  - Does not apply to health care professionals in the context of providing care
- Prohibits employers, employment agencies, and labor organizations from using genetic information when making decisions about employees' or applicants' hiring, promotion, or eligibility or selection for training programs or apprenticeships
- Prohibits employers, employment agencies, and labor organizations from requesting, requiring, or purchasing genetic information
  - Allows acquisition of genetic information by these groups when:
    - Offering a health service program (requires prior, knowing, voluntary, and written authorization)
    - The employee provides written authorization
    - Information used to monitor biological effects of toxic substances in the workplace (employer must provide written notice, AND either obtain prior, knowing, voluntary, and written authorization OR be acting to comply with federal or state laws)
  - Allows an employer to obtain genetic information when:
    - They inadvertently requested or required family medical history
    - They offered health or genetic services, and individual provided authorization
    - Identity of specific employees not disclosed
    - Employer requested information to comply with the Family and Medical Leave Act
    - Purchased publicly available documents that may have included family medical histories
- Treats genetic information as part of an individual's confidential medical record, and requires employer to maintain separate forms or files for genetic information if they obtain it
  - Disclosure is prohibited except when disclosure is:
    To the individual or employee at their request
    - To an occupational or other health researcher (must comply with part 46 of title 45, Code of Federal Regulations, pertaining to protection of human subjects)
    - In response to a court order (must give employee notice and enough time to challenge the order)
    - To government officials investigating compliance with Title II
- Establishes a commission to review the science of genetics and make recommendations on whether enforcement of "disparate impact" should be added to legislation (in order to protect individuals from situations where an employer unwittingly violated the law, and as a result disproportionate adverse effects are experienced by some individuals)

Finally, GINA would prohibit insurers from denying enrollment or adjusting premiums based on genetic information in the individual market. All of these prohibitions would be implemented by specific federal agencies, which would have the authority to issue specific penalties in cases of noncompliance.

Title II of GINA seeks to prevent genetic discrimination in the workplace by prohibiting the acquisition of genetic information by employers as well as its use in making decisions. Employers would not be able to access genetic information because it would prohibit employers, employment agencies, and labor organizations from requiring or requesting that an individual submit to a genetic test.

The new legislation would also prohibit employers from using the results of such a test in making decisions about hiring, promotion, or selection for training programs. GINA would achieve this by requiring that employers treat genetic information as part of an employee's confidential medical record, kept separate from employment files and protected from disclosure except in certain instances. The bill does, however, specify situations in which an employer could obtain genetic information in order to carry out necessary duties or comply with other laws such as the consensual genetic monitoring of the biological effects of toxic substances in the workplace.[26]

GINA Is Supported by a Broad Coalition

GINA is supported by virtually the entire scientific and medical community. Both researchers and physicians fear that the much talked about era of personalized medicine—where pharmaceuticals and therapies can be tailored to the specific genetic makeup of the patient — will not be realized without the protections that it offers, as people opt out of participation in research and patients forgo genetic tests. A February 14, 2008 editorial in the magazine *Nature* declared "it's hard to imagine a more worthy cause…or a more important legacy for this Congress."[27]

Advocacy organizations representing a range of interests have also coalesced around the issue of genetic discrimination and declared their support for GINA. These groups come from the patient advocacy, academic, business, and professional communities. As stated previously, a survey conducted by the Genetics and Public Policy Center demonstrates that three quarters of Americans support legislation to ban the practice of genetic discrimination by employers and insurance companies.

Such support has translated into strong political support in Congress. Previous votes in Congress demonstrate bipartisan support for GINA, which was passed unanimously by the Senate in 2003 and 2005. When the House of Representatives approved the measure last year it was by the considerable margin of 420 to 3, and the Senate version of the bill currently has 36 cosponsors drawn from both political parties. President Bush also declared that he supports the legislation and will sign it if it reaches his desk.[28]

Yet in a statement issued after the passing of H.R.1424, the administration stated in spite of its support for preventing the misuse of genetic information, "the Administration has both substantive and process objections to the rule."[29] They state that they oppose the lack of a clear firewall between the section dealing with insurance and the section dealing with employ-

ers. It would also like to see a clearer definition of the legislation's relationship with HIPAA, and the statues under which employees settle health benefits disputes.[30] Only the small group of Senators holding the legislation, it seems, stand in the way of enactment. To understand their concerns requires a look at industry worries over the proposed legislation.

Industry Concerns

The main opposition to genetic nondiscrimination legislation has come from the industries it seeks to regulate—business and insurance interests. Representatives from these groups maintain that genetic information is not being used in decision-making, and express concerns that the enactment of new regulations will place an excessive burden on business.[31] They point to the state and federal laws detailed above to support the argument that genetic discrimination is already prohibited, despite the shortcomings of these policies. They also have somewhat legitimate concerns that new regulations could lead to an increase in the number of lawsuits that they face. Multiple states have passes similar legislation and have not experienced the flood of lawsuits that many GINA opponents anticipate.

Another concern of insurance companies is that the information imbalance between the companies and their policyholders could result in the insurance companies ultimately going out of business. They feel that this would be the inevitable result of policyholders concealing a predisposition from their insurers, resulting in unanticipated higher payouts when the disease or condition finally manifests itself.

These concerns are unfounded. The true threat of concealed genetic information arises when patients obtain genetic tests privately and do not inform their doctors because they are afraid that their insurance premiums will go up because they posses a certain gene as was recently detailed in a harrowing *New York Times* piece.[32] This could ultimately prove even more costly for insurance companies because patients cannot receive proper treatment from doctors who do not know about their genetic predispositions, leading patients to become sicker than if their doctors had known. And this would lead to more expensive care down the road.

From an innovation standpoint, it is important that we remember that genetic testing will only become more commonplace, and that the age of personalized medicine will not be able to take off unless patients are confident that their genetic information will not be misused. It is better that these legislative provisions are put in place as the personalized medicine industry grows rather than after it has become set in its ways.

Indeed, in the United Kingdom a moratorium is already in place on insurance company usage of all but the most reliable genetic information such as tests for Huntingdon's disease. The United Kingdom, however, with its universal coverage, does not have the same problem that the United States does with our 47 million uninsured citizens. The United Kingdom also has a Genetics and Insurance Committee in their Department of Health that assesses genetic tests for Technical, Clinical, and Actuarial Relevance.

Clearly, Great Britain has a better understanding of how genetic nondiscrimination and the quality of genetic tests are intertwined. As a result of this oversight committee and the United Kingdom's universal coverage, the usage of genetic infor-

mation by insurance companies does not have the same discriminatory effect that it would in the United States.[33] Therefore, the United States must act sooner rather than later—not just because of our problem with health insurance coverage but also because of the private genetic testing industry which is growing at a rapid pace.

Moreover, this new industry is making questionable claims in its advertising and offering tests of uncertified reliability. Thus, much like the usage of genetic information, the quality of genetic testing is an aspect of the genetic age that will need to be brought under proper control in order for patients to truly feel secure in the knowledge that they are benefiting from innovation.

Chances of GINA Enactment

For some time, the hold on the legislation has kept Senate Majority Leader Reid from placing the legislation on the Senate calendar. Knowing this, Sen. Reid is now trying to override the hold and get the bill to the floor for a vote.

The legislation has broad support from legislators on both sides of the political aisle, especially in the House. This is evidenced by GINA passing by wide margins both as a stand-alone bill (420 to 3) and as part of the Paul Wellstone Mental Health and Addiction Equity Act (H.R.1424) (268 to 148). Ultimately, GINA will need to pass as a stand-alone bill since a conference on H.R. 1424 and its GINA-excluding Senate counterpart seems unlikely.

The stand-alone passage, however, depends on whether that small group of Senators can be persuaded to give up their hold. Alternatively, Senate Majority Leader Reid could seek out 59 of his fellow Senators to vote to override the hold. Since the Senators who are holding the bill do not seem likely to budge without significant compromise, it is imperative that 59 other Senators follow Reid's lead.

Post-GINA: The Next Steps

Whether or not GINA becomes law, there will continue to be a gaping hole in the regulatory framework for genetic testing. Fortunately, legislation recently introduced by Congress—specifically the "Genomics and Personalized Medicine Act of 2007" and the "Laboratory Test Improvement Act of 2007"—give the relevant regulatory agencies the power to regulate genetic tests and the labs that carry them out. Ultimately, whether through legislation or simply new regulatory protocols, this testing hole can easily be patched by four measures that will allow for the federal oversight of genetic tests, the labs that conduct them, the transparency of their results, and the advertising of direct-to-consumer genetic tests.

One measure would be to have the Centers for Medicare and Medicaid Services, or CMS, create a "specialty" for genetic testing laboratories. The second measure would be to expand the FDA's jurisdiction to include the regulation of lab-developed tests in addition to pre-manufactured test "kits" that already fall under its jurisdiction. The third measure entails the HHS creating a mandatory genetic test registry so that the clinical validity of all genetic tests is transparent for the public. And finally, the FDA and FTC should collaborate on the enforcement of Section 5 of the FTC Act in order to curtail false or misleading advertising by genetic testing companies.

CMS and the Creation of a Genetic Test Specialty

Currently, the CMS certifies clinical laboratories pursuant to the Clinical Laboratory Improvements Act of 1998. Under CLIA's enforcement ambit, CMS designates some kinds of lab tests as "high-complexity" tests and requires the labs that conduct them to undergo "periodic proficiency testing" in addition to the basic requirements that all labs must follow. This means that labs would have to show that they can accurately perform the test and interpret the results.

The problem is that not all "high-complexity" tests are required to undergo "periodic proficiency testing" because they are not all designated as "specialties." Genetic testing is one such "high-complexity" test, but CMS has repeatedly neglected to create a genetic testing "specialty". Therefore, labs that conduct genetic tests are not required to enroll in formal proficiency testing programs.[34] Labs can enroll voluntarily, but formal proficiency testing programs only exist for the small fraction of genetic tests that overlap with other specialties. This is because of the lack of a designated specialty area for genetic testing and the consequent lack of a federal requirement for proficiency testing of genetic tests.[35]

Enhanced genetic testing regulation was first recommended by a joint task force of the National Institutes of Health and the Department of Energy in 1997. This was followed by a recommendation from an advisory committee at the Centers for Disease Control, which explicitly recommended a genetic testing specialty. The Department of Health and Human Services eventually published a Notice of Intent in the Federal Register in May 2000.

Of the 57 responses it received, 93 percent were supportive of a genetic testing specialty. The Proposed Rule described in the Notice of Intent would ensure the "analytic validity" of genetic tests—that the test can accurately find the gene for which it is looking—as well as require that laboratory directors assess "clinical validity," or the connection of test results to health recommendations.

That second provision was a more controversial proposal since it was unclear as to where and how lab directors should obtain data about the clinical validity of their tests. The consensus among the 23 out of 57 comments opposed to documenting clinical validity was that even though documenting analytical validity was well within a lab director's duty, documentation of clinical validity would be too burdensome for lab directors to carry out. Others who commented felt that lab directors should have some role in assessing clinical validity, but that it should consist of nothing more than reviewing existing medical and scientific literature on the tests.

Yet the New York State Program for lab test regulation, which is more stringent than CLIA, fully supported this recommendation since it has been successful with its own regulatory protocol for assessing clinical validity of "home brew" genetic tests. Overall, then, the New York model might be considered a realistic and useful protocol from which a similar federal model could be built. Given the other conflicting comments however, its success would depend on clear communication of the protocol's details.

No federal regulations on any of the above matters—from the creation of a genetic testing specialty to requiring documentation of clinical validity—has yet to be instituted or even proposed. There are multiple reasons for this. First of all, in the transition from the Clinton administration to the Bush administration, CMS stalled on the creation of a genetic specialty until September 2005. At that point CMS announced it would establish the genetic testing specialty category sometime during 2006. Nevertheless, during the summer of 2006, CMS changed its position, claiming that the specialty lacks sufficient "criticality" for a new regulation and that the existing ones are adequate.[36]

There may be more to it than that, however. CMS seems unwilling to be burdened with creating a new specialty for genetic testing, according to a recent article in *Nature Biotechnology*. In the February 2008 article, a CMS official is quoted as saying, "It's not like we're in some total dead end because CLIA is limited to analytic validity…clinicians can rely on their own judgment…"[37] More remarkably, in a newsletter from the American Association of Clinical Chemistry, Judy Yost, the Director of CMS's Division of Laboratory Services, is quoted as saying, "[W]e really don't have any evidence that we need more stringent requirements than those that already exist." She also suggested, "[M]aybe we can look at some

ways [of regulating] that are less burdensome and time consuming.[38]"

Consequently, as an alternative measure, CMS has offered to add genetic analytes—the chemical substances whose presence are demonstrated in a genetic test—to the list of 83 analytes for which labs must undergo proficiency tests for analytical validity under CLIA.[39] The problem with this recommendation is that this list, which has not been updated since 1992, would still not require the assessment of the *clinical* validity of the genetic tests that use the new analytes. It would only require the assessment of the *analytical* validity of the tests.[40]

Moreover, the existing 83 analytes already fall into other specialty categories, so if new "genetic testing" analytes were to fall into those existing categories too, then a genetics testing lab would not have to undergo proficiency testing for those new analytes unless it was also certified for those other specialty categories. Indeed, this would be a band-aid policy modification to an already grossly inadequate regulatory regime.

A 2006 survey by the Genetics and Public Policy Center found that one-third of high-volume laboratories are not certified in any specialty, and that the specialty certifications that are most commonly held by genetic testing labs are of questionable relevance to genetic testing. This survey also discovered that labs that fail to perform proficiency testing on all of their tests are eight times more likely to report multiple deficiencies.[41]

In the same survey, 73 percent of lab directors approved of the creation of a specialty category for genetic testing.[42] Therefore, the only proper measure is to establish a new specialty category for genetic testing and to implement limited requirements for clinical validity—such as those in place in New York State—to be part of proficiency testing for genetics labs.[43] Ultimately, though, CMS at the very least will need to stop waffling on the establishment of a separate genetics testing specialty in order to ensure the quality of genetic testing.

Hopefully, the release of the report by HHS's Secretary's Advisory Committee on Genetics, Health, and Society at the end of April 2008 will make the need for a separate specialty category crystal clear to the CMS. If this does not convince them, then legislation (S.976) proposed by Sens. Barack Obama (D-IL) and Richard Burr (R-NC) would charge CMS with establishing the specialty category for molecular and biochemical genetic tests.

FDA Regulation of Lab-Developed Tests

Since CMS only regulates labs, there still remains a need to regulate the actual tests. Responsibility for this regulatory gap lies at the feet of the FDA. Currently, the FDA does not oversee genetic testing since most labs do not use pre-manufactured genetic testing "kits," instead using tests developed in-house, or lab-developed tests, known as LDTs or "home-brews." Incidentally, it is the labs themselves which determine whether a new test should be developed as a "kit" or a "home-brew."

Not surprisingly, most elect not to develop them as "kits." As a result, there are currently only a dozen test kits that are approved by the FDA.[44] Interestingly, the FDA *does* have the jurisdic-

tion to oversee "home-brews" but has chosen not to. The FDA refers to this as "enforcement discretion," which they are exercising due to a lack of resources and a reluctance to interfere in the rapidly developing field of genetic testing.[45]

The FDA, however, does regulate certain chemical ingredients used in LDT genetic testing called "analyte-specific reagents," or ASRs. These small molecules can be made by a lab itself or by a manufacturer for sale to labs. According to the Genetics and Public Policy Center, however, manufactured ASRs can only be sold to laboratories that are approved to do high-complexity tests. What's more, ASR-based home-brew tests can only be ordered by a health care provider, though it remains unclear whether these tests must be ordered by the patient's physician or simply a physician employed by the lab. Nor does the FDA regulate the claims of the tests that use the ASRs.[46]

As of July 2007, however, the FDA moved into the territory of LDT regulation by issuing draft guidelines for the oversight of so-called in vitro diagnostic multivariate index assays, or IVDMIAs, which are a special subset of LDTs that use an algorithm on lab data in order to generate recommendations for diagnosis, prevention, or treatment of a disease. These guidelines have yet to be finalized, and only one IVDMIA has undergone FDA pre-market review.

This subset of tests "include[s] those used to diagnose and guide treatment decisions for breast cancer, prostate cancer recurrence, cardiovascular disease, and Alzheimer disease.[47]" In other words, this limited form of LDT regulation only applies to genetic tests for conditions whose genetic components have been thoroughly studied and would not affect newer genetic tests.

This is problematic from the industry's point-of-view since the companies that make IVDMIAs would like to see all genetic tests subjected to the same scrutiny. This is because a company could manufacture a genetic test that is not an IVDMIA (and therefore not subject to FDA approval) yet still make the same claims as the manufacturer of the FDA-approved IVDMIA. This creates unfair competition in the marketplace and would ultimately hurt the credibility of the genetic testing industry in the long run in addition to putting consumers at risk by providing them with unreliable tests.

The Laboratory Test Improvement Act of 2007 (S. 736), introduced by Sen. Edward Kennedy (D-MA), would grant the FDA the authority to regulate LDTs as medical devices. Most of the LDTs would be classified as Class II medical devices but the FDA could move them to the more stringent Class III or less stringent Class I under certain conditions.[48] The Obama-Burr bill (S. 976) would commission the Institute of Medicine to make recommendations to the HHS Secretary, who would then implement a "decision matrix" that would clarify the regulatory roles of the FDA and CMS in genetic testing. This might not be necessary, however, if the SACGHS report spurs enough regulatory action from HHS following the release of its recommendations.

Genetic Testing Database

Another important aspect of the age of genetic medicine is the relationship between the consumer and the test in terms of access and information. When it

comes to access, different states have different laws that determine how a patient gets access to genetic testing.

This is because of the legal confusion caused by CLIA. According to CLIA, patients need a "written or electronic request for patient testing from an authorized person" (42 C.F.R. § 1241(a)), but it is up to the states to define who is an "authorized person." Some states allow direct-to-consumer laboratory testing without restriction (26 states plus DC), others forbid DTC tests altogether (13 states), and 11 permit it only for specified categories of tests, which usually exclude genetic tests[49] (see table, "Survey of Direct-to-Consumer Testing Statutes and Regulations" on page 29.)

Unfortunately, this current patchwork of state laws can still be circumvented by the almost 30 internet-based DTC genetic testing companies that currently exist.[50] Although these types of regulations are best left under the jurisdiction of state law, the federal government can still take specific measures to ensure that states, health care providers, and consumers have the right information about the analytical and clinical validity of genetic tests.

The problem with the current lack of reliable and transparent information about genetic tests is that DTC genetic testing companies exploit the naiveté of consumers who are unaware of how the fine details of a genetic test can have huge implications for the clinical relevance of the results. This becomes especially confusing when some DTC companies attempt to differentiate themselves from "lifestyle" and "nutrigenomic" testing—the genetic tests that purport to give nutritional, exercise, and lifestyle advice based on genetic tests and have recently been condemned in a GAO report—by only providing "more established" genetic tests for diseases such as breast cancer or cystic fibrosis.

Therefore, there must be a way of ensuring that genetic tests have clinical validity, meaning that the tests results should have some connection to a person's current or future health.[51] Although this might seem like we are recommending that genetic testing should be singled out for special treatment, we are actually recommending that it be put on par with other kinds of medical tests. When over-the-counter HIV tests came out, for example, both the kit sold to consumers and the lab where they sent their specimen were regulated by the FDA. That is for one disease, however. Genetic tests can apply to hundreds of diseases and conditions, and the number of available tests is increasing very rapidly.[52]

HHS's first regulatory priority, then, should be to create a mandatory genetic test registry. This recommendation has already received strong support from multiple industrial, professional, and advocacy groups. Many groups argue it should be hosted by GeneTests, though a few groups felt that the federal government should host it through the FDA, CMS, NIH, or some other new body.

CMS, however, commented by noting that it does not collect test-specific data, but would still be willing to cooperate with GeneTests and cross-reference to their database.[53] Indeed, CMS is not properly equipped to house the database, but other agencies under HHS have experience with databases, such as the FDA, CDC, and NIH, and could easily collaborate with GeneTests.[54]

Overall, this recommendation is largely uncontroversial because of its broad support and would do the genetic testing industry much good by instilling public confidence in genetic tests. Therefore, it should be established as quickly as possible, which seems likely given its support among the comments to the upcoming SACGHS report.[55] Thus it is imperative, at the very least, for the HHS to fund an expansion of the GeneTests database and make it mandatory for labs and companies to register their tests with it.

Honest and Accurate Advertising

Finally, federal regulators need to better police the marketing and advertising of direct-to-consumer genetic tests. This will require a more aggressive collaboration between CMS, FDA, and Federal Trade Commission to ensure these companies do not oversell the clinical validity of their tests, and that they adequately convey the limitations of specific tests.[56]

Here's the state of play. At the February 12 meeting of the Secretary's Advisory Committee on Genetic Testing, FTC representative Matthew Daynard said that the current FDA-FTC collaboration is "working." Yet it is clear that DTC industry remains a wild frontier. According to the Genetics and Public Policy Center, most of the nearly 30 DTC companies they investigated claimed to be CLIA-certified, but the FTC does not hold them to this claim since CMS does not make the list of certified labs public.

Additionally, some of the genetic testing companies, such as 23andMe, claim that their tests are for "research purposes" only, and are therefore exempt under CLIA. Under CLIA, however, labs that return tests to subjects (which 23andMe does) are not considered "research" labs. Multiple complaints have been filed with the FTC about DTC genetic testing and the FTC has issued a consumer alert (http://www.ftc.gov/bcp/edu/pubs/consumer/health/hea02.shtm), but the agency has yet to take any direct action against multiple false and misleading claims.[57]

Therefore, the FTC should utilize information from CMS and collaborate with the FDA to create guidelines for DTC genetic testing companies to follow so that they adequately convey the scientific limitations of their tests. The FTC should also take action against any DTC companies that make false or misleading claims about their tests. This should allow for a proper balance among the needs for consumer protection, freedom of commercial speech, scientific innovation, and scientific integrity.

Conclusion

If the age of genetic medicine is to become legitimate and useful to the general public, then laws must be put in place to ensure that genetic information does not act as a basis for discrimination. And regulatory guidelines must be established to ensure that genetic tests are valid, reliable, and relevant.

GINA will take care of the former by preventing employers from using genetic information to discriminate against employees for the purpose of hiring, training, or promoting. The proposed legislation will also prevent insurance companies from using genetic test results as a basis for denying coverage or increasing premiums.

These legislative stipulations will greatly increase patients' sense of security with regard to genetic testing. They will allow patients to acquire information about their health and deal with it at their own and their physician's discretion. Under this statutory framework, patients, physicians, and the public at-large will see genetic testing as a revolutionary tool for diagnosing and treating disease and not as a liability.

This is why it is essential that the Senate follow the lead of the House of Representatives by passing GINA. Members of the Senate who support the legislation must work actively with Senate Majority Leader Reid to secure all 60 votes necessary to override the hold and pass the bill.

In addition, other measures that are currently within the law must be taken to ensure reliable regulatory oversight of the genetic testing industry. The creation of a genetic test registry along with the regulation of genetic testing labs by CMS, laboratory-developed tests by the FDA, and DTC advertising by the FTC will add legitimacy to the burgeoning genetic testing industry. By making innovation and entrepreneurialism accountable to patients, providers, and consumers, the federal government can take genetic testing beyond the realm of novelty and help to usher in the medical revolution that the science and practice of genetic testing truly deserves.

Appendix

Genetic Testing Regulation

Key Findings of this Paper

Lab Regulation

- Although genetic testing is designated as "high-complexity," it is not required enroll in proficiency testing since a "specialty area" does not exist for genetic testing.
- One-third of high-volume laboratories are not certified in any specialty
- Labs that fail to perform proficiency testing on all of their tests are eight times more likely to report multiple deficiencies.
- 73 percent of lab directors surveyed approved of the creation of a specialty category for genetic testing.

Test Regulation

- Although it has the jurisdiction to do so the FDA has chosen not to regulate laboratory- developed tests.
- The FDA does regulate pre-manufactured genetic test "kits," certain chemical ingredients used in LDTs, and in vitro diagnostic multivariate index assays, or IVDMIAs.

Genetic Testing Access

- Some states allow direct-to-consumer laboratory testing without restriction (26 states plus the District of Columbia), others forbid DTC tests altogether (13 states), and 11 permit it only for specified categories of tests, which usually exclude genetic tests.
- According to CLIA, patients need a "written or electronic request for patient testing from an authorized person" (42 C.F.R. § 1241(a)), but it is up to the states to define who is an "authorized person."

Honest and Accurate Advertising

- Of the nearly 30 DTC companies investigated by the Genetics and Public Policy Center, most claimed to be CLIA-certified, but the FTC does not hold them to this claim since CMS does not make the list of certified labs public.
- Some DTC genetic testing companies claim that they are exempt from CLIA certification because their tests are only for "research purposes," but under CLIA, labs that return results to consumers are not considered "research" labs.

Genetic Testing Regulation

Key Recommendations of this Paper

Lab Regulation

- The Centers for Medicare and Medicaid Services, or CMS, should stop delaying and create specialties for biochemical and molecular genetic testing so that genetic testing labs will be required to undergo formal proficiency testing for CLIA certification.
- If the SACGHS report does not convince CMS to do this, then the Senate should move ahead with passing the provision in S. 976 which would force CMS to do so.

Test Regulation

- The FDA should subject all lab-developed genetic tests to an approval process. This would create a level playing field for all genetic tests whether they are pre-manufactured "kits," IVDMIA tests, or lab-developed tests. The provisions in S. 736 would be adequate since it would designate LDTs as Class II medical devices but allow the FDA to make them Class I or Class III if needed.

Genetic Testing Access

- HHS should immediately create a mandatory genetic testing registry—hosted by GeneTests—to which all genetic testing labs should submit data. Other agencies accustomed to building databases should collaborate with GeneTests in order ensure transparency, public access, and utilization of the data in regulatory measures.

Honest and Accurate Advertising

- The FTC should utilize information from CMS and collaborate with the FDA to create guidelines for DTC genetic testing companies to follow so that they adequately convey the scientific limitations of their tests.
- The FTC should also take action against any DTC companies that make false or misleading claims about their tests.

SURVEY OF DIRECT-TO-CONSUMER TESTING STATUTES AND REGULATIONS

JURISDICTION	DTC PERMITTED?	CITATION TO STATUTE OR REGULATION	COMMENTS
Alabama	No	Ala. Admin. Code r. 420-5-8-.01(2)(l),(o) Ala. Admin. Code r. 420-5-8-.04(5)(d)(3)	While state law does not address DTC testing directly, an official in the Alabama Department of Public Health stated that DTC testing is prohibited under state law, since the Administrative Code limits ordering tests to a licensed physician or other "authorized person."
Alaska	Yes	None identified	State law is silent on the issue. An official in the Alaska Department of Public Health stated that no law prohibits any person from ordering a laboratory test.
Arizona	Limited	Ariz. Rev. Stat. § 36-470 Ariz. Admin. Code R9-14-102	State law lists specific tests that may be ordered directly by consumers. Other than those specified tests, only health professionals authorized by law are permitted to order tests, and laboratories must report results only to the person who ordered the test. An official at the Arizona Department of Health Services confirmed that DTC testing is limited to a few specified tests.
Arkansas	Yes	None identified	An official in the Arkansas Division of Health Facility Services confirmed there are no laws in Arkansas that address this issue.
California	Limited	Cal. Bus. & Prof. Code § 1241 Cal. Bus. & Prof. Code § 1246.5 Cal. Health & Safety Code § 120917 Cal. Health & Safety Code § 123148 17 Cal. Code. Reg.1053.5	DTC testing is expressly authorized only for specified tests: "pregnancy, glucose level, cholesterol, occult blood, and any other test for which there is a test for a particular analyte approved by the federal Food and Drug Administration for sale to the public without a prescription in the form of an over-the-counter test kit," as well as HIV tests. A test approved only as an over-the-counter collection device may not be conducted pursuant to this section.* An official at the California Department of Health and Human Services confirmed that DTC testing is limited.
Colorado	Limited	Colo. Rev. Stat. § 12-36-106(3)(u) Colo. Rev. Stat. § 12-36-106(1)	An official at the Colorado Department of Public Health & Environment noted that under Colorado law, the definition of the practice of medicine does not include the provision of laboratory tests to individual patients. Therefore, he stated, DTC testing is understood to be permitted. The official pointed to a section of the Colorado Revised Statutes that excludes from the practice of medicine the provision of laboratory results to a licensed physician, other than histopathology and cytology test results. While this provision of the law is somewhat ambiguous, the state official interprets it to mean that Colorado does not allow laboratories to provide histopathology and cytology tests directly to consumers.
Connecticut	No	Regs., Conn. State Agencies §19a-36-D29(a) Regs., Conn. State Agencies §19a-36-D32(a)	Laboratories may accept specimens only upon request of licensed physician or other persons authorized by law to make diagnoses. Laboratories may report findings only to the licensed provider that ordered the test. Laboratories may provide results to lay persons upon written request of the provider who ordered the test. An official at the Connecticut CLIA Laboratory Program confirmed that DTC testing is not permitted.
Delaware	Yes	None identified	State law is silent on the issue. An official with the Delaware Department of Public Health Laboratories stated that Delaware permits DTC testing without limitations.
District of Columbia	Yes	D.C. Code § 44-211	D.C. law does not directly address DTC testing. The law does permit patients to request, in writing, access to or copies of the results of their laboratory tests. The law also states that all clinical laboratory results shall be reported to the requesting physician, but that when there is no requesting physician the laboratory shall report the test results directly to the patient and recommend that the patient forward the results to his or her physician. An official with the D.C. Department of Health stated that there is a D.C. clinical laboratory licensing law but it will not be implemented until regulations are promulgated; currently only the federal CLIA law is being enforced.
Florida	Limited	Fla. Stat. Ann. § 483.181(1),(2) Fla. Stat. Ann. § 483.288 Fla. Stat. § 483.314	A clinical laboratory may examine human specimens at the request only of a licensed practitioner or other person authorized by law to use the findings of clinical laboratory examinations. The results of a test must be reported directly to the licensed practitioner or other authorized person who requested it. An individual forwarding a sample of the individual's own blood to a clinical laboratory collected using an FDA approved home access HIV test kit shall be considered a person authorized to request this test. An official with the Florida Agency for Health Care Administration, Laboratory Licensing Unit, explained that only "medical" laboratory tests fall under these laws. Paternity tests, for example, do not. The official noted that it is unclear whether genetic tests that screen for a predisposition for Alzheimer's Disease or cancer, for example, would qualify as "medical" tests under these laws, as a clinician receiving these results would not necessarily make any medical decisions based on them.*

SURVEY OF DIRECT-TO-CONSUMER TESTING STATUTES AND REGULATIONS *(continued)*

JURISDICTION	DTC PERMITTED?	CITATION TO STATUTE OR REGULATION	COMMENTS
Georgia	No	Ga. Code Ann. § 31-22-4(a),(c)	Tests may be ordered only by a "licensed physician, dentist, or other person authorized by law to use the findings of laboratory examinations." Test results may be reported only "to or as directed by the licensed physician, dentist, or other authorized person requesting such test." An official at the Georgia Department of Human Resources confirmed that DTC testing is not permitted.
Hawaii	No	Weil's Code of Hawaii Rules § 11-110-12(b),(c) Weil's Code of Hawaii Rules § 11-110-33(6)	Tests may be ordered only by a "person authorized by law to receive and interpret laboratory results." Test results may be reported only to a person authorized by law to receive and interpret laboratory results or to a referring laboratory. According to an official at the Hawaii Department of Health, new regulations, which as of 6/13/07 had not been signed by the governor but which will probably go into effect in July 2007, will specify who "authorized persons" are. The new regulations will allow designees of authorized persons (e.g. individual patients authorized by their physicians) to order tests directly from laboratories.
Idaho	No	IDAPA 16.03.14.350.07	A provision in the state administrative code pertaining to hospital laboratories states that "orders for tests shall be made only by those practitioners legally authorized to diagnose, treat and prescribe." An official with the Idaho Bureau of Laboratories confirmed that DTC testing is not permitted.
Illinois	Limited	210 Ill. Comp. Statutes 25/7-101 210 Ill. Comp. Statutes 25/9-101	State law provides that tests may be ordered only by physicians, other health professionals listed in the statute, and police officers. Test results must be provided only to the authorized person who requested it. A state official with the Illinois Department of Public Health explained that there is now a rule allowing DTC testing, but only for CLIAwaived tests. However, this rule has not been approved by an Advisory Board, as the department no longer has an Advisory Board, so it is unclear what would happen if the rule were challenged.
Indiana	Yes	None identified	State law is silent on the issue. An official with the Indiana State Department of Health stated that DTC testing is permitted.
Iowa	Yes	None identified	State law is silent on the issue. An official with the Iowa CLIA Laboratory Program confirmed that there is no state law on the issue.
Kansas	Yes		State law is silent on the issue. An official at the Kansas Department of Health & Environment confirmed that Kansas is a direct access state.
Kentucky	No	Ky. Rev. Stat. § 333.150 Ky. Rev. Stat. § 333.190 Ky. Rev. Stat. § 333.240	State law provides that a "medical laboratory shall examine human specimens only at the request of a licensed physician, podiatrist, dentist, or other person authorized by law to use the findings of medical laboratory examinations. The results of a test shall be reported directly to the licensed physician, dentist, or other authorized person who requested it." An official with the Kentucky Office of Inspector General confirmed that DTC testing is prohibited.
Louisiana	Yes	None identified	State law is silent regarding authorization for DTC testing. An official with the Louisiana Department of Health and Hospitals stated that there are no state laws regulating laboratories and that anyone is permitted to order a laboratory test.
Maine	Limited	Me. Rev. Stat. Ann. tit. 22, § 2031-A Me. Rev. Stat. Ann. tit. 22, § 2030 Me. Rev. Stat. Ann. tit. 22, § 2031 CMR 10-144-256(9)	In general, "a medical laboratory shall examine specimens only at the request of a licensed physician or other person authorized by law to use the findings of laboratory examinations. However, a medical laboratory may examine specimens without a physician referral for a limited number of laboratory services to be determined by rules adopted by the department." These services include tests for (a) glucose for patients who have been previously diagnosed as having diabetes; (b) pregnancy; (c) colon cancer; and (d) cholesterol.* An official at the Maine CLIA Program confirmed that DTC testing is limited.
Maryland	Limited	Md. Health Gen. Code § 17-202.1 COMAR 10.10.01.03 COMAR 10.10.06.02 COMAR 10.10.06.04 COMAR 10.10.06.12	In general, tests can be ordered only by authorized persons listed in the statute, which does not include consumers, and results must be reported directly to the ordering individuals. Certain specified "health awareness tests" (tests approved by the Secretary to be performed at a temporary laboratory), such as cholesterol tests, may be provided DTC. An official at the Maryland Department of Health & Mental Hygiene confirmed that DTC testing is limited.

SURVEY OF DIRECT-TO-CONSUMER TESTING STATUTES AND REGULATIONS *(continued)*

JURISDICTION	DTC PERMITTED?	CITATION TO STATUTE OR REGULATION	COMMENTS
Massachusetts	Limited	Mass. Gen. Laws ch. 111D, §§ 4,8 105 CMR 180.010 105 CMR 180.043 105 CMR 180.280 105.CMR.180.290	In general, tests may be ordered only by physicians or other authorized persons listed in the statute, and test results may be reported only to the authorized person who requested the test, unless the authorized person requests that the result be sent to the patient. The law provides an exception for tests conducted pursuant to "health promotion screening programs," for the purpose of "promoting health awareness and education among the general public by early detection of disease and/or associated risk factors." Health promotion screening tests are "not used for the purpose of providing clinical diagnosis or treatment to patients." A state official at the Massachusetts Department of Health and Human Services explained that such exceptions are limited to eight tests, including for pregnancy and cholesterol.*
Michigan	No	Mich. Comp. Laws § 333.17001 (1) (f) Mich. Comp. Laws § 333.17020 (1)	The law defines the "practice of medicine" to include "diagnosis …by diagnostic test." Another law that requires informed consent for genetic testing refers to "a physician or an individual to whom the physician has delegated authority." According to an official with the Michigan Department of Community Health, DTC testing is prohibited because ordering tests and receiving results is part of the practice of medicine. However, the official stated that the prohibition does not apply to tests that are categorized as waived under CLIA.
Minnesota	Yes	None identified	State law is silent on the issue. An official with the Minnesota Department of Health confirmed that there are no state limitations on DTC testing.
Mississippi	Yes	None identified	State law is silent on the issue. An official with the Mississippi Department of Public Health confirmed that there are no state licensure regulations for clinical laboratory testing.
Missouri	Yes	None identified	State law is silent on the issue. An official with the Missouri Department of Health and Senior Services confirmed that DTC testing is permitted.
Montana	Yes	None identified	State law is silent on the issue. An official with the Montana CLIA program confirmed that DTC testing is permitted.
Nebraska	Yes	R.R.S. Neb. § 71-1,104.01	State law is silent on the issue. An official with the Nebraska State Health & Human Services Program confirmed that DTC is permitted. However, a statute that relates to genetic testing states that a physician, or person to whom a physician has delegated authority "shall not order a predictive genetic test" without first fulfilling informed consent requirements. The state official explained that a physician has an obligation to make sure that the patient is informed before he orders a predictive genetic test, but if the patient takes it upon himself to order the test, whether directly from a laboratory or through an intermediary, informed consent is not required.
Nevada	Limited	Nev. Rev. Stat. Ann. § 652.190	In general, a laboratory may examine specimens only at the request of a licensed physician or any other person authorized by law to use the findings of laboratory tests and examinations. However, if the examination can be made with a testing device or kit which is approved by the Food and Drug Administration for use in the home and which is available to the public without a prescription, the laboratory may examine the specimen at the request of any person. In general, the laboratory may report the results of the examination only to: (a) the person requesting the test or procedure; (b) a provider of health care who is treating or providing assistance in the treatment of the patient; (c) a provider of health care to whom the patient has been referred; and (d) the patient for whom the testing or procedure was performed. An official at the Nevada State Health Division confirmed that DTC testing is limited.
New Hampshire	No	N.H. Admin. Rules He-P 817.15(a).	Regulations provide that laboratories may perform testing only at the "request of a physician, dentist, chiropractor, court of law or any other person authorized by state statute to order and receive laboratory tests." An official with the New Hampshire CLIA program stated that the rules indirectly limit the ordering of tests to licensed practitioners, but that individuals have the right to access all their medical records including laboratory test results directly from the laboratory performing the test.
New Jersey	Limited	N.J. Stat. Ann. § 45:9-42.42 N.J.A.C. § 8:44-2.2 N.J.A.C. § 8:44-2.7	In general, tests may be ordered only by a "licensed physician, dentist, or other person authorized by law to use the findings of laboratory examinations and shall report only to those authorized by law to receive such results," although patients can also request copy. According to an official at the New Jersey Department of Health, the only tests that are exempt are original CLIA-waived tests, such as dipstick urinalysis, fecal occult, and pregnancy tests.*

SURVEY OF DIRECT-TO-CONSUMER TESTING STATUTES AND REGULATIONS *(continued)*

JURISDICTION	DTC PERMITTED?	CITATION TO STATUTE OR REGULATION	COMMENTS
New Mexico	Yes	None identified	State law is silent on the issue. An official with the New Mexico state CLIA program confirmed that there are no state laws prohibiting DTC testing.
New York	Limited	N.Y. Pub Health Law § 576-b N.Y. Pub. Health Law § 577 10 NYCRR § 19.1(j) 10 NYCRR § 58-1.7 10 NYCRR § 58-1.8 10 NYCRR § 63.3(e)	In general, tests may be ordered only by licensed physicians "or other persons authorized by law to use the findings of laboratory examinations in their practice or the performance of their official duties." Consumers are not listed among those authorized. Test results cannot be sent directly to patients except with written consent of the physician or authorized person, except blood type and RH factor can be given in writing to the patient without written consent. DTC testing is permitted for tests that have been approved by the Food and Drug Administration for direct, over-the-counter sale to consumers. An official with the New York State Department of Health confirmed that DTC testing is not permitted, other than for certain tests relating to the blood supply, such as HIV and Hepatitis C tests.
North Carolina	Yes	N.C. Gen. Stat. § 130A-148 N.C. Admin. Code tit. 10A, chapter 42	With the exception of HIV tests, which can be ordered only by licensed physicians, state law is silent regarding DTC testing. An official with the North Carolina CLIA program confirmed that other than HIV tests, physicians' orders are not required.
North Dakota	Yes	None identified	State law is silent on the issue. An official with the North Dakota Department of Health stated that laboratory testing facilities should establish their own policies to address DTC testing.
Ohio	Yes	None identified	State law is silent on DTC testing, other than a law specifically allowing individuals to request an HIV test from a public health clinic. Under scope of practice laws specific to each profession, there are limitations as to what kinds of tests practitioners can order, but these laws do not explicitly prohibit individuals from requesting a test from a lab. According to an official at the Ohio Department of Health, The Ohio Medical Board has objected to stores selling "doc in a box" testing kits, but so far no laws or rules have been passed to prevent it. Nevertheless, the official stated that there is some ambiguity in the law, as well as in a 1980 Medical Board opinion possibly implying a limitation on DTC tests.
Oklahoma	Yes	None identified	State law is silent on the issue. An official with the Oklahoma State Department of Health stated that she was aware of no state law prohibiting DTC testing.
Oregon	Limited	Or. Rev. Stat.§ 438.430 Or. Admin. R. 333-024-0050 Or. Admin. R. 333-024-0375 Or. Admin. R. 333-024-0395	In general, tests may be ordered only by "physician, dentist, or other person authorized by law to use the findings of laboratory examinations." The phrase "other person authorized…" has been interpreted by several practitioner boards to include different types of licensed practitioners, but not consumers. Regulations require written consent of physicians or other authorized persons to report test results to patients. DTC testing is permitted for certain specified tests including substance abuse testing, hemoglobin, glucose, fecal occult blood, pregnancy, and cholesterol. An official at the Oregon State Public Health Laboratory confirmed that DTC testing is limited in Oregon.
Pennsylvania	No	28 Pa. Code § 5.41 28 Pa. Code § 5.47	Tests may be ordered only by licensed "member[s] of the healing arts" or "other persons authorized by statute" and results may be sent only to the person ordering the test. An official at the Pennsylvania Department of Health confirmed that DTC testing is not permitted in Pennsylvania.
Rhode Island	No	Rules and Regulations for Licensing Clinical Laboratories and Stations R-23-16.2-C&S/Lab, §1.2 R-23-16.2-C&S/Lab, §10.2 R-23-16.2-C&S/Lab, §13.2	Tests may be ordered only by licensed physicians or other authorized personnel, defined as "health professionals working under the auspices of a physician or other licensed health care professional acting within his/her scope of practice." Patients may only have direct access to their laboratory results with written permission from their physician. An official at the Rhode Island Department of Health confirmed that DTC testing is not permitted in Rhode Island.
South Carolina	No	None identified	No laws specifically address DTC testing by consumers. However, an official with the South Carolina Department of Health & Environment Control stated that it is understood in South Carolina that patients cannot directly order laboratory tests. Under CLIA, "The laboratory must have a written or electronic request for patient testing from an authorized person… Authorized person means an individual authorized under State law to order tests or receive test results, or both." There is no law or rule in South Carolina listing who "authorized persons" are, but in practice laboratories have not accepted orders from individual consumers.
South Dakota	Yes	None identified	State law is silent on the issue. An official with the South Dakota Department of Health stated that she was aware of no state law prohibiting DTC testing.

SURVEY OF DIRECT-TO-CONSUMER TESTING STATUTES AND REGULATIONS *(continued)*

JURISDICTION	DTC PERMITTED?	CITATION TO STATUTE OR REGULATION	COMMENTS
Tennessee	No	Tenn. Code Ann. § 68-29-121	According to the Tennessee Code, "No person, except patients who are performing tests on themselves by order of their physician, shall examine human specimens without the written request of a physician or . . . other health care professional legally permitted to submit to a medical laboratory a written request for tests appropriate to that professional's practice, or the written request of a law enforcement officer . . . ." Test results "shall be reported directly to the physician… or other health care professional who requested it."
Texas	Yes	None identified	State law is silent on the issue. A state official at the Texas Board of Medical Examiners confirmed that as far as he knew, there were no state limitations on DTC testing.
Utah	Yes	None identified	State law is silent on the issue. An official with the Utah Department of Health stated that she was aware of no law prohibiting DTC testing and that there was an internal legal opinion stating that ordering a test, performing the test, and giving the results of that test to a person does not constitute the "practice of medicine."
Vermont	Yes	None identified	State law is silent on the issue. An official with the Vermont Department of Health stated that Vermont has no laws or regulations addressing DTC testing or regarding who is authorized to request testing or receive test results.
Virginia	Yes	Va. Code Ann. § 8.01-581.18:1	State law provides that when lab tests are conducted at the request of someone other than a physician, the laboratory report to the patient must state in bold type that the patient has the responsibility to contact a physician for test consultation and interpretation. An official at the Virginia Department of Health confirmed that DTC testing is allowed, and noted that the Virginia General Assembly had just passed a new law, which will become effective July 1, 2007, that allows physicians to request that laboratories send physician-ordered test results directly to patients. The law provides immunity to physicians for failing to act on the results of a laboratory test if the test is not requested or authorized by them, other than in limited situations such as when the results are provided to the physician by the person tested with a request for consultation.
Washington	Yes	Wash. Admin. Code § 246-338-010 Wash. Admin. Code § 246.338-070	State law provides that "test reports must… be released only to the "authorized persons or designees" and defines "authorized person" as "any individual allowed by Washington state law or rule to order tests or receive test results." An official with the Washington State Department of Health stated that nothing in Washington State law prohibits DTC testing.
West Virginia	Yes	None identified	State law is silent on the issue. An official at the West Virginia Department of Health confirmed that state law does not prohibit DTC testing.
Wisconsin	Yes	Wis. Adm. Code HSS 165.16 Wis. Adm. Code HSS 165.17	Law provides that clinical laboratories "shall examine specimens only at the request of persons or agencies authorized or allowed by law to submit specimens" and "shall report specimen findings to persons authorized or allowed by law to receive such reports." However, an official with the Wisconsin CLIA program stated that Wisconsin does not have any regulations that would prohibit DTC testing; she stated that the cited provision no longer applies to clinical labs.
Wyoming	No	Wyo. Stat. § 33-34-107 Wyo. Stat. § 33-34-108	Tests may be ordered only by a physician, dentist, "or other persons authorized by law to use the findings of laboratory examinations." Test results may be reported only "to or as directed" by the person who ordered the test. An official at the Wyoming Office of Healthcare Licensing and Surveys confirmed that DTC testing is not permitted.

Scope and Methodology

This chart was compiled based on a survey of state statutes and regulations and state government officials in order to determine whether state law permits direct ordering of laboratory tests by consumers and the delivery of test results from clinical laboratories directly to consumers. Federal regulations governing clinical laboratories under the Clinical Laboratory Improvement Amendments of 1988 (CLIA), which is implemented through a CLIA program in each state, leave the decision regarding the permissibility of direct to consumer (DTC) testing up to state law. Federal regulations require the laboratory to have a "written or electronic request for patient testing from an authorized person" (42 C.F.R. § 1241(a)). However, the regulations do not define "authorized person." Thus it is up to each state to determine who is an authorized person.

In order to determine whether individual states permit DTC testing, individuals listed as the state CLIA representative were contacted and asked whether state law permitted direct ordering of tests by consumers from laboratories and direct delivery of test results to consumers by laboratories. Traditional LEXIS research of state statutes and regulations, attorney general opinions, and in some instances case law, was also undertaken using a variety of search terms. Some state web pages were searched if a government representative suggested it or if other avenues did not yield information.

The search strategy EXCLUDED laws and regulations related to newborn screening, DNA testing for criminal justice purposes, specific health care facilities such as hospitals and outpatient facilities, genetic discrimination, reportable diseases, reimbursement, paternity testing, non-diagnostic testing generally, or waived testing (although some information regarding waived testing was obtained).

Column 2 of the chart ("DTC permitted?") reflects a synthesis of both state statutes and regulations and the opinions of state government officials. In cases where the law is ambiguous, the state official's view was considered to be the final answer; thus states with very similar statutory or regulatory

* These states explicitly exempt certain entities from restrictions on DTC testing. These entities may include laboratories operated by the United States government, public health laboratories, and laboratories maintained exclusively for research and teaching purposes that do not involve patient or public health service.

Source: Genetics and Public Policy Center, Survey of Direct-to-Consumer Testing Statutes and Regulations, June 2007, available at http://www.dnapolicy.org/resources/DTCtable_Feb2008.pdf.

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About the Authors

Michael Rugnetta

Michael Rugnetta is Fellows Assistant at the Center for American Progress and a May 2007 graduate of the University of Pennsylvania with a BA in Political Science and Cognitive Science.

Jonathan Russell

Jonathan Russell is a Spring 2008 Intern at the Center for American Progress and a senior at UCLA majoring in Microbiology, Immunology, and Molecular Genetics.

Jonathan Moreno

Jonathan Moreno is the David and Lyn Silfen University Professor at the University of Pennsylvania and Senior Fellow at the Center for American Progress.

Acknowledgements

Special thanks to Kathy Hudson for her knowledge and support, and to the Genetics and Public Policy Center for use of its research and data.

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1333 H Street, NW, 10th Floor
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