Protecting the Marks of Life: Genetic Testing and the Personalization of Health Care

Michael L. Wilson

American University Washington College of Law

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Mark and Jennifer Jones recently gave birth to Sarah, a seemingly healthy newborn baby girl. During the pregnancy, the Jones’ physician learns that both Mark and Jennifer have relatives who have been diagnosed with cystic fibrosis. Cystic fibrosis is a hereditary disease, and the Jones’s physician recommends that Sarah be tested for the genetic abnormalities that lead to the disease later in life. A small sample of Sarah’s blood is taken and sent to the hospital laboratory. The hospital laboratory analyzes 87 of the most common genetic mutations that can lead to Cystic Fibrosis. Unfortunately the tests conclude that Sarah, although not currently displaying any symptoms, will contract the disease early in her life. After receiving these results, Mark, Jennifer, the Jones’ doctor, and a genetic counselor discuss the results. The doctor reviews all the medical options for a young child who has cystic fibrosis, and stresses how a well-balanced, high-calorie, high-protein diet can help manage many of the symptoms. The Jones’ take the doctor’s advice to heart, and follow his advice in the care and early upbringing of Sarah.1

The above scenario is an example of a genetic test performed through routine medical practice and is presented, along with examples of other beneficial genetic tests, on the United States Department of Health and Humans Services’ (HHS) Personalized Health Care Initiative website.2 This is an example of an ideal scenario where a genetic test can be performed in the course of everyday medical practice, and in this case, is a routine genetic test performed on all newborn infants in many states.3 The benefits of performing a test for cystic fibrosis are undoubtedly of enormous and can lead to careful lifestyle planning and a much improved and prolonged life for the individuals affected.4

In contrast, what if the genetic testing described above were performed with a larger or alternative focus in mind? What if, instead of, or in addition to cystic fibrosis, genetic testing reveals that Sarah will have a sixty percent chance of developing a severe mental illness such as schizophrenia by the time she is thirty? What if genetic testing reveals if she will have a seventy percent likelihood of developing Alzheimer’s by the time she is seventy? What if there is not a cure or well-proven treatment to reduce the effects of these debilitating conditions? Sarah could end up marked for life.

Many of the fears associated with ‘genetic marking’ have played out in science fiction works—primarily based on the fear that genetic testing could predict the potential intelligence, strength, or talents of a particular individual and thus evolve into a new system of discrimination and class distinction.5 The real fear still persists that Sarah, or individuals like her who receive genetic testing, might be discriminated against because of a genetic propensity to develop one or more of a vast number of hereditary diseases in their life.6

Perhaps an individual may exercise a right not to be informed about non-curable genetic diseases or they may decline to know about any genetic abnormalities whatsoever. But can an individual or the parent of an individual decline all testing? If the testing is performed, and the individual declines to be informed of the results, what might happen if the information is somehow disseminated to others, i.e., employers or insurance companies or others who may exploit such information for criminal purposes? What about the possibilities that the genetic diseases are never manifest?

As the practices and procedures of medicine are evolving into a new ideology based on the treatment of the individual pursuant to a ‘personalized health care’ approach, there has never been a more important time to ensure that the identities and private information of those whose genes are being tested are thoroughly respected and safe-guarded.

This article will examine the progress being made toward the development of personalized health care
medical systems based on the use of genetic testing, the current and pending laws required to protect the genetic privacy of every human being, and the effects of these systems on the American health care system in general.

I. The Personalized Health Care Initiative

The Personalized Health Care Initiative will improve the safety, quality and effectiveness of healthcare for every patient in the U.S. By using “genomics”, or the identification of genes and how they relate to drug treatment, personalized health care will enable medicine to be tailored to each person’s needs. Healthcare that is proactive, instead of reactive, gives the patient the opportunity to become more involved in their own wellness.7

On March 23, 2007, former Secretary of Health and Human Services, Mike Leavitt, identified a strategy for achieving gene-based medical care combined with the use of health information technology, something he referred to as “Personalized Health Care” (the Initiative).8 Secretary Leavitt commented that the “initiative has the potential to transform the quality, safety and value of health care for patients in the future.”9 The idea behind this initiative is to take advantage of scientific breakthroughs resulting from the human genome combined with recent technological advancements to exchange and manage medical information. This will result in an increased ability to provide correct treatment to each individual patient at just the right time.10 Secretary Leavitt continued,

Gene-based medicine can help individuals identify their particular susceptibilities to disease while they are well and take effective preventive steps. In the future, it will help detect the onset of disease much earlier, enabling treatment to prevent disease progression, and can help bring about medical products that are tailored more precisely to the needs of each individual... In the future, we’ll understand diseases at a new level...We’ll know them as gene- or molecular-based diseases. And that will give us new kinds of treatments that will be effective for both the very specific condition and the individual patient.11

Upon announcing this important initiative, Secretary Leavitt further identified the implementation steps that the Federal Government was already taking.12 He also emphasized that there is much work remaining to build a system capable of delivering effective personalized health care.13 In identifying the steps that HHS is taking to lay the foundation for the Initiative, Secretary Leavitt established that “HHS is engaged in a broad review of the implications for privacy protection as health information technology is increasingly adopted, including needs for genetic information, and the anticipated effect on the confidentiality, privacy and security of individually identifiable health information.”14

HHS seeks to advance the Initiative through two guiding principles:

Provide federal leadership supporting research addressing individual aspects of disease and disease prevention with the ultimate goal of shaping preventive and diagnostic care to match each person’s unique genetic characteristics.

Create a “network of networks” to aggregate anonymous health care data to help researchers establish patterns and identify genetic “definitions” to existing diseases.15

With or without a federal initiative, it seems inevitable that the practice of medicine is undoubtedly on course to shift from a broad disease prevention and treatment approach to a personalized approach where each individual is treated based on his unique conditions and needs—and, principally, genetics. The advantages the Initiative offers include specific funding for the shift in medical practice and the benefit of Congressional oversight to assure that protective measures are put in place for the protection of privacy.

II. Privacy Concerns

Privacy concerns in genetic testing and health care are not new. As a result of modern advances in genetic testing, individuals fear not only what they might discover about themselves but what others, like employers or health insurance providers, might discover about them.16 Due to recent advancements in technology many of these past fears are perhaps closer than ever before.

The Personalized Medicine Coalition (PMC), “an independent, non-profit group that works to advance the understanding and adoption of personalized medicine for the ultimate benefit of patients,”17 came out in support of the Initiative.18 However, the PMC quickly identified many of the privacy concerns that must be overcome in order for the Initiative to come to fruition.19 In identifying these obstacles, the PMC pointed out that “[s]everal surveys have been conducted to gauge public opinion around the use and protection of genetic information ... [t]he surveys revealed that more than two-thirds of the public is...
concerned about potential misuse of genetic information.” 20 About one-third of the public are of the opinion that if legal protections are not put in place, concerns revolving around privacy could prevent individuals from utilizing or participating in any genetic research.21 The concern is that greater technological advances will be made without equal advances in protective measures.

PMC expounds upon the data gathered in the surveys by identifying the specific concerns of the general population:

PMC believes that all genetic information, including family history, deserves strong and enforceable protections against misuse in health insurance and employment, and PMC supports passage of the Genetic Information Nondiscrimination Act. The benefits of personalized medicine can only be fully realized when the fear of genetic discrimination, and its actual practice, are eliminated from the healthcare system.22

The concerns presented by PMC and many other groups who cautiously support the advancement of personalized medical care are just now being addressed.

III. The Genetic Information Nondiscrimination Act

On May 21, 2008, Congress passed, and former President George W. Bush signed into law, the Genetic Information Nondiscrimination Act of 2008 (GINA), as a solution for many of the concerns associated with genetic testing and personalized health care.23

GINA is modeled after Title VII of the 1964 Civil Rights Act. 24 GINA protects employees, job applicants and family members by prohibiting employers and health insurers from requesting, requiring, or even buying genetic information about them. Additionally, GINA strictly prohibits health insurers from purchasing genetic information for underwriting purposes.25

a. Employment Discrimination

The Equal Employment Opportunity Commission (EEOC) has been charged with promulgating regulations to enforce GINA. In the realm of employment discrimination, GINA brings in a whole new world of enforcement for the Equal Employment Opportunity Commission.

Particular difficulty in enforcing GINA revolves around six loopholes or exceptions to its prohibition on the use or collection of genetic information.26 These include: the inadvertent acquisition of medical information (the so called water cooler exception), health or genetic services, genetic monitoring of the biological effects of toxic substances, Federal or state Family and Medical Leave Acts, compliance, commercially and publicly available records, and law enforcement.27 These exceptions leave significant leeway for an employer to obtain information and use that information to discriminate against an employee. The position of the EEOC, however, is that an employment decision based on genetic testing is in violation of the law. 28 The idea is that any test which purports to predict future disabilities, whether it is accurate or not, is unlikely to be relevant to the employee’s present ability to perform his or her job.29

Another issue is that the EEOC has very little experience with regulating genetic information.30 One notable case took place in 2001, when the EEOC filed its first lawsuit challenging genetic testing. The case took place in the Eastern District of Wisconsin and the parties’ settled for $2.2 million.31 The allegation presented by the EEOC was that the company had “violated the ADA by requiring dozens of employees to provide blood samples in medical exams after they submitted claims for work-related carpal tunnel syndrome.”32 The blood from the medical exams was secretly used in tests to determine if an employee had any possible genetic predisposition for carpal tunnel syndrome.33

With the passage of GINA, the number of employee genetic discrimination cases may rise significantly. Though the number of U.S. companies that are conducting medical tests of employees is dropping, according to surveys by the American Management Association, at least some companies were conducting tests that might be in violation of GINA.34 The American Management Association found that three percent of companies reported medical testing for breast or colon cancer, two percent for sickle-cell anemia, and one percent for Huntington's disease; all of which can have genetic links.35 In addition, these surveys found that fifteen percent of companies collected family medical histories which can reveal hereditary genetic predispositions for specific diseases.36 Employers need to take urgent measures to ensure that employees and all necessary parties are sufficiently instructed on the nondiscriminatory measures within GINA and adequately warned about the consequences of violating GINA.37

b. Insurance Discrimination

GINA fills in the gaps of current federal law such that all health insurers—whether governmental, private, group or individual—would be forbidden to discriminate on the basis of genetic information. Health insurers may not use genetic information to determine eligibility or set premiums. They cannot use genetic information to impose enrollment restrictions or adjust premium or contribution amounts. Health insurers may not require or even request genetic testing or test results, except as necessary for treatment, payment or health care operations. This includes requesting, requiring or purchasing genetic information prior to enrollment.38

Though GINA’s health insurance protection provisions appear to be near watertight, it will be important for lawmakers to keep an eye on insurance companies to ensure they do not navigate around the provisions of GINA. Careful attention will be required to assure GINA’s exceptions are not exploited at the expense of otherwise qualifying individuals.

The passage of GINA is a step in the right direction to protect private information that may be collected in the course of practicing personalized health care, but problems and questions persist. One concern associated with the privacy of genetic information collected in the course of personalized health care is that the information could still find its way into the hands of an employer or insurance provider through one of the loopholes.

IV. Beyond GINA’s Reach

While necessary privacy protections must be, and are being put in place, greater measures must be taken in order to overcome the general public mistrust of genetic testing. The measures must both resolve and dispel public concerns and misconceptions revolving around genetic testing.

a. The Right Not to be Tested Right Not to Know
What about an individual’s right not to be tested or right to not know? While much remains to be seen regarding this particular issue, legal precedent says that “adults are free to refuse even potentially beneficial testing and treatment... children [however,] can be treated without their [parent’s] consent (and over their parents’ refusal) to prevent serious imminent harm.” Currently, there is a great discord among the states as to whether genetic testing is mandatory or can be refused. Some states have no provision on refusing genetic testing, while others provide criminal penalties for parents who refuse to have their children genetically tested.

“The idea behind mandatory newborn screening is a benevolent one—to try to ensure that all children get the benefits of screening for PKU and hypothyroidism, for which early treatment can make a dramatic difference in the child’s well-being by preventing mental retardation.” However, little evidence suggests that it is necessary for a newborn screening program to be mandatory to ensure that children are screened. Rather, evidence suggests that a voluntary program is more effective and reaches a higher percentage of children.

Though it may be difficult to comprehend where such a requirement would be initiated, making adults undergo genetic testing for any reason would appear to be even less effective and may only create more distrust in the system. Further, for adults who are tested, many may wish to exercise a right not to know the results of their tests.

[Given a patient a right to refuse genetic testing, or its results, is justified as vindicating a patient’s autonomy, a ‘basic bioethical principle.’... Though such choices are often justified by a rational interest in remaining free of the psychological harm that might follow from receiving test results, the right to assess that harm and make a choice whether to know lies solely with the patient.

Another consideration is whether the individual being tested, or the parent of the child who is tested, is capable of living with the knowledge that he, or his child, is plagued with a genetic identity that will likely lead to disease. Parents who are carriers of genetic diseases may feel desperate and guilty for passing on a disease to their children. “Studies have shown that knowing that one is at risk for genetic conditions or even learning that one does not have a defective gene strongly affects self-perception and life experiences.” Many individuals, for example, suffer from depression when they learn that they have a gene responsible for causing Huntington disease (HD)—some have gone so far as to commit suicide. “Not surprisingly, many who were at risk [for HD] and discover they do not carry the mutation feel liberated. But, after having lived with a sense of being at risk, some have difficulty adjusting to the knowledge that they will not develop HD.”

At the very least, regulations should be considered to allow individuals, including children (through parents), to exercise the right not to be tested for genetic disorders for which there is no cure or effective treatment.

b. The Use of Genetic Information in Criminal Proceedings

Some concerns loom as to what might be done with the genetic information collected. What if genetic propensities were introduced as evidence in criminal trials? This type of concern is found in the area of neuroscience and is associated with analyzing images of the brain with an MRI.

What if you could do a brain scan and determine to a high probability whether a criminal defendant was a psychopath, with, for example, a 60-70 percent chance of recidivism within five years instead of only 20-30 percent? Would that make a difference to a judge or a jury? What if you were a juror in a capital case in the sentencing phase? Would you want to know if someone is a psychopath or not if it affects his odds of committing another murder? How would we want to use that information? What if you can say that... particular 12-year-olds will be psychopaths while the others won’t be? What do you do with the children you are confident will be psychopaths?

This type of ethical dilemma is further implicated by advancements in genetic testing. What if you could determine that an individual will likely have a propensity (aside from simply having XY sex chromosomes as opposed to XX sex chromosomes) to commit some form of violent crime based on the presence of certain DNA structures? Certainly it provides a basis for taking preventive measures by implementing lifestyle adjustments—much like the result of discovering the likely risk for certain diseases like cystic fibrosis. Should this information ever be presented in a court of law, or disseminated to police officers? These are questions that must be answered by the judiciary, or perhaps more preferably by Congress, before these problems come to light.

c. A Revival of Eugenics?

In the 1927 case Buck v. Bell, the United States Supreme Court upheld a Virginia statute that allowed for the forced sterilization of ‘feeble minded’ and epileptic individuals that were committed to state institutions. Specifically, the hearing procedure conducted before sterilization could be performed was found to satisfy the Fourteenth Amendment because it did not deny equal protection to inmates in state institutions. In his majority opinion, Justice Holmes wrote:

It is better for all the world, if instead of waiting to execute degenerate offspring of crime, or to let them starve for their imbecility, society can prevent those who are manifestly unfit from continuing their kind. The principle that sustains compulsory vaccination is broad enough to cover cutting the Fallopian tubes. Three generations of imbeciles are enough.

What if genetic testing were used today for the same or similar purpose? Though genetic testing is unlikely to lead to forced sterilization of those who are ‘genetically unfit,’ a more realistic possibility is de-facto sterilization where health care is denied to those considered unfit to reproduce. Although this example is extreme, and would be unlikely to arise under GINA, it illustrates the possibility that the results of genetic testing might need to be protected from health care providers who have no apparent need for the information.

V. Further Action Necessary to Implement Personalized Health Care

In additional to privacy concerns, the implementation of an effective personalized health care plan will require other important measures. Many of these measures were laid out by the Personalized Medicine Coalition. These include educating physicians and other health care professionals who will be responsible for treating patients and implementing sufficient technology information systems to aid in the sharing of information.
produced, through genetic testing, the costs of treating certain illnesses and focused on the individual. As more accurate information about a patient is implemented could significantly contribute to the realization of this goal.

Another area of concern is establishing sufficient health care information technology (HIT) systems such that information can be easily communicated between the appropriate parties.

PMC actively supports the creation of a national health information network that enables the interoperable exchange of digital biomedical information securely between a diverse set of stakeholders in the healthcare ecosystem. This infrastructure should also take into account the unique needs of the basic, clinical and translational research community. PMC supports the examination of potential incentive structures to induce investment in HIT by all healthcare providers, from solo practitioners to large hospitals. The implementation of adequate HIT systems raises various concerns. Effective measures, including legislation, must be put in place to ensure the protection of information that is stored or transferred over such systems.

A final concern and stated goal of the HHS Initiative is ensuring the accuracy and validity of genetic tests. Specific verification and testing measures require implementation. Further, penalties for laboratories that continually produce wrong results require contemplation.

Among the remaining steps of implementing the Initiative, great care and concern still needs to be made for privacy. As new measures are taken and new technological feats are reached, careful analysis of privacy protection needs to be made at each milestone. Still, it is appropriate to consider a far reaching privacy measure at this early stage. Namely, Congress should consider creating penalties for any party who sells the genetic information of another or who seeks the genetic information of another to make a profit or with malicious intent.

VI. Personalized Health Care, GINA, and Health Care Reform

President Barack Obama declared his commitment to ensuring that comprehensive health care reform is passed within this year with the stated goal of controlling rising health care costs. Given the President’s determination, carefully assuring that a personalized health care initiative is implemented could significantly contribute to the realization of this goal.

a. Cost Savings Under the Initiative

The Initiative will ensure that the practice of medicine evolves into a practice focused on the individual. As more accurate information about a patient is produced, through genetic testing, the costs of treating certain illnesses and conditions may be greatly reduced. The further use of typical hit and miss treatment strategies could be completely abolished and replaced with more narrow and individualized treatment plans. This necessarily leads to a much more effective and cost efficient system. No more time or money need be wasted in ‘attempting’ to treat individuals.

b. Why GINA Further Necessitates Coverage for All

GINA now makes it illegal for health insurance companies to raise insurance premiums for, or otherwise discriminate against, the class of individuals found to have a genetic propensity for certain illnesses or for the class of individuals who have a family history for certain illnesses. Insurers might choose to quadruple the premiums of or refuse to sell a policy to the third class of individuals—those who are diagnosed with illnesses through routine medical care. Moreover, if a member of this third class “is enrolled in an employer-sponsored group health plan, insurers could raise the rates for everyone in the group.”

Researchers have argued that:

In making such distinctions, GINA is emblematic of this country’s piecemeal and inconsistent approach to health care policy, which makes little sense and leaves many Americans without access to care or in danger of financial ruin if they seek care. Our recent history is replete with examples of similar half-measures in health policy. The Emergency Medical Treatment and Active Labor Act (EMTALA) of 1986 ensures that neither the poor nor the sick can be denied emergency medical treatment, but it leaves those without insurance completely on their own when it comes to follow-up care. So when a patient presents at the emergency room with a myocardial infarction with ST-segment elevation, she will receive a lifesaving coronary-artery stent, but she may not be able to afford Plavix (clopidogrel) — which she must take to avert in-stent restenosis — and may not have access to follow-up care, which might enable her to modify her risk factors for heart disease. Medicare might help if the patient is 1 day past her 65th birthday, but not if she is 1 day shy of it. Medicaid might help if her income is lower than the qualifying threshold in her state, but not if she earns $1 more.

Rather than leave out individuals who may have a genetic predisposition for a disease but only find out through routine medical care, the researchers argue that the better solution is outright prohibition of medical underwriting—a prohibition on setting health insurance premiums based upon any health care information.

Moreover, to ensure that the costs of bad health are shared equitably, all Americans would have to be in the same risk pool. This would mean enacting a health insurance mandate either for employers or, if health insurance could be made affordable, for individuals — and specifying a minimum set of benefits that everyone would be required to have. Given the growing disparity between the cost of modern medicine and the incomes of many Americans, enforcing such a mandate would be difficult. Even with income-based subsidies, an individual mandate could place an undue financial burden on many families. Nonetheless, bringing everyone into the same risk pool is an important long-term goal.
The goals of comprehensive health care reform, as stated by President Obama, may go a long way in ensuring that health care costs for any individual do not escalate out of control.

VII. Conclusion

The practice of medicine is on the verge of moving into a new frontier of personalized health care. The HHS Initiative indeed promises to enshrine a revolutionary approach to the practice of medicine where the focus is on the individual needs of each patient as determined by genetic screening. As the practice of medicine shifts into this new frontier, it is important that the government ensure the protection of each individual’s genetic privacy.

GINA provides a substantial step towards protecting an individual’s private genetic information against the possibilities of being used for employment or health care insurance discrimination. The beginning steps have been taken, but much needs to be done to ensure that each individual maintains his or her rights regarding genetic testing and the right to not be tested or the right to not know test results. Further, it is of vital importance that Congress keeps a close eye on the progress being made within this field and on the efforts of those who would seek to thwart the laws.

As medicine moves into this new realm of personalized health care, it would be a huge advantage for all parties involved, especially the federal government, to take advantage of the opportunities that personalized health care offers. Specifically, as health care reform is considered and established, the cost benefits of carefully implementing personalized health care initiatives cannot be ignored.

The words of Professor Henry T. Greely of Stanford University Law School apply: “although this stuff is really interesting, we need make sure it works before we use it. Let’s make sure we understand what it can and can’t do. And, as a society, we don’t do patience very well.”64 Let us be patient and careful in protecting the rights of all mankind.

2 Id.
4 Id.
5 See, e.g., GATTACA (Paramount Pictures 1997) (depicting genetic engineering that results in the creation of genetically superior human beings who automatically qualify as members of a higher class).
9 Id.
10 Id.
11 Id.
12 Id.
13 Id.
20 Id.
21 Id.
22 Id.
25 Id.
26 EEOC Urged to Address Exceptions to Prohibited Use of Genetic Info, FED. HUMAN RES. WEEK, June 15, 2009.
27 Id.
28 Smith, supra note 24.
29 Id.
30 Id.
31 Id.
32 Id.
33 See id. (referencing EEOC v. Burlington Northern and Santa Fe Railway Co., 440 U.S. 930 (1979)).
34 Smith, supra note 24.
35 Id.
36 Id.
37 Id.
40 Id.
41 Id.
42 Id.
43 Id.
44 Id.
46 Sonia M. Suter, Disentangling Privacy from Property: Toward a Deeper Understanding of Genetic Privacy; 72 GEO. WASH. L. REV. 737, 774 (April 2004).
47 Id. at 775.
48 Id. (citing Lori B. Andrews, A Conceptual Framework for Genetic Policy: Comparing the Medical, Public Health, and Fundamental Rights Models, 79 WASH. L.Q. 221, 241 n. 186 (2001)), (“The incidence of suicide among those with HD is four times that of the general Caucasian population...Studies suggest, however, that the incidence of suicide in those found to carry the HD allele is similar to the incidence in those at risk for HD who have not been tested,”) (citing Janet K. Williams et al.,
Redefinition: Coping with Normal Results from Predictive Gene Testing for Neurodegenerative Disorders, 23 RES. NURSING & HEALTH 260, 261 (2000)).


52 Id. at 207.

53 Id. (citing Jacobson v. Massachusetts, 197 U.S. 11 (1905)).

54 Personalized Medicine Coalition, supra note 17.

55 Id.


59 Id.

60 Id.

61 Id.

62 Id.

63 Id.

64 Greely, supra note 50, at 710.