Spreading Education and Awareness about the Genetic Information Nondiscrimination Act (GINA)

A look into how the lack of knowledge of genetic privacy is affecting genetic testing and an attempt to inform doctors and patients about it

Tag Words: Genetic testing, GINA, Education, Genetic counseling, Healthcare, Prevention

Authors: Melissa Koger, Daniel McGrane, Nicole Salvatore with Julie M. Fagan, Ph.D.

Summary
The Genetic Information Nondiscrimination Act (GINA) was enacted in 2008 to protect patients undergoing genetic testing from insurance companies and employers from using their test results against them. This act is extremely important in giving people confidence in seeking out testing when needed, however many healthcare professionals and patients have no or little idea about this act. We intend to raise awareness of this act by creating an educational video, revising the HIPAA form to include GINA Awareness, publishing information on various websites, and writing letters to medical websites to persuade them to put information about the act on their websites. This way, people who are already visiting these sites could gain information about the act and then be more confident in seeking out the genetic testing that is already available for these conditions.

Video Link: http://youtu.be/ZG8g8L50oTk

Genetic Discrimination (MK)

Genetic testing is an invaluable tool used to help diagnosis and allow for effective treatment of genetic illnesses. Unfortunately, most individuals are hesitant to undergo genetic testing due to discrimination, psychological changes, and cost. The public needs to be more aware of the protections that are in place to alleviate these fears. In order to educate the public, one must also educate the physicians.

Benefits of Genetic Counseling (NS)

Genetic Counseling is a clinical service used to identify genetic predispositions to assess risk for future diseases, diagnose patients currently affected with hereditary conditions and potentially provide personalized treatment options for people and assist in reproductive decisions. It has an educational aspect and a therapeutic aspect, where the genetic counselor’s job is to educate the patient on their genetic predispositions and then possibly help in establishing a treatment plan.¹ There are currently about 1500 tests that are available to patients that genetic
counselors can offer. These tests include prenatal tests for Down Syndrome and other trisomy’s, adult-onset disease testing like for Huntington’s and Alzheimers, and cancer gene tests for BRCA 1 and 2 for breast cancer and Lynch Syndrome associated genes for Colorectal cancer.

The idea behind genetic counseling is to allow for preventative care versus palliative care. Studies have shown that preventative care is considerably less money than treating the disease. With many genetic disorders with childhood-onset, early intervention is possible to slow or stop the progression of the disease. People with a predisposition for cancers can undergo surgeries, like a mastectomy or colectomy, that can near-eliminate their risks for developing that type of cancer. For hypertrophic cardiomyopathy, there is a 40-60% detection rate with testing. This means that more than 50% of people with the predisposition for this could undergo preventative care to save their lives. With this condition, you don’t know you have it until it is usually too late.

More times than not, patients are referred to genetic counselors by their primary care physician, oncologist or obstetrician. This referral system is based off the presumption that these doctors should know the signs of genetic conditions or inherited predispositions in families. In Freedman’s and Wilderoff’s study of US physicians’ attitudes toward genetic testing, it was found that many physicians have a great deal of uncertainty in regards to the availability of genetic services, guidelines for these services, insurance discrimination, the utility of genetic results and which health care professionals are able to perform genetic testing. This confusion could lead to physicians misinforming their patients about testing or not offer it to them at all.

Because genetic testing is such a controversial issue right now, there is a large spectrum of attitudes towards genetic counseling. “Insurance discrimination has been reported as the single most important reason why patients choose not to undergo genetic testing,” This is understandable of patients, but if physicians also do not understand the insurance protections for genetic testing, then the rate of genetic testing would further diminish. “Many people are afraid that their genetic information will be used against them and are unwilling to participate in medical research or be tested clinically, even when they are at substantial risk for serious disease,” says Francis Collins, the current director of the National Institute of Health. In Freedman and Wilderoff’s study, it was found that more than 50% of physicians in the US believe results from a genetic test are hard to keep confidential and a majority of them believe a positive result on a test will affect insurance coverage. It was also found that one-third of physicians don’t believe genetic counseling services are regularly available to them. This again lowers the amount of patients being referred. If the education, awareness, and public understanding of genetic counseling improved, more patients would be willing to undergo testing.

**Personalized Medicine Coalition (PMC)** (MK)
According to Huizenga, Lowstuter, Banks, Lagos, Vandergon & Weitzel, education of non-cancer genetic professionals seems to be of utmost importance to increase the number of patients to be genetically tested. GINA and health care laws in general need to be incorporated into the curriculum for the continuing medical education requirements for physicians.

One example of a seminar was given by the Personalized Medicine Coalition (PMC). The PMC presented a seminar at the American Association of Clinical Chemistry Annual Meeting for continuing medical education credit for healthcare professionals discussing personalized medicine. Personalized medicine is defined as “using molecular analysis to achieve optimum medical outcomes in the management of a patient’s disease or disease predisposition, personalized medicine promises to introduce a new standard of healthcare.” DNA analysis for genetic testing is a pivotal instrument for personalized medicine.

Generally speaking, physicians diagnose a disease based on the patient’s symptoms and possibly some routine blood work, and then select a drug that might help. If symptoms persist, then the drug may be switched and possibly switched again, while the disease increases in severity (Fig. 1). This is known as reactive medicine, which we currently use and is ineffective. It should also be noted that by using this protocol, only the symptoms are being treated and not the disease itself.

---

**Figure 1. Source: Abrahams, 2007**
On the other hand, personalized medicine uses genetic testing before diagnosis and asymptomatic (Fig. 2) or immediately after (Fig. 3) to help prevent the onset of the disease or allow administration of the “right” drug, respectively. Preventive medical care determines an individual’s predisposition from family history and then orders a genetic screening to verify inheritance (Fig. 2). If an increased risk is reported, then preventive measures and monitoring can be implemented for early detection or to impede the progression of the disease (Fig. 2).

Efficient medical care utilizes DNA analysis promptly after diagnosis in order to prescribe the right medication or treatment depending on the particular genetic markers that were mutated (Fig. 3). In most cases, this protocol will treat the disease at its source and not just the symptoms, leading to a decrease in disease severity (Fig. 3). In fact, 34% of women with breast cancer had a reduction in chemotherapy when they were genetically tested for specific markers just prior to treatment. Therefore, not only is genetic testing used as a preventative measure, but also specifies a correct course of treatment. Some viable targets are listed in Table 1. There are seven different treatments for breast cancer alone and even in the cancer’s recurrence; there are two different methods for chemotherapy depending on the genes affected.

<table>
<thead>
<tr>
<th>Variable Target</th>
<th>Therapy/Prevention</th>
<th>Disease</th>
</tr>
</thead>
<tbody>
<tr>
<td>BCR-abl; c-KIT</td>
<td>Gleevec/Imatinib</td>
<td>Cancer/Chronic Myelogenous leukemia</td>
</tr>
<tr>
<td>BRCA 1/2</td>
<td>Surveillance; tamoxifen; prophylactic surgery</td>
<td>Breast and ovarian cancer</td>
</tr>
<tr>
<td>EGFR</td>
<td>Tarceva, Iressa</td>
<td>Lung cancer</td>
</tr>
<tr>
<td>Estrogen receptor</td>
<td>Tamoxifen</td>
<td>Breast Cancer</td>
</tr>
<tr>
<td>HER-2/neu receptor</td>
<td>Herceptin/Trastuzumab</td>
<td>Breast Cancer</td>
</tr>
<tr>
<td>PML-RAR alpha</td>
<td>Tretinoin/All trans retinoic acid</td>
<td>Acute Myelocytic Leukemia</td>
</tr>
<tr>
<td>Oncotype DX: 16-gene profile</td>
<td>Chemotherapy protocols</td>
<td>Breast Cancer recurrence</td>
</tr>
<tr>
<td>MammaPrint 70-gene profile</td>
<td>Aduvant chemotherapy</td>
<td>Breast Cancer recurrence</td>
</tr>
<tr>
<td>TPMT</td>
<td>Mercaptopurine</td>
<td>Acute Lymphocytic Leukemia</td>
</tr>
</tbody>
</table>
TruGene®-HIV 1 Genotyping | Anti-retroviral drugs | HIV virus drug resistance

Table 1. Source: Abrahams, 2007

The PMC discussed their policy initiatives including, 1) the importance of educating patients on genetic privacy as a well-informed patient will make proper treatment decisions; 2) FDA regulations of market approved personalized drugs; 3) reimbursement for genetic testing and incentives for insurance companies; 4) Research & Development (R&D) business incentives to develop personalized medicine; and 5) an investment of government funding for research. The PMC promotes education to multiple agencies to ensure the public’s awareness in genetic information as well as their health and well-being.

Hypertrophic Cardiomyopathy: A Case Study (DM)

To look in depth at a particular condition, Hypertrophic Cardiomyopathy (HCM) is a condition that offers particular promise as a target for genetic screening and personalized medicine. HCM is the most common genetic heart disorder affecting 1 in 500 and is the leading cause of sudden heart failure in the young. This condition is caused by an abnormal thickening of heart muscle potentially leading to degraded function and, in some cases, sudden loss of function. The clinical progression is extremely varied and can range from lifetime asymptomatic status to progressive exhaustion to arrhythmia to death. HCM can be extremely difficult to diagnose with only phenotypic tests and early symptoms can be misdiagnosed, especially in the young, as more common and much less fatal conditions such as asthma or bronchitis. This lack of early diagnosis can result in sudden heart failure as the “sentinel” event. The overall prognosis for sufferers of HCM is only a 25% of reaching 75 and 1% mortality per year on average. HCM is a heterogeneous condition with over 1400 different genetic mutations over 9 different alleles identified as a cause of HCM. The condition is passed down in an autosomal dominant pattern.

HCM is an ideal condition to diagnose with genetic testing. As mentioned before, it can be difficult to diagnose and can effectively be asymptomatic before causing sudden heart failure. In addition, the autosomal inheritance pattern creates family histories that can be used to target testing on high risk individuals. The well-defined set of genes associated with the condition also creates a well-studied target for genetic testing. Identification of individuals with HCM causing alleles allows preventative targeted treatment. Even in individuals without symptoms, preventative treatment based solely on genetic predisposition has been shown to be beneficial. Treatment with angiotensin-converting enzyme on asymptomatic genetic carriers found a significant decrease in rate of death by the endpoint of the test. Genetic testing can also be used to decide on a course of treatment. Not all alleles for HCM are equally dangerous and some require more extreme preventative measures. The MYH7-R453C mutation, for instance, has been linked to 50% mortality by the age of 40. Some HCM causing mutations have been
identified as so dangerous that a preventative cardverter defibrillator is recommended due to a high chance of sudden cardiac failure. The targeted screening at the 9 alleles responsible for HCM combined with treatment targeted for the particular disease-causing allele offers an example of the promise of personalized medicine and genetic screening.

Genetic testing for HCM is also useful for identifying individuals with a family history who are not carriers of a disease carrying allele. In the pre-genetic screening era, patients with a family history of HCM were recommended to have an echocardiogram every 3 years for the duration of their lives to identify the possible appearance of symptoms. A genetic test could eliminate the half of patients who have a family history but who were not carriers of any disease carrying allele and do not suffer from increased risk of HCM. Identifying patients not needing care would improve the quality of life of these patients, both financially and emotionally, while also conserving medical resources. The cost of ECMs is $650 per test with tests being administered every 3 years. Meanwhile, a genetic test would currently cost only $3000 with the cost of genetic tests likely to decrease greatly in coming years given past trends.

**Fears among Consumers (MK)**

*Genetic Discrimination (MK)*

Many people are fearful of being discriminated against by their health insurance and employer due to their genetic information. The exact prevalence of genetic discrimination is difficult to determine. However, the term has become a household name which is propagated by social stigmas thus, ensuring that the fears remain. They fear that if diagnosed with a life-long ailment, their insurance companies will deny coverage as it will be an added expense to pay for treatments or may raise premiums in order to save a few pennies. However, early detection and prevent and reduce financial costs long term and save insurance companies money. On the other hand, “avoidance of genetic tests because of discrimination fears [can] ultimately…cost thousands of dollars for additional detection tests and treatment.” Employers may deny job placement or dismissal based on genetic predisposition. This will lead to an endless spiral. With no health insurance, you will have to pay all medical expenses out-of-pocket, but there is no household income because you do not have a job.

The good news is insurance companies cannot use one’s genetic information against them. There are 28 states that have passed genetic privacy legislature and there are also federal laws, especially the Genetic Information Nondiscrimination Act of 2008 (GINA) established to protect individuals from discrimination.

*Cost (MK)*

Currently, cost ranks number one on why people do not get genetically tested. The cost varies depending on the number of genes screened. For example, the BRCA1/BRCA2 gene test for breast cancer can range from $300-$3000. This may be a financial hardship for some
families, but if the results come back with an increased risk of being diagnosed with breast cancer, you can save upwards to $100,000 cost of late-stage treatments.\textsuperscript{19}

\begin{center}
\begin{tabular}{|c|c|c|c|}
\hline
\textbf{Stage at Diagnosis} & \textbf{Initial} & \textbf{Continuing} & \textbf{Terminal} \\
\hline
Local & $7,993 & $151 & \\
Regional & $10,481 & $44 & $17,794 \\
Distant & $12,461 & $14,468 & $11,958 \\
\hline
\end{tabular}
\end{center}

\begin{center}
\begin{tabular}{|c|c|c|c|}
\hline
\textbf{Stage at Diagnosis} & \textbf{Initial} & \textbf{Continuing} & \textbf{Terminal} & \textbf{Total} \\
\hline
Local & $15,759 & $3,907 & $19,665 \\
Regional & $20,406 & $641 & $34,794 \\
Distant & $22,127 & $9,422 & $92,547 \\
\hline
\end{tabular}
\end{center}

*Treatment phases are initial (first 6 months); terminal (final 6 months before cervical cancer-related death); and continuing (of indefinite duration).

\textbf{Figure 4: Source: Helms & Melnikow, 1999}

Early detection is vital in preventing a tremendous financial burden as well as averting the agony of losing the life of a loved one. According to Figure 4, early detection of cervical cancer can save approximately $73,000 compared to late-stage diagnosis depending on the length of the Continuing Treatment Phase.\textsuperscript{20} By testing positive for cervical cancer, Pap smears can be collected in higher frequency to identify minor abnormalities before they become malignant.

**Psychological Changes (MK)**

Many individuals may not want to know their percentage chance of being genetically predisposed to Huntington’s Disease or Cystic Fibrosis due to the stigma associated with genetic testing, implying it is terminal. Not many people desire knowledge of their death. According to Marteau and Croyle\textsuperscript{21}, people are more likely to seek genetic testing if the disorder is curable. 10% receive DNA analysis for Huntington’s Disease which has no treatment; 50% for breast cancer which has some possibility of prevention and treatment; and 80% get tested for familial adenomatous polyposis, which is completely curable.\textsuperscript{21} On the other hand, studies have shown there is a slight increase in distress within the first six months after testing positive, but have a decrease in distress to pretest levels at 12 months.\textsuperscript{22}

Initially, genetic testing is perceived as formidable, which would give many sleepless night, when, in fact, it is the complete opposite. The distress changes for carriers or non carriers over 12 months after being genetically tested for Huntington’s Disease from five studies reviewed by Broadstock, Michie, & Marteau.\textsuperscript{22} All five studies had no change in distress from 0
to 6 months and two reported a decrease at 12 months for carriers. The non carriers experienced an immediate decrease within the first month, but returned to the baseline in the 3 to 6 month range.

**GINA Introduction (NS)**

GINA is the Genetic Information Nondiscrimination Act. It was signed in 2008 by President Bush and was in full effect by the end of 2009. The act involves protecting patients from genetic discrimination through employers and insurance companies. The act prohibits use of an individual’s genetic information for insurance eligibility or premium rates, prohibits insurers or employers from requiring genetic testing and prohibits use of genetic information by employers in decisions like hiring, firing and assignments. These guidelines apply to family medical history, carrier testing, prenatal genetic testing, predictive testing (i.e. BRCA testing) and tumor analysis. GINA does not provide protections for conditions that have already manifested in the patient, life, disability or long-term insu...ers, employers with fewer than 15 employees, military personnel and federal employees. With the new health reform, it is expected that groups not covered by GINA will be covered under the new legislation, like ones with already manifested genetic conditions. The law also does not require that health-care professionals counsel patients on the implications of this act. This is often problematic because patients will not want to get tested because of insurance fears and doctors aren’t mentioning the protections of GINA to these patients.

The main reason for this act being passed was for the fear that as genetic technologies got more advanced and we could test for more conditions, genetic discrimination would be a serious issue in the United States. Genetic counselors report that patients at one point were paranoid, not wanting to call from work, not wanting to give their address, etc. Genetic counselors also note that many people get the notion of genetic discrimination from the media mostly. They also found that patients seeing prenatal or pediatric genetic counselors knew less about genetic discrimination than patients seeing an adult-onset disease counselor.

**Raising GINA Awareness**

**Advertisement Failures (MK)**

Recently, there have been direct-to-consumer advertisements for at home genetic testing kits that may have increased awareness of getting screened, but there have been several misconceptions within these advertisements. According to *Limitations of Direct-to-Consumer advertising for Clinical Genetic Testing*, direct-to-consumer advertisements have educational value but “it is limited by the complexity of genetic information, the complicated social context, and the lack of consensus regarding the desirability of testing.” Not one answer is black and white, but filled with multiple shades of grey. A positive result does not mean you will definitely develop the disorder, but environmental factors can influence the outcome, good or bad. This is difficult to express to patients in layman’s terms, let alone in an advertisement.
Advertisements might allude to cures for the disease by using metaphors for genes such as a “code” that can easily be repaired. Phrases such as “save your baby’s life” or “testing for BRCA will dispel fear” are inaccurate and misinforms the public of the complexity of genetic testing. At home testing also completely bypasses the healthcare professionals, who are more equipped to explain the results and its implications versus the description on the back of the box. They can also refer the individual to seek genetic counseling.

These advertisements are very one-sided and either promotes fear or hope; both of which are misleading. They need to be more informative about the added benefits as well as the increased risks associated with genetic testing. Current direct-to-consumer advertisements do not mention genetic discrimination or the psychosocial risks involved and especially GINA. The poster attached can provide a simple and yet effective way of communicating the pros and cons of genetic testing and GINA.

According to Huizenga, Lowstuter, Banks, Lagos, Vandergon & Weitzel, education of non-cancer genetic professionals seems to be of utmost importance to increase the number of patients to be genetically tested. GINA and health care laws in general need to be incorporated into the curriculum for the continuing medical education requirements for physicians.

**National Peripheral Arterial Disease Public Awareness** (MK)

Public awareness is essential to informing clients of various diseases as shown by the countless commercials for breast cancer, Crohn’s Disease, lung cancer, etc. In regards to hypertension, two surveys conducted from 1976 to 1980 (pre-education) and 1988 to 1994 (post-education), awareness increased 43 percent and treatment increased by 71 percent with 29 percent of the population having their hypertension under control. From 1983 to 1995, cholesterol testing increased from 35 to 75 percent, saving 70-80 million people. Hirsch et al. stated that “[e]ducation of the public, linked to education of clinicians, can result in real world health gains.” The same mentality can be applied to genetic testing. Awareness begins with physicians to better educate their patients for the betterment of their health and longevity.

**Treatment-Focused Genetic Testing (TFGF)** (MK)

Newly diagnosed women with breast cancer were given either a standard informational pamphlet or a decision aid pamphlet and their knowledge and desire to be tested was evaluated. The decision aid pamphlet resulted in the group being more informed, increased knowledge, and a decrease in decision conflict. The pamphlet was one-page bifold material that contained information of TFGF, its purpose, why a woman might consider TFGF, what it entails, and the positive and negative outcomes and their implications. The effectiveness of the material was evaluated by the group of women. It was concluded that the amount of information given was not too overwhelming, was clearly laid out, and had high readability due to the question and answer format.
Educational materials should be presented in a doctor’s office for a more effective way to deliver information to the patient. Even though, the decision aid pamphlet worked well in increasing knowledge, while decreasing fears on its own; the women preferred to receive the TFGF pamphlet during a face-to-face consultation. 26 This would help facilitate a discussion between the patient and the doctor, who would reinforce the material or clarify any misconceptions. 26 That is, if the physician was also well aware of the topic to provide sound advice.

**Sun Awareness Poster** (MK)

The effectiveness of three posters, illustrating sun protection and skin cancer were evaluated in a dermatology office. The most observed ad was the sex appeal ad (67.8%), followed by the informative ad (49.2%), then the parental ad (35.8%) 27 (Fig.5). It was also concluded that posters are not strong on their own, but works well in conjunction with physician’s advice. 27 The posters initiate the conversation 27, which was also concluded previous by Meiser et al. 26

![Sun Awareness Poster](image)

**Fig. 5: Source: Jung, Senthilselvan, & Salopek, 2010**

**Health Insurance Portability and Accountability Act (HIPAA)** (MK)

There are consent clauses written in the HIPAA form to protect your medical records, such as “We will not make any other uses or disclosures of your health information unless you sign a written ‘authorization form.’ The content of an ‘authorization form’ is determined by federal law.” A revision of the form can concisely describe GINA as well as eliminate fears of discrimination. It may read, “Under the Genetic Information Nondiscrimination Act of 2008 (GINA), we cannot disclose genetic information and family history to your health insurance
Recent Alterations to GINA and HIPAA (DM)

While GINA and HIPAA are currently the main laws that govern the privacy of health information, including genetic, several alterations and extensions to their provisions have occurred in the more recent Patient Protection and Affordable Care Act, commonly referred to as Obamacare, and the HITECH Act.

The PPACA through its limitations on criteria for premium setting and healthcare denial provides some of the same protections as GINA but does not fully address the need for protection from genetic discrimination. When it fully takes effect in 2014, it creates specific sets of criteria that individual and small group health insurance companies are allowed to adjust premiums based on. As genetic predisposition is not included among these criteria, PPACA increases the protection on sufferers of genetic conditions for individuals covered by these plans. These protections do not extend to all forms of insurance and self-insured group or large group health insurance is excluded from coverage by this regulation. 28 PPACA does forbid exclusion based on preexisting conditions among these two varieties of health insurance. It does not, however, forbid higher premiums on individuals with preexisting conditions, leaving possible genetic discriminations that must be covered by GINA.

The Health Information Technology for Economic and Clinical Health Act, the HITECH Act, which was passed in 2009 as part of the American Recovery and Reinvestment Act is a more sweeping reform of medical information laws. This act was extensive and aimed at improving the quality of healthcare information technology. 29 The bills stated intention was to:

“(1) ensures that each patient’s health information is secure and protected, in accordance with applicable law;

(2) improves health care quality, reduces medical errors, reduces health disparities, and advances the delivery of patient- centered medical care;

(3) reduces health care costs resulting from inefficiency, medical errors, inappropriate care, duplicative care, and incomplete information;

(4) provides appropriate information to help guide medical decisions at the time and place of care;

(5) ensures the inclusion of meaningful public input in such development of such infrastructure;
(6) improves the coordination of care and information among hospitals, laboratories, physician offices, and other entities through an effective infrastructure for the secure and authorized exchange of health care information;

(7) improves public health activities and facilitates the early identification and rapid response to public health threats and emergencies, including bioterror events and infectious disease outbreaks;

(8) facilitates health and clinical research and health care quality;

(9) promotes early detection, prevention, and management of chronic diseases;

(10) promotes a more effective marketplace, greater competition, greater systems analysis, increased consumer choice, and improved outcomes in health care services;

(11) improves efforts to reduce health disparities.”

In accordance with fulfilling goals 1 and 9, the HITECH act contains provisions that will significantly alter and extend both HIPAA and GINA. Foremost is formation of the HIT Standards Committee charged with crafting regulations to control health information technology for the purpose of creating a specific consistent harmonious set of laws that reconcile GINA, HIPAA, PPACA, and all previous laws governing health information technology. While this board was charged with delivering regulations to do so within 90 days of passage of the law in 2009, the board has still not done so as of Nov 6, 2012 and the final set of regulations that will encompass GINA and HIPAA are still under review and unreleased.

There are known alterations to current law. Several interim rules were released on Feb 10, 2010 including the HITECH Act Enforcement Interim Final Rule. This rule added criminal fines to breaches of health information privacy, including in cases where the violation could not be shown to have done any harm or if the breaching entity was not aware that they were violating the law. Which were cases not previously covered. While the unfinished nature of these new regulations limits the ability to know exactly the end state of healthcare information regulation, it also provides a unified board crafting the new standards for HIPAA forms that can be appealed to in order to draw up clearer and more patient friendly standards for providing information about genetic discrimination standards.

Community Action: Public Awareness Campaign

The main mission is to educate your physicians and staff and thus your patients about the protections of GINA and ease genetic-testing associated fears in order to amplify the number of genetic tests for the betterment of the patient’s health and well-being.

Step One: Physician Education (MK)
The first initiative should be educating your physicians and staff about the benefits and the few limitations of GINA. The National Coalition for Health Professional Education in Genetics has published a short, six-page document summarizing GINA without the use of excessive legal jargon for easier comprehension. “A Discussion Guide for Clinicians” details how physicians can better communicate with their patients about the advantages of GINA. This document can be distributed to your staff such as any other internal memo as a hard copy or electronically.

The second phase involves formal continuing education for your doctors and physician’s assistants, similar to the PMC seminar discussed in detail earlier. Partners HealthCare Center for Personalized Genetic Medicine, the Harvard Medical School, and the Harvard Business School presents their annual Personalized Medicine Conference in Boston, MA on November 28-29, 2012. The 8th annual conference will increase “…knowledge to patient care by using genetic and genomic information in diagnosis, prognosis and treatment. The goal of personalized medicine is to provide the right diagnosis and treatment to the right patient at the right time at the right cost.” This will enable your doctors to receive official education from healthcare professionals from around the world. Appendix A displays a list of future conferences in the United States with their corresponding event website.

Step Two: Patient Education (MK)

As education of the patients is closely correlated to the education of the clinicians, it is important to increase doctor-patient communication. This can be done indirectly by the patient initiating the genetic testing conversation. There are an abundant number of commercials and pamphlets advertising for new marketable drugs, but they advise that they should consult with their doctor. This type of advertising helps commence a medical discussion. An informational poster and decision aid pamphlet will be used as a conversation starter that can be posted in the waiting room or in the individual examination rooms. The poster has been provided, see Appendix B. The pamphlet should depict accurate information about GINA and genetic testing, including the pros and the cons in a question and answer format. This is something different from current advertisements, which promote “cures” through genetic testing and other fallacies. It allows the patient to gain both perspectives that will make them curious enough to ask their doctor questions; who will be better equipped to answer them with their previous education.

The second way to provide information to your patients is through the Health Insurance Portability and Accountability Act (HIPAA) form. There are consent clauses written in the HIPAA form to protect your medical records, such as “We will not make any other uses or disclosures of your health information unless you sign a written ‘authorization form.’ The content of an ‘authorization form’ is determined by federal law.” A revision of the form can describe GINA as well as eliminate fears of discrimination. See Appendix C.

Step 3: General Public Awareness (NS)
The last part of this plan is to get this information out to the general public through media outlets such as magazines, blog-like websites, and YouTube. To encourage popular magazines, such as *Prevention Magazine*, to publish articles about GINA would assist in creating widespread knowledge about this act. This method would reach people who are not necessarily going to the doctor’s office right now, but may need to know about this in the future or can even pass it on to a friend who may have no idea. We have written and suggested a feature article on GINA and then also asked to try to always mention GINA whenever they have an article about some sort of genetic condition or testing in the future. *Prevention Magazine* is one of the largest circulated magazines in the world and is all about living healthy and preventing health problems. We chose to write this magazine because the purpose of genetic testing is to prevent disease, and GINA is helping to promote genetic testing.

We also are posting on websites about genetic testing laws, such as Yahoo Answers, a very common site for people with questions of all sorts to look for answers, and also responding to our question to give people the correct answers. This way, if someone asks a similar question, they will be referred to our questions and see the answer that we have provided. This is another simple way to spread information about GINA. This site is also frequented by younger people, so to start giving out this information early will hopefully change attitudes about genetic testing earlier on in life. Lastly, we hope to create an informative video to post on Youtube as another media outlet for people to see. Lastly, we will send out a letter to our senators requesting that they change the HIPAA form to include information regarding GINA and genetic testing.
References


http://www.partneringforcures.org/2012/2012-registration.php

http://www.personalizedmedicinecoalition.org/events


<http://www.govtrack.us/congress/bills/111>
Appendices

Appendix A: Upcoming Genetic and Personalized Medicine Seminars.

<table>
<thead>
<tr>
<th>Date</th>
<th>Event</th>
</tr>
</thead>
<tbody>
<tr>
<td>November 13, 2012</td>
<td>Personalized Medicine Awareness Day</td>
</tr>
<tr>
<td></td>
<td>Hyatt Hotel</td>
</tr>
<tr>
<td></td>
<td>Greenville, SC</td>
</tr>
<tr>
<td></td>
<td>SCBIO (Organizer)</td>
</tr>
<tr>
<td></td>
<td><strong>Event Website</strong></td>
</tr>
<tr>
<td>November 27, 2012</td>
<td>Board of Directors Meeting</td>
</tr>
<tr>
<td></td>
<td>Hotel Commonwealth</td>
</tr>
<tr>
<td></td>
<td>Boston, MA</td>
</tr>
<tr>
<td></td>
<td>500 Commonwealth Avenue</td>
</tr>
<tr>
<td></td>
<td>Personalized Medicine Coalition (Organizer)</td>
</tr>
<tr>
<td>November 28-30, 2012</td>
<td>Partnering for Cures</td>
</tr>
<tr>
<td></td>
<td>Grand Hyatt</td>
</tr>
<tr>
<td></td>
<td>New York, NY</td>
</tr>
<tr>
<td></td>
<td>FasterCures (Organizer)</td>
</tr>
<tr>
<td></td>
<td><strong>Event Website</strong></td>
</tr>
<tr>
<td></td>
<td>Omni Shoreham Hotel</td>
</tr>
<tr>
<td></td>
<td>Washington, DC</td>
</tr>
<tr>
<td></td>
<td>National Pharmaceutical Council (Organizer)</td>
</tr>
<tr>
<td></td>
<td><strong>Event Website</strong></td>
</tr>
<tr>
<td>December 3, 2012</td>
<td>Improving the Efficiency and Effectiveness of Genomic Science</td>
</tr>
<tr>
<td></td>
<td>Translation: A Workshop</td>
</tr>
<tr>
<td></td>
<td>Beckman Center</td>
</tr>
<tr>
<td></td>
<td>Irvine, CA</td>
</tr>
<tr>
<td></td>
<td>Institute of Medicine (Organizer)</td>
</tr>
<tr>
<td></td>
<td><strong>Event Website</strong></td>
</tr>
<tr>
<td>December 4-6, 2012</td>
<td>The Cancer Genome Summit</td>
</tr>
<tr>
<td></td>
<td>Boston, MA</td>
</tr>
<tr>
<td></td>
<td>Hanson Wade (Organizer)</td>
</tr>
<tr>
<td></td>
<td><strong>Event Website</strong></td>
</tr>
<tr>
<td>January 28-29, 2013</td>
<td>Personalized Medicine World Conference</td>
</tr>
<tr>
<td></td>
<td>Computer History Museum</td>
</tr>
<tr>
<td></td>
<td>Mountain View, CA</td>
</tr>
<tr>
<td></td>
<td>Silicom Ventures (Organizer)</td>
</tr>
<tr>
<td></td>
<td><strong>Event Website</strong></td>
</tr>
<tr>
<td>August 5-7, 2013</td>
<td>International Conference on Predictive, Preventive and Personalized</td>
</tr>
<tr>
<td></td>
<td>Medicine &amp; Molecular Diagnostics</td>
</tr>
<tr>
<td></td>
<td>Chicago-North Shore OMICS Group (Organizer)</td>
</tr>
<tr>
<td></td>
<td><strong>Event Website</strong></td>
</tr>
</tbody>
</table>

Source: Personalized Medicine Coalition, 2012; OMICS Group, 2012
Appendix B: GINA Poster

Don’t be in the dark about genetic testing

Genetic Information Nondiscrimination Act (GINA) can protect you

Pro:
- Family history and genetic testing are protected from your employer and health insurance
- Can provide insight to genetic disorders that are asymptomatic.
- Distress minimizes within the first month of results.
- Genetic testing isn’t a “cure” but it allows for early preventative care and detection.
- The cost is insignificant. $3000 is better than over $100,000

Con:
- GINA doesn’t protect against life, disability, and long term insurance.
- Cannot protect you once you are diagnosed with the disease.
- Cost of the depends on a multitude of factors, including how many genes are tested and the number of people tested and can range from $300-$3000, all out of pocket.

Consult your doctor if you should be genetically tested.

Be aware that everything isn’t 100%, but a little knowledge is better than no knowledge.
Appendix C: Letter to Senators Robert Menendez and Frank R. Lautenberg of NJ Regarding HIPAA Forms

We are writing you today in the hopes of your amending the legislation American Recovery and Reinvestment Act of 2009 § 1, 111 U.S.C. (2009) in regards to the HIPAA form requirements, which can help alleviate some of the fear of genetic discrimination by creating awareness about GINA, or the Genetic Information Nondiscrimination Act. This is a very important piece of legislation that prevents genetic discrimination from insurance companies and employers, but when you try asking around about it, many people have no idea about it. This ignorance is what is allowing the fear of genetic discrimination to persist, even now when people are protected from it. As you may know, genetic testing is on the rise, now with 1500 tests available to consumers. The tests vary from looking for the BRCA mutation for breast cancer, to looking for Down syndrome in pregnant women, to looking for markers to create a personalized treatment plan. The purpose of this letter is not to create awareness about testing however. We believe most people know about the variety of testing and make their own decision if properly informed about the costs and benefits. However, many Americans are misinformed and do not seek treatments due to fears about genetic discrimination.

We are seniors at Rutgers University with our professor, Julie M. Fagan, and it saddens us to see people turn away testing that could be so beneficial to them. Through research, we have even found that many doctors, themselves are not aware of the protections offered by GINA. We think a good solution to this problem would be to put a description of GINA on HIPAA forms. This way when people are actually at the doctor’s office, they will know to ask about it. There is of course the problem of whether or not patients actually read the HIPAA form. In some of the doctor offices we contacted, there has been a shift to having paperwork digitalized for the convenience of the patient to view at their leisure.

Because each doctor’s office has their own HIPAA form and there is no real standard form, we have thought of two options. Below, is a paragraph that could be added on to any HIPAA form, so that offices would not need to change their entire form. Also, we have attached a template HIPAA form that could be adopted by practices that already have included information about GINA.

“Under the Genetic Information Nondiscrimination Act of 2008 (GINA), we cannot disclose genetic information and family history to your health insurance provider and employer without your written consent. This is not limited to life, disability, and long term insurance. Coverage may change due to diagnosis of illness.”

We think this is an issue that needs to be address in the Senate because this problem is a nationwide issue. We hope that my letter to you today has helped you see the need out there for awareness about GINA. We are trying to do our part in the New Brunswick area by creating posters for doctor’s offices to display and providing doctor’s with a list of conferences in
personalized medicine that cover GINA for their continuing education credits. We have even created a PSA on GINA so that more of the public will get a chance to learn about GINA, which you can view at http://youtu.be/ZG8g8L50oTk. But we believe that the most important group of people to target with this awareness is the patients who are already in the doctor’s office for one reason or another. If they see this small paragraph on the form they are signing, perhaps it will change their perspective on testing and they will take advantage of testing, or even pass it on to a friend who may be on the fence about genetic testing. We need your help in getting this into legislature so that this becomes a requirement for HIPAA forms.

There is no reason to fear genetic testing anymore because everyone can be protected under GINA. Testing can lead to prevention of conditions, which can help out a lot of people of this great nation. Thank you for your time and consideration.

Sincerely,

Nicole Salvatore
nicsal@gmail.com
School of Environmental and Biological Sciences
Rutgers, The State University of New Jersey

Daniel McGrane
dgrane@gmail.com
School of Environmental and Biological Sciences
Rutgers, The State University of New Jersey

Melissa Koger
mkoger89@gmail.com
School of Environmental and Biological Sciences
Rutgers, The State University of New Jersey

Julie M. Fagan, Ph.D.
Associate Professor
School of Environmental and Biological Sciences
Rutgers, The State University of New Jersey
84 Lipman Dr.
New Brunswick, NJ 08901
fagan@rci.rutgers.edu
Appendix D: Revised HIPAA Form (DM)

**HIPPA/GINA CONFIDENTIALITY NOTICE**

**THIS NOTICE DESCRIBES HOW MEDICAL AND GENETIC AND**
**DRUG AND ALCOHOL RELATED INFORMATION ABOUT YOU**
**MAY BE USED AND DISCLOSED AND HOW YOU CAN GET ACCESS**
**TO**

**THIS INFORMATION. PLEASE REVIEW IT CAREFULLY.**

**General Information**

Information about your treatment and care, including payment for care, is protected by three federal laws: The Health Insurance Portability and Accountability Act of 1996 ("HIPAA"), the Confidentiality Law, and the Genetic Information Nondiscrimination Act of 2008 ("GINA"). Under these laws the program may not disclose information about your medical data or genetic information to a non-authorized party.

Your written consent must be obtained before your information can be disclosed to parties outside of treating physicians. For example, your doctor must obtain your written consent before it can disclose information to your health insurer in order to be paid for services. Generally, you must also sign a written consent before the treating organization can share information for treatment purposes or for health care operations. However, federal law permits the program to disclose information in the following circumstances without your written permission:

1. To organization staff for the purposes of providing treatment and maintaining the clinical record;
2. Pursuant to an agreement with a business associate (e.g. Clinical laboratories, pharmacy, record storage services, billing services);
3. For research, audit or evaluations (e.g. State licensing review, accreditation, program data reporting as required by the State and/or Federal government);
4. To report a crime committed on the organization’s premises or against program personnel;
5. To medical personnel in a medical/psychiatric emergency;
6. To appropriate authorities to report suspected child abuse or neglect;
7. To report certain infectious illnesses as required by state law;
8. As allowed by a court order.

Before the program can use or disclose any information about your health in a manner which is not described above, it must first obtain your specific written consent allowing it to make the disclosure. Any such written consent may be revoked by you in writing. (NOTE: Revoking a consent to disclose information to a court, probation department, parole office, etc. may violate an agreement that you have with that organization. Such a violation may result in legal consequences for you.) Disclosed genetic information cannot be used in a discriminatory manner.

**Your Rights**

• Under HIPAA you have the right to request restrictions on certain uses and disclosures of your health and treatment information.
• Under HIPAA you also have the right to inspect and copy your own health and treatment information maintained by the program, except to the extent that the information contains
psychotherapy notes or information compiled for use in a civil, criminal or administrative proceeding or in other limited circumstances.
• Under HIPAA you also have the right, with some exceptions, to amend health care information maintained in the program’s records, and to request and receive an accounting of disclosures of your health related information made by the program during the six (6) years prior to your request.
• Under GINA, disclosed genetic information cannot be used by health insurance companies to set rates or deny coverage.
• Under GINA, disclosed genetic information cannot be used by an employer in any discriminatory fashion. It cannot affect employment status, advancement, or treatment. If an employer receives knowledge of genetic information without consent, through a relationship with the health insurance provider for instance, the nature and cause of the genetic information breach must be disclosed.

**The Use of Your Information**
In order to provide you with the best care, the program will use your health and treatment information in the following ways:

• Communication among treating physicians and staff for the purposes of treatment needs, treatment planning, progress reporting and review, staff supervision, incident reporting, medication administration, billing operations, medical record maintenance, discharge planning, and other treatment related processes.
• Communication with Business Associates such as clinical laboratories (blood work, urinalysis), food service (special dietary needs), agencies that provide on-site services (lectures, group therapy) long term record storage.

**The Health Organization’s Duties**
The program is required by law to maintain the privacy of your health information and to provide you with notice of its legal duties and privacy practices with respect to your health information. The program is required by law to abide by the terms of this notice. The program reserves the right to change the terms of this notice and to make new notice provisions effective for all protected health information it maintains. The program will provide current patients with an updated notice, and will provide affected former patients with new notices when substantive changes are made in the notice.

**Complaints and Reporting Violations**
Patients have the right to make a complaint about the Confidentiality and Privacy of their Health Information. The patient may also register a complaint with the:

**Office for Civil Rights**
U.S. Department of Health and Human Services,
Jacob Javits Federal Building
26 Federal Plaza--Suite 3313
New York, New York, 10278
Voice Phone (212) 264-3313.
FAX (212) 264-3039.
TDD (212) 264-2355
OCR Hotlines-Voice: 1-800-368-1019

You will not be retaliated against for filing such a complaint.
Violation of the Confidentiality law by a program is a crime. Suspected violations of the
Confidentiality Law may be reported to the United States Attorney in the district where the violation occurs.

CONFIDENTIALITY NOTICE

I, ____________________________ have received a copy of the Confidentiality Notice, and it has been explained to me.

____________________________________________________________________
Signature Date

____________________________________________________________________
Parent/Guardian Signature (if necessary) Date
Appendix E: Letter to Editor (MK)

Increasing awareness of the Genetic Information Nondiscrimination Act to patients and physicians for early detection of illnesses

To Whom It May Concern:

I am Melissa Koger, a senior at Rutgers University majoring in Animal Science Pre-vet with a minor in Endocrine Physiology and Health Sciences. I am looking to spread awareness of the Genetic Information Nondiscrimination Act through education of physicians and patients.

In today’s economy, when many people are being laid off and insurance for their families is a major concern, the fear of being denied coverage because of a known pre-existing genetic condition will keep many people from the screening, which is now available to treat their conditions before it becomes a major medical concern. Genetic Information Nondiscrimination Act (GINA) protects a patient’s genetic information and family history from being released to health insurance companies as well as employers.

However, not many patients, nor physicians, are aware that such protection exists and will withhold screening out of fear of genetic discrimination. I, myself, was not aware of GINA before conducting research on genetic discrimination and I feel more at ease about undergoing genetic testing for certain diseases that are prevalent in my family. Unfortunately, not every American is going to research this topic on their own; therefore, there needs to be measures taken to spread awareness of GINA. By knowing the percentage chances of having breast cancer or cardiovascular disease early in one’s life can save thousands of dollars, and more importantly, their life.

I, therefore, recommend that you should send information to your affiliated medical groups to have their offices include a provision for GINA on the HIPAA form and to increase awareness of GINA for both the doctor and the patient through continuing medical education seminars of genetic privacy and informational advertisements. I have included a poster that can be displayed in their office that will allow awareness and further discussion between patient and doctor.

Genetic testing is not something that should be feared anymore, but used as a meaning of hope within scientific reason. This should be applied to both the patient and the doctor and of course your own employees. As a doctor, it is our duty to save lives and genetic testing will do just that. Thank you for your time and consideration. If you have any questions or concerns, please feel free to contact me via email at mkoger@eden.rutgers.edu. I look forward to hearing from you.

Sincerely,

Melissa Koger
Appendix F: Letter to the Editor (NS)
Letter to the Editor – Prevention Magazine

I am currently a senior at Rutgers University studying Genetics and Psychology. I also am shadowing a genetic counselor and what I have found is that many families coming in for counseling have no idea about the laws that protect them from genetic discrimination. As genetic technology progresses, clinicians will be able to test more people for more diseases and give them better answers and solutions. This can only happen though if people are willing to come to counseling. The fear of genetic discrimination by employers and insurance companies is ranked second on the list of why consumers don’t pursue genetic testing. However, in 2008 the Genetic Information Nondiscrimination Act (GINA) was put into law to protect people from genetic discrimination by prohibiting use of an individual’s genetic information for insurance eligibility or premium rates, prohibits insurers or employers from requiring genetic testing and prohibits use of genetic information by employers in decisions like hiring, firing and job assignments. GINA’s purpose is great, but if no one knows about it, it can’t do any good!

With that being said, awareness and understanding of this law is crucial to its success. There are over 1500 genetic tests available to patients but with the fear of discrimination in the back of their mind, they are less likely to utilize these tests. Increasing public awareness of this act could encourage more people to undergo testing when needed. The purpose of most testing is to prevent disease, rather than have to treat the disease later on. Preventative care is usually much less expensive than treating a lifelong condition. For example, the genetic test for the breast cancer genes, \textit{BRCA1} and \textit{BRCA2}, can range around $300 for patients. Late-stage treatment for breast cancer can be upwards of $100,000. With the results of the test, patients can begin increased screening and possible measures such as preventative surgeries. This is the greatest benefit to having this genetic information. Another instance where this information would be crucial is with Hypertrophic Cardiomyopathy, a heart condition that usually presents no symptoms until a sudden death. This is one particular case that with genetic testing, we can directly prevent deaths in affected patients through lifestyle changes and increased surveillance.

\textit{Prevention Magazine} is one of the largest magazines in the world and a renowned source for healthy lifestyle tips. To feature an article on this act and then refer to it whenever a subsequent article speaks of some sort of genetic predisposition would help generate awareness and understanding. Studies have shown that many general physicians don’t know about GINA or don’t fully understand it, so we need to educate the consumer directly as best we can. Most genetic conditions are not curable but some are preventable, and genetic testing is the first step to prevention. Preventing health problems is your goal – it’s in your name! With your help in creating public awareness in your magazine of the GINA act, we can start saving more and more lives and improving the quality of life to many.

Thank you for your time and consideration,
Nicole Salvatore
Rutgers University