J Genet Counsel (2014) 23:891–902
DOI 10.1007/s10897-014-9743-2

PROFESSIONAL ISSUES

Genetic Information, Non-Discrimination, and Privacy Protections in Genetic Counseling Practice

Anya E. R. Prince · Myra I. Roche

Received: 4 April 2014 / Accepted: 4 July 2014 /

Abstract The passage of the Genetic Information Non Discrimination Act (GINA) was hailed as a pivotal achievement that was expected to calm the fears of both patients and research participants about the potential misuse of genetic information. However, 6 years later, patient and provider awareness of legal protections at both the federal and state level remains discouragingly low, thereby, limiting their potential effectiveness. The increasing demand for genetic testing will expand the number of individuals and families who could benefit from obtaining accurate information about the privacy and anti-discriminatory protections that GINA and other laws extend. In this paper we describe legal protections that are applicable to individuals seeking genetic counseling, review the literature on patient and provider fears of genetic discrimination and examine their awareness and understandings of existing laws, and summarize how genetic counselors currently discuss genetic discrimination. We then present three genetic counseling cases to illustrate issues of genetic discrimination and provide relevant information on applicable legal protections. Genetic counselors have an unprecedented opportunity, as well as the professional responsibility, to disseminate accurate knowledge about existing legal protections to their patients. They can strengthen their effectiveness in this role by achieving a greater knowledge of current protections including being able to identify specific steps that can help protect genetic information.

Keywords Genetic counseling · Genetic Information Non-Discrimination Act · Privacy · Genetic information · Insurance · Employment

Introduction

Significant advances in genomic technology are rapidly expanding the number and scope of genetic tests available both for diagnosing existing disorders and for predicting treatable ones before the onset of symptoms. Public awareness of genetic testing options has been most recently heightened by news stories reporting Angelina Jolie's decision to have prophylactic surgery after her testing revealed a *BRCA1* gene mutation (Jolie 2013) and by the recent, controversial Supreme Court ruling on gene patents (AMP et al. v. Myriad Genetics, Inc et al. 2013). There is growing interest in and demand for genetic testing as treatment options expand, the cost of using newer sequencing technologies declines, the insurance coverage for testing widens, and the population for whom testing is recommended broadens.

Ironically, as genetic testing becomes an increasingly powerful diagnostic and prognostic tool, health care providers and their patients remain wary of the potential of genetic testing to trigger discrimination. Limited awareness of the true scope of legal protections afforded by legislation including the Genetic Information Nondiscrimination Act (GINA) persists and is still fueling fears of genetic discrimination by both patients and their health care providers nearly 6 years after the law's passage (Huntsman Cancer Institute Survey 2013). Genetic counselors can play an influential role in increasing awareness about these legal protections, both because they are more knowledgeable about them than most other health care providers and because their patients can derive direct benefits from this knowledge. Correcting patients' common misconceptions about this topic is, in and of itself, an admirable goal

---

A. E. R. Prince · M. I. Roche
Center for Genomics and Society, School of Medicine, University of North Carolina, Chapel Hill, NC, USA

M. I. Roche
Department of Pediatrics and Department of Genetics, School of Medicine, University of North Carolina, Chapel Hill, NC, USA

M. I. Roche (✉)
University of North Carolina, CB# 7240, Room 326 A MacNider, Hall Chapel Hill, NC 27599-7240, USA
e-mail: myra_roche@med.unc.edu

but genetic counselors could further expand their influence by learning how their patients can take specific steps to help prevent discrimination. Yet counselors may find it difficult to attain an adequate understanding of the legal protections, and their limitations, because the combination of state and federal laws have created a patchwork of protections that vary between individuals and their family members depending upon their circumstances. Though these laws continue to have lingering gaps, patients and their health care providers, including genetic counselors, could benefit from a greater knowledge of the breadth of protections that are currently in place.

Patient fear of genetic discrimination has been reported with the application of genetic technologies to patient care (Lapham et al. 1996; Hall et al. 2005; Allain et al. 2012). Despite widespread apprehension that genetic information will inevitably be misused, there is limited, convincing, empirical evidence that discrimination on the basis of genetic information has occurred (Hall et al. 2005; Pollitz et al. 2007). Whether the lack of evidence stems from under reporting, confusion about what constitutes illegal discrimination, or if it is a true reflection of the situation, is unclear (Sharpe and Carter 2006). The discrepancy between the magnitude of patient concern over potential misuse on one hand and the limited evidence of its occurrence on the other hand, may leave genetic counselors uncertain as to which and how much information they should provide to their patients about the potential for discrimination (Pamarti 2011). This uncertainty can be encapsulated by the following questions: How can a genetic counselor best summarize the legal protections and their caveats and yet acknowledge the limited evidence of discriminatory practices in a time-sensitive manner and without causing patient distress? Which resources can a genetic counselor recommend to a patient who expresses concerns about discrimination? What actions can a patient take if he or she experiences discrimination?

The goal of this paper is to illustrate elements of the legal protections against genetic discrimination that are applicable to issues that arise during a counseling session. We first summarize research findings about fears of genetic discrimination among health professionals and the public, review their understandings of the laws banning discrimination, and describe the current practice of discussing the possibility of discrimination during a counseling session. We then present three genetic counseling cases to highlight ways that issues of genetic discrimination can arise during a session and provide the relevant background information on the applicable legal protections.[1]

Genetic Discrimination and Genetic Counseling Practice

Fears of Genetic Discrimination

As genetic technologies have become integrated into clinical care, patients and health care providers have consistently raised alarms about how certain actors – most notably insurers and employers – could potentially use genetic information (Pollitz et al. 2007; Bombard et al. 2012). In 2000, reasoning that health care professionals could be expected to be more knowledgeable than patients about the validity of the potential threat, Matloff et al. conducted a survey of cancer genetic professionals and found that 26 % would use an alias for genetic testing because of their concern about discrimination (Matloff et al. 2000). By contrast, in a 2013, post-GINA version of the study, these percentages had plummeted almost tenfold; from 26 to 3.2 % (Matloff et al. 2014).

It remains to be seen if and to what extent patients' fears of discrimination might also be alleviated by increased awareness of existing laws. In a post-GINA study, many patients still favored anonymous testing out of fear of discrimination related to life insurance (42.7 %), health insurance (30 %), or employment (29.1 %)(Ader et al. 2009). Fears of discrimination have been reported most commonly when the symptoms of a genetic condition begin in adulthood but they appear to have little influence on genetic testing decisions made in prenatal and pediatric settings. Possible explanations for this difference could be that children are typically symptomatic when tested, and, if they have health insurance, they are usually covered under their parents' policies or by the state. Furthermore, their employability is not usually a pressing concern (Hall and Rich 2000). Prior to federal legal protections, evidence that fears of discrimination were scaring patients away from clinical genetic testing and from participating in genetic research (Hall and Rich 2000; Hadley et al. 2003), led to efforts that, in 2008, resulted in the passage of GINA.

Broadly speaking, GINA prohibits employers and health insurance companies from discriminating against an individual based on his or her genetic information. Importantly, these entities are not allowed to collect genetic information in order to use it to raise premium rates, deny coverage, or make adverse employment determinations. Health insurance companies are permitted to request limited genetic information when it involves their decision about whether or not to pay for a medical procedure (GINA 2008).

Overall, GINA has greatly improved protections for many individuals in the US not only by prohibiting some forms of genetic discrimination but, also, although this facet remains less well-recognized, by transferring to patients a much greater control over who has access to their genetic information. Despite these significant gains, the prudent genetic counselor will paint a balanced picture of the current legal landscape – acknowledging both the gaps in the law as well as the

---

[1] This article presents general information about the law in order to educate genetic counselors about legal protections regarding genetic discrimination. It is not legal advice. Professional legal advice should always be sought before any legal action is taken. Application of the law may vary across situations because it is dependent on individually specific circumstances and on the applicable state and federal law.

uncertainty about how often genetic discrimination occurs. But counselors should also be careful not to undersell the law's substantial benefits.

Genetic Counselor, Public, and Physician Knowledge of GINA

During a session, genetic counselors attempt to provide a balanced portrayal of both the benefits and the gaps of existing protections; however, the crazy quilt of laws is complicated and requires general knowledge about the law as well as its specific provisions. Genetic counselors are quite well informed about GINA's general protections. A recent survey by Pamarti reported that 99.3 % knew that GINA protects against health insurance discrimination; however, many fewer were knowledgeable about specific details of the law (Pamarti 2011). For example, only 44.2 % of the 257 counselors in this survey knew that GINA does not apply to symptomatic individuals and only 33.8 % knew about the implications for direct-to-consumer genetic testing (Pamarti 2011). Thus, although genetic counselors are aware that GINA offers protection, they may not fully appreciate some of the potential applications to specific situations that they may encounter in their practice. Additionally, because the aforementioned survey only measured counselor knowledge about GINA's anti-discrimination provisions, it did not assess what they knew, or didn't know, about the act's privacy protections; a less well-recognized facet of the law that has direct applications to individuals with a family history of a genetic condition.

Genetic counselors may have some knowledge gaps about GINA's specific protections but a much higher percentage of genetic counselors are aware of the law's existence and its general provisions as compared to people in the general population (AMA 2013). In the previously described survey, genetic counselors estimated that only about 15 % of their patients were aware of GINA prior to their discussion of it during the counseling session (Pamarti 2011). This limited public awareness is corroborated by other surveys that directly measured public knowledge of either the existence of GINA or the existence of laws protecting the privacy of genetic information. In 2006, **prior** to the passage of GINA, in a general population survey administered by Cogent Research, 18 % of 1,000 respondents believed that there were laws to protect the privacy of genetic information (Cogent 2010). Astoundingly, their 2010 survey, conducted **after** the passage of GINA, showed that even **fewer** (16 %) believed that protective laws existed. Likewise, in 2011, an online survey of the general public found that only 8.8 % of 295 respondents had ever heard of GINA (Huang et al. 2013). Similarly, in striking contrast to the public's increasing knowledge about genomic advances, knowledge about the social implications of genetic testing, such as the potential impact on the ability to obtain health insurance, has lagged far behind (Haga et al. 2013).

Even within a population for whom GINA would be expected to be highly relevant, many remain unaware of it. In one study, fewer than half of the asymptomatic individuals who had an expanded allele for Huntington Disease (HD) were familiar with the law, a far fewer number than the three quarters of them who were familiar with the Health Insurance Portability and Accountability Act (HIPAA) (Dorsey et al. 2013). An Australian study of those at risk for HD found a similar lack of awareness about legislation that prevents employers and health insurers from accessing and using genetic information in that country (Goh et al. 2013).

Family physicians appear to have a level of knowledge about GINA that lies between that of genetic counselors and the general public. In a 2010 study of family practitioners, 54.4 % said they were unaware of GINA, 35.2 % knew about GINA, but had no knowledge about any specific features, and 10.3 % had basic knowledge of GINA and its specific protections (Laedtke et al. 2012).

Given the relatively high levels of knowledge about GINA among genetic counselors and the relatively low levels among some physicians and the general public, genetic counselors could serve as a valuable source of information about the implications of both the privacy and nondiscrimination protections of the law.

Discussing Genetic Discrimination

Genetic counselors have an unparalleled opportunity and ability to disseminate accurate knowledge of existing protections of genetic information to their patients. Despite this opportunity, Pamarti found that fewer than half of the 257 counselors surveyed reported discussing GINA during a session (Pamarti 2011). In this sample, counselors only discussed the law if a patient specifically inquired about discrimination (Pamarti 2011). The same study showed that, perhaps not surprisingly, cancer genetic counselors reported discussing the possibility of genetic discrimination with their patients more often than counselors in other specialties; 68 % as compared to 28 % in pediatric and 11 % in prenatal (Pamarti 2011).

Given the amount and complexity of genetic information that is typically conveyed during a session, suggesting that balanced information about legal protections and their limitations also merits inclusion may seem unrealistic. It may also be viewed as an unnecessary diversion given the lack of empirical evidence of discrimination. A concise discussion about the existence and scope of legal protections need not be a major focus of the session, but the failure to describe a realistic picture of the current legal landscape surrounding genetic information can cause future harm to patients and their families. There are several organizations and websites to which patients can be referred that provide more detailed information about GINA and the gaps in the law (Resources). Referring patients to these sources can help genetic counselors balance the time constraints of a session with their responsibility to present accurate information. It is important to realize that even when patients do not ask questions about genetic discrimination, they may still have concerns. Simply discussing basic information about GINA has been reported to lower patient fears about potential discrimination (Allain et al. 2012). Therefore, combining a brief overview of the current legal protections of genetic testing with a referral to resources that describe the gaps and the limitations of the law could be an efficient method that, at the very least, introduces patients to the existence of the law and its general provisions. Depending upon their circumstances, some patients may need more comprehensive information.

Genetic Discrimination Post-GINA

There continues to be anecdotal stories of genetic discrimination but data on the use or misuse of genetic information in employment and insurance are lacking and few additional empirical reports of genetic discrimination have been published in the 6 years since GINA became law. It is not clear if this sparse amount of data is due to lack of genetic discrimination overall or lack of collected evidence. Additionally, there are likely many more violations of GINA's privacy provisions, in contrast to its anti-discrimination protections, in part because the public and provider awareness of these aspects is even lower. There have been several studies exploring the existence of genetic discrimination in life insurance, but due to limited methodological rigor and the few number of subjects studied, the validity of the conclusions remains uncertain (Joly et al. 2013). Despite lack of empirical evidence that discrimination is occurring, fear of genetic discrimination remains a barrier to the uptake of genetic testing, even in a post-GINA world (Allain et al. 2012). Therefore, the discussion between counselors and patients about the legal protections that exist remains both necessary and beneficial.

Case Studies

The following cases illustrate some common questions and fears that genetic counseling patients may have regarding the potential for discrimination and summarizes the relevant legal background. *We use these cases to highlight particular features of the legal protections; however similar real life situations may have different outcomes if an individual's insurance or employment falls under legal exceptions, since the determination of whether state or federal laws apply depends on individual circumstances.*

Case study 1

A 38-year-old woman calls a genetic counselor because her mother, maternal aunt, and maternal grandmother all had breast cancer. She would like to schedule an appointment for risk assessment and to discuss options for genetic testing but is worried about the possibility of genetic discrimination if information about her family history is entered into her medical record. What information does the genetic counselor need to address this concern?

Legal Protections

Most genetic counselors know that GINA regulates how some employers and health insurance companies can use genetic information. They may not, however, fully appreciate how broadly GINA defines some crucial terms. "Genetic information," as defined by GINA, includes not just genetic test results, but also family medical history, use of genetic services – such as genetic counseling –, and participation in genetic research (GINA 2008). Therefore, those employers and health insurance companies regulated by GINA are banned from using the woman's family medical history or the fact that she had a consultation with a genetic counselor to do the following: raise her premium rates, deny her health insurance, make adverse employment decisions against her, or otherwise discriminate against her.

GINA's definition of "family member" is also very broad, and includes first, second, third, and fourth degree relatives – all the way back to great, great-grandparents, and includes first cousins once-removed (CFR 2013). An individual's genetic information, therefore, includes manifested conditions in any of these relatives.

In this case, the counselor could reassure the woman that employers and health insurers regulated by GINA would be banned from discriminating against her because of her relatives' diagnoses of breast cancer. Additionally, her session with a genetic counselor would also be classified as "genetic information", so the appointment itself – regardless of whether she decides to have genetic testing or not – is also protected information that cannot be used to discriminate. There are situations, especially when medical records are requested, in which an employer or health insurance company can obtain genetic information, including family history. These circumstances will discussed further later, but, in all situations, even if a covered entity **learns** of genetic information, it cannot **use** this information to discriminate.

Case Study 2

A couple consults with a genetic counselor because the woman, who is 15 weeks pregnant, had fragile X testing. Her results showed that she is a carrier and has a pre-mutation of

78 CGG repeats in the *FMR1* gene. The couple is worried that her employer or health insurer may be able to use the results of the test to discriminate against her even though she has no signs of premature ovarian insufficiency (POI); a condition associated with carrier status. They are also concerned about potential discrimination against the fetus, should they decide to have prenatal testing and find out the fetus has inherited the expansion. What information does the genetic counselor need to address these concerns?

Legal Protections

GINA includes a specific provision to emphasize that the genetic information of a fetus is considered part of the genetic information of the pregnant woman (GINA 2008). Therefore, in this situation, any genetic information discovered during prenatal testing would be considered the mother's genetic information under GINA. After the baby is born, any testing done during pregnancy would also still be considered his or her own genetic information.

The association of expanded repeats in the *FMR1* gene with an increased risk for POI as well as the fragile X tremor and ataxia syndrome (FXTAS) highlights one of the legal thresholds or limitations of GINA. Although the law protects against discrimination on the basis of genetic information, this protection does not extend to "manifested conditions". The genetic information is protected under the law, even if symptoms begin, but the symptoms themselves are not protected. For example, in this case, the woman's carrier status is protected genetic information. However, if she begins to have symptoms of either premature menopause or FXTAS, GINA would no longer protect her from being discriminated against because of these symptoms. Even after her symptoms develop, however, covered employers or health insurance companies could not cite her carrier status as the reason for an adverse decision. As genomic sequencing becomes more commonly performed, this category of individuals, those who are asymptomatic but who are at risk for multiple phenotypes, could become more prevalent as the pleiotrophic effects of genomic variants become increasingly recognized (Kocarnik and Fullerton 2014).

To determine whether a covered employer or health insurer could use, for example, the woman's premature menopause symptoms to legally discriminate, it is necessary to look to other laws. The Patient Protection and Affordable Care Act (ACA) currently makes it illegal for health insurers to deny health insurance or raise premiums based on a pre-existing condition (Patient Protection and Affordable Care Act of 2010) for adults. Genetic information is explicitly not considered a pre-existing condition under GINA (GINA 2008). Therefore, protections under GINA and the ACA meet at the point when a person manifests symptoms that could reasonably lead to diagnosis (Fig. 1). A health insurer would be prohibited from using the woman's carrier status to discriminate under GINA, but also would be prohibited from using her symptoms to discriminate under the ACA.

Legal protections in the employment arena are less comprehensive as compared to those applicable to health care. The Americans with Disabilities Act (ADA) protects against discrimination on the basis of a disability. In order for medical symptoms to be protected under this law, they must meet specific criteria. A "disability" is defined as "a physical or mental impairment that substantially limits one or more major life activities, a record of such an impairment, or being regarded as having such an impairment" (ADA 1990; ADAAA 2008). Some symptoms and conditions will not fall under the definition of disability if they do not create a substantial limitation for the individual. Therefore, for some conditions, a gap remains between the legal protections of GINA and the ADA against employment discrimination (Rothstein 2008).

The symptoms of FXTAS and POI could potentially fall under the ADA, although this depends upon how the symptoms affect the individual. For example, an ataxia may substantially limit the major life activity of walking and POI may substantially limit the major life activity of reproduction. However, in the early stages, the symptoms may not reach the level of a substantial limitation and therefore not yet rise to the level of a disability under the ADA. It is possible that, in this circumstance, the woman's employer could legally fire her based on early symptoms, although legal counter-arguments could be made under the ADA, especially under the 'regarded as' portion of the definition (Rothstein 2008).

Case study 3

After his 49-year-old father died of liver failure, a 20-year-old man becomes convinced that the cause was undiagnosed hemochromatosis and wonders about his own risk of this condition. He asks his primary care physician about the option of genetic testing and the physician orders *HFE* gene testing. The results showed that the man is homozygous for the deleterious Cys282Tyr mutation.

Since requesting the test, the man has read about the possibility of discrimination based on the results of genetic testing. He is now concerned because he is applying for a new job and he doesn't want a prospective employer or health insurance company to discriminate against him based on the results of his genetic test. He has told his physician that he doesn't want to know his results until he gets a new job. The primary care provider calls a genetic counselor for advice. What information does the genetic counselor need to address these concerns?

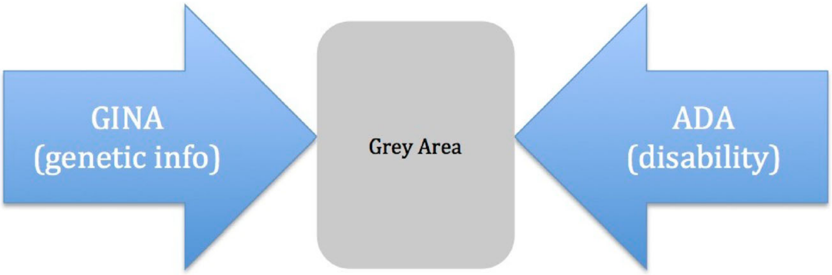

Fig. 1 Application of Gina and the ADA in the continuum from asymptomatic genetic information to manifested disease

Legal Protections

Although heralded as "the first civil rights bill of the new century" (CGF 2008), GINA extends the definition of anti-discrimination far beyond society's colloquial meaning of the concept. GINA bans covered health insurers and employers, not just from using genetic information to harm an individual – in most instances it also **prevents** these actors from **collecting** genetic information in the first place (GINA 2008). Genetic information includes the results of a genetic test, such as the *HFE* gene testing in this case. It is important to note that the definition of genetic testing itself extends beyond single gene, highly penetrant disorders – even though these are the examples that are most often used. A genetic test is defined as "an analysis of human DNA, RNA, chromosomes, proteins, or metabolites that detects genotypes, mutations, or chromosomal changes" (GINA 2008). This definition would extend to many other situations such as learning about carrier status for an autosomal recessive disorder. Additionally, the definition does not depend upon when the genetic test was done or who performed it, so direct-to-consumer genetic tests, tests ordered by physicians and other healthcare professionals, and tests completed prior to the passage of GINA, in 2008, all would be protectable under the law.

The term "collection" in GINA encompasses requesting, requiring, or purchasing an individual's genetic information – including any family medical history. Although covered health insurance companies are generally prohibited from collecting genetic information, in reality, as described above, they often gain access to genetic information through requests for medical records. GINA requires that all requests for medical records state that no genetic information should be included in the request, unless it is directly related to a payment determination (GINA 2008). The law then places the onus on the healthcare professional to redact out all genetic information from the copy of the medical record to be submitted to the insurer. Redaction does not mean removal of the information from the original medical record. Rather, it is the removal or masking of information from the copy of the medical record that is being transmitted to the requesting insurance company or employer. Redaction probably occurs infrequently due to the voluminous amounts of genetic information, including family medical history, sprinkled throughout medical records.

Patients could collaborate with healthcare professionals to attempt to limit the amount and type of genetic information inadvertently given to health insurers by focusing on redacting information that is of particular concern to the individual. Ideally, it has been suggested genetic information could be kept in a separate section of the medical record that is not provided to health insurers or employers that are covered by GINA and requires a separate consent to obtain (Prince 2012).

It is important to appreciate GINA's expansive definition of genetic discrimination for two reasons. First, the broader conceptualization of what counts as discrimination under the law means that genetic discrimination for which an individual could bring a complaint likely occurs at a much higher rate than is currently acknowledged. Imagine the difference in the response to a survey question asking individuals whether or not they have ever been denied insurance, fired from a job, or otherwise adversely affected based on their genetic information versus one that asks if a health insurance representative or employer has ever requested genetic test results or, even more likely, family history information from them. The second question is much more likely to garner a positive response; however, both instances are equally illegal and actionable under GINA. Both health professionals and the general public generally remain unaware that it is illegal for some entities to request genetic information, including family medical history and, furthermore, that in those instances, individuals can legitimately decline these requests thereby protecting the privacy of this information.

Secondly, the prophylactic ban on collection of genetic information by covered health insurers and employers places the patient in an unusual position of power. Lawsuits are incredibly time consuming, costly, and – especially in the case of employment and health insurance – very difficult for a plaintiff to win. In part, this is because it is relatively easy for an employer or health insurance company to invent reasons for a denial that mask the true, underlying reason of genetic discrimination. In one example, an insurance company denied a woman health insurance coverage because it was stated that her weight was slightly too low and she took birth control. The insurance denial also mentioned that she had had a prophylactic surgery – indicating that the stated reasons of low weight and being on birth control could have been proxies for genetic discrimination. The recent changes banning health insurance denials based on pre-existing conditions under the ACA now make this type of proxy reasoning unlikely to be effective in the health insurance arena.

Proxy genetic discrimination remains a risk in the employment cases– especially if an individual is considered to be an "at-will" employee. In this type of employment, an employee can be fired for "any reason or no reason" – as long as it is not a discriminatory reason (Guz 2000). This rule makes employment cases very difficult for employees to win since a savvy employer can easily hide a discriminatory intent for the adverse decision. In the case described above, it would be difficult for the man to know if he had been victim of genetic discrimination if he was not hired for a job for which he applied. Employers do not generally tell a person why he or she was not hired, so discriminatory intent can be very difficult to prove.

The enforcement mechanisms for health vs. employment claims under GINA are different. Individuals who believe that they have been discriminated against in employment can file a complaint with the Equal Employment Opportunity Commission (EEOC). Once individuals have exhausted the EEOC's administrative process, they can litigate in federal court. In some cases, the EEOC will litigate a complaint that has been filed on behalf of the individual. For example, the EEOC recently settled with Founders Pavilion, a nursing and rehabilitation center, and the company agreed to pay $370,000 because they collected family history as part of a medical exam for new hires (EEOC 2014).

It is much more difficult to uncover the number and types of complaints about genetic discrimination in health insurance because GINA's enforcement provisions are tied to state-specific procedures. If an individual feels that a health insurance company has violated his or her rights, he or she can file a complaint with the state department of insurance. Every state has a different agency and mechanism for these complaints, making it difficult to gather comprehensive data. The current evidence of genetic discrimination in health insurance remains anecdotal, just as it was prior to GINA. However, given the broad definition of genetic information and the ban on collecting genetic information, genetic discrimination – as defined by GINA – likely occurs much more often than people realize or report.

Realistically, patients may opt not to enforce their legal rights because of the hassles and cost of appeals and litigation. An individual may decide that this process is too costly, both financially and emotionally, as compared to a monthly premium rate increase. Avoiding legal action is an understandable decision for many individuals – and unfortunately in some cases, a necessity, when the cost of litigation is prohibitive. Therefore, knowledge that GINA bans the collection of genetic information is an important and powerful tool for individuals that enable them to help prevent genetic discrimination from occurring in the first place.

In contrast to genetic status, the nature of the bias in most forms of discrimination is generally readily apparent – one can often tell an individual's race and gender, and sometimes even a person's religion or disability, simply by looking at them. However, genetic information, in the absence of manifested symptoms, is never obvious just from looking at an individual. Therefore, if an individual can prevent an employer or health insurance company from obtaining information about his or her genetic status, he or she can prevent the possibility of subsequent genetic discrimination based on that information.

One of the most practical steps individuals can take is simply to refuse to answer general questions about their genetic information – including family medical history – that is asked by a covered health insurance representative or employer. Sometimes, even though questions about family medical history are not asked on the application, a representative from the company may ask these questions over the phone if they have not been properly trained on the law. Similarly, a covered employer may ask about family medical history or other genetic information during medical examinations or in other situations. The questions in both of these instances would likely be illegal; however they are still routinely being asked. The man concerned about his genetic test result for hemochromatosis could simply refuse to answer requests by a health insurance company or by his potential employer if either is a "covered entity". This action stops the company from gaining access to his genetic information and therefore prevents genetic discrimination before it can occur.

Some Exceptions to GINA's Ban on Collection of Genetic Information

It is important to note that there are several exceptions that allow companies to collect genetic information. As stated above, health insurance companies are permitted to request genetic information if it involves their decision about whether or not to pay for a medical procedure. For example, if the cost of the genetic testing for hemochromatosis was billed to his health insurance, the company can ask the man for family medical history, such as the father's liver disease, to show that testing was medically necessary. Similarly, if a woman's BRCA sequencing is negative and her genetic counselor recommends BART testing, the health insurance company could request the initial test results. In these cases, the insurer can only ask for the minimum amount of information necessary to make their determination. Additionally, these insurers are not permitted to use the collected genetic information to discriminate.

The employment setting represents a less protected environment than that of health insurance and there are several additional exceptions to the prohibition on collecting genetic information. These exceptions include inadvertent acquisition, a voluntary disclosure by the individual as requested by a “wellness” program, any publicly available information, disclosure via a family and medical leave request, information requested for law enforcement purposes, and requests made as a part of a company’s toxic substances monitoring. For the most part, individuals can most easily prevent an employer from gaining access to genetic information in each of the first three exceptions by not discussing genetic information at work, refusing to answer questions about genetic information, including family history, during enrollment and participation in wellness programs, and by limiting the amount of genetic information publicly available, such as that posted on social media.

Genetic counselors can educate patients by explaining the circumstances under which they do not have to provide information about genetic tests and family medical history to covered health insurers and employers. With this knowledge, patients can take simple, specific steps to help prevent genetic discrimination before it occurs. In the case above, the man can decline to answer questions about his genetic information, including his family history, to potential employers, and, if he insists on posting information about his genetic status on social media sites and blogs, he should, at the very least, restrict the accessibility of others to these sites. These steps can help to protect him against discrimination based on genetic information in the employment and health insurance settings.

Case Study 3 Revisited

After hearing about the steps he can take to limit the amount and nature of the genetic information disclosed to a prospective employer, the man says, “Great because the job I really want is with a small start up company that has only 10 employees.” What information does the genetic counselor need to address this statement?

Legal Protections

GINA prohibits only **certain kinds** of entities, namely **some** health insurers and **some** employers, from using genetic information to discriminate against individuals (GINA 2008). Many private health insurers in the US are included under GINA’s umbrella and some that are excluded, such as the Federal Employee Health Benefits Plan, Tricare, Veteran’s Health Benefits, and the Indian Health Service, have their own restrictions against use of genetic information. Since these health insurers are group plans, they do not take any medical information, including genetic information, into account when setting rates and eligibility. GINA does **not** extend to insurance companies that provide life, long-term care, or disability insurance **nor** does it apply to other entities such as education or licensing.

Many employers, including state, local, and some private employers, are included under GINA but the law does **not** apply to federal government employees and members of the military that have their own rules about what constitutes genetic discrimination (NHGRI 2014). Laws covering federal employees broadly ban employment discrimination based on genetic information but they do not include the privacy protections of GINA. Military rules are less protective and allow some use of genetic information in employment decisions, such as the military’s prerogative to decide upon service placement based on genetic susceptibilities to disease (NHGRI 2014; Baruch and Hudson 2008). For example, some branches use the results of genetic testing to make specific assignments to avoid adverse events (Baruch and Hudson 2008). Members of the military can refer to their employment policies to determine whether their branch provides information regarding genetic information, discrimination, and employment.

In the private sector, employers with fewer than fifteen employees do not have to comply with GINA. This segment accounts for about 15 % of the US workforce leaving a substantial minority of workers without federal-level protections against genetic discrimination in employment (SBA 2011). Some of these workers are still protected against genetic discrimination at the state level and several states extend the employment protections to include businesses with fewer than fifteen employees (NCSL 2008). Therefore, these individuals have some state protections against genetic discrimination but these are typically not as broad as the federal level protections.

State Laws Covering Gaps in GINA

GINA creates a baseline of protection and, importantly, **does not pre-empt** stronger state laws. Therefore, individuals who work for a private employer with more than 15 employees may have the choice to file a complaint under either state or federal law. However, state laws are typically not as robust as GINA and the protections and the enforcement mechanisms against infractions vary greatly. For example, the size of employer that must comply with state statutes varies and is state law-dependent. Most notably, many state laws that “protect” against genetic discrimination in employment do not include the powerful prohibition that GINA has against the collection of genetic information. Some states do incorporate the broader protections by legislating that the entities that must comply with state law, must also comply with GINA.

As an example, if the man in this case was applying to a Californian employer with only ten employees, GINA would not apply, but Cal-GINA, a recently passed state law, may be applicable (Cal-GINA 2011). Cal-GINA applies to employers with five or more employees and, although it bans genetic discrimination, it does not prohibit those employers from collecting genetic information. Therefore the man would not enjoy the broader privacy protections of GINA in this case, but he would still be protected from genetic discrimination. He can still take steps to limit a prospective employer's access to his genetic information, such as limiting public access via social media, but a small business employer could be allowed to ask about his genetic information directly. Importantly, however, even if he were asked for this information directly, in California, the small business employer would be banned from using that information to discriminate against him.

A comprehensive discussion with genetic counseling patients about their legal protections becomes even more difficult because of the familial nature of genetic information. If a patient works for a large employer who offers health benefits, it may be tempting for the genetic counselor to paint broad-brush strokes and briefly note that GINA protects him or her against genetic discrimination. However, there are two serious flaws in this approach. First, it is likely that an individual, especially a younger patient, will switch jobs, be covered under different insurance companies, and/or move across state lines during his or her lifetime. These changes could affect his or her current legal protections because of gaps in the law. Secondly, the genetic information of an individual could impact others in the family and it is likely that some of these relatives will have different legal protections based upon where they live or who employs them.

Genetic counselors can consult guides such as the Council for Responsible Genetics (CRG) to identify the protections afforded by a specific state. Determining whether or not specific state laws apply to employers that operate across state lines is often very complicated.

Case Study 2 Revisited

After the delivery of their son, the couple call the genetic counselor and explain that they want to obtain life insurance so that they can be assured that their children will be provided for should anything happen to them. They ask if the woman's risk for POI and FXTAS will be considered pre-existing conditions in their life insurance application. In addition, the man's father has recently developed symptoms of Alzheimer disease (AD). They have learned about the option of *APOE* testing and wondered about the implications if the man has this testing. What information does the genetic counselor need to address this statement?

Legal Protections

One of the most notorious gaps in GINA is that it does not apply to three types of insurance that individuals with genetic conditions may greatly desire; namely, life, long-term care, and disability insurances. Patients who discover they have a predisposition for cancer, AD, or other chronic illness are likely to seek insurance coverage to pay for nursing home care or to provide for their family when they pass away. At the same time, life, long-term care, and disability insurance companies are likely to seek information about an applicant's risk level in order to make the best economic decisions for the company. In the vast majority of cases, these insurances can legally use genetic information in coverage decisions and could even require that an individual take a genetic test before deciding whether or not to cover them (Schultz 2013).

Although it is true that GINA does not apply to these three types of insurances, some state laws regulate the use of genetic information in these arenas (NCSL 2008). All laws, however, are not created equally and it is important for patients and genetic counselors to refrain from equating the existence of a law with adequate protection. For the most part, states only **regulate the use** of genetic information in these insurances – **not ban the use**. For example, some state laws simply require that insurance companies show actuarial justification for charging different premium amounts or for denying coverage (NCSL 2008). Actuarial justification requires insurers to show that their premium rates are reasonable given their expected costs – a task that is fairly straightforward if an individual has a genetic pre-disposition to a health condition because of the implication that expected costs will be higher. Therefore the requirement of actuarial justification does not protect individuals in the same way that the public commonly conceptualizes the word "protection".

In other states, the laws regulating the use of genetic information in life, long-term care, and disability insurance simply requires "informed consent" from the enrollee when and if the insurance company requires a genetic test (NCSL 2008). These laws do **not** prevent insurers from gathering genetic information and making coverage decisions based upon the information. For example, New Jersey's law prohibits 'unfair' genetic discrimination in life insurance but this legally translates to requiring actuarial justification to use genetic information and obtaining "informed consent" from the individual prior to performing a genetic test (New Jersey Code 2008).

Finally, GINA broadly defines "genetic information" to include family medical history, use of genetic services, and participation in genetic research (GINA 2008). However, most state laws were passed prior to GINA and so define "genetic information" much more narrowly; namely, as genetic test results (NCSL 2008). California's law is an exception and includes family medical history in its expansive protection against discrimination in life, long-term care, and disability insurance (Cal-GINA 2011).

Unfortunately, the patchwork of state laws in life, long-term care, and disability insurance provides little concrete protection for individuals in these arenas. As new state laws continue to be passed, patients and genetic counselors must look carefully at the protections and should not assume that the laws are as comprehensive as GINA.

Options for Access to Supplemental Insurance

The lack of comprehensive protection at the state level, unfortunately, creates a difficult decision for individuals who are considering testing and yet are concerned about the possibility of genetic discrimination. One often-advised option is to secure coverage prior to having genetic testing although this approach has limitations as described in the next section. Additionally, if an individual is denied life, long-term care, or disability insurance, he or she should check the relevant state law to see if it is possible to appeal the decision (CRG resource). Although state laws vary, individuals have won appeals for denials based on genetic information, especially in states with more protective coverage, like California. Finally, policy makers at the federal and state level are increasingly considering legislation to improve access to these insurances for individuals with genetic conditions. Individual experiences and stories can be invaluable information to share with policy makers to increase their understanding about how current industry practices are affecting the public.

In the case study above, life, long-term care, and disability insurance companies may be able to deny the couple coverage based on genetic information – depending on which state they live in. In this situation, the genetic counselor could advise the couple to find out more about their state law by referring to a credible resource (CRG, NCSL, or consulting with an attorney specializing in insurance law). It could be more difficult for the man to secure insurance due to his family history of AD because his case appears to present a stronger actuarial justification for increased costs. The man could consider getting insurance coverage prior to genetic testing, although in many states, the insurers would be allowed to ask about the family history, ask about genetic test results, and in some situations, require him to have genetic testing before making coverage determinations.

Fraudulent Information

When completing insurance applications or otherwise providing information to insurance companies, it is important that individuals be warned against committing fraud or lying on their applications. Unfortunately, it is not uncommon for patients to be incorrectly advised that, as long as a genetic test result is not in the medical records, they can state to an insurance company that they have not been tested. This ill-advised tactic can create substantial problems for them in the future. For example, if a long-term care insurer discovers that an individual committed fraud on an application, they can likely revoke the coverage and past reimbursements. If this discovery occurs after an individual has been in a nursing home for a number of years, it can result in a considerable financial obligation that the patient will then owe to the facility since the insurance is very likely to retroactively revoke past reimbursement payments.

Similarly, although GINA shields individuals from disclosing genetic information to health insurers and employers in most cases, it does not sanction fraud. If, in violation of GINA, a covered entity asks for genetic information, the appropriate response would be not to lie about testing results or family history, but rather to choose not to answer these questions.

Table 1 Protections of the collection and use of Genetic Information (GI) by entity

A. Health Insurance (HI)

Private HI
- GINA usually applies
- Collection of GI is banned except for payment decisions
- Use of GI to discriminate is banned

Group HI through military, federal or state government
- GINA does not apply
- Collection of GI may be allowed
- No medical information, including GI used for rates and eligibility

B. Employment

Private employer
- 15 or more employees
  - GINA applies
  - Collection of GI generally banned with few exceptions
  - Use of GI to discriminate is banned
- Employed in a state with an applicable law
  - Both GINA and state law may apply
  - Determine best jurisdiction to file complaint
- Fewer than 15 employees
  - GINA does not apply, but state law may apply

State or local government employer
- GINA applies
- Collection of GI generally banned with few exceptions
- Use of GI to discriminate is banned

Military or federal government employer
- GINA generally does not apply
- Collection of GI is not banned
- Use of GI to discriminate under some circumstances (military)
- Federal employees, see Executive Order 13145; file complaints via Equal Opportunity Officer;

Military employees, see employee manual

C. Life, Long-term Care, or Disability Insurance (Supplemental Insurance)
- GINA does not apply
- State law may provide some protections
- Possible appeal of denials if no fraudulent information given

Conclusion

It is a promising sign that there have been increasing numbers of both state and federal laws passed to protect individuals from genetic discrimination. The resulting patchwork of legislation, however, creates important gaps relevant for genetic counselors and their patients (Table 1). The limited awareness of these laws, by both the public and health care professionals, greatly restricts their potential effectiveness. It is crucial that patients have access to credible information about the existing laws, as there may be actions they can take to help protect their genetic information and lower their risk of genetic discrimination, thereby preventing future harm to themselves and their families. Genetic counselors have both the unprecedented opportunity and the professional responsibility to disseminate accurate knowledge of existing legal protections to their patients. By acquiring additional knowledge of how these protections might apply to their practice, genetic counselors could help ease some unfounded concerns about possible discrimination and enlighten patients about actions they can take to help protect their genetic information, wherever possible.

Acknowledgments The authors wish to thank Catherine Fine, MS, CGC for her helpful comments and suggestions in the preparation of this manuscript.

Ms. Roche is supported, in part, by the National Human Genome Research Institute of the National Institutes of Health, Grants Number P50HG004488 and U01HG006487 with additional support from the UNC Cancer Research Fund and the UNC Lineberger Comprehensive Cancer Center.

Ms. Prince is supported by the National Human Genome Research Institute of the National Institutes of Health, Grant Number P50HG004488.

Conflict of interest Author Anya E. R. Prince declares no conflict of interest.

Author Myra I. Roche declares no conflict of interest.

Human and Animal Rights No animal or human studies were carried out by the authors for this article.

References

Ader, T., Susswein, L. R., Callanan, N. P., & Evans, J. P. (2009). Attitudes and practice of genetic counselors regarding anonymous testing for BRCA1/2. *Journal of Genetic Counseling, 18*(6), 606–617.

Allain, D. C., Friedman, S., & Senter, L. (2012). Consumer awareness and attitudes about insurance discrimination post enactment of the genetic information Nondiscrimination Act. *Familial Cancer, 11*(4), 637–644.

Americans with Disabilities Act of 1990 (ADA), Pub. L. 101-336. (1990).

Americans with Disabilities Act Amendments Act (ADAAA) of 2008, Pub. L. 110-325. (2008).

American Medical Association (AMA). (2013). *Report 7 of the Council on Science and Public Health: Genetic Discrimination and the Genetic Information Nondiscrimination Act*. Available at: http://www.ama-assn.org/resources/doc/csaph/a13csaph7.pdf (last visited March 26, 2014).

Association for Molecular Pathology (AMP) et al. v. Myriad Genetics, Inc. et al., 569 U.S. 12-398. (2013).

Baruch, S., & Hudson, K. (2008). Civilian and military genetics: nondiscrimination policy in a Post-GINA World. *The American Journal of Human Genetics, 83*(4), 435–444.

Bombard, Y., Palin, J., Friedman, J. M., Veenstra, G., Creighton, S., Bottorff, J. L., & Hayden, M. R. (2012). Beyond the patient: the broader impact of genetic discrimination among individuals at risk of Huntington disease. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 159B*(2), 217–226.

Cal-GINA, Senate Bill No. 559. (2011). (codified in scattered sections of the California code).

Coalition for Genetic Fairness (CGF). GINA: An Overview. November 2008. Available at: http://www.geneticfairness.org/ginaresource_overview.html. Accessed 10 July 2014

Cogent Research. 'Cogent Genomics, Attitudes & Trends study (CGAT).' Survey. (2010). Summary available at: http://www.councilforresponsiblegenetics.org/GeneWatch/GeneWatchPage.aspx?pageId=324 (last visited March 26, 2014).

Code of Federal Regulations (CFR), Definitions Specific to GINA, 29 C.F.R. §1635.3. (2013).

Dorsey, E. R., Darwin, K. C., Nichols, P. E., Kwok, J. H., Bennet, C., Rosenthal, L. S., Bombard, Y., et al. (2013). Knowledge of the Genetic Information Nondiscrimination act among individuals affected by Huntington disease. *Clinical Genetics, 84*(3), 251–257.

Equal Employment Opportunity Commission (EEOC). (2014). Founders Pavilion Will Pay $370,000 to Settle EEOC Genetic Information Discrimination Lawsuit [Press release]. Retrieved March 26, 2014 from http://www.eeoc.gov/eeoc/newsroom/release/1-13-14.cfm.

Genetic Information Nondiscrimination Act of 2008 (GINA), Pub. L. No. 110-233. (2008). (codified as amended in scattered sections of 26, 29, and 42 U.S.C.).

Goh, A. M., Chiu, E., Yastrubetskaya, O., Erwin, C., Williams, J. K., Juhl, A. R., & Paulsen, J. S. (2013). Perception, experience, and response to genetic discrimination in Huntington's disease: the Australian results of The International RESPOND-HD study. *Genetic Testing and Molecular Biomarkers, 17*(2), 115–121.

Guz v. Bechtel National, Inc., 24 Cal. 4th 317,8 P.3d 1089, 100 Cal. Rptr. 2d 352. (2000).

Hadley, D. W., Jenkins, J., Dimond, E., Nakahara, K., Grogan, L., Liewehr, D. J., Steinberg, S. M., et al. (2003). Genetic counseling and testing in families with hereditary nonpolyposis colorectal cancer. *Archives of Internal Medicine, 163*(5), 573–582.

Haga, S., Barry, W., Mills, R., Ginsburg, G., Svetkey, L., Sullivan, J., & Willard, H. (2013). Public Knowledge of and attitudes toward genetics and genetic testing. *Genetic Testing and Molecular Biomarkers, 17*(4), 327–335.

Hall, M. A., & Rich, S. S. (2000). Patients' fear of genetic discrimination by health insurers: the impact of legal protections. *Genetics in Medicine, 2*(4), 214–221.

Hall, M. A., McEwen, J. E., Barton, J. C., Walker, A. P., Howe, E. G., Reiss, J. A., Power, T. E., et al. (2005). Concerns in a primary care population about genetic discrimination by insurers. *Genetics in Medicine, 5*(7), 745.

Huang, M. Y., Huston, S. A., & Perri, M. (2013). Awareness of the US Genetic Information Nondiscrimination Act of 2008: an online survey. *Journal of Pharmaceutical Health Services Research, 4*(4), 235–238.

Huntsman Cancer Institute. 'Measuring Public Perception about Cancer Prevention, Treatment and Research.' Survey. (2013). Available at http://healthcare.utah.edu/cancer-poll (last visited March 26, 2014).

Jolie, A. (2013). My Medical Choices. *The New York Times*, p. A25.

Joly, Y., Feze, I. N., & Simard, J. (2013). Genetic discrimination and life insurance: a systematic review of the evidence. *BMC Medicine, 11*, 25.

Kocarnik, J., & Fullerton, S. (2014). Returning pleiotropic results from genetic testing to patients and research participants. *Journal of the American Medical Association, 311*(8), 795–796.

Laedtke, A. L., O'Neill, S. M., Rubinstein, W. S., & Vogel, K. J. (2012). Family physicians' awareness and knowledge of the Genetic Information Non-Discrimination Act (GINA). *Journal of Genetic Counselin\g, 21*(2), 345–352.

Lapham, E. V., Kozma, C., & Weiss, J. O. (1996). Genetic discrimination: perspectives of consumers. *Science, 5287*(274), 621–624.

Matloff, E. T., Shappell, H., Brierley, K., Bernhardt, B. A., McKinnon, W., & Peshkin, B. N. (2000). What would you do? Specialists' perspectives on cancer genetic testing, prophylactic surgery, and insurance discrimination. *Journal of Clinical Oncology, 12*(18), 2484–2492.

Matloff, E. T., Bonadies, D. C., Moyer, A., & Brierley, K. L. (2014). Changes in specialists' perspectives on cancer genetic testing, prophylactic surgery and insurance discrimination: then and now. *Journal of Genetic Counseling, 23*(2), 164–171.

National Conference of State Legislatures (NCSL). (2008, January). Genetic Employment Laws. Retrieved March 26, 2014 from http://www.ncsl.org/research/health/genetic-employment-laws.aspx.

National Conference of State Legislatures (NCSL). (2008). Genetics and Life, Disability, and Long-term Care. Retrieved March 26, 2014 from http://www.ncsl.org/research/health/genetic-nondiscrimination-laws-in-life-disability.aspx.

National Human Genome Research Institute (NHGRI). (2014). Genetic discrimination. Retrieved March 26, 2014, from http://www.genome.gov/10002077#al-3.

New Jersey Code, NJS § 17B:30-12. (2008).

Pamarti, A. (2011). *Genetic Information Nondiscrimination Act (GINA) and Its Affect on Genetic Counseling Practice: A Survey of Genetic Counselors*. Unpublished Masters Thesis. Brandies University, Massachusetts.

Patient Protection and Affordable Care Act, 42 U.S.C. § 18001 et seq. (2010).

Pollitz, K., Peshkin, B. N., Bangit, E., & Lucia, K. (2007). Genetic discrimination in health insurance: current legal protections and industry practices. *Inquiry, 44*(3), 350–368.

Prince, A. (2012). Genetic information and medical records – a cautionary tale for patients, healthcare professionals, and insurance companies. *Health Lawyer, 5*(24), 29–33.

Rothstein, M. A. (2008). GINA, the ADA, and genetic discrimination in employment. *Journal of Law, Medicine, & Ethics, 36*(4), 837–840.

Schultz, D. (2013). It's Legal for Some Insurers to Discriminate Based on Genes. National Public Radio [Internet], Retrieved April 2, 2014 from: http://m.npr.org/news/Health/169634045.

Sharpe, N. F., & Carter, R. F. (2006). *Genetic testing: Care, consent, and liability* (p. 153). New Jersey: John Wiley & Sons.

Small Business Administration (SBA). (2011). Firm Size Data. Retrieved March 26, 2014 from http://www.sba.gov/advocacy/849/12162.

Further Reading

Council for Responsible Genetics (CRG). State Laws on Genetic Privacy. http://www.councilforresponsiblegenetics.org/geneticprivacy/map_statelaw.html.

Equal Employment Opportunity Commission (EEOC) – Where individuals must file a complaint of employment discrimination.

Gina Help: www.ginahelp.org - Website offering information about GINA and the gaps in the law.

Cancer Legal Resource Center – Organization has a free national telephone assistance line where patients can ask questions about cancer and genetic-related legal issues.

National Human Genome Research Institute - https://www.genome.gov/10002328.

Patient Advocate Foundation – Organization offers assistance with insurance appeals as well as general resources for patients.