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Confidentiality & the Risk of Genetic Discrimination: What Surgeons Need to Know

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Abstract

In the past decade, federal and state laws have been passed to provide legal protections against genetic discrimination as it pertains to employment, health coverage and rates. Much of the public and a notable portion of medical providers are not aware of the legislation surrounding genetic discrimination and unnecessary concerns about genetic privacy can get in the way of opportunities to deliver the optimal medical care. Patient health information including genetic testing and family history are protected under Health Insurance Portability and Accountability Act (HIPAA) and the Genetic Information Nondiscrimination Act (GINA). Additional protections are afforded through the American with Disabilities Act, state laws, and the Affordable Care Act. Communicating a genetic test result back to a patient is important for medical management decisions. Physicians also have a duty to warn patients of genetic findings and that the patient's relatives are at risk from a genetically transferable condition. Medical care providers cannot, however, inform relatives in the absence of patient permission. Because management of a patient with a positive genetic test extends to the family members, it is important to empower and provide the tools for the patient to communicate with relatives. A discussion of family implications includes benefits and challenges to the larger family, maintaining confidentiality in light of sharing information with family members, and disclosure of result if patient dies. Emerging issues include the use of whole genome sequencing for both germline and tumor DNA and confidentiality in the era of social media.

Keywords

genetic discrimination; privacy; GINA; HIPAA; genetic testing; legal; hereditary cancer

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Introduction

Fears about genetic discrimination plague patients and providers alike. These concerns can arise throughout the genetic testing process: before testing, after results are received, and when patients are deciding what type of cancer screening or risk reduction measures they should take based on their results. Some of the major concerns expressed by patients and providers around genetic discrimination include:

- "Could I lose my health insurance if my genetic test comes back positive?"
- "If I have genetic testing, will my insurance premiums increase?"
- "What if my employer finds out I'm at high risk for cancer – could I be fired or demoted?"
- "Should I recommend genetic testing for my patient if it could cause him/her problems obtaining health insurance?"

When genetic testing for hereditary cancer risk first became clinically available in the mid-1990s, comprehensive legal protections prohibiting genetic discrimination did not exist. Both patients and clinicians were wary of the potential for genetic discrimination.

The current state of affairs in the U.S. is quite different. Both federal and state laws are now in place to prohibit many forms of genetic discrimination. Despite this, many patients and clinicians are unaware of these protections. A 2010 study conducted by Parkman et al. used questions added to the Behavioral Risk Factor Surveillance System (BRFSS) survey in four states to assess public knowledge regarding legal protections from genetic discrimination.[1] Only 13.3% – 19.1% of respondents indicated that they were aware of laws (such as the Genetic Information Nondiscrimination Act, GINA) that "prevent genetic test results from being used to determine health insurance coverage and costs".[1] In 2009, Laedtke et al. sent surveys to 1500 members of the American Academy of Family Physicians assessing their knowledge of GINA and their concerns regarding genetic discrimination.[2] Of the 401 physicians who responded, over half (54.5%) were not aware of GINA and 44% were "highly concerned" about their patients' potential risk for genetic discrimination in health insurance.[2]

In this article, we will review the current legal protections against genetic discrimination, how they can affect both patients and their families, and perspectives on how current protections will be applied to an ever changing genetic testing landscape. We will focus on how this information can be used by surgeons to help reassure their patients regarding the protections that exist and also educate them about loopholes where safeguards are not currently in place.

Current Legal Framework in the United States regarding Genetic Discrimination and Genetic Information Privacy

The Americans with Disabilities Act (ADA)

The Americans with Disabilities Act was passed in 1990.[3] The primary purpose of the ADA is to prevent discrimination against individuals with disabilities in the workplace and to set enforceable standards for accessibility in public and commercial buildings, transportation, and communication services (specifically TDD/telephone relay services).[3,4] It provides some limited protections regarding genetic discrimination with regard to hereditary cancer predispositions to individuals employed by an employer with 15 or more employees.[3] Some state laws also ban employers from discriminating against individuals on the basis of disability. If an individual has a genetic disease which causes symptoms that significantly impair a person's ability to perform one or more functions, then their disease qualifies as a disability under the ADA.[3,5–7] This would then afford an individual protection from employment discrimination under the ADA, as long as they are able to perform the duties of their job with reasonable accommodations. Some hereditary cancer syndromes can be associated with cognitive impairment (such as *PTEN* hamartoma tumor syndrome), which may classify as a disability for some individuals.[8,9] Others may have experienced debilitating effects following their cancer treatment which could potentially rise to the level of a disability.[10,11] However, most individuals with hereditary predispositions to cancer do not have disease effects which rise to the level of disability.

Confidentiality and Health Insurance Portability and Accountability Act (HIPAA)

HIPAA is the Federal Health Insurance Portability and Accountability act of 1996. The primary goal of the law is to make it easier for people to keep health insurance (Title 1, Portability), protect the confidentiality and security of healthcare information and help the healthcare industry control administrative costs (Title II Administrative Simplification) [12]. In 2013, the HIPAA privacy rule was modified to prohibit most health plans from using or disclosing genetic information (individual or family) for underwriting purposes [12]. This includes determining eligibility, benefits under the plan, coverage, and premiums.

The portability section of HIPAA provides rules for continuity in health insurance coverage for individuals and their families if they change jobs. It limits restrictions that a group health plan can place on benefits for preexisting conditions. Health plans cannot consider pre-existing conditions if there is no more than a 63-day lapse in coverage.

The HIPAA Privacy Rule provides federal protection for individually identifiable health information. This includes information in patient health record, conversations with care providers, billing information, and information that the patient is seen at the clinic. It has three major components: how data is protected, when it can be disclosed, and the patient's rights to this information.

Data Protection—The HIPAA Security Rule establishes national security standards for protecting health information that is held or transferred in electronic form. In general these

laws apply to “covered entities” which include health plans, most health care providers, business associates and subcontractors of covered entities, and healthcare clearinghouses.

Disclosure—Disclosure of health information is permitted for treatment, care and payment. It can also be disclosed to others the patient identifies as involved with health care termed personal representatives. It can be disclosed to protect the public’s health (e.g. contagious conditions), and police reports (e.g. gunshot wounds).

Patient rights—Patients have the right to see and obtain a copy of health records, have corrections added, receive notice of how your information may be used and shared, provide a report on when and why health information was shared, and can file a complaint with the health provider, insurer or U.S. Government.

Organizations who do not have to follow Privacy and Security Rules—Examples of other organizations who do not have to follow Privacy and Security Rules include: Life insurers, schools, workers compensation carriers, law enforcement, many state agencies like child protective services. An employer can also ask for a doctor’s note or other information if an employer needs information to administer sick leave, worker’s compensation, wellness programs, or health insurance. However, the health care provider can’t release this without authorization from the patient.

State laws prohibiting genetic discrimination—Prior to the enactment of the Genetic Information Nondiscrimination Act (GINA) in 2008, many states had laws prohibiting genetic discrimination (Slaughter 2008, Prince 2014, NCSL website).[6,7] One difficulty with these laws is that they differ from state to state. Some laws only extend protection to people with individual health care policies and not group policies, or vice versa.[13] Other laws are very narrow – for example, only focusing on requiring patient consent before genetic information can be shared with his/her health insurance company.[13] Since hereditary cancer predispositions are by nature a family affair – the proband initially seeking genetic testing could live in a state with strong legislative protections while their potentially at-risk relatives could reside in states with no protections.[6] This presented quandaries for patients whose families were spread throughout the US before GINA provided a baseline level of protection across the nation.

As of 2014, 48 states plus Washington D.C. have laws in place which prohibit forms of genetic discrimination in health insurance.[14] Regarding employment protections, 35 states plus Washington D.C. have laws prohibiting some types of genetic discrimination.[14] Of note, some state laws provide stronger protections than GINA, or prohibit genetic discrimination in other areas not covered by GINA (such as life insurance or long-term disability insurance). GINA is explicitly written so that the most comprehensive law in effect for an individual person (whether that be GINA or a stronger local law) takes precedence.[15] For a complete list of the state protections regarding genetic discrimination in health insurance enacted prior to GINA, please see the list compiled by the National Conference of State Legislatures.[13]

Genetic Information Nondiscrimination Act (GINA)—The Genetic Information and Non-Discrimination Act (GINA) was the culmination of 13 years of debate at the federal level regarding the best way to provide genetic discrimination protections to the American public.[7,15–17] The first such bill was introduced to congress in 1995 and not until May 21, 2008 was the final bill, GINA, enacted; GINA's protections fully came into effect over the course of the next three years. Lauded as the "first major civil rights bill of the century" by Senator Edward Kennedy, GINA provides the majority of the American public a baseline level of protection against genetic discrimination in the workplace and health insurance realms.[16]

As its name suggests, GINA involves regulations regarding the collection and use of genetic information as well as prohibitions on genetic discrimination.[15,17] GINA specifically applies to health insurance plans and employers (with a few exceptions noted later). It is important to note that GINA does not provide legal protections against genetic discrimination in other types of insurance underwriting or enrollment, including life insurance and short/long term disability insurance.[6,15,17]

GINA defines genetic information broadly – including not only genetic test results, but genetic services (such as documentation of a patient meeting with a genetic counselor, and family health history as well (see Boxes 1 and 2).[17] With few exceptions (see Box 3), it prohibits health insurance plans and employers from requesting, requiring, or collecting genetic information on an individual or their family members (out to 4th degree relatives).[15,17] One exception is that a health insurance plan may request the results of an individual's genetic test (or family history) to determine coverage of a procedure. Also, if no genetic testing has been performed and an individual is requesting a specific procedure because of their family history of cancer, a health insurance plan may request that genetic testing be completed. This could apply to a woman with a *BRCA1* mutation requesting that her insurance cover an annual breast MRI or prophylactic mastectomy due to her high lifetime risks for breast cancer. Her insurance company may request to see a copy of her *BRCA1* genetic test results in order to confirm that she is truly at high risk for breast cancer and that the requested screening/surgery is warranted. Under GINA, employers are also allowed to collect genetic information in certain limited circumstances – for example, collecting family health history as part of a workplace wellness program.[17]

Box 1

GINA's protections regarding genetic information privacy

Individual and group health insurance plans cannot:

- require an individual to undergo genetic testing for underwriting or enrollment purposes (i.e. determining premiums, starting/terminating coverage, etc.)
- request genetic information (genetic test results, information on genetic assessment services pursued by the patient, or family history information) for underwriting or enrollment purposes

Most employers cannot:

- request an individual to undergo genetic testing for hiring, termination, promotion, or placement decisions
- request information (genetic test results, information on genetic assessment services pursued by the patient, or family history information) on an individual for hiring, termination, promotion, or placement decisions

Box 2

GINA's protections from genetic discrimination

Health Insurance:

Most individual and group health insurance plans **cannot**:

- use an individual's genetic information for underwriting or enrollment purposes (i.e. determining premiums, starting/terminating coverage, etc.)

Most individual and group health insurance plans **can:**

- request genetic information (i.e. genetic test results, family history, etc.) for the purpose of determining coverage of a specific procedure/claim (i.e. a cancer screening, prophylactic surgery, etc.)

Employers:

Most employers with more than 15 employees cannot use an employee's genetic information for hiring, termination, promotion, or placement decisions

Box 3

Insurance providers and employers who do not need to comply with GINA

Health insurance providers: Federal government employees, Military, Veteran's Administration, Indian Health Services

Employers: Military, Federal government, Employers with fewer than 15 employees

Other forms of insurance: Life insurance, Short/long-term disability insurance

Regarding health insurance protections, GINA prohibits most health insurance policies from using an individual's genetic information for underwriting purposes.[6,17] Under GINA, a person's current health insurance policy cannot be terminated due to genetic information, nor can their premiums be raised.[15,17] When an individual is applying for health insurance coverage or changing policies, genetic information cannot be used to decide whether or not that individual will be covered. GINA applies to most group and individual health insurance policies – some notable exceptions include health insurance provided through the military, Veteran's administration, Indian Health services, or to Federal employees.[17] Many of these organizations have other protections in place that are similar to GINA, but may have some restrictions or gaps in protection. If an individual has served in the military for at least six

months and is later found to have a genetic condition, they are typically still eligible for health insurance benefits[18] The Veteran's Administration follows a similar policy – veterans generally cannot be denied benefits through the VA on the basis of a genetic disease that was diagnosed after the individual started military service.

GINA also provides protections against genetic discrimination in the workplace. Most employers with 15 or more employees fall under GINA's provisions. Under GINA, employers are not allowed to use genetic information in hiring, firing, or promotion decisions. The military does not fall under GINA's employment provisions, but as mentioned above has some similar protections in place. The military reserves the right to potentially use an individual's genetic information to assist with duty assignments.[18] Some genetic testing can be required by the military, such as mandatory testing for sickle cell anemia.[18] Individuals employed by the Federal government are not covered under GINA either. However, they have protections under Executive Order 13145 (To Prohibit Discrimination in Federal Employment Based on Genetic Information), issued by President Clinton in 2000.[19] While employers with fewer than 15 employees are exempt from GINA's provisions, prospective employees are not required to disclose genetic information in most situations.

One important nuance of GINA is that is written specifically to provide protections against genetic discrimination based on the use of a person's genetic information and not symptoms of their disease.[17] For example, a 40 year-old man who was recently diagnosed with Lynch syndrome, but has never developed cancer, would be protected from genetic discrimination with regard to health insurance and employment in most cases. However, if he later developed colon cancer, he could become at risk for adverse changes to his health insurance or employment on the basis of his cancer diagnosis. His employer and health insurance policy could not cite Lynch syndrome as the cause for their actions, as this is still protected genetic information under GINA, but they could use his colon cancer diagnosis as the reason for their actions. There are many other laws that can protect patients from this discrimination on the basis of health status. As explained in detail in the next section, the Affordable Care Act now prevents most individuals from experiencing discrimination in health insurance underwriting on the basis of pre-existing conditions, such as a cancer diagnosis.[20] The Americans with Disabilities Act prohibits most employers from using an individual's health status against them unless it is compromising his/her work duties in a way which cannot be resolved through reasonable accommodations.[3] Other programs available to workers to help when they are unable to perform their job due to medical problems include FMLA (Family and Medical Leave Act) and short/long term disability coverage.

GINA's interaction with the Affordable Care Act (ACA)—With the passing of the Affordable Care Act in 2010, GINA's protections were strengthened in an important way: by stating that individuals could not face health insurance discrimination on the basis of a pre-existing health condition, individuals whose hereditary cancer syndrome had manifested could now be protected.[5,20] This manifestation could include a malignancy (such as breast cancer in a woman with a *BRCA1/2* mutation) or a pre-malignant lesion (such as a colon polyp in an individual with Familial Adenomatous Polyposis) or a benign feature (macrocephaly in a man with Cowden syndrome). While GINA prohibited health insurance

discrimination on the basis of genetic test results, once an individual manifested symptoms of the cancer predisposition, GINA alone could no longer protect the individual from facing potential health insurance discrimination since the symptoms could be considered a pre-existing condition.[5,17] Now that the ACA outlaws the use of a person's pre-existing conditions to determine health insurance coverage or premium determination, both pre-symptomatic and symptomatic individuals are protected.[20] Through ACA, health insurance no longer needs to be tied to the employer. This relieves some concerns of losing health insurance because of genetic information when employed by a company of 15 or less employees. However, the enactment of the ACA does not make GINA irrelevant – GINA still provides important protections against genetic discrimination in employment. Given the mixed political sentiment regarding the ACA, some patients may be wary of relying on its protections regarding genetic discrimination. Given the low level of public awareness of GINA in comparison to the ACA, some patients can conflate the two when clinicians are discussing their protections. It is often beneficial to highlight to patients that GINA is an entirely separate piece of legislation, with wide support in Congress and among the public.

Wading through the misconceptions to deliver optimal medical care

Now that we have discussed the current legal protections against genetic discrimination, as well as the existing gaps in coverage, how can this information be applied to patients' surgical needs? Asking a few basic questions of your patients regarding their health insurance and employment can assist you in determining whether or not GINA's protections will apply. The following vignettes serve to highlight ways in which surgeons can help their patients navigate questions regarding genetic discrimination – see Table 1 for points to consider when addressing these patients' situations:

1. Before genetic testing occurs

   A 30 year-old woman presents to your clinic due to her family history of cancer – her paternal aunt and paternal grandmother were both diagnosed with breast cancer in their mid-30s/early 40s and passed away from metastatic disease. Her father has recently developed pancreatic cancer at age 60. Your patient is very worried about developing cancer and is interested in prophylactic mastectomy to reduce her breast cancer risk. She has considered genetic testing previously, but had heard she would lose her health insurance if she pursued it.

2. After genetic testing

   A 40 year-old woman recently tested positive for a *BRCA1* mutation after her sister was diagnosed with breast cancer at 38. She is interested in pursuing a prophylactic salpingo-oophorectomy and bilateral mastectomy as soon as possible. However, she fears that if her employer found out that she's at high risk for cancer, she would be let go, and is thus worried about taking time off to have the surgeries.

3. When symptoms are present

   A 20 year old male is referred to you for colectomy – he was recently found to have hundreds of adenomatous colon polyps. He is currently covered by his parents'

health insurance plan. His new diagnosis makes him worry about his ability to obtain his own health insurance policy later in life.

Table 1 helps to summarize the key points related to genetic discrimination protections and gaps that apply to each of the above patients, as well as some additional points regarding management. Most patients can be reassured that the combined protections of the laws described earlier in this article will protect them from genetic discrimination in the areas of health insurance and employment. In a busy surgical practice, an in-depth, lengthy discussion of every legal protection and loopholes is not feasible, nor should it be necessary, for all patients. A brief explanation will suffice in most instances. When your patient has a question that requires more research or they possibly fall into a gap in genetic discrimination protection, local genetics specialists are an excellent source of current knowledge and information. For example, some states have laws restricting use of genetic information to determine life, disability, and long-term care insurance. Individuals who are very concerned about obtaining life insurance should consider obtaining a policy prior to genetic testing, but understand that life insurance can use family history to determine eligibility. Box 4 includes the listing for the National Society of Genetic Counselors' website, where local genetic counselors specializing in cancer genetics can be found. This box also lists other resources which may assist your patients with additional information.

Box 4

Resources for providers and patients

National Conference of State Legislatures – List of state laws regarding genetic discrimination: http://www.ncsl.org/research/health/genetic-nondiscrimination-in-health-insurance-laws.aspx

GINAhelp.org – patient friendly information on GINA, its protections, and gaps: http://www.ginahelp.org/

HIPAA policies – summary of HIPAA and its protections: http://www.hhs.gov/ocr/privacy/hipaa/understanding/index.html

National Society of Genetics Counselors – resource for identifying genetic counselors in your local area: http://www.nsgc.org

Family Implications

Management of a patient with a positive genetic test extends to the family members as well. In most cases, the genetic cause of the patient's condition is inherited from the mother or father. As most hereditary cancer predispositions are inherited in an autosomal dominant manner, siblings and biologic children typically have a 50% chance of carrying the mutation with more extended family members having a risk as well. A few hereditary cancer predispositions have other patterns of inheritance, including MAP (*MUTYH*-associated polyposis) which is autosomal recessive and *SDHD* mutations causing hereditary paraganglioma/pheochromocytoma syndrome which demonstrate a maternal imprinting effect. Another exception is when a patient has a *de novo* mutation - a mutation that arose in

that individual shortly after conception but was not inherited from a parent. Other articles in this issue have addressed these inheritance patterns. Risk perception can also influence who patients inform about their genetic test results. Multiple studies have shown that individuals with a *BRCA1/2* mutation are more likely to inform their female relatives about the mutation than their male relatives, due to higher lifetime cancer risks seen in women who have a *BRCA1/2* mutation.[21,22] This is in spite of the fact that men have increased cancer risks associated with *BRCA1/2* mutations and are just as likely to pass the mutation on to their children. It is important for clinicians to help educate their patients about the inheritance pattern of the syndrome within their family to assist in correctly identifying at risk relatives.

The clinical provider has an obligation to maintain the confidentiality of his/her patient's genetic test result under HIPAA guidelines. In an ideal situation, the patient is willing to share the genetic results and has the tools to properly communicate this to family members who may be at risk for carrying the genetic mutation. This becomes an ethical challenge when the individual tested does not share the information with family members.

Key points to empower family communication

- Physician has a duty to warn the patient that their relatives are at risk from a genetically transferable condition [23].
- Patient should tell family members about their genetic test results because it is not just their individual result - it affects the health of their biologic relatives.
- Patient should be provided with a copy of the test report so that it can be easily shared with family members without having to sign medical record release forms.
- When an individual presents for genetic testing for a known syndrome within his/her family, an official copy of one of their relative's positive genetic test results is needed to ensure the individual's test are ordered correctly and in the most cost-effective manner.
- Identify a plan for communicating genetic information upon patient's death.
- Ultimately it is the patient's choice of what information they will share and what procedures they will undergo.

Benefits and challenges to the larger family

The benefits of germline genetic testing are that if the mutation is known, conclusive genetic testing can be offered to family members at a fraction of the cost and medical management will be specific to their genetic status. When the specific mutation is known, the appropriate technology can be applied so that a false negative result can be avoided. Mutation carriers will be educated on appropriate cancer screening and risk reduction and non-carriers can avoid unnecessary worry and procedures.

Most patients elect to communicate their test results to relatives. A 2013 study showed that *BRCA1/2* mutation carriers shared their test results with 73% of their at-risk first and second degree relatives.[22] Family dynamics, however, can present challenges that can get in the way of sharing results. Some patients may be estranged or have lost contact from some or all

of their relatives, inhibiting disclosure of test results. Some patients may have cultural factors that influence their willingness to share genetic test results or undergo genetic testing.

Conversely, a patient may be eager to share their genetic test results, but may run into resistance from family members. Some individuals do not want to know if they have a hereditary cancer risk, or they may wish to block other relatives, like children, from learning the information.[26] A recent review by Sharaf et al in 2013 summarized eight published studies looking at uptake of genetic testing for Lynch syndrome among relatives of individuals who had tested positive.[27] Importantly, each of the studies covered in this review paper recruited patients during the mid-1990s to mid-2000s.[27] Genetic testing uptake reported among first degree relatives in these reviewed studies ranged from 34% – 52%.[27] Some studies that did not delineate between uptake in first degree or extended relatives showed higher uptake rates, up to 75%.[27] These numbers may improve with the more recent improved legal protections regarding genetic discrimination.

A genetic diagnosis also has the potential to change family relationships in positive and negative ways.[28] Some individuals experience guilt and worry about whether or not they could have passed the condition on to their children.[28,29] Patients sometimes experience conflict with family members over their decision to pursue genetic testing or certain management decisions (i.e. prophylactic surgery).[28,29] Relatives who are found not to carry the mutation can also experience guilt – wondering why they were spared from having increased cancer risks. Genetic counselors and other clinicians are trained to help patients prepare for a range of potential reactions from their family members.

Maintain confidentiality in light of sharing information with family members

Does a physician have a duty to warn patients' relatives of their risk for a hereditary condition? [3031]. Two legal cases *Pate v. Threlkel (1995)* and *Safer v. Estate of Pack* (1996) found that the physician has a duty to inform the patient that their genetic condition was transferable to offspring and *Safer v. Estate of Pack* extended the duty to warn to members of the patient's immediate family [23]. Under the HIPAA privacy rule which followed these legal decisions, divulging the genetic result to third parties without patient's permission is not allowed, so the physician is left with relying on the patient to facilitate this communication.

Disclosure of result if patient dies

One may consider genetic information belonging to the family, especially after the death of a patient, however the laws surrounding privacy change over time and are by no means consistent across different jurisdictions [32]. There is a fine balance between respecting the privacy and wishes of the deceased versus the health interests and needs of the family. The obstacle has been that the HIPAA privacy rule applies to both alive and deceased and the interpretation continues to evolve. In 2013 HIPAA was modified to extend privacy protection of health information for 50 years after death, but considered the needs of family and care takers. Disclosure is now permitted to family members or others involved in an individual's care before death, even when this person is not the designated personal

representative [33]. The exceptions are if the deceased has expressed otherwise or if the practitioner is uncomfortable doing so. In order to disclose genetic results upon patient death, the providers must make reasonable assurances that the requester is a relative or involved in the care through documents, past interactions, or reasonable discretion, and check for patient's preferences about disclosure.

Interestingly, a survey of research biobank participants showed that 52% would want their results returned to their nearest biologic relative after death, 30% would designate someone other than a biological relative and 9% would not want results disclosed after death[34].

Emerging issues

Genome sequencing

With the increasing access to affordable whole genome sequencing, the genomes of patients and their tumors are being sequenced as standard clinical care [35].

Sequencing of the tumor will become increasingly common and important to precisely target chemotherapy. Sequencing of the tumor can reveal germline genetic predispositions as well. This may include mutations in cancer syndrome genes which explain the origins of the cancer as well as completely unrelated but medically actionable findings such as mutations leading to cardiac failure.

Sequencing of individual genomes is being requested clinically when there is concern of an underlying condition in patients or their children [36,37]. It can also be requested by individuals for preventative measures or even curiosity through their physician. A time may be coming when everyone will be sequenced at birth and use this information to manage their care throughout their lifetime [38]. Although this would create a broader opportunity for miss use, the HIPAA, GINA, and ADA laws in place should still offer protection.

Now, with newer HIPAA rules, the patient has rights to access their medical records, including complete access to the DNA sequence report and interpretation. Additionally, the ordering physician has a duty to warn the patient of the findings. One challenge is management and reporting of medically actionable incidental findings, that is, findings that are unrelated to the original reason for performing sequencing, are unexpected, and have immediate implications for clinical management. Up to 5% of the time, incidental findings result from sequencing, and clinical providers should be prepared to help the patient manage these through education and referral [36]. Ideally the risk of these findings should be discussed prior to sequencing. Engaging a genetic counselor early in the process of genome sequencing (tumor or germline) is of enormous benefit in that they can address these unanticipated risks, educate, and facilitate communication.

Social media/scenarios

Under GINA, federal protections are in place that disallow intentional acquisition of genetic information and this includes social media and web sites [39]. The key to this is "intentional" which opens the door to different levels of interpretation. It is not uncommon for employers and prospective employers to review the internet and social media sites when considering

someone for a position. This could lead to unintentional acquisition of genetic information which, legally, they could not act on. However, the burden of proof that an employer intentionally viewed this information and/or used it for an employment decision, is on the individual who suspects such a violation. Patients concerned about confidentiality of their genetic information should practice caution when sharing on the internet.

Conclusions

Overall, significant protections are currently in place for individuals residing in the U.S. related to genetic discrimination in employment and health insurance. Other countries have also pursued legislation prohibiting genetic discrimination. While reviewing all countries' protections/gaps is beyond the scope of this article, it is encouraging that the worldwide community is considering this important issue. As genetic testing becomes a greater part of all individuals' healthcare management, current laws are likely to change to address our evolving understanding of genetics. We encourage all clinicians to reach out to local genetics specialists to keep abreast of the evolving legal landscape surrounding genetic discrimination.

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Key points

- Federal and state laws are now in place to prohibit many forms of genetic discrimination including health insurance eligibility, coverage, and rates and employment.
- Patient health information including genetic testing and family history are protected under HIPAA and GINA.
- Some groups are not covered by current regulations.
- Physicians have a duty to warn patients that they and their relatives are at risk from a genetically transferable condition, but must rely on the patient to communicate with family members.

Table 1

Patient vignettes – Current protections and additional considerations

	Patient #1	Patient #2	Patient #3
Benefit to using genetic information as part of surgical decisions	Genetic testing could help clarify patient's cancer risks.	Prophylactic salpingo-oophorectomy and bilateral mastectomy are reasonable risk-reducing surgeries for a woman with a *BRCA1* mutation.	Colectomy is warranted in this patient based on his colonic polyp phenotype.
	Insurance may be more likely to cover prophylactic surgery if she is found to have a specific hereditary cancer predisposition.		
Legal protections from discrimination	Patient is most likely protected by GINA, HIPAA, ACA, and applicable state laws.	GINA makes it illegal for most employers to use her *BRCA1* mutation status to terminate her employment.	The ACA makes it illegal for most health insurance companies to use his clinical diagnosis of polyposis as a pre-existing condition to deny him health insurance coverage in the future.
	Her health insurance policy would be prohibited from using her genetic test results to alter or terminate her coverage.	In most situations, her employer cannot require her to reveal information regarding her *BRCA1* positive status.	
Exceptions to legal protection	If patient receives her health insurance through one of the entities not covered by GINA (table 3), there may be other protections in place that would apply (on the state level or through her insurance provider).	If patient is in the military, or works for an employer with fewer than 15 employees, she could be at risk for genetic discrimination by her employer.	The ACA should allow him to receive coverage by a health insurance plan in the future (either through an employer or via an individual policy).
Medical management options	Patient's father would be the ideal person in the family to first pursue genetic testing.	Salpingo-oophorectomy is recommended as there is no current effective screening for ovarian cancer.	Genetic testing could potentially clarify his type of polyposis (likely FAP or MAP).
		Breast cancer screening with mammogram and MRI is a reasonable alternative.	Medical insurance more likely to cover additional screening for associated cancers (upper GI, etc.) when genetic diagnosis can be made.
Other considerations	If her father tests positive for a hereditary cancer predisposition, genetic testing for your patient will be more targeted, less costly, and more likely covered by her insurance	Filing for FMLA for her surgery recovery period would further help to secure her employment.	