REPORT 7 OF THE COUNCIL ON SCIENCE AND PUBLIC HEALTH (A-13)
Genetic Discrimination and the Genetic Information Nondiscrimination Act
(Reference Committee E)

EXECUTIVE SUMMARY

Objectives. Genetic discrimination and the fear of it have negative effects on the delivery of clinical care. The Genetic Information Nondiscrimination Act (GINA), passed in 2008, is intended to protect individuals from genetic discrimination by health insurers and employers. However, GINA left unaddressed a number of areas in which individuals may experience genetic discrimination, and certain populations are not protected by its provisions. Other federal and state laws provide a patchwork of varied protections. Given the rapid advance of genomic technologies that are transforming health care, consistent and robust protections against genetic discrimination are needed and will help to foster patient trust and engagement in cutting-edge genomic based care. The Council has undertaken this review to briefly examine genetic discrimination and GINA and to identify gaps in protection and necessary steps toward strengthening protections.

Data Sources. Literature searches were conducted in the PubMed database for English-language articles published between 2000 and 2013 using the search terms "genetic discrimination," "genetic information nondiscrimination act" and "GINA," for the purpose of identifying articles detailing the history and recent cases of genetic discrimination, the impact that fear of genetic discrimination has on clinical care, the protective provisions of GINA and other laws, and assertions for strengthening protections. To capture reports that may not have been indexed on PubMed, a Google search was also conducted using the same search terms. Additional articles were identified by manual review of the references cited in these publications. The Library of Congress, Government Printing Office, and state databases were consulted for legislative language.

Results. Well-documented instances of genetic discrimination have occurred in recent history, and fears about genetic discrimination have led to refusal to undergo genetic testing among patients. This can result in serious health implications for individuals for whom genetic testing would be beneficial. Health care professionals also have expressed concern that their patients could experience discrimination after undergoing genetic testing. Many health care providers and patients are not aware of current protections against genetic discrimination, suggesting that efforts toward educating all health care professionals about protections are warranted. In addition, current protections must be strengthened since the shortcomings of GINA and other federal laws along with the inconsistency in state laws leave many patients vulnerable to genetic discrimination and misuse of their genetic information.

Conclusions. Patient care is negatively impacted by fear of genetic discrimination. GINA has afforded important protections, and increased awareness of it may reduce the fear. However, GINA leaves individuals vulnerable to discrimination in areas such as life, long-term care, and disability insurance, and does not extend to certain sectors of the population. The Council believes that the increasingly common uses of genetic information both inside and outside of the clinical setting and the difficulty in maintaining the privacy of individuals' genetic information, combined with the negative impact of the fear of genetic discrimination on patient care, make it essential that robust and comprehensive protections against genetic discrimination and misuse of genetic information be enacted.

Action of the AMA House of Delegates 2013 Annual Meeting: Recommendations in Report 7 of the Council on Science and Public Health Adopted in lieu of Resolution 511, and Remainder of Report Filed.

REPORT OF THE COUNCIL ON SCIENCE AND PUBLIC HEALTH

CSAPH Report 7-A-13

Subject:	Genetic Discrimination and the Genetic Information Nondiscrimination Act
Presented by:	Sandra A. Fryhofer, MD, Chair
Referred to:	Reference Committee E (Lawrence K. Monahan, MD, Chair)

INTRODUCTION

Genetic discrimination and the fear of it have negative effects on the delivery of clinical care. The Genetic Information Nondiscrimination Act (GINA), passed nearly five years ago, is intended to protect individuals from genetic discrimination by health insurers and employers.[1] GINA was hailed as the "first major civil rights bill of the new century,"[2] and indeed, the fear of genetic discrimination appears to have lessened among some patients since its passage.[3] However, GINA left unaddressed a number of areas in which individuals may experience genetic discrimination; it does not extend to life, long-term care, or disability insurance, and certain populations are not protected by its provisions. Other federal and state laws provide a patchwork of varied protections.

Given the rapid advance of genomic technologies that are transforming health care, the Council believes that consistent, robust protections against genetic discrimination are needed and will help to foster patient trust and engagement in care that while considered cutting-edge, has already become standard for an increasing number of medical conditions and treatments. The Council has undertaken this review to briefly examine genetic discrimination and GINA and to identify gaps in protection and necessary steps toward strengthening protections.

METHODS

Literature searches were conducted in the PubMed database for English-language articles published between 2000 and 2013 using the search terms "genetic discrimination," "genetic information nondiscrimination act" and "GINA," for the purpose of identifying articles detailing the history and recent cases of genetic discrimination, the impact that fear of genetic discrimination has on clinical care, the protective provisions of GINA and other laws, and assertions for strengthening protections. To capture reports that may not have been indexed on PubMed, a Google search was also conducted using the same search terms. Additional articles were identified by manual review of the references cited in these publications. The Library of Congress, Government Printing Office, and state databases were consulted for legislative language.

THE GENETIC INFORMATION NONDISCRIMINATION ACT

In 2008, after 13 years of effort on the part of many advocacy organizations including the American Medical Association (AMA), Congress passed GINA nearly unanimously.[1] Then-President George W. Bush signed it into law on March 21, 2008. GINA addresses discrimination in

---

Action of the AMA House of Delegates 2013 Annual Meeting: Recommendations in Report 7 of the Council on Science and Public Health Adopted in lieu of Resolution 511, and Remainder of Report Filed.

two areas, health insurance and employment. A summary of GINA's provisions can be found in the Table. Title I of GINA prohibits group and individual health insurers from using a person's genetic information in determining eligibility or premiums and prohibits health insurers from requesting or requiring that a person undergo a genetic test in order to collect genetic information on that person for underwriting decisions.[1] Title II of GINA prohibits employers from using a person's genetic information in making employment decisions such as hiring, firing, job assignments, or any other terms of employment; and prohibits employers from requesting, requiring, or purchasing genetic information about a person or their family members.[1]

For the purposes of GINA, "genetic information" is defined as a person's genetic test results, the genetic test results of a person's family members (up to and including fourth-degree relatives), any manifestation of a disease or disorder in a family member, and participation of a person or family member in research that includes genetic testing, counseling, or education.[1] A "genetic test" refers to any test that assesses genotypes, mutations, or chromosomal changes;[1] for example, tests to detect hereditary breast or colorectal cancer mutations, examination of the genetic properties of a tumor, tests to diagnose a genetic disease such as Huntington's, and carrier screening for disorders such as cystic fibrosis (CF).[2] Examples of tests that are not considered to yield genetic information are complete blood counts, cholesterol tests, and liver-function tests.[2]

Importantly, GINA does not prohibit health insurance underwriting or employment decisions based on current health status, including manifest disease of a genetic nature. Rather, it is intended to protect individuals with a genetic predisposition to disease that has not manifested, whether or not an individual has knowledge about that predisposition based on his or her own genetic test results or the genetic test results or manifestation of disease in a family member. GINA is based on the premise that it is unfair for a health insurer or an employer to make a decision about an individual based on a condition that may or may not actually develop in the future.[4] Therefore, GINA is protective only before genetic conditions become manifest. Once a person is symptomatic, GINA is no longer protective.

GENETIC DISCRIMINATION

Genetic discrimination is considered the differential and adverse treatment of asymptomatic individuals based solely on their or their family members' actual or presumed genetic characteristics.[5]

Cases of Genetic Discrimination

Well-documented instances of genetic discrimination have occurred in recent history. For example, in the 1970s, some states began to mandate sickle cell anemia screening for African-Americans. However, inadequate education and counseling about sickle cell disease resulted in confusion about the difference between carrying the sickle cell trait and having sickle cell disease.[6] Healthy carriers of the sickle cell trait suffered adverse employment actions, and a stigma developed that African-Americans were inherently more susceptible to genetic disease than were members of other ethnic and/or racial groups.[6]

In 2001, the Equal Employment Opportunity Commission (EEOC) filed a claim against Burlington Northern Santa Fe for testing its employees who developed carpal tunnel syndrome for a rare genetic condition that is sometimes causal of the syndrome.[7] Employees examined by company physicians were not told that the blood being drawn during the examination was being used for genetic testing. An employee who refused testing was threatened with termination.[7]

Several cases of health insurance discrimination have been published.[8-12] Below are examples.

- Two children who were carriers of a mutation that causes alpha-1 antitrypsin deficiency were denied coverage by their mother's health insurance company even though they would never develop the disease (alpha-1 antitrypsin deficiency is a recessive disease, so carriers who have only one copy of the mutation will not develop disease).
- A young boy who was a carrier of a mutation for Long QT Syndrome was denied coverage under his father's health insurance policy because of his "pre-existing condition," even though his condition was not manifest.
- A young woman who had undergone prophylactic mastectomy and hysterectomy was denied coverage when her health insurance company requested her medical records and discovered that she carried a *BRCA1* mutation associated with an increased risk of breast cancer.

Since the enactment of GINA's health insurance and employment provisions, only a modest number of genetic discrimination complaints have been filed under its provisions; in 2012, 280 cases of genetic discrimination were filed out of nearly 100,000 total discrimination cases filed.[13,14] It is possible that the small number of cases reflects the effectiveness of GINA at discouraging the practice of genetic discrimination in the health insurance and employment sectors, or alternatively, discrimination continues to occur but is unrecognized or unreported, possibly because awareness of GINA is low.

Fear of Genetic Discrimination

Fears about genetic discrimination have led to refusal to undergo genetic testing among patients.[15-18] This can result in serious health implications for individuals for whom genetic testing would be beneficial. Even among those who do undergo genetic testing, many withhold test results from their physicians, and some request that their results be placed in a "shadow chart" or withheld entirely from their medical record.[18-20] This lack of information can have detrimental effects on future care of the patient; treating physicians unfamiliar with the patient will have no record of genetic test results unless volunteered by the patient.

A majority of health care professionals surveyed also have expressed concern that their patients could experience discrimination after undergoing genetic testing.[21,22] Survey data demonstrate that those with the strongest concern about genetic discrimination are more likely not to refer patients to genetics professionals (medical geneticists and genetic counselors), effectively preventing their patients from receiving optimal care.[23]

Fear of genetic discrimination, on the part of both patients and physicians, also has detrimental effects on research. Potential research participants have refused to be part of genetic studies because of fear that their genetic test results might not remain confidential.[16]

Only a few studies assessing fear of genetic discrimination after the passage of GINA have been completed, but collectively, they find that despite the existence of GINA, fear has persisted among some groups. In a post-GINA survey of individuals who had considered genetic testing for hereditary breast and ovarian cancer, 60% indicated that they were worried about health insurance discrimination and 28% were worried about employment discrimination; 52%, 33%, and 34% were worried about life, disability, and long-term care insurance discrimination, respectively.[3] In another study, structured interviews with 64 patients at risk for genetic diseases revealed that they often did not trust how laws would work in real world circumstances, and would consider withholding genetic information or ask for it not to be included in their medical record.[19,20]

Like patients, some health care providers continue to worry about genetic discrimination after the passage of GINA. In a survey of family physicians, 49%, 44%, and 42% were "highly concerned" about discrimination in life, health, and long-term care insurance, respectively.[24] More than 80% of obstetrician-gynecologists and oncologists also report that they are very or somewhat concerned about genetic discrimination.[25]

Among patients reporting fear of genetic discrimination, improved knowledge of GINA and its protections appears to lessen the fear. After receiving information about GINA, more than half of individuals who had considered genetic testing for hereditary breast and ovarian cancer reported that the information made them less worried about genetic discrimination.[3] However, unlike patients, knowledge of GINA does not appear to lessen the fear of genetic discrimination among physicians. In a survey, family physicians who were knowledgeable about GINA reported being no less concerned about genetic discrimination than were family physicians with little or no knowledge about it.[24] More research is required to examine this finding as it may be the result of several factors, including doubt about the real-world utility of GINA's current protections or a belief that GINA's current protections are inadequate.

PHYSICIAN ROLE IN PROTECTING AGAINST GENETIC DISCRIMINATION

Genomic-based technologies are becoming an increasingly routine part of medical care. Every newborn, with few exceptions, undergoes a panel of genetic tests (which is continually expanding) at birth to detect inherited conditions that are vitally important to treat early in life.[26] Several clinical guidelines now include genetic testing, and the safe and effective use of many drugs requires knowledge of the patient's genotype.[27] Genetic tests are available for risk assessment, diagnosis, and/or management of nearly 3,000 diseases,[28] and whole-genome sequencing is gaining traction as a useful clinical tool.[29] Genomic data is also increasingly common in non-clinical applications. Direct-to-consumer genetic testing companies analyze customers' DNA to reveal information about non-medical traits, and genealogy services analyze customers' DNA samples to deliver information on genetic ethnicity.[30,31] With more frequent use of technologies that involve analysis of patients' genomic information, the potential for misuse and discrimination grows. In a troubling recent example, an 11-year old boy who carries a mutation for CF was reportedly ordered by school administrators to transfer to a different school for the protection of another student with CF, even though carriers do not pose a threat to those with CF or to anyone else.[32]

Physicians have historically advocated for measures to safeguard against the inappropriate use of patients' medical information, in part because use of such information to harm or penalize patients deters patients from seeking needed medical treatment. Fears of inappropriate use of medical information also undermine the truthful and accurate communication between patients and physicians essential to the provision of quality medical care.

Physician Knowledge of Protections Against Genetic Discrimination

A majority of physicians report being concerned about genetic privacy,[33] yet a gap in physician knowledge about GINA exists. For example, only approximately 10% of family physicians report being aware of GINA's existence and have a basic understanding of its protections.[24]

Although knowledge of GINA does not appear to reduce concerns about genetic discrimination among family physicians,[24] awareness of protections may have lessened the fear of genetic discrimination among other health care professionals. In a recent study, cancer genetics professionals who are familiar with protections afforded by federal laws other than GINA (the study was conducted before GINA's passage) reported less concern about genetic discrimination than did non-genetics professionals who were unfamiliar with protections.[34] This may reflect the importance given to genetic information by cancer genetics professionals, but it also suggests that efforts toward educating all health care professionals about protections are warranted. Such education could lead to more appropriate referral for genetic services and increased uptake of genetic testing among patients, ultimately resulting in better patient care.[34] Education of consumers and patients is also important because fear of discrimination may prevent individuals from speaking to their physicians about genetic testing in the first place.[3]

Physicians have a duty to keep their patients' genetic information confidential, yet dilemmas arise when such information has consequences for the patient's family members. Many physicians feel obligated to inform and/or treat relatives who may be at risk.[20] AMA Ethical Opinion E-2.131, "Disclosure of Familial Risk in Genetic Testing," states that physicians in this situation should counsel patients on the implications of genetic information for their relatives, and identify circumstances under which they would expect patients to notify relatives about their own genetic test results. A basic understanding of the protections afforded by anti-discrimination laws is needed for physicians who will likely get questions about potential misuse of genetic information from patients and relatives.

ADEQUACY OF CURRENT PROTECTIONS

The persistent concern about genetic discrimination among some health care professionals and patients is not unreasonable given the shortcomings of GINA. While GINA prohibits discrimination by health insurers, it does not extend to life, long-term care, or disability insurance. Additionally, some groups are not afforded GINA's protections. For example, employers with less than 15 employees are exempt from GINA's employment discrimination provisions.[35] Also, patients obtaining care through the Veterans Health Administration (VHA) and the Indian Health Service also are not protected by GINA, nor are federal civilian employees participating in the Federal Employee Health Benefits Program or U.S. military members participating in the Tricare program.[35] These exceptions exist because GINA amended existing health insurance and employment laws that do not apply to the aforementioned groups.[35] Some protections for these groups are afforded by Executive Orders (for federal civilian employees) or by internal policies similar to the protections afforded by GINA (U.S. military and VHA).[35]

In addition to GINA, other laws only partially protect against genetic discrimination in the health insurance realm. The Health Insurance Portability and Accountability Act (HIPAA) of 1996 specifically lists genetic information as protected health information and explicitly states that a genetic risk factor for disease cannot be considered a preexisting condition.[36] HIPAA prevents health insurers from increasing the cost of an individual's insurance discriminatorily, but insurance companies may raise an employer's group premiums based on the genetic information of its employees as a whole.[6] HIPAA also does not apply to the use of genetic information for individuals who purchase health insurance independently.[6] The Affordable Care Act's (ACA) protection against denial of health insurance due to preexisting conditions does not strengthen GINA's protections, since genetic information is not considered a preexisting condition under the ACA.[37]

In the employment realm, the Americans with Disabilities Act (ADA) prohibits employment discrimination based on a disability, the history of a disability, or a perceived disability.[38] However, it is not clear whether the ADA protects against genetic discrimination in employment decisions.[39] EEOC guidelines appear to conflict with court decisions that suggest genetic test results may be used in employment decisions.[6]

Adding complexity to the shortcomings of GINA is the patchwork of state laws addressing genetic discrimination. Slightly fewer than half of U.S. states have laws providing additional protection against discrimination in aspects of life, long-term care, and disability insurance, as well as in other areas, that are not present in GINA.[6,40] For example, California law prohibits genetic discrimination in such areas as housing, mortgage lending, education, life insurance and elections.[41] Arizona statute prohibits the use of genetic information in the underwriting of life and disability insurance policies.[42] In contrast, many states' protections are no more strict than those afforded by GINA.[40] Importantly, in states that provide more comprehensive protections than those provided by GINA, GINA does not preempt state law.

The shortcomings of GINA and other federal laws along with the inconsistency in state laws leave many patients vulnerable to genetic discrimination and misuse of their genetic information. Further, physicians are placed in the difficult position of explaining to patients confusing genetic discrimination protections that vary by state and by individual circumstance.

A very important additional consideration is how difficult it has become to maintain the privacy and security of genomic information. In October 2012, the Presidential Commission for the Study of Bioethical Issues concluded that efforts to de-identify such information are exceptionally challenging and will gradually become impossible.[43] Indeed, in January 2013, a group of scientists demonstrated that the genetic information provided by individuals who had been assured anonymity can in fact be re-identified.[44-46] Therefore, given the rapid uptake of genomic-based technologies in both the clinical setting and outside the clinic, there is a pressing need to move quickly to mitigate inappropriate uses of genomic information. It is often asserted that the important protections that GINA currently provides should be extended to cover other areas in which individuals could experience genetic discrimination, such as in life, long-term care, and disability insurance coverage.[6,43,47]

AMA POLICY ON GENETIC DISCRIMINATION

AMA policy and Ethical Opinion relating to genetic discrimination is listed in Appendix I. Briefly, AMA policy explicitly supports prohibitions on the use of genetic information in the context of health insurance. Policy H-185.972, "Genetic Information and Insurance Coverage," states that health insurance providers should be prohibited from: 1) using genetic information to deny or limit any health benefit coverage; 2) establishing differential rates or premium payments; 3) requesting or requiring collection or disclosure of genetic information; and 4) releasing genetic information without express prior written authorization of the individual. Policy H-165.856, "Health Insurance Market Regulation," similarly states that an individual's genetic information should not be used to determine his or her health insurance premium. Ethical opinions further address genetic information as it relates to genetic discrimination in health insurance. E-2.135, "Insurance Companies and Genetic Information," and E-2.137 "Ethical Issues in Carrier Screening of Genetic Disorders," state that genetic testing results should not be shared with health insurers or other third parties, and that health care providers should ensure that genetic testing results are removed before fulfilling requests to share medical records.

In the employment context, AMA policy is silent. However, Ethical Opinion E-2.132, "Genetic Testing by Employers," states that it is generally inappropriate to exclude workers with genetic risks of disease from the workplace because of their risk, and that the use of genetic testing to make employment decisions can result in unfair discrimination.

Lengthy AMA policy generally addresses patient privacy and confidentiality (H-315.983, "Patient Privacy and Confidentiality"), stating that genetic information should be kept confidential and should not be disclosed to third parties without the explicit informed consent of the tested individual. It further directs the AMA Board of Trustees to monitor and support federal legislation that will afford patients protection against discrimination on the basis of genetic testing.

AMA Legislative Principles

Early in 2013, the AMA Council on Legislation studied the issue of genetic discrimination and developed a set of legislative principles that could guide AMA advocacy activities in the absence of explicit AMA policy. The complete text of the principles can be found in Appendix II. Briefly, the principles state that prohibitions on genetic discrimination are essential to advancements in medical knowledge and clinical care, and it is part of a physician's duty to safeguard against the inappropriate use of patient medical information for non-medical purposes and to promote open and honest patient-physician communications. The principles further state that comprehensive federal protections against genetic discrimination are needed since patients remain at risk of discrimination in a broad array of areas. The AMA Board of Trustees approved the principles in March of 2013.

CONCLUSIONS

The AMA has been a strong opponent of discrimination based on genetic information, in part because patient care is negatively impacted by fear of such discrimination. GINA has afforded important protections, and increased awareness of it may reduce the fear. However, GINA leaves individuals vulnerable to discrimination in areas such as life, long-term care, and disability insurance, and does not extend to certain sectors of the population. Physicians are impeded in the delivery of care when patients are not forthcoming about genetic information or ask for measures such as withholding genetic information from medical records. Physicians also may be expected to be unreasonably fluent in detailed legal nuances of current protections. The Council believes that the increasingly common uses of genetic information both inside and outside of the clinical setting and the difficulty in maintaining the privacy of individuals' genetic information, combined with the negative impact of the fear of genetic discrimination on patient care, make it essential that robust and comprehensive protections against genetic discrimination and misuse of genetic information be enacted. Such protections would benefit physicians, the research community, and most importantly, patients.

RECOMMENDATIONS

The Council on Science and Public Health recommends that the following statements be adopted and the remainder of this report be filed:

1. That our American Medical Association (AMA) strongly oppose discrimination based on an individual's genetic information. (New HOD Policy)

2. That our AMA pursue and support legislation intended to provide robust and comprehensive protections against genetic discrimination and misuse of genetic information. (Directive to Take Action)

3. That our AMA support education for health care providers and patients on the protections against genetic discrimination currently afforded by federal and state laws. (New HOD Policy)

Fiscal note: $10,000

REFERENCES

1. The Genetic Information Nondiscrimination Act of 2008. http://www.gpo.gov/fdsys/pkg/PLAW-110publ233/html/PLAW-110publ233.htm. Accessed 03-27-12.

2. Hudson KL, Holohan MK, Collins FS. Keeping pace with the times--the Genetic Information Nondiscrimination Act of 2008. *N Engl J Med*. 2008;358(25):2661-3.

3. Allain DC, Friedman S, Senter L. Consumer awareness and attitudes about insurance discrimination post enactment of the Genetic Information Nondiscrimination Act. *Fam Cancer*. 2012;11(4):637-44.

4. Prince AE, Berkman BE. When does an illness begin: genetic discrimination and disease manifestation. *J Law Med Ethics*. 2012 Fall;40(3):655-64.

5. Rothstein MA, Anderlik MR. What is genetic discrimination, and when and how can it be prevented? *Genet Med*. 2001;3(5):354-8.

6. Barken, J. Judging GINA: Does the Genetic Information Nondiscrimination Act of 2008 Offer Adequate Protection? *Brooklyn L Rev*. 2009;75:545-77.

7. National Human Genome Research Institute. Cases of genetic discrimination. http://www.genome.gov/12513976. Accessed 03-27-13.

8. National Partnership for Women and Families on behalf of the Coalition for Genetic Fairness. Faces of Genetic Discrimination. 2009. http://www.nationalpartnership.org/site/DocServer/FacesofGeneticDiscrimination.pdf. Accessed 03-27-13.

9. Billings PR, Kohn MA, de Cuevas M, et al. Discrimination as a consequence of genetic testing. *Am J Hum Genet*. 1992;50(3):476-82.

10. Reilly PR. Genetic risk assessment and insurance. *Genet Test*. 1998;2:1-41.

11. Volpe LC. Genetic testing and health insurance practices: an industry perspective. *Genet Test.* 1998;2:9-12.

12. Pollitz K, Peshkin BN, Bangit E, Lucia K. Genetic discrimination in health insurance: current legal protections and industry practices. *Inquiry*. 2007;44:350-68.

13. Equal Employment Opportunities Commission. Genetic Information Nondiscrimination Act Charges. http://www.eeoc.gov/eeoc/statistics/enforcement/genetic.cfm. Accessed 03-27-13.

14. Equal Employment Opportunities Commission. Discrimination Charge Statistics FY 1997 Through FY 2012. http://www.eeoc.gov/eeoc/statistics/enforcement/charges.cfm. Accessed 04-14-13.

15. Lapham EV, Kozma C, Weiss JO. Genetic discrimination: perspectives of consumers. *Science*. 1996;274(5287):621-624.

16. Hadley DW, Jenkins J, Dimond E, et al. Genetic counseling and testing in families with hereditary nonpolyposis colorectal cancer. *Arch Intern Med*. 2003;163(5):573-582.

17. Peterson EA, Milliron KJ, Lewis KE, Goold SD, Merajver SD. Health insurance and discrimination concerns and BRCA1/2 testing in a clinic population. *Cancer Epidemiol Biomarkers Prev*. 2002;11(1):79-87.

18. McKinnon W, Banks KC, Skelly J, et al. Survey of unaffected *BRCA* and mismatch repair (MMR) mutation positive individuals. *Fam Cancer*. 2009;8(4):363-9.

19. Klitzman, R. Views of Discrimination Among Individuals Confronting Genetic Disease. *J Genet Counsel*. 2010;19:68–83.

20. Klitzman R. Exclusion of Genetic Information From the Medical Record: Ethical and Medical Dilemmas. *JAMA*. 2010;304(10):1120-1.

21. Freedman AN, Wideroff L, Olson L, et al. US physicians' attitudes toward genetic testing for cancer susceptibility. *Am J Med Genet A*. 2003;120A(1):63-71.

22. Nedelcu R, Blazer KR, Schwerin BU, et al. Genetic discrimination: the clinician perspective. *Clin Genet*. 2004;66(4):311-17.

23. Lowstuter KJ, Sand S, Blazer KR, et al. Influence of genetic discrimination perceptions and knowledge on cancer genetics referral practice among clinicians. *Genet Med*. 2008;10(9):691-98.

24. Laedtke AL, O'Neill SM, Rubinstein WS, Vogel KJ. Family physicians' awareness and knowledge of the Genetic Information Non-Discrimination Act (GINA). *J Genet Couns.* 2012;21(2):345-52.

25. Heald B, Melvin A, Madeo AC. Presented Abstracts from the Thirty First Annual Education Conference of the National Society of Genetic Counselors (Boston, MA, October 2012): Physicians & GINA: Awareness & Knowledge of Antigenetic Discrimination Laws Amongst Obstetrician-Gynecologists & Oncologists. Presented by Nehory BM, Ford JM, Kingham K. *J Genet Couns.* 2012;21(6):884-984.

26. National Newborn Screening and Genetics Resource Center. National Newborn Screening Status Report. January 2013. http://genes-r-us.uthscsa.edu/sites/genes-r-us/files/nbsdisorders.pdf. Accessed 03-28-13.

27. Personalized Medicine Coalition. The Case for Personalized Medicine, 3rd Edition. http://www.personalizedmedicinecoalition.org/sites/default/files/files/Case_for_PM_3rd_edition.pdf. Accessed 03-28-13.

28. GeneTests database. Growth of Laboratory Directory. 2012. http://www.ncbi.nlm.nih.gov/projects/GeneTests/static/whatsnew/labdirgrowth.shtml. Accessed 03-28-13.

29. American Medical Association Council on Science and Public Health. Clinical Application of Next-Generation Genomic Sequencing. 2012. http://www.ama-

assn.org/resources/doc/csaph/i12-csaph4-nextgengenomicsequencingsummary.pdf. Accessed 03-28-13.

30. 23andMe. DNA Ancestry Service. https://www.23andme.com/ancestry/. Accessed 04-14-13.
31. Ancestry.com. AncestryDNA. http://ldna.ancestry.com. Accessed 04-14-13.
32. Council for Responsible Genetics, Genetics Watchdog. Colman Chadam, California Boy Ordered To Transfer Schools For Carrying Cystic Fibrosis Gene, Goes Back To Class; November 8, 2012. http://www.councilforresponsiblegenetics.org/blog/post/Colman-Chadam-California-Boy-Ordered-To-Transfer-Schools-For-Carrying-Cystic-Fibrosis-Gene-Goes-Back-To-Class.aspx. Accessed 04-02-13.
33. Klitzman R, Chung W, Marder K, et al. Attitudes and practices among internists concerning genetic testing. *J Genet Couns*. 2013;22(1):90-100.
34. Huizenga CR, Lowstuter K, Banks KC, Lagos VI, Vandergon VO, Weitzel JN. Evolving perspectives on genetic discrimination in health insurance among health care providers. *Fam Cancer*. 2010;9(2):253-60.
35. Genetics and Public Policy Center, National Coalition for Health Professional Education in Genetics, Genetic Alliance. GINA: A Discussion Guide for Clinicians. http://www.nchpeg.org/documents/GINA_discussion_guide_2june10.pdf. Accessed 03-28-13.
36. U.S. Department of Health and Human Services. Summary of the HIPAA Privacy Rule. http://www.hhs.gov/ocr/privacy/hipaa/understanding/summary/. Accessed 03-28-13.
37. Feldman EA. The Genetic Information Nondiscrimination Act (GINA): Public Policy and Medical Practice in the Age of Personalized Medicine. *J Gen Intern Med*. 2012;27(6):743-6.
38. U.S. Department of Justice. Americans With Disabilities Act of 1990, As Amended. http://www.ada.gov/pubs/ada.htm. Accessed 03-28-13.
39. Hudson KL. Prohibiting Genetic Discrimination. *N Engl J Med*. 2007;356(20):2021-3.
40. National Human Genome Research Institute. Genome Statute and Legislation Database. http://www.genome.gov/PolicyEthics/LegDatabase/pubsearch.cfm. Accessed 03-28-13.
41. California Senate Bill 559. California Genetic Information Nondiscrimination Act. http://www.leginfo.ca.gov/pub/11-12/bill/sen/sb_0551-0600/sb_559_bill_20110906_chaptered.pdf. Accessed 03-28-13.
42. Arizona State Legislature. Title 20, Section 448. Insurance, Unfair Discrimination. http://www.azleg.state.az.us/FormatDocument.asp?inDoc=/ars/20/00448.htm&Title=20&DocType=ARS. Accessed 03-28-13.
43. Presidential Commission for the Study of Bioethical Issues. Privacy and Progress in Whole Genome Sequencing. 2012. http://bioethics.gov/cms/sites/default/files/PrivacyProgress508_1.pdf. Accessed 03-28-13.

44. Gymrek M, McGuire AL, Golan D, Halperin E, Erlich Y. Identifying personal genomes by surname inference. *Science*. 2013;339(6117):321-4.

45. Rodriguez LL, Brooks LD, Greenberg JH, Green ED. Research ethics. The complexities of genomic identifiability. *Science*. 2013;339(6117):275-6.

46. Bohannon J. Genealogy databases enable naming of anonymous DNA donors. *Science*. 2013;339(6117):262.

47. Walker LE, Rothstein MA. Are Genetic Discrimination Laws Up to the Task? Medscape Genomic Medicine. August 8, 2012. http://www.medscape.com/viewarticle/768560. Accessed 03-28-13.

Table. Details of the Genetic Information Nondiscrimination Act (GINA). Adapted from Hudson et al., 2008.[2]

What GINA does
Prohibits group and individual health insurers from using a person’s genetic information in determining eligibility or premiums
Prohibits an insurer from requesting or requiring that a person undergo a genetic test
Prohibits employers from using a person’s genetic information in making employment decisions such as hiring, firing, job assignments, or any other terms of employment
Prohibits employers from requesting, requiring, or purchasing genetic information about persons or their family members
Enforced by the Department of Health and Human Services, the Department of Labor, and the Department of Treasury, along with the Equal Opportunity Employment Commission; remedies for violations include corrective action and monetary penalties
What GINA does not do
Does not prevent health care providers from recommending genetic tests to their patients
Does not mandate coverage for any particular test or treatment
Does not prohibit medical underwriting based on current health status, including manifest disease of a genetic nature
Does not cover life, disability, or long-term-care insurance
Does not apply to members of the military or federal civilian employees
Key terms
“Genetic information” includes information about:
A person’s genetic tests
Genetic tests of a person’s family members (up to and including fourth-degree relatives)
Any manifestation of a disease or disorder in a family member
Participation of a person or family member in research that includes genetic testing, counseling, or education
“Genetic tests” refers to tests that assess genotypes, mutations, or chromosomal changes
Examples of tests that would yield protected genetic information are:
Tests for *BRCA1*/*BRCA2* (breast cancer) or *HNPCC* (colon cancer) mutations
Classifications of genetic properties of an existing tumor to help determine therapy
Tests for Huntington’s disease mutations
Carrier screening for disorders such as cystic fibrosis, sickle cell anemia, spinal muscular atrophy, and fragile X syndrome
Examples of tests that would not yield protected genetic information are:
Routine tests such as complete blood counts, cholesterol tests, and liver-function tests

Appendix I. AMA Policy and Ethics Opinions Relating to Genetic Discrimination

H-185.972 Genetic Information and Insurance Coverage

AMA believes: (1) Health insurance providers should be prohibited from using genetic information, or an individual's request for genetic services, to deny or limit any health benefit coverage or establish eligibility, continuation, enrollment or contribution requirements. (2) Health insurance providers should be prohibited from establishing differential rates or premium payments based on genetic information or an individual's request for genetic services. (3) Health insurance providers should be prohibited from requesting or requiring collection or disclosure of genetic information. (4) Health insurance providers and other holders of genetic information should be prohibited from releasing genetic information without express prior written authorization of the individual. Written authorization should be required for each disclosure and include to whom the disclosure would be made. (BOT Rep. 15, I-96; Reaffirmed: CMS Rep. 8, A-06; Reaffirmed in lieu of Res. 102, A-10)

H-315.983 Patient Privacy and Confidentiality

(1) Our AMA affirms the following key principles that should be consistently implemented to evaluate any proposal regarding patient privacy and the confidentiality of medical information: (7) Genetic information should be kept confidential and should not be disclosed to third parties without the explicit informed consent of the tested individual. (17) Our AMA Board of Trustees will actively monitor and support legislation at the federal level that will afford patients protection against discrimination on the basis of genetic testing. (BOT Rep. 9, A-98; Reaffirmation I-98; Appended: Res. 4, and Reaffirmed: BOT Rep. 36, A-99; Appended: BOT Rep. 16 and Reaffirmed: CSA Rep. 13, I-99; Reaffirmation A-00; Reaffirmed: Res. 246 and 504 and Appended Res. 504 and 509, A-01; Reaffirmed: BOT Rep. 19, I-01; Appended: Res. 524, A-02; Reaffirmed: Sub. Res. 206, A-04; Reaffirmed: BOT Rep. 24, I-04; Reaffirmed: BOT Rep. 19, I-06; Reaffirmation A-07; Reaffirmed: BOT Rep. 19, A-07; Reaffirmed: CEJA Rep. 6, A-11; Reaffirmed in lieu of Res. 705, A-12)

H-165.856 Health Insurance Market Regulation

Our AMA supports the following principles for health insurance market regulation: (4) Strict community rating should be replaced with modified community rating, risk bands, or risk corridors. Although some degree of age rating is acceptable, an individual’s genetic information should not be used to determine his or her premium; (CMS Rep. 7, A-03; Reaffirmed: CMS Rep. 6, A-05; Reaffirmation A-07; Reaffirmed: CMS Rep. 2, I-07; Reaffirmed: BOT Rep. 7, A-09; Res. 129, A-09; Reaffirmed: CMS Rep. 9, A-11; Reaffirmed in lieu of Res. 811, I-11; Reaffirmed in lieu of Res. 109, A-12; Reaffirmed in lieu of Res. 125, A-12; Reaffirmed: Res. 239, A-12)

E-2.132 Genetic Testing by Employers

As a result of the human genome project, physicians will be able to identify a greater number of genetic risks of disease. Among the potential uses of the tests that detect these risks will be screening of potential workers by employers. Employers may want to exclude workers with certain genetic risks from the workplace because these workers may become disabled prematurely, impose higher health care costs, or pose a risk to public safety. In addition, exposure to certain substances in the workplace may increase the likelihood that a disease will develop in the worker with a genetic risk for the disease. (1) It would generally be inappropriate to exclude workers with genetic risks of disease from the workplace because of their risk. Genetic tests alone do not have sufficient predictive value to be relied upon as a basis for excluding workers. Consequently, use of the tests would result in unfair discrimination against individuals who have positive test results. In addition, there are other ways for employers to serve their legitimate interests. Tests of a worker’s actual capacity to meet the demands of the job can be used to ensure future employability and protect the public's safety. Routine monitoring of a worker's exposure can be used to protect workers who have a genetic susceptibility to injury from a substance in the workplace. In addition, employees should be advised of the risks of injury to which they are being exposed. (2) There may be a role for genetic testing in the exclusion from the workplace of workers who have a genetic susceptibility to injury. At a minimum, several conditions would have to be met: (a) The disease develops so rapidly that serious and irreversible injury would occur before monitoring of either the worker's exposure to the toxic substance or the worker's health status could be effective in preventing the harm. (b) The genetic testing is highly accurate, with sufficient sensitivity and specificity to minimize the risk of false negative and false positive test results. (c) Empirical data demonstrate that the genetic abnormality results in an unusually elevated susceptibility to occupational injury. (d) It would require undue cost to protect susceptible employees by lowering the level of the toxic substance in the workplace. The costs of lowering the level of the substance must be extraordinary relative to the employer's other costs of making the product for which the toxic substance is used. Since genetic testing with exclusion of susceptible employees is the alternative to cleaning up the workplace, the cost of lowering the level of the substance must also be extraordinary relative to the costs of using genetic testing. (e) Testing must not be performed without the informed consent of the employee or applicant for employment. (IV) Issued June 1991 based on the report "Genetic Testing by Employers," adopted June 1991 (JAMA 1991; 266: 1827-1830).

E-2.135 Insurance Companies and Genetic Information
Physicians should not participate in genetic testing by health insurance companies to predict a person's predisposition for disease. As a corollary, it may be necessary for physicians to maintain separate files for genetic testing results to ensure that the results are not sent to health insurance companies when requests for copies of patient medical records are fulfilled. Physicians who withhold testing results should inform insurance companies that, when medical records are sent, genetic testing results are not included. This disclosure should occur with all patients, not just those who have undergone genetic testing. (IV) Issued June 1994 based on the report "Physician Participation in Genetic Testing by Health Insurance Companies," adopted June 1993; Updated June 1996.

E-2.137 Ethical Issues in Carrier Screening of Genetic Disorders
All carrier testing must be voluntary, and informed consent from screened individuals is required. Confidentiality of results is to be maintained. Results of testing should not be disclosed to third parties without the explicit informed consent of the screened individual. Patients should be informed as to potential uses for the genetic information by third parties, and whether other ways of obtaining the information are available when appropriate. Carrier testing should be available uniformly among the at-risk population being screened. One legitimate exception to this principle is the limitation of carrier testing to individuals of childbearing age. In pursuit of uniform access, physicians should not limit testing only to patients specifically requesting testing. If testing is offered to some patients, it should be offered to all patients within the same risk category. The direction of future genetic screening tests should be determined by well-thought-out and well-coordinated social policy. Third parties, including insurance companies or employers, should not be permitted to discriminate against carriers of genetic disorders through policies which have the ultimate effect of influencing decisions about testing and reproduction. (IV, V) Issued June 1994 based on the report "Ethical Issues in Carrier Screening for Cystic Fibrosis and Other Genetic Disorders," adopted June 1991.

Appendix II. AMA Legislative Principles on Genetic Discrimination and Surreptitious Testing (Approved by the Board of Trustees in March 2013)

1. Physicians support efforts to prohibit genetic discrimination broadly as well as surreptitious testing, because they are essential to advancements in medical knowledge and clinical care, and because part of a physician's duty is to safeguard against the inappropriate use of patient medical information for non-medical purposes and promote open and honest physician-patient communications.

2. Comprehensive federal protection against genetic discrimination is needed because patients remain at-risk of discrimination in a broad array of areas such as life, long-term care, and disability insurance as well as housing, education, public accommodations, mortgage lending, and elections.

3. Federal law should not preempt state laws that provide a greater level of protection against genetic discrimination.