Published in final edited form as:

Ann Intern Med. 2019 May 21; 170(10): 717–721. doi:10.7326/M18-2666.

Cases in Precision Medicine: Concerns about Privacy and Discrimination After Genomic Sequencing

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A 33 year-old healthy woman hears about an executive health program that offers exome sequencing as a component of its health assessment. The service provides information about medically actionable results and carrier status for recessive conditions with reproductive implications to participants and their physicians, and she is curious about her genes. She consults her internist about the implications of enrollment. She is aware that if a mutation is detected, it could have implications for her siblings, with whom she is not close, and their children. In addition, she holds a responsible position in a major corporation that she fears may be in jeopardy if it becomes known that she has a mutation that increases her risk of a serious disorder. Moreover, she has concerns about the potential impact of the results on her insurability. Before undergoing testing, she wants to know what protections exist for the confidentiality of the test results that are returned to her internist, including whether her internist will be obligated to reveal them to other members of her family, and whether her employer and insurers could use the information against her interests.

What Privacy Protections in the U.S. Apply to Genetic Information?

This concerns of this hypothetical patient about the privacy of her genetic information and the ways in which it might be used to her disadvantage are by no means unusual. Patients (1) and research participants (2) have indicated that privacy of genetic test results is an important concern, particularly with respect to insurance coverage. Physicians seem to share that worry, with nearly 80% of physician-respondents in one study who had ordered genetic testing within the previous six months reporting that they were somewhat or very concerned about their patients' genetic privacy (3). A comparable proportion wanted to know what they could do to better protect it. A recent systematic review of the literature relating to genetic privacy found that although most of the general public, patients and professionals worried about genetic privacy, the reasons for their concerns remained unclear. (4)

In addition to the ethical duties of a physician to guard the privacy of patients' medical information, which are embodied in every contemporary code of medical ethics, there are also statutory, regulatory and common law requirements in the U.S. (laws of other countries

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are not addressed here) for physicians to maintain the confidentiality of health information. The federal Health Insurance Portability and Accountability Act (HIPAA) Privacy Rule (the Privacy Rule) (5), which became operational in 2003, is the main statutory basis for protecting a patient's health information. Under the Privacy Rule, most uses and disclosures of protected health information (PHI) other than for treatment, payment or health care operations require a written authorization from the patient—although access to certain goods or benefits, such as long-term care insurance or disability benefits, may be dependent on PHI being released. Genetic information generated for the purpose of health care is deemed to be part of a person's health information and therefore is protected under the Rule.

Although HIPAA overrides state laws that are less protective of patient privacy, states can provide additional protections beyond those in the Privacy Rule. Approximately half of the states have policies governing genetic privacy *per se*, recognizing a privacy interest in genetic information that transcends concern for the privacy of other medical information, but there is considerable variation in the protections afforded their citizens. The result of the variation in state laws is that there is no standard or comprehensive approach to the protection of genetic information in the U.S. (6). As an example, if our patient lived in New York State, she would be covered by Section 79-L of the New York State Civil Rights Law, which provides that all records, findings, and results of any genetic test are deemed confidential and may not be disclosed to any person or organization without the specific written consent of the patient. This would include even those circumstances under which HIPAA might permit unauthorized disclosure or if such results have been obtained for research purposes. In contrast, in those states without specific laws governing the privacy of genetic information, the HIPAA Privacy Rule is controlling.

What is the Responsibility of the Executive Health Program with Regard to Patient Privacy and Communication of Results?

An executive health program offering genomic testing services has an obligation to obtain adequate informed consent from the patient, which requires prior provision of information regarding how and to whom test results will be reported and under what circumstances results can be disclosed to others (7). The program has the responsibility to convey the promised results (here medically actionable variants and recessive carrier status) and their interpretation to the patient, but it will usually be advisable to obtain the patient's consent to communicate the information to his/her primary care physician as well. Although the HIPAA Privacy Rule allows communication of PHI to another provider involved in the patient's treatment without patient authorization, state laws, as noted above, may require explicit consent of the patient and the physician's ethical obligations will usually mandate such consent as well.

An internist or other physician with ongoing responsibility for a patient's care will be in the best position to situate the findings in the context of the patient's medical and family history, which can minimize the risk of misinterpretation of test results and inaccurate assessment of disease risk. In addition, the primary care physician can ensure that needed followup occurs. However, many internists and other primary care physicians do not have sufficient expertise

to interpret and assess risk conferred by genetic variants and to develop plans for further diagnostic workup or ongoing surveillance tailored to a patient's needs (3). In the absence of such expertise, a physician should refer the patient to a medical geneticist or genetic counselor for advice with respect to choices for disease management or prevention (8).

What is a Physician's Responsibility If a Patient's Genetic Testing Reveals information That Is Relevant to the Patient's Relatives?

Notwithstanding the medical profession's commitment to preserving patient privacy, exceptions have long been recognized, including in circumstances in which the failure to disclose information may result in harm to third parties (9). Since the advent of genetic testing, it has been acknowledged that non-disclosure of results to potentially affected relatives might fall into this category. Hence, a series of professional and scholarly bodies have considered whether and when disclosure of genetic results might be permissible without a person's consent.

In 1983, the President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research suggested that, when a patient refuses to inform an at-risk relative, disclosure would be permissible under the following conditions: 1) reasonable efforts to elicit voluntary consent to disclosure have failed; 2) there is a high probability that harm will occur if the information is withheld and the disclosed information will actually be used to avert harm; 3) the harm that would result to identifiable individuals would be serious; and 4) appropriate precautions are taken to ensure that only the genetic information needed for diagnosis and/or treatment of the disease in question is disclosed (10). A decade later, a committee of the Institute of Medicine adopted a similar proposal, suggesting that there could be disclosure to relatives when the genetic disorder is highly penetrant and would be treatable or preventable (11). Similarly, the American Society of Human Genetics has suggested that disclosure to family members may be encouraged, but there must be "exceptional" circumstances to justify non-consensual disclosure, i.e., 1) attempts to encourage disclosure by the patient have failed; 2) the harm is likely to occur and is serious and foreseeable; 3) the at-risk relative is identifiable; 4) either the disease is preventable or treatable or early monitoring will reduce the genetic risk; and 5) the harm that may result from non-disclosure outweighs the harm of disclosure (12).

The law has been inconsistent in its guidance to physicians regarding their duties to nonpatient family members, especially when the implications of patients' genetic test results are unclear (13). There is limited case law in the U.S. that deals with physicians' common law duty to warn at-risk relatives, with just two frequently cited cases addressing disclosure of genetic risk *per se*. In *Safer v. Estate of Pack* (14), the daughter of a man who died from metastatic colon cancer secondary to polyposis of the colon—which is usually transmitted as an autosomal dominant condition—sued the estate of his physician for failing to inform her of the risk that she too might develop the disorder. Her suit was filed after she was diagnosed with polyposis and had already developed colon cancer, which by then was metastatic. The New Jersey appellate court that considered the case held that a physician has a duty to warn those known to be at risk of a genetic disorder; such duty may not always be fulfilled by

simply warning the patient, but may require a warning to the relative(s) at risk themselves. (This decision was later effectively overruled by the enactment in New Jersey of a broad genetic privacy statute.) In contrast, the Florida Supreme Court took a different approach in *Pate v. Threlkel* (15), a case involving the daughter of a woman who had been treated for medullary thyroid carcinoma, probably due to multiple endocrine neoplasia type 2, an autosomal dominant condition. The daughter sued her mother's physician after she too was diagnosed with medullary carcinoma of the thyroid for his failure to inform her that she had a 50% risk of developing the condition. The court held that a duty to warn a relative at risk exists if a reasonably prudent physician would do so, but that the duty can be fulfilled if the physician warns the patient of the risk to his or her relatives, with the expectation that the patient will inform such relatives, thus fulfilling the physician's obligation of confidentiality. A recent survey of medical malpractice litigation in the U.S. indicates that the question of whether physicians have a duty to warn their patients' relatives of genetic risks, at least by informing the patients and recommending that they warn their relatives, remains a litigated and unsettled issue, with most courts holding that there may be such a duty, but with the contours of that duty still very much uncertain. (16)

The foregoing cases and position statements pre-date the adoption of the HIPAA Privacy Rule, which would likely preempt the state law considerations on which these court rulings were based. The Privacy Rule does have several "public interest" exceptions to its non-disclosure policy, including when it is necessary for "preventing or controlling disease, injury or disability," such as in situations of infectious diseases, domestic violence or child abuse or neglect (17). However, additional exceptions to the non-disclosure rules relating to family members were introduced in the 2013 Privacy Rule amendments. To the extent that an individual's genetic information is needed for the treatment purposes of a family member, a health care provider is permitted to disclose such information, "subject to any agreed-upon restriction," at the request of another provider who is treating other family members who are seeking to identify their own genetic health risks, provided that the individual has not requested and the health care provider has not agreed to a restriction on such disclosure (18). The Office for Civil Rights, which oversees compliance with the Privacy Rule, has made clear that the physician does not have to agree to the requested restriction (19), thereby effectively giving the physician veto power over the wishes of the patient. This interpretation is controversial, since it would seem to undermine the physician-patient relationship and contradicts the commonly assumed ethical duty of a physician to maintain patient confidentiality, but it appears to represent current regulatory law (20).

Given the ethical and legal uncertainties surrounding the question of disclosure of genetic information to relatives, the best guidance may come from the American Medical Association's Council on Ethical and Judicial Affairs. In its opinion on disclosure of familial risk in genetic testing, the Council does not see finding and notifying family members as a physician's duty, but recommends that physicians inform patients in advance of what they expect patients to disclose to their families and subsequently be available to assist in this communication (21). In certain circumstances, consultation with an attorney may be helpful to a physician.

Are There Protections Against Discrimination by Insurers, Employers, or Other Entities Based on a Person's Genetic Test Results?

Discrimination in employment and insurance coverage have been major concerns of persons asked to consider genetic testing (22). These concerns led to the adoption of the federal Genetic Information Nondiscrimination Act of 2008 (GINA), which protects against discrimination on the basis of genetic information in employment and the provision of health insurance coverage. GINA defines genetic information broadly to include both the results of genetic testing and family history of disease, but not an individual's manifest disease or condition—i.e., a condition that has been diagnosed, is being treated, or from which an individual is experiencing symptoms.

With regard to employment, Title II of GINA provides protections against genetic discrimination in the workplace. Specifically, an employer may not use genetic information in making decisions regarding hiring, promotion, terms or conditions of employment, privileges, compensation or termination. In addition, an employer may not request, require or purchase genetic information about an individual employee or the employee's family members. Subsequent, controversial regulatory changes promulgated by the EEOC allow employers to request genetic information in connection with employee wellness programs that may offer reduced premiums or enhanced benefits of health insurance as incentives, although GINA's ban on use of any information that is provided for employment decisions remains (23). The question of what constitutes a permissible incentive (as opposed to a coercive means of forcing employees to divulge health information) in employer wellness programs is currently being litigated, but for the time being a federal district court's decision vacating the EEOC wellness regulations does not change the current regulatory posture. (24) In any case, proving that employment decisions were made on the basis of genetic information is a daunting challenge for many employees, leading many advocates to argue that the wellness program exception constitutes a major breach in GINA's protective wall.

In the area of insurance, Title I of GINA provides protections against discrimination in health insurance—but not with regard to other types of insurance. Specifically, group health plans, health insurance issuers (including HMOs), and issuers of Medicare supplemental policies may not require individuals to provide their genetic information or the genetic information of a family member to the insurer for eligibility, coverage, underwriting, or premium-setting decisions, nor may a health insurer require an individual to undergo a genetic test as a condition of coverage. Despite passage of GINA, surveys show that large portions of the public remain unaware of these protections, and that their lack of awareness may influence decisions not to undergo genetic testing (25). Of note, GINA does not include protections against genetic discrimination in life, disability or long-term care insurance, or any other insurance product. Insurers typically request access to medical records for applicants desiring many types of insurance and, other than for health coverage, are free to use information in the medical records (including the results of genetic tests) in their underwriting decisions.

How Should Her Internist Advise This Patient?

Internists and other physicians approached by patients such as the one described above about the legal protections for information generated by genome sequencing for clinical purposes can provide both reassurance and cautionary warnings. Protections that exist for medical information in general, such as the HIPAA Privacy Rule, as well as laws in some states that provide additional protections for genetic information, should be reassuring with regard to the privacy of genetic data. Although current regulatory interpretations of HIPAA appear to permit the non-consensual disclosure of genetic information on request of the health care providers of relatives whose medical treatment may be affected if they share a patient's genetic results, physicians have the discretion not to disclose that information. The internist in our case should make clear to the patient his or her policy on this issue. Additional reassurance should come from the knowledge that GINA generally precludes the acquisition and use of genetic information, broadly defined, by employers and health insurers. However, our patient should also be told that other kinds of insurance are not protected by GINA and that insurers may request access to medical records as a condition of coverage. Patients themselves will need to weigh the risks of generating genomic data against the direct benefits to reach a decision about whether to have exome sequencing.

Acknowledgements:

This publication was supported by the National Center for Advancing Translational Sciences, National Institutes of Health (NIH), through Grant Number UL1TR001873. Dr. Appelbaum received additional support from the following grants from the National Human Genome Research Institute, NIH: P50HG007257, RM1HG007257, and U01HG008680.

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Box 1:**Key Points**

- Patients often have concerns about the privacy and confidentiality of genetic test results and whether they could negatively impact their employment and/or insurance coverage.
- Privacy of genetic information generated for clinical purposes is protected by the federal Health Insurance Portability and Accountability Act—HIPAA. Some state laws provide additional protections, but there is no uniform approach.
- In limited circumstances, when patients fail to do so, it may be ethically appropriate to inform at-risk relatives or their clinicians of test results. However, the HIPAA Privacy Rule has been interpreted to preclude non-consensual disclosure for this purpose, except on request of a relative's treating clinician.
- The federal Genetic Information Nondiscrimination Act of 2008 (GINA) protects against discrimination on the basis of genetic information in the workplace and in health insurance coverage—but not for life, disability, long-term care, or any other insurance product.
- Physicians can offer patients considering genetic testing reassurance about privacy and discrimination, but with some reasons for caution as well. Ultimately patients will need to weigh the risks of generating genomic data against the benefits for them or for medical research.