

OVERVIEW: There is a growing awareness and interest among the public and policymakers of the important role genetics can play in providing medical care. Health care providers, especially primary care physicians, are likely to face increasing expectations that they will use genetic technologies to diagnose and treat their patients.

One important, but sometimes overlooked, technique is the use of family history to assess disease risk and refer patients for genetic testing where appropriate. Yet the expanded use of family history poses novel and complex ethical and legal issues. Although many of these issues are not unique to family history, advances in our understanding and use of genetic information present new challenges to existing frameworks.

This guide will highlight the core ethical and legal challenges providers must consider when utilizing family history. At this point, there is little agreement as to a physician's ethical and legal obligations. For those issues where there is consensus, the respective ethical and legal conclusions may conflict with each other. This is particularly true in reference to a physician's duty to warn a patient's relative of familial or genetic risks.

Thus, the guide is designed to introduce, rather than to resolve, the most important legal and ethical issues likely to arise when using family history in clinical practice. Links to additional materials are provided should you wish to explore further the issues raised in this guide.

WHAT IS FAMILY HISTORY? Family history reflects genetic, environmental, and behavioral factors that run in families. As such, it can be used with other risk factors to assess a patient's disease risk. Family history is also considered a common form of genetic screening and is often a precursor for referrals for more sensitive testing using DNA or other biomarkers.

The clinical benefits from using family history include early identification and treatment of persons at higher risk for certain diseases. Expanded use of family history can assist physicians in identifying and linking risks and symptoms affiliated with predominantly genetic conditions. Family history can also provide a forum for physicians to discuss the importance of prevention and protective behaviors, including lifestyle changes, with patients.

WHAT KINDS OF ETHICAL AND LEGAL CHALLENGES MAY CLINICIANS FACE?

Because family history is directly relevant to both patients and relatives, it challenges many of the tenets of traditional biomedical ethical models by bringing the family's interests into the doctor/patient relationship. Furthermore, patients and relatives may have conflicting opinions or obligations regarding the collection and disclosure of family history data.

DUTY TO DIAGNOSE: Physicians may face liability for failing to collect a family history and, consequently, failing to diagnose a genetic condition. This is likely to become more of an issue as the collection and use of family history as a precursor for genetic testing is further recognized as standard clinical practice for treating primary care patients.

DUTY TO WARN: In collecting a patient's family history, a physician may become aware of relatives' disease risks. It is generally believed that patients have an ethical obligation to

disclose familial risks to their relatives. Patients may nonetheless refuse disclosure, thus limiting a relative's ability to seek care.

Ethically, physicians must evaluate whether the harms resulting from non-disclosure outweigh those created by breaching patient confidentiality. Consensus is limited on a physician's ethical duty to directly warn at-risk relatives; however, the perspectives fall into several camps:

- 1. Impermissible:** A physician's direct disclosure of familial risk to relatives is unallowable. It is the patient's moral obligation to inform relatives of genetic and familial risks. Regardless of whether a patient complies with their responsibilities, a physician who informs the relatives may irreparably damage the patient/doctor relationship.
- 2. Consent:** Physician disclosure is restricted, requiring at a minimum patient consent.
- 3. Ethically Justifiable:** There are conditions under which direct physician disclosure may be justified. These vary but generally include: (1) all efforts to encourage patient disclosure have failed; (2) harm to the relatives from non-disclosure is imminent and serious; (3) at-risk relatives are identifiable; (4) harm from failing to disclose outweighs harms from disclosing; (5) information that is disclosed is the minimum necessary to allow for appropriate protective actions by the relatives.

Legally, some states have found that providers may have a duty to warn at-risk parties of imminent and serious harm, possibly including genetic risk. The three cases which have addressed a physician's duty to warn of genetic risk have reached inconsistent results. In general, this duty is discharged by warning the patient of the need to disclose familial risk. Family members do not have a legal duty to warn each other of such risks.

PATIENT CONFIDENTIALITY: Providers who directly disclose family history or genetic risk to relatives without patient consent may be liable for breaching their duty of confidentiality. This could result in state licensing board sanctions on providers for failure to maintain patient confidentiality, in addition to potential civil liability.

The HIPAA Privacy Rule governs the flow of protected health information by covered entities including physicians. The disclosure of family history information to a relative without patient authorization may, in very limited circumstances, be permissible where required by law or where there is a serious threat to health and safety in the absence of disclosure for which disclosure would lessen the threat.

One potential complication under HIPAA is when a physician sends a patient's medical record containing familial medical information to another clinician. Trying to eliminate references to family histories in a medical record prior to transfer could impose an impractical administrative burden, but physicians may need to make sure that privacy protections are maintained. Additionally, in those cases where familial risk information is scattered throughout medical records, eliminating references to such info could potentially compromise patient care.

INFORMED CONSENT: As with any other clinical encounter, physicians must ensure the knowing and voluntary consent of patients. Informed consent in the context of family history

means ensuring that patients understand how the information they provide will be used along with the clinical and non-clinical (i.e., discrimination) implications of having such knowledge for themselves and their relatives.

Clinical guidelines suggest that physicians inform patients of: (1) their responsibility for disseminating familial risk; (2) who else is at-risk; (3) options for assistance if needed; and (4) the assessment's implications for themselves and their relatives. Physicians should also provide the patient sufficient time to internalize their new-found risks and duties. But there is no legal consensus on the specific informed consent requirements.

RIGHT NOT TO KNOW: Clinicians must also consider a patient's, or relatives', right not to know family history or genetic risks. Knowledge of disease risks can be harmful. In contrast, ignorance could prevent patients or relatives from acting on potentially life saving information. Limiting a person's informed decision making capacity may also contradict our conception of autonomy, while overriding a patient's explicit knowledge preferences could have similar negative consequences. Although there is debate as to whether a physician should presume that patients do or do not want to know genetic risks, it is advisable to ask patients about their knowledge preferences and to inquire about what their relatives want to be told.

DISCRIMINATION: Physicians must also be aware of the potential for discriminatory and stigmatizing harms for patients or relatives possessing genetic risk information. Although the actual instances of documented genetic discrimination are rare, studies suggest that the public perception of genetic discrimination may be a barrier to genetic testing and related counseling.

Physicians can help patients by: (1) correcting misconceptions patients have regarding the role of genetics in determining disease; (2) discussing the extent of third party access to family history or genetic test results; (3) explaining how genetic information can be used in making informed decisions about their care; and (4) offering pre-testing consultations for genetic tests to inform patients as to the implications and risks/benefits of a given test for themselves and their relatives.

Virtually all states have enacted legislation specifically targeting insurance and employment discrimination but protections against genetic discrimination vary significantly across states:

Insurance: State laws address discrimination, including the inability to obtain coverage, changes in the terms and conditions of coverage, and termination of coverage.

Employment: Although few courts have addressed employer based genetic discrimination, many states have enacted legislation to limit employers' ability to collect, use, and disclose genetic information. On the federal level, the EEOC indicated that it will use the Americans with Disabilities Act (ADA) to deal with employer genetic discrimination. In 2000, EEOC prohibited use of genetic and family history information in public employment decisions; it is unclear if the ADA will decide that a genetic predisposition or disease constitutes a disability.

LINKS & RESOURCES: For more information about the laws in your state, visit the National Conference of State Legislatures website at: <http://www.ncsl.org/programs/health/genetics/ndishlth.htm>