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Exclusion of Genetic Information From the Medical Record:

Ethical and Medical Dilemmas

Robert Klitzman, MD
Division of Psychiatry, Law, and Ethics, and HIV Center, Columbia University, New York, New York

Increasingly, physicians and patients face dilemmas of whether to exclude genetic information from medical charts, posing critical challenges for practice, research, policy, and education. Physicians and patients are obtaining more genetic information, yet medical records are rapidly becoming electronic, threatening confidentiality. Tensions thus arise between potential medical benefits vs social risks of including information. Use of genetic testing is rapidly increasing through clinicians and direct-to-consumer marketing. Direct-to-consumer tests may be definitive or show only slightly increased disease probabilities, but with advances may have increasing clinical utility. Several institutions have also discussed including whole genome data in medical records. Genetic discrimination has occurred with α_1-antitrypsin deficiency, Huntington disease, and other mutations,[1] although the extent remains unclear,[2] partly because such discrimination can be subtle or difficult to prove. Patients may be passed over for promotion or marginalized, but not fired.[3]

The Health Insurance Portability and Accountability Act (HIPAA) protects medical information in certain contexts and the 2008 Genetic Information Nondiscrimination Act (GINA) protects genetic test results in the absence of symptoms, but these laws do not prevent discrimination in many realms (eg, life, disability, or long-term care insurance). Many patient advocates are concerned that under GINA, discriminating against patients and paying fines may cost companies less than covering them—as racial, sex, and age discrimination continue, despite legislation. Federal health care reforms of 2010 should broaden coverage of preexisting conditions but have yet to be implemented, and advocates are concerned that reforms might simply raise premiums.

Patients' Views

Recent interviews conducted with patients confronting several genetic diseases highlight several challenges,[3,4] including concerns about storing genetic information. Patients often did not trust how laws would work in real-world circumstances; whether subtle/indirect discrimination, difficult to demonstrate, might occur.[3] These interviews, along with other, anecdotal evidence, suggest that many patients thus remain wary of providing results to physicians, assuming that all information will be entered into medical records. Hence, some patients may withhold information. Moreover, information, once in charts, may be virtually impossible to remove. Some support groups run by patient organizations have encouraged

Corresponding Author: Robert Klitzman, MD, Columbia University, 1051 Riverside Dr, Unit 15, New York, NY 10032 (rlk2@columbia.edu).

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patients to ask physicians to exclude genetic information from the medical record. Some patients may not reveal information to their physicians until they are assured it will not appear in their medical record.

Physician Decisions

The American Medical Association Code of Medical Ethics states that “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. . . . Physicians who withhold . . . results should inform insurance companies that, when medical records are sent, genetic testing results are not included.”[5]

However, health care systems may not allow this option of stating that some information is excluded, and physicians may face adverse legal and institutional consequences. Insurers may insist on obtaining the data, arguing that such data are relevant, and may not cover fees for patient care unless all information is received. Moreover, physicians, in stating that genetic information may be excluded, might inadvertently alert insurers that a serious mutation exists.

Physician sensitivity and approaches concerning these issues appear to vary. Some might voluntarily exclude information, keep separate, parallel “shadow” charts (without telling insurers that sent records exclude information), or encode information, using broad generic terms, not specific diagnoses.[6] However, office staff may not appreciate this need for nondocumentation and inadvertently forward information.

Although patients may want to exclude information that involves potential harm to a third party (eg, relatives), physicians may be obliged to not only document but also report that information to external agencies. Judges have ruled that physicians sometimes have duties to warn third parties about genetic risks.[7] Hence physicians, in not documenting the genetic information or ways they are handling the situation (eg, whether they or patients are disclosing that information to family members), may be endangering themselves. Physicians might disguise information and justify this behavior,[8] but as a result they could face ethical and legal problems.

Questions emerge about whether physicians can or should also exclude other kinds of genetic information (eg, family members’ genetic test results or risks), and if so, when and how; and whether to then also inform insurers that medical records exclude these data. Alternatively, physicians, concerned about liability or discounting discrimination, may document information, even refusing patients’ requests for exclusion.

Omitting genetic information has medical risks. With certain genetically associated syndromes such as myotonic dystrophy and breast cancer, excluded information may pose problems if patients are seen by other physicians who can access only the information documented in the medical record. For rare disorders, clinicians may overlook diagnoses and order additional, unnecessary tests to rule out more common but absent diseases. Breast cancer mutations can affect treatment decisions. In omitting or disguising information, physicians may also devalue the medical record, undermining patients’ and physicians’ trust in it and each other, and reinforcing fears that genetic tests indeed cause discrimination. Moreover, increasing health care system fragmentation makes the medical record one of the only vehicles for continuity in care.

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Future Directions

Attention and research are needed to address questions such as how often and when patients request excluding genetic information and also when physicians omit that information or record it in code or separate charts; what the legal status of shadow charts are in different states; how insurers do or will respond; how much patients and clinicians value completeness of medical records; and how often patients experience or have concern about genetic discrimination.

Ethical dilemmas emerge about when physicians should exclude certain genetic information and how to decide whether to exclude it or not. Physicians ordinarily have no contractual duty to give information to patients' employers, but they face tensions between medical benefits and social risks (eg, possible stigma/discrimination) of including information. One can argue that information, if irrelevant to patients' health, can be omitted. But views of relevance, based on probabilities of future medical events, can be uncertain. How to resolve clinician, insurer, and patient disagreements is also unclear.

Professional and public education is crucial. Some physicians have a poor understanding of genetics,[9] but given genetic counselor shortages, need to become more sensitive to these areas, prepare to counsel patients about these issues, and be open to patient requests, realizing that patients may withhold information. Physicians should obtain informed consent regarding these options by educating patients about the risks and benefits of disclosing genetic information, the scope and limitations of current policies, safeguards and guidelines, the choices available, and the advantages and disadvantages to recording genetic information in the medical record. Before promising confidentiality, clinicians should ensure that patients understand its limitations.[10] Deciding whether to chart direct-to-consumer testing and, if so, when raises additional concerns. Physicians can also consider suggesting that patients contemplate obtaining disability, life, or long-term care insurance before testing.

These choices pose potential risks and benefits, embodying difficult-to-weigh uncertainties. Yet presenting these considerations can help patients and physicians think through these options together. Physicians should realize that patients' preferences will vary with specific diseases and genetics and extent of patients' discrimination concerns.

Current guidelines should be revisited; threats to confidentiality remain real. Careful, sensitive implementation of GINA can reduce discrimination fears, but legislation is needed to cover other forms of insurance and increase fines if employers or health care insurers violate GINA.

In electronic medical records, 2-tiered systems for sensitive vs other information may help, but details remain to be determined (eg, who exactly would have access). Health, disability, and life insurers may well still demand full information. How well such complex systems will work or assuage patients' concerns is unclear. Other kinds of sensitive (eg, psychiatric) information pose related, although sometimes differing, issues also requiring attention and further research.

Increasing threats to confidentiality will surely perpetuate desires of patients and physicians to exclude information. Clinicians should recognize and address, not ignore, these challenges to attend closely to these critical concerns.

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