The new world of genomic testing, families and privacy

Can physicians and researchers share information with family members who may be at risk for heritable diseases?

BY SUSAN M. WOLF, JD

As genomic testing—including genome and exome sequencing—becomes more established and affordable, an increasing number of physicians are encountering genomics in their practice. Sequencing of cancer patients and their tumors is becoming an accepted part of oncology practice. Sequencing is used in other specialties as well, for example to aid in the diagnosis of pediatric patients with puzzling neurodevelopmental anomalies. As an increasing number of patients undergo genomic analysis, physicians will face challenging privacy issues.

Like other medical information, genetic data is typically regarded as private, and physicians have stringent responsibilities to protect it, as a matter of law, ethics and institutional policy. But families are beginning to come forward and ask whether genomic information about an individual family member may have implications for their own health. This may present physicians with a new version of an old dilemma: Do they protect the privacy of the patient's genomic information, or do they share that information with relatives?

This problem first surfaced with the rise of traditional genetic testing. Genomic analysis can yield much more information revealing multiple genetic risks. Some of those risks are serious and some may be lowered or even eliminated through clinical intervention. This means that information about genetic variants may be highly important to relatives, especially since first-degree biological relatives commonly share 50 percent of their genes.

Here's a possible scenario: A physician learns that her patient has a *BRCA1* gene mutation, conferring increased risk for breast and ovarian cancer. In the course of a consultation, the doctor recommends that the patient share this information with close relatives, so they can consider being tested. The patient replies that she's not willing to disclose the mutation because she's estranged from her family. What is the doctor to do? Do family members have a right to know?

The physician's role

In the 1990s, lawsuits were filed claiming that physicians had a "duty to warn" patients and relatives about hereditary disease risk. The case law that emerged from those suits was inconsistent. The Supreme Court of Florida ruled in 1995 that it was enough for the physician to warn a patient that a disease is heritable, leaving the responsibility with the patient to share that information with family members. However, in a 1996 case from New Jersey, the court found a broader responsibility to warn at-risk relatives. The court required that "reasonable steps be taken to assure that the information reaches those likely to be affected."[1,2]

These cases unleashed a flurry of commentary. Overwhelmingly, legal and policy authorities urged the importance of continuing to respect the confidentiality of patient information. In 1996, the Health Insurance Portability and Accountability Act (HIPAA) was passed. HIPAA protects the privacy of health information for 50 years after an individual's death. That protection applies to patients' genetic and genomic information as well.

In 1998, the American Society of Human Genetics (ASHG) released guidelines for health care professionals, in which they recognized that genetic information is "personal—yet simultaneously familial" and "raises new and profound questions about … legal and moral obligations to disclose genetic information to at-risk relatives." The guidelines specifically focused on what doctors should do if a patient refuses to disclose relevant genetic information to at-risk relatives. The ASHG asserted that:

- Patient confidentiality should be paramount and a breach of confidentiality should be highly exceptional.
- Confidentiality is not absolute: "ethical, legal, and statutory obligations may . . . permit physicians to disclose otherwise confidential information."

Under these guidelines, doctors must stay within a narrow zone. They have the privilege to warn, but only in cases "where attempts to encourage disclosure on the

part of the patient have failed; where the harm is highly likely to occur and is serious and foreseeable; where the at-risk relative(s) is identifiable; and where either the disease is preventable/treatable or medically accepted standards indicate that early monitoring will reduce the genetic risk."[3] The ASHG thus recognized a privilege to warn in these narrow circumstances, not a duty to warn.

In the case of the patient refusing to share information about a *BRCA1* mutation with family members, the physician would be on firmer ground encouraging the patient to reconsider her refusal, rather than reaching out directly to family members.

No established guidance available to researchers

All of this means clinicians have some guidance regarding what genomic data they can disclose and to whom. The same cannot be said for researchers, who collect data to address broad scientific questions, rather than to determine a course of treatment for one patient. Only now are policies emerging to guide researchers and biobanks about what to do with individual research results as well as unexpected "incidental findings"—discoveries that researchers may stumble upon in performing their primary analysis and consider offering to the research participant because of the potential health importance for that individual.[4,5]

Adding to the challenge is the fact that these same issues may arise after the death of the research participant. Genomic research commonly involves archiving data and specimens for long periods of time to facilitate continued research. Especially in the case of cancer, the individual whose genome was sequenced may die, leaving relatives concerned about their own risk.

Let's say a cancer researcher finds that a subject who died carried a genomic variant placing him at higher risk of colorectal cancer or a variant for malignant hyperthermia that indicated a potentially catastrophic reaction to a commonly used anesthetic. What should the researcher do with this information? The researcher may have no relationship with surviving family members and may not even be a clinician. In any case, the researcher likely promised to protect the confidentiality of the subject's health information when obtaining informed consent for participation in the research.

Moving forward

When the debate over the duty or privilege to warn family members of potential genetic risk first began, the human genome had not yet been sequenced. Now that sequencing and other forms of genomic testing are readily available—both in research and increasingly in clinical care—we can-

The consortium's contributions

Since 2005, the University of Minnesota's Consortium on Law and Values in Health, Environment and the Life Sciences has collaborated with scholars across the country on a series of National Institutes of Health (NIH)-funded projects analyzing the issues involved in the return of genomic results and incidental findings to research participants. The resulting publications have had a major impact on the national debate.

When the consortium first started working on these issues, there was little discussion about how to handle findings that might be important to individual research participants when conducting genomic research. Researchers analyzing scans using MRI and other technologies had already recognized that incidental findings were inevitable in research and had begun to devise consensus guidelines for grading the urgency of such findings and managing them. Consortium work built on that early foundation to create guidance for genetic and genomic findings, which was published in a dedicated issue of the *Journal of Law, Medicine & Ethics* in 2008.

Our next project funded by the NIH grappled with the reality that genomic data and specimens are often archived in biobanks and large, shared databases for long-term research use all over the world. This raises the potential for research results and incidental findings to be uncovered at the biobank or data archive or in subsequent research by teams that may be far removed from the source individual. That work resulted in a special issue of *Genetics in Medicine* in 2012.

Our latest NIH research, with colleagues at the Mayo Clinic and the University of California, San Francisco, addresses return of results and incidental findings to family members, including after the death of the research participant. As part of that project, we co-directed a workshop on return of results from biobanks at the Brocher Foundation in Switzerland in November 2013.

NIH Director Francis Collins has declared the issue of incidental findings "one of the thorniest current challenges in clinical research."[1] In 2013, the Presidential Commission for the Study of Bioethical Issues devoted an entire report to the problem.[2] The NIH now dedicates substantial funding to these issues and has formed the Clinical Sequencing Exploratory Research (CSER) Consortium to speed progress.[3] The University of Minnesota's Consortium participates actively in CSER work and collaborates with scholars all over the world on these high-impact issues.

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not ignore the tension between protecting individual privacy and facing the reality of shared risk in biological relatives. U.S. privacy law is about protecting individuals, but genetics is about more than individuals; is it about kin.

Efforts are under way to craft policy sensitive to these new realities. At the University of Minnesota's Consortium on Law and Values in Health, Environment and the Life Sciences, we are engaged in a multi-year project funded by the National Institutes of Health to study these issues and create new guidelines (see p. 33).[6] In partnership with researchers at the Mayo Clinic and the University of California, San Francisco, we have convened a multidisciplinary group of experts from the United States and Canada to draft consensus recommendations on the return of results to family members, both before and after the research participant's death. Those recommendations will be published in the fall in a symposium issue of the *Journal of Law, Medicine & Ethics,* which will feature 15 articles stemming from our national conference on families and genomic privacy last November.

Central to the project recommendations is the need to face this issue and plan for it. Researchers inviting individuals to participate in genomic research need to be clear on the privacy protections for the individual's genomic data both before and after their death. Researchers can ask prospective participants whether they wish to have data shared with family members and who they trust to make decisions about information-sharing after they can no longer make those decisions themselves. Clinicians, too, can broach these issues with patients. Both can seek counsel on the complex questions of family access under state and federal law.

As genomics increasingly becomes part of the practice of medicine, these issues will become more and more important. Families, researchers and clinicians need to start talking about who will get access to genomic results, including after an individual dies. We plan who gets our property after death. We need to start planning who will have access to our genomic data. MM

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