GENETIC TESTIN

Genetic discrimination? The idea was unheard of a decade ago. Now it looms as a real possibility. Healthcare professionals and legal experts tackle some tough issues.

Genetic research has been likened by many to Pandora's Box. Now that the lid of that metaphorical box has been lifted, there's no turning back. One of the unalterable effects, some fear, is that individual privacy has been compromised.

"In the early '90s, when the Human Genome Project began, some wise people perceived that the biological research could have a negative impact on individual privacy and the possibilities of discrimination," said Barbara Fuller, JD, RHIA, branch chief for policy, education, and outreach for the National Human Genome Research Institute at the National Institutes of Health, Bethesda, Md., during a recent presentation to the Healthcare Information and Management Systems Society.

Whether all of the fears will be realized remains to be seen. In the meantime, genetic research has spawned genetic testing, and such testing is increasingly being used, especially for breast cancer. No doubt, genetic information gathered from testing can be a powerful tool when it comes to preventive measures and management of the disease. According to Rebecca D. Pentz, PhD, professor of hematology and oncology in research ethics in the Winship Cancer Institute at Emory University, Atlanta, Ga., genetic testing will someday be a standard part of every oncologist's practice.

In the meantime, medical professionals and lawmakers grapple with troubling questions. The most troubling question involves genetic disenfranchisement—more commonly referred to as genetic discrimination—because many fear the potential misuse of genetic information.

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BREAST CANCER PREDISPOSITION

Of the nearly 200,000 women diagnosed with breast cancer yearly in the United States, approximately 5% to 10% have a hereditary form of the disease. In the mid-1990s, scientists identified the inherited alterations, or mutations, in genes that cause some women to be more predisposed to developing breast cancer. These gene mutations have been labeled BRCA1 (or breast cancer 1) and BRCA2 (or breast cancer 2). Underscoring what physicians recognized over the years—that breast cancer runs in families—scientists found that cancers associated with either of these mutations runs highest in families with a breast cancer history, and a woman's chances of developing breast cancer is increased if she inherits either of the altered genes. Women who inherit the mutations have a 36% to 85% risk of developing cancer.

Genetic testing for BRCA1 or BRCA2 involves testing a blood sample in the laboratory. A positive result indicates that the individual has inherited a mutation in BRCA1 or BRCA2 and, therefore, has an increased risk of developing breast cancer. That's not to say that person will actually develop breast cancer. It indicates only that the probability is higher for that individual.

"There is only a 60% to 80% chance they will get cancer in their lifetime," points out Duane W. Superneau, MD, chief of the section of medical genetics at the Ochsner Clinic, New Orleans, La., "and we aren't really sure about those probabilities yet. We know it increases your risk, but it doesn't mean you're going to get cancer."

GENETIC DISCRIMINATION

The capability of genetically testing for breast cancer gave rise to fears of genetic discrimination, specifically in the form of workplace discrimination or healthcare insurance discrimination. With the former, a woman is treated differently by an employer because she tested positive for a risk-increasing genetic alteration. With the latter, someone who tested positive for BRCA1 or BRCA2 is denied coverage for medical expenses, dropped from current coverage, or unqualified for new insurance.

– Images courtesy of the National Human Genome Research Institute

Illustration of cells and chromosomes

Protecting Breast Can

IG ON TRIAL

By Dan Harvey

Thus far, no landmark case involves such types of discrimination, but the potential for abuse exists. "We haven't run into anything yet, but we are always worried about it," says Pentz. "There is a potential for discrimination with all kinds of healthcare information, not just genetic testing."

Some protection is provided by existing legislation, including the following:

• Americans with Disabilities Act (ADA) of 1990 — Many observers see this act as the most likely way to protect against genetic discrimination in the workplace. It does not directly address genetic information, but it does provide protections against disability-related genetic discrimination. Above all, it prohibits discrimination against a person who is regarded as having a disability. However, it neither protects against discrimination based on unexpressed genetic conditions nor protects prospective employees from requirements or requests to provide genetic information when offered a job.

• The Health Insurance Portability and Accountability Act (HIPAA) — HIPAA is the only federal law that directly addresses the issue of genetic discrimination. The act prohibits group health plans from using genetic information as a reason to deny coverage or to charge more for coverage for someone currently without disease. Also, it clarifies that genetic information for someone currently without disease cannot be considered a preexisting condition. At the same time, it does not prevent employers from refusing to offer health coverage as part of their benefits or prevent insurance companies from requesting genetic information.

• HIPAA National Standards to Protect Patients' Personal Medical Records — Released in 2000 by Health and Human Services, the regulation provides the first comprehensive federal protection for health information privacy. Protection includes limiting the nonconsensual use and release of private health information, giving patients rights to access their medical records and to know who else has accessed them, restricting most disclosure of health information to the minimum needed for the intended purpose, establishing new criminal and civil penalties for improper use or disclosure, and establishing new requirements for access to records by researchers and others. However, while HIPAA covers medical records maintained by healthcare providers, health plans, and healthcare clearinghouses, the standards are not specific to genetic information.

"There is a potential for discrimination with all kinds of healthcare information, not just genetic testing," says Rebecca D. Pentz, PhD, professor of hematology and oncology in research ethics in the Winship Cancer Institute at Emory University, Atlanta, Ga.

Several bills more specific to the issue of genetic discrimination have been introduced within the past 10 years. The major concerns addressed by these bills are that insurers could use genetic information to deny, limit, or cancel insurance policies or that employers could use genetic information as a reason to fire workers or to screen potential employees. The bills include the following:

• H.R. 293, Genetic Information Health Insurance Nondiscrimination Act of 1999 — Sponsored by Rep John E. Sweeney (R-N.Y.), the act would amend the Public Health Service Act and Employee Retirement Income Security Act of 1974 to prohibit health insurers and group health plans from discriminating against individuals on the basis of genetic information.

• H.R. 602, introduced by Rep Louise McIntosh Slaughter (D-N.Y.), pending in the 107th Congress, which is essentially the same bill as H.R. 306, the Genetic Information Nondiscrimination in Health Insurance Act

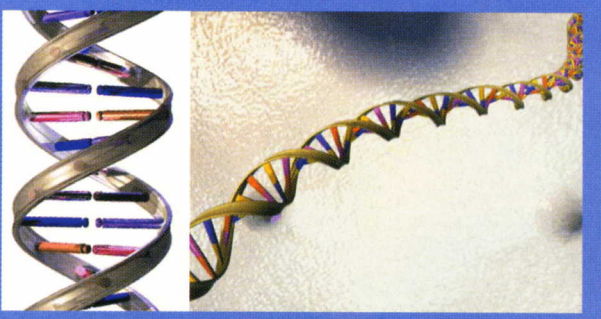

Illustration of a double helix

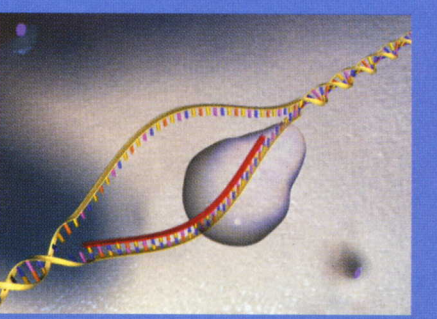

Illustration of a gene

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