At-Home Genetic Tests Pose Ethical Dilemmas

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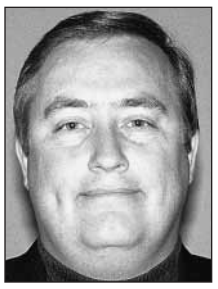
WASHINGTON — The use of at-home genetic tests raises ethical dilemmas for both patients and physicians, several speakers said at a forum sponsored by the Johns Hopkins University Genetics and Public Policy Center.

"Let's be practical," said Richard T. Scott Jr., M.D., of Reproductive Medicine Associates of New Jersey in Morristown. "I'm fortunate because I'm a subspecialist—when I meet with my patients, I usually get an hour. But now you're an internist—you have 7-10 minutes. The patients are going to walk in with their letter [summarizing their test results], and you've got 7 minutes to get through that, it's going to create a problem."



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DR. SCOTT

In general, "things which move patients toward being better informed so they have a better understanding of their circumstances and make better decisions in their life, including treatment decisions, have to be good," Dr. Scott added. "But when it comes to putting these tests together, the real question will be what tests are you going to offer, and how are you going to decide if those tests are appropriate?"

Whether physicians want them or not, at-home genetic tests are out there; many are for fertility- or gynecologic-related conditions. One company, San Francisco-based DNA Direct, offers tests for *BRCA* mutations, thrombophilia, hemochromatosis, cystic fibrosis, and α_1-antitrypsin deficiency. Prices for the tests range from $199 to $3,311 and include a "personalized report" of the results and phone consultation with a genetic expert.

Many of the tests are done with a cheek swab; the sample is sent through the mail. For tests that require a blood sample, the customer is directed to a federally certified lab that collects the sample and ships it in the manner required, according to Ryan Phelan, DNA Direct's founder and CEO. In either case, the customer remains anonymous throughout the process.

Michael Mennuti, M.D., chair of the obstetrics and gynecology department at the Hospital of the University of Pennsylvania in Philadelphia, also expressed concerns. "When you have laboratory tests available to patients who have a problem, and you report a normal result [on the first test], then the patient's next question is, 'Well, what is causing this problem, and what's the next test?' " said Dr. Mennuti, who is also president-elect of the American College of Obstetricians and Gynecologists. "Here you're communicating with someone who has an employer-employee relationship with the laboratory. There is a potential for a conflict of interest."

Customers have several reasons for using at-home tests, Ms. Phelan said. "One is access to a test that their physician did not know about, did not choose to offer, or did not recommend. Number two is to get it interpreted by experts who actually understand the nuances. And third is insurance discrimination or worry about insurance discrimination."

Physicians' lack of genetic knowledge is a real issue, said R. Alta Charo, professor of law and medical ethics at the University of Wisconsin at Madison. When she worked for the now-defunct Federal Office of Technology Assessment about 15 years ago, the office surveyed physicians about which men they would allow to donate sperm for their patients.

"We discovered a woeful lack of understanding of basic genetics," she said. "They would screen out perfectly healthy men who had first-degree relatives with hemophilia—apparently not recognizing that if they were healthy they couldn't have the hemophilia mutation—but would allow in men who had parents with Huntington's chorea and who were young enough not to have presented [with symptoms]."

Although medical schools have since made great strides in these areas, "I would suspect that there is still a relative lack of sophisticated understanding of modern genetics prevalent among many physicians," Ms. Charo said.

Some panelists expressed concern about lack of regulation of at-home genetic tests. Gail Javitt, policy analyst at the Genetics and Public Policy Center, noted that state laws on at-home tests vary greatly. "Some states require a health care provider to order a test and get the results back; other states don't require this," she said. But "even when a provider's involvement is required, it does not necessarily have to be the patient's personal physician. It can be a provider associated with the company offering the test."

As for the actual tests, there is little government oversight, she noted. "Most genetic tests are provided by clinical laboratories, and the laboratory director decides when to offer the tests and what tests to offer. . . . Of the more than 800 tests currently out there, only a handful are regulated by the [Food and Drug Administration]."

Steve Gutman of the FDA's Office of In-Vitro Diagnostics, who was in the audience, said that the FDA is "not disinterested" in this area. The agency "is not unaware of very colorful array of forces that are operating here, and it does not have easy answers. The unanswered question is how the FDA might move from the small set of commercial tests it now regulates to broader regulation of building blocks of home-brewed tests or the home-brewed tests themselves. I can't make any promise about timelines, but I can make a promise that the last chapter hasn't been written. It's a novel in progress." ■

POLICY & PRACTICE

The Chosen Profession

"Be a physician" is the most common career advice that Americans give young adults, according to a Gallup poll of 1,003 adults aged 18 years and older. Of those who responded to the survey, 20% recommended that young women become doctors, while 17% suggested medicine as a career for young men. By comparison, only 11% and 8% suggested that women and men choose careers in computers, respectively. Nursing continues to be viewed as a women's profession: 13% thought women should choose nursing, but that choice did not even make the top five careers for men. Medicine has always been cited as a top career choice for men, although the percentages have been rising steadily over the years for women, as more pursue careers as physicians. "These poll results offer great encouragement for a profession facing a diversity gap and a workforce deficit," said Jordan Cohen, M.D., president of the Association of American Medical Colleges.

Pay-for-Performance Shortfalls

The "pay-for-performance" style of reimbursement system is still largely untested and is not designed to reap cost savings, "particularly since most of the quality measures it targets are measures of underuse," Meredith B. Rosenthal, Ph.D., of Harvard School of Public Health, Boston, said during testimony before a subcommittee of the House Committee on Education and the Workforce. Also, there is little guidance in the literature for purchasers and health plans to reference when they set out to design pay-for-performance programs. Coordination among payers in using these measures is needed, she said. "If only a few of the many payers that a provider contracts with are paying for performance, or if each payer focuses on a different measure set, the effects of pay for performance may be dulled." She suggested that Congress fund more research by the Agency for Healthcare Research and Quality to identify approaches that would improve this method's cost-effectiveness and increase the likely gains in quality of care.

Monitoring Health Fraud

The Federal Bureau of Investigation is not monitoring its spending on health care fraud investigations as carefully as it should, according to a report from the Government Accountability Office. The report, requested by the chairman of the Senate Finance Committee, Chuck Grassley (R-Iowa), found that some agents who previously were assigned to work on health care fraud had been shifted to counterterrorism activities. The GAO said it had been told by the FBI that the bureau wasn't too concerned about not spending enough because most of the time such spending was "historically far in excess" of the budgeted amount. "However, once FBI began to shift agent resources away from health care fraud investigations, agent[s] . . . charged to health care fraud investigations fell below the budgeted amounts." The GAO recommended that the FBI improve its monitoring capability and establish better reporting procedures. The bureau said it already has taken steps in that direction.

Illinois Malpractice Bill

Another state has taken steps to curb rising malpractice costs. In May, the Illinois General Assembly approved legislation to place caps of $500,000 per physician and $1 million per hospital on noneconomic damages. The legislation also calls for increased physician scrutiny by posting disciplinary actions and malpractice lawsuit outcomes on the Internet, and requires insurers to release actuarial data during public hearings called to review rate increases. Steve Schneider, vice president of the American Insurance Association, Midwest Region, took issue with this last provision, indicating it would "send the wrong message to insurers who may be considering entering the market." At press time, Gov. Rod Blagojevich (D) was expected to sign the bill into law.

Medicaid Commission Formed

To strengthen Medicaid, the Department of Health and Human Services established an advisory commission to identify reforms necessary to stabilize the program. The commission must submit two reports to HHS Secretary Mike Leavitt. The first, due Sept. 1, will outline recommendations for Medicaid to save $10 billion over the next 5 years, targeting potential long-term enhancements and performance goals. The second, due Dec. 31, 2006, will make recommendations to help ensure Medicaid's long-term sustainability, addressing issues such as expanding coverage while still being fiscally responsible, and providing long-term care to those in need. Secretary Leavitt plans to appoint up to 15 voting members to the commission with expertise in health care policy, finance, or administration.

Gender-Difference Research Stalled

Research into gender differences is receiving limited funding at the National Institutes of Health, according to the Society for Women's Health Research (SWHR). Grants awarded to study gender differences make up a small percentage of the total number of NIH grants, and none of the NIH institutes had devoted more than 8% of its funded grants to research on gender differences from 2000 to 2003, according to a report from SWHR. "We looked at NIH research grants awarded between 2000 and 2003 and found that across all institutes, an average of just 3% of grants focused on sex differences," Sherry Marts, Ph.D., SWHR vice president for scientific affairs and the study author, said in a statement. SWHR officials said they had hoped to see increasing levels of funding for gender-related research, but they are encouraged that some NIH institutes have established mechanisms to foster such research.

—Jennifer Silverman