

IN EVERY ISSUE

Home-Brewed Gene Tests

BY DIANE COLE

A growing number of companies are directly offering to assess a person's DNA—testing for everything ranging from risk of hereditary breast and ovarian cancer to cystic fibrosis to how quickly a person metabolizes caffeine.

For cancer patients and family members worried about their genetic legacy, access to these tests is only a mouse click away. But the Food and Drug Administration is taking steps to regulate the budding industry of what it calls in vitro diagnostic multivariate index assays.

The FDA already regulates genetic test kits sold to hospitals because they fall under the realm of medical devices, but genetic tests developed and performed by a single laboratory were previously considered lab services. The FDA released draft guidelines in early September that create a new category for the multivariate index assays, which involve multiple genes and complex data analysis. The Centers for Medicare and Medicaid Services currently regulate multigene assays under the Clinical Laboratory Improvement Amendments of 1988.

For consumers worried about confidentiality, these tests can be ordered online, making “do-it-yourself” part of the appeal. But lack of medical oversight is part of the problem. Steve Gutman, MD, director of the FDA's Office of In Vitro Diagnostic Device Evaluation and Safety, says consumers should be wary because lab results are not always reliable, many tests lack scientific data gained only from clinical trials and even reliable test results are difficult to interpret without the help of a trained medical professional. While companies like Sciona Inc. or Genelex Corp. offer DNA tests for things like nutritional predispositions, other companies deal with tests for serious diseases, including cancer.

“These [cancer gene] tests can be complicated and very nuanced, and do not lend themselves well to self-testing in the same way as a test for cholesterol or pregnancy,” Dr. Gutman says. “Tests purchased using the Internet should be approached with care.” Though insurance often covers most or all of the cost of genetic testing for appropriate candidates, the at-home tests can set consumers back anywhere from \$100 to thousands of dollars.

San Francisco-based DNA Direct, a company formed in March 2005, offers

direct-to-consumer tests for a variety of illnesses. Its cancer tests look for 23 DNA markers associated with colorectal cancer and precancerous polyps, BRCA1 and BRCA2 for breast and ovarian cancer and the tamoxifen 2D6 gene, which reveals the effectiveness of tamoxifen in a specific breast cancer patient. DNA Direct has performed about 1,000 tests since 2005, and about 15 percent of the patients who contact DNA Direct are referred by a medical practitioner, says DNA Direct's clinical director Elissa Levin, who is a genetic counselor.

“Before we engage anyone in the testing process, they go through a comprehensive genetic counseling session by phone,” usually 45 minutes to an hour long. For most tests, she says the results are delivered by phone, and a detailed, secure report is made available online. Subsequent counseling sessions and the option to send the report to the primary care doctor for follow-up care are also offered.

Despite the test result—positive or negative—genes only provide limited predictability for cancer risk. Many cancers are caused by interactions of more than one gene, as well as environmental factors, such as diet, smoking and a variety of cancer-causing exposures. Patients, with the assistance of a medical professional, must take all factors into consideration to determine and understand their individual risk.

Before taking any direct-to-consumer test, experts advise making sure the tests offered by a given company are research-backed, medically accepted procedures. Patients should confirm that trained, board-certified genetic counselors work for the company and are available before, during and after the testing process. And it's critical to check the bona fides of the lab conducting the test by seeing if it follows standards set by the CLIA.

In addition to mail-order tests, other tests ordered by physicians, such as the well-known OncotypeDX™ that measures risk of breast cancer recurrence, also fall into the category of tests the FDA plans to regulate.

To find a genetics professional in your area, contact the National Cancer Institute at 800-422-6237 or visit www.cancer.gov/search/geneticsservices.