The Wide, Wild World of Genetic Testing

By ANDREW POLLACK

A MEDICAL journal in March published a study suggesting that drinking coffee can raise the risk of heart attack, but only for people with a gene that makes them slow metabolizers of caffeine. Experts called the finding intriguing, but said it needed to be validated by others and its health implications better understood.

Still, Consumer Genetics, a company formed only a month earlier, is already advertising a genetic test that purports to tell consumers whether they can continue to enjoy their morning jolt.

That is how fast things can move in the rapidly expanding, chaotic and largely unregulated world of direct-to-consumer genetic testing, an industry at the confluence of two of the greatest of recent technological achievements — the Human Genome Project and the Internet.

With a few mouse clicks, consumers can order tests that promise to tell them if they are at risk for particular diseases, to trace their ancestry back to the time of Genghis Khan, to help choose which antidepressant would be best for them, to identify the sex of their fetus as few as five weeks into pregnancy and to give advice on diet or exercise.

About two dozen companies, most started in the last few years, now offer tests, which generally cost from just under $100 to several hundred dollars. Usually, consumers receive a kit requiring them to swab the inside of their cheek to obtain a DNA sample, which they mail back to the company.

Some of the companies say their business is growing quickly. "People are beginning to take a whole lot of interest in preserving their own health," said Kim Bechthold, chief executive of NeuroMark Genomics, which offers a test that it says determines whether a person is at risk of depression after experiencing life's traumas.

But other companies are struggling. And there are other clouds over the industry.

Critics say that many tests are unproven, unnecessary or even "modern-day snake oil," as Senator Gordon H. Smith, Republican of Oregon, said in July when he was chairman of a hearing on home DNA tests. The Federal Trade Commission has issued an alert to consumers that some tests "lack scientific validity and others provide medical results that are meaningful only in the context of a full medical evaluation."

Even when the tests are valid, some genetics experts say, they are so complex that consumers need a doctor or even a genetic counselor to help them interpret the results.

The quality of service can also be questionable. As of early this month, Consumer Genetics did not have the caffeine test ready, even though it was advertised on its Web site. And one customer who ordered a test from NeuroMark in May did not receive a test until mid-August, despite numerous e-mailed promises from the
company. Ms. Bechthold said the company had been overwhelmed by demand after its test was mentioned in an article.

Some of the tests are well validated, and some service is fastidious. Some testers say that it is a matter of judgment when a test has been sufficiently validated, and that consumers have the right to information.

“This is really all about consumer choice at the end of the day,” said Ryan Phelan, chief executive of DNA Direct, a San Francisco company that offers generally well-established tests, like for genetic mutations that cause cystic fibrosis.

The history, strategy and financial wherewithal of the companies offering at-home tests vary. Sciona, a Boulder, Colo., company that offers diet advice based on genetic variations, has attracted $20 million from biotechnology venture capitalists. Sorenson Genomics, which offers ancestry testing through its Relative Genetics and GeneTree Web sites, was bankrolled by James LeVoy Sorenson, a medical-device entrepreneur.

But many companies appear to operate on a shoestring. Theral Timpson, president of Consumer Genetics, said his company, in Sunnyvale, Calif., had not raised much money yet and hoped to support itself by selling tests in the meantime. Mr. Timpson, who once was a marketing manager for a company selling labware, said his company was formed after he and a business partner met someone who had developed a test to determine the sex of a six-week-old fetus.

Then there are companies like CyGene Laboratories and DNAPrint Genomics, which were developing other technologies but began offering consumer tests, apparently as a more immediate way to bring in revenue.

Jorge A. Leon, president of Leomics Associates, a diagnostics industry consulting firm, estimated it would require $2.5 million to set up a government-certified testing lab and a Web site for marketing. But a garage lab could be started with a few hundred thousand dollars, he said. And many of the consumer testing companies farm out their work to a lab rather than set up their own.

Once a scientific paper about the effect of a genetic variation is published, a test for the variation can usually be set up in months. In some cases there are patent rights that must be licensed, in other cases not. Or a patent might be granted years after the first scientific publication and companies already offering the test might be asked to stop.

Most of the companies involved in direct-to-consumer DNA testing are private, so it is difficult to judge how they are doing. Sorenson Genomics contends that it does more than $10 million a year in consumer genealogy testing. “Our business has doubled in just six months,” said Doug Fogg, the company’s chief operating officer.

Genelex, one of the oldest companies in the business, has been offering direct-to-consumer paternity tests since the mid-1990’s. Howard Coleman, its founder, had worked as a contractor and ski instructor before going back to school to study molecular biology.

Last year, the company, which is privately held and based in Seattle, had $2.2 million in revenues, Mr. Coleman said. It sold about 2,000 paternity tests and 500 each of pharmacogenetic and nutrigenetic tests. Sales of the latter two tests, which use information on certain genes to recommend medicines or a certain diet, are doubling every year, he said.
But the regulatory filings of companies that are publicly traded paint a more dismal picture. GeneLink, of Jersey City, said that sales in the first half of this year dropped 70 percent, to $83,337, because it did not have enough money for marketing and sales. It had only $82,000 in cash.

In some cases, genetic tests are used to sell “personalized” nutritional supplements or skin creams. Some Sciona tests are sold by Market America, which also sells supplements. And the test developer Interleukin Genetics is majority owned by the parent company of Amway.

The nutrigenetic tests were criticized at the Senate hearing. The Government Accountability Office said that it had found “personalized” supplements costing $1,200 a year with the same ingredients as vitamins that sold for $35 a year at a store.

Such criticisms could increase the pressure for regulation. “I think it’s impossible to discern which companies are reputable and which are reprehensible,” said Kathy Hudson, director of the genetics and public policy center at Johns Hopkins University.

Tests offered by a single laboratory usually do not need approval from the Food and Drug Administration before they can be sold. But the F.D.A. is showing signs of changing its stance.

Medical testing laboratories are regulated by Medicare, but that regulation is intended to determine only if a lab can detect a genetic variation reliably. It is not concerned whether the variation really does mean a person is at risk of depression or shouldn’t drink coffee.

Some people in the industry say they welcome some regulation, though requiring every test to be pre-approved by the F.D.A. could stifle development. Voluntary industry standards may be one solution.

“We do look forward to developing appropriate standards for this business,” said Rosalynn Gill-Garrison, a founder at Sciona.