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DNA tests a mouse click away



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Sussing out secrets from your DNA can start with a home kit.

Cancer tests

Many women who qualify for provincially funded genetic testing for breast and ovarian cancer are not getting it, says Dr. Steven Narod, the Canada

Research Chair in Breast Cancer.

Currently a woman's doctor must refer her to a genetics centre. But in a study of women with invasive ovarian cancer, which qualifies for testing, only 20 per cent had been referred.

Generally, genetic testing should be considered for families with multiple cases of breast cancer and/or ovarian cancer, especially when they're in close relatives over generations.

Aletta Poll, genetics counsellor at the Familial Breast Cancer Clinic at Women's College Research Institute, says several types of cases qualify for testing, with no family history:

Women who develop breast cancer under the age of 35.

Male breast cancer.

Invasive serous ovarian cancer.

The testing looks for mutations in the BRCA1 or BRCA2 genes that can increase a woman's lifetime chance up to 75 per cent of developing breast cancer, and from 25 to 40 per cent of developing ovarian cancer.

Narod suggests that a woman who meets the provincial criteria get a referral to a genetics centre or call his clinic at 416-351-3765.

Nancy J. White

Home genetics tests are a growing field that can help determine cancer risks, baby gender and even how humans metabolize caffeine

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LIVING REPORTER

The cheek swab kit came in the mail.

Carol Morgan, who had breast cancer, ordered it over the Internet from a genetic testing company, wanting to be sure she'd fully benefit from the drug tamoxifen prescribed by her doctor. Genetic variations may influence how tamoxifen is metabolized.

Her cancer centre in Victoria, B.C., didn't test for it, so she paid \$300 (U.S.) to DNADirect, a San Francisco-based Internet company, swabbed the inside of her cheek and mailed off her buccal cells.

"It felt a little odd to do, no question," says Morgan, 43, who found out she's a good metabolizer. "But it was just another stone you uncover to make sure you're doing the right thing."

The genetics revolution, or at least the promise of it, is available for a price on the Internet.

Want to know your risk for diabetes, heart disease, colon cancer? How you process antidepressants? Your optimal diet plan? Gender of a fetus? DNA tests are a mouse click away.

There's even a test for caffeine metabolism – slowpokes supposedly run increased health risks.

Sussing out secrets from your DNA is a growing field: as researchers discover more genetic links, marketers develop more swab-yourself tests.

But critics of this brave new world of consumer genetics voice concerns about a lack of counselling, lab quality and possibly false promises.

"It's not just about consumer choice," says Trudo Lemmens, associate professor in law and medicine at the University of Toronto, who thinks a regulatory approval system is needed. "It's about too much potentially misleading information, about creating problems in the public system based on wrong information."

In Britain, several scientific groups are calling for an official body to monitor the tests, to weed out valuable services from what one scientist called "genetic gobbledygook."

In this country, Health Canada does not regulate genetic testing services that involve a sample being sent to a lab.

While the science is solid for some tests, it's still shaky on many others.

"It's 1492 and a half. Columbus has just come back. That's equivalent to where we are," says Roderick McInnes, scientific director of the Institute of Genetics, part of the Canadian Institutes of Health Research. "We're just beginning to realize the potential."

Out of approximately 25,000 genes, scientists don't know what about half of them do, he explains, and they're just starting to learn how they work together.

But if there's a gene for patience, it's not widely inherited.

This is the age of empowerment, with people demanding information, pronto. Especially if it affects their health.

Besides her tamoxifen question, Morgan also wanted to know if she carried an inherited breast cancer risk. She had developed the disease fairly young, age 42, and her grandmother had had it. Mutations on genes BRCA1 or BRCA2 could mean Morgan ran a higher risk of ovarian cancer and a second breast cancer. The Victoria resident didn't qualify for the provincially funded gene test because her risk of having the mutation was too low.

So she went back to DNADirect. She paid \$3,500 (U.S.) for the blood test and received genetic counselling over the telephone before and afterwards. The lab results came back negative.

"Seven per cent is significant to me," explains Morgan, a veterinarian. "Risk is one of those things that's in the eye of the beholder."

Risk, indeed. While some diseases are determined by a single gene mutation, most illnesses are much less straightforward. Gene variations signify only some risk.

So what do you need to get the disease? "The wrong environment and probably other predisposing gene variants," answers McInnes. "It's a mix, a witch's brew."

Not all Internet companies offer genetic counselling, leaving the consumer alone to ponder the percentages.

Without professional advice, McInnes says, some people may overreact, joining the ranks of what's known as "the worried well." Others may fall victim to genetic determinism, giving up on prevention, convinced their destiny is cast in their DNA.

Even with clearer genetic links, a test may not be the best bet.

McInnes points out that researchers have pinpointed several gene variants that increase a person's risk of age-related macular degeneration to as high as 90 per cent.

The eye disease affects about 35 per cent of people over age 75 in Western countries and can lead to blindness. So should people be genetically tested?

Not at this time, says McInnes. "The value of this tidal wave of genetic information is insight into diseases where we didn't have a clue," he says. "Long-term, whether it's 10 or 30 or 50 years, it will translate into prevention and treatment that will impact the disease. There's huge faith in this."

Another area of huge promise is pharmacogenomics, the interplay between genes and drugs. For some Internet companies, drug response tests are already popular sellers. Genelex, a web-based Seattle firm, offers screening for five genes that it maintains will help a doctor predict response to many drugs.

"It's like looking below the water line of adverse drug reactions," says Howard Coleman, Genelex CEO. "Add the genetics and you get a better view of what's going on for people, especially those taking multiple medications."

Toronto resident Ann Elizabeth Carson purchased Genelex's drug response testing for \$1,000 (U.S.). After a severe adverse reaction years ago, she's avoided pharmaceuticals, but Carson, 79, author of *My Grandmother's Hair* and a poetry collection, *Shadows Light*, worries that someday she may need drugs. Her test results showed that she metabolized some drugs too slowly, some too fast and others barely at all.

With help from a Genelex counsellor, she's identified a couple of drugs she can likely handle. But the testing was limited and, she says, her case is extreme and likely may involve allergies.

"I feel validated," says Carson, a retired psychoanalyst, who wishes doctors were able to use genetic drug-response testing. "It's helping steer me to get the best care I can get."

Some companies sell response tests to particular drugs, such as warfarin, marketed as Coumadin, and widely used for blood thinning. Last August, the U.S. Food and Drug Administration updated warfarin labelling to explain that a person's genetic makeup might influence reactions. Health Canada is updating its list of drugs with warfarin to include genetic information.

Yet not even the warfarin story is complete, according to an editorial in *The New England Journal of Medicine*, on March 6. It refers to an accompanying article on new findings about the genetic response to the drug.

The editorial is entitled: "Pharmacogenomics – Ready for Prime Time?"

"Perhaps not quite yet," answer the authors, adding that gene-based studies will shed more light.

David Cole, head of the division of chemistry and genetics at Sunnybrook Health Sciences Centre, foresees a day when a doctor can electronically log onto information to target the optimal drug at the optimal dose for that patient.

Says Cole: "It's the wave of things to come."

Like so much in genetics.