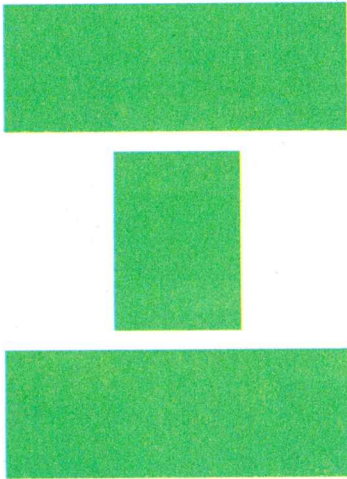


READ THIS BEFORE YOU BUY A GENETIC TESTING KIT

These products are widely accessible, are a snap to use, and promise answers to pressing questions about your family, your health, and your origins. But while at-home DNA testing is a scientific marvel, it can also offer an incomplete picture of your risk of various diseases, reveal unwelcome truths about relatives, get your ancestry wrong, and compromise your privacy.

BY CATHERINE ROBERTS ■ ILLUSTRATIONS BY BEN SHMULEVITCH



IN SPRING 2003, excitement about the future of genetic science and its potential to dramatically advance the course of disease prevention and treatment was running high.

That April, the National Institutes of Health (NIH) announced that researchers had completed the first full sequence of the human genome—the biological instructions for building the human body, decoded from 3 billion twisted strands of DNA.

The hope was that pinpointing the identity and location of every gene in the body would lead to an understanding of the inherited risks for diseases such as cancer, improve care for conditions like diabetes, and help scientists develop targeted therapies for disorders such as Alzheimer's, which had eluded effective treatment for years.

It was a “historic moment,” Francis Collins, MD, director of the NIH, told a congressional subcommittee at the time. “It is our sincere belief that the newly created discipline of genomics will make a profound difference to the health and well-being of all the people of this world,” he said.

Almost two decades later, genomics has led to critical progress in medical science, particularly in identifying

individuals' genetic predispositions to diseases such as breast cancer.

This knowledge, however, is also being used toward somewhat less profound ends. An endeavor that originally involved billions of government dollars and collaboration among thousands of scientists around the globe has yielded not just medical progress but also a slew of consumer products in the form of at-home genetic tests.

The Rise of DIY Gene Testing

For a relatively modest fee, anyone can purchase a direct-to-consumer (DTC) genetic test kit. Users spit into a tube or swab the inside of their mouth to obtain DNA (molecules that form the building blocks of genes), mail the sample off, and receive an analysis a few weeks later.

The kits are wildly popular: About 1 in 5 Americans has taken a DTC genetic test, according to an October 2020 Consumer Reports nationally representative survey of 2,000 U.S. adults. And brands such as 23andMe and Ancestry are household names.

Manufacturers offer a variety of possibilities, including matching you with unknown relatives, telling you which country your ancestors came from, revealing your risk of certain illnesses, and even determining which diet is best for you.

But while these tests may offer potentially valuable genealogical or medical insights, experts say results may be easy to misinterpret or could be based on a misapplication of the science. “If you go in there thinking that this test is going to tell you who you are, you're going to be wrong,” says Wendy Roth, PhD, an associate professor of sociology at the University of Pennsylvania in Philadelphia.

A genetic test can also leave you with information you'd prefer not to have about your family or about your risk for an incurable disease. And once data about your genes is shared, it can be sold

or even potentially used to discriminate against you (see “The Privacy Risks of Genetic Tests,” on page 38).

DIY Tests vs. Doctor Tests

We all have small differences in our DNA code, known as variants, which genetic testing can reveal. Some variants are benign, some are linked to certain traits or illnesses, and many others are largely a mystery, their significance unknown.

When a doctor recommends genetic testing as part of your care, they're usually aiming to answer a specific question about your health, says Kyle Brothers, MD, an associate professor in the department of pediatrics at the University of Louisville in Kentucky. He says they will typically order tests focused only on that particular question, such as your inherited risk of cancer. If you send away your saliva for DTC genetic testing, however, the lab will look at a whole range of variants that have been linked—sometimes quite loosely—to a number of traits, some not related to your health at all.

Think of it this way: When your doctor orders genetic testing, it's akin to fishing for a particular fish, in a part of the ocean where it's known to live. A DTC test is more like throwing a net into the ocean and seeing what comes back.

Here are four common claims from the manufacturers of these products, whether they deliver, and what to know about their potential pitfalls.

CLAIM **1** **THE TESTS CAN FIND FAR-FLUNG RELATIVES**

HOW IT WORKS To determine whether you might be related to another person in its customer database, a DTC testing company considers how many identical



segments of DNA you have in common and the length of those segments. The closer the relative, the more large DNA segments you'll share.

"For close relationships, those predictions can be made very accurately," says Debbie Kennett, a genetic genealogist in the U.K. who helps people construct family trees using DTC testing and traditional methods, such as digging into historical records.

For instance, "relative matching" tests will always work with a parent or full sibling, she says. The further away a relative is on the family tree, however, the less certain the results. "If you share, say, 25 percent of your DNA, then there's a wider range of possible relationships," Kennett says.

THE DRAWBACKS While a test might unearth a previously unknown cousin

who becomes your new best friend, you could also learn something negative or even shocking about a family member or relationship. In CR's survey, 9 percent of respondents who used these tests said their reports contained unsettling information, such as news that someone thought to be a biological relative—like a father—isn't genetically related.

An unexpected discovery about a relative is one of the most frequent surprises for users of DTC genetic tests, says Anita DeLongis, PhD, a professor in the department of psychology at the University of British Columbia in Vancouver, who is studying the effects these products have had on people's lives. Many of her study subjects, she says, "had not anticipated learning a family secret."

CLAIM 2 TESTING CAN UNCOVER WHERE YOUR ANCESTORS ARE FROM

HOW IT WORKS Companies such as Ancestry and 23andMe say they can tell you the regions—and sometimes even the countries—where your ancestors originated, based on your DNA. In CR's survey, curiosity about ancestry was the most common reason for taking a DTC genetic test, listed by about two-thirds of respondents.

Finding relatives involves comparing your DNA directly with other people's in a database, but estimating your genetic ancestry is more roundabout. One common strategy looks for variations in your genetic code that have been found to be statistically more likely in people from certain regions. The testing company then gives you an estimate of the percentage of your DNA that comes from such areas. For example, your result might show that 40 percent of your DNA comes from Europe and 60 percent comes from Asia.

THE DRAWBACKS It's hard for testing to pin down genetic ancestry accurately for several reasons. Different test makers may rely on different sets of data, including, often, some from their own customers—which can skew findings. Also, the fewer samples a company has from a certain region, the more limited its ability to tell you whether you have ancestors from that area. On the flip side, as more people take one company's test, its pool of information should become larger and better. That can cause the strange phenomenon of having your genetic ancestry test results change over time as a manufacturer widens its "reference database."

Another concern: The databases often have genetic details on people whose families came from a particular region in the recent past. But ancestry

stretches back much further than, say, our grandparents. “It’s not always clear that the population you’re being compared to is the same one that was in that location several hundred or thousand years ago,” says the University of Pennsylvania’s Wendy Roth.

It’s important to remember, too, that although a given gene variant may be more common among one group of people, it can also appear in others. So finding a particular variation in your genetic code doesn’t definitively place you in any one regional, ethnic, or

racial group. What all this means for consumers, according to Mwenza Blell, PhD, a biosocial medical anthropologist at Newcastle University in the U.K., is that genetic ancestry tests are closer to palm reading than science.

This testing also has an unintended social consequence, experts say. Because country-of-origin labels closely overlap with commonly used ethnic and racial labels, the test can reinforce the mistaken idea that your ethnicity or race is encoded in your genes.

In reality, the mapping of the human genome demonstrated that all people

are more than 99 percent the same genetically, regardless of race or ethnicity. This finding represented a major milestone for scientists and historians who had been working to dismantle the idea that racial categories—an invention of modern history—were biological rather than social.

The emergence of DTC genetic testing and its focus on genetic ancestry has proved to be a major and unexpected setback to this work, Blell says, by reinscribing notions of biological differences among races.

A 23andMe representative told CR that

THE PRIVACY RISKS OF GENETIC TESTS

WHAT HAPPENS TO YOUR genetic data after you mail your sample to a private company for analysis? We don’t exactly know, because few laws regulate how companies can use your information and what they must do to keep it private and secure, according to Consumer Reports’ privacy experts. “An individual’s most personal information is being bought, sold, and traded without clear understanding or consent,” says Justin Brookman, CR’s director of privacy and technology policy.

Federal protection is limited. The Genetic Information Nondiscrimination Act prevents employer discrimination based on genetic information—meaning you can’t be fired because of,

for instance, a genetic problem that predisposes you to cancer. But that covers only genetic conditions that aren’t yet symptomatic.

The Affordable Care Act bars health insurers from refusing applicants or charging more based on a preexisting condition—which includes any illness discovered as the result of genetic testing, says Ellen Clayton, MD, a professor of pediatrics, law, and health policy at Vanderbilt University Medical Center in Nashville, Tenn. A handful of states have also enacted laws protecting some aspects of genetic privacy.

THE GAPS IN THE LAW

Even with existing laws, DTC genetic testing companies have substantial flexibility in controlling what happens to your data once they receive it, Brookman says.

That can be problematic. Unlike your credit card number, your genetic data can’t be changed if it’s stolen or if it’s shared without your consent.

Law enforcement agencies have already used services that allow people to publicly post their genetic data—in an effort to find relatives—to help solve high-profile cold cases. This is a controversial practice among privacy experts and consumers, who might not want to unwittingly help put a relative behind bars.

Also concerning is that genetic data could, at least theoretically, be used in determining your rate for life, long-term-care, or disability insurance in almost every state. And if you seek damages for a work-related injury, your employer or its insurer could try to use your

genomic data to challenge your claim for compensation.

Another possibility is that mortgage lenders might require a genetic test before deciding whether to approve a mortgage, says Mark Rothstein, a lawyer and a professor at the University of Louisville School of Medicine in Kentucky, in order to weed out applicants who, for instance, have an inherited risk of Alzheimer’s disease.

THE PROTECTIONS WE NEED

With a few exceptions, when it comes to your privacy, DTC genetic test makers have to abide only by the data privacy rules in their own privacy policies, which they can change at will. CR believes lawmakers should pass regulations to make results from all genetic testing private by default. These laws should also require strict safeguards to prevent data theft and the potential reidentification of anonymized data, Brookman says.

it has worked to dispel these notions, “including noting in our reports the results are based on recent ancestor locations.” Ancestry did not provide a comment on this by press time.

CLAIM

3

GENETIC TESTS CAN REVEAL YOUR RISK FOR CERTAIN DISEASES

HOW IT WORKS Some manufacturers say that their tests, using research about variants associated with various conditions, can help assess whether you are more likely to develop a disease such as breast cancer or Alzheimer’s.

The company 23andMe does this using technology for which it has received approval from the Food and Drug Administration, allowing the firm to sell the test directly to you. Others use what’s known as physician-mediated tests. Once you purchase such a kit, the company enlists a doctor—generally one who is associated with the test maker and does not meet with you directly. This lets the manufacturer offer the test without FDA approval, as long as it is analyzed in a federally certified lab, says Katie Stoll, executive director of the Genetic Support Foundation, a nonprofit that provides genetic counseling services.

THE DRAWBACKS The picture of your disease risk that you get from a DTC genetic kit might be less complete than that of a test you get as part of your regular medical care.

In contrast to at-home testing, your own healthcare providers will generally work with you closely to determine whether you need certain tests, based on your health history and your family’s. They’ll also pair you with licensed genetic counselors, who can help ensure that you’re tested for all potentially relevant genetic variants and that your testing will be covered by insurance.

And DTC disease-risk tests might not include a wide range of important gene variations. Take, for instance, 23andMe’s test for variants of the BRCA1 and BRCA2 genes, which have been linked to a higher risk of breast and ovarian cancer. The company’s test looks for three specific variants that have been well-studied and are most common among people of Ashkenazi Jewish descent. But thousands of additional variants of these same genes are also known to cause breast and ovarian cancer, including certain variations that occur more frequently in other populations, according to genetic counselor Kelly Ormond, a professor in the department of genetics at Stanford University School of Medicine in California.

What this means: A negative result may provide a false sense of relief because it doesn’t look at all the variants, Ormond says. Also, while DTC test makers often offer genetic counseling, a company counselor will be less familiar with your medical history than a counselor your doctor refers you to, Brothers says.

AncestryHealth, which takes the physician-mediated approach for health tests, told CR it helps customers understand their results, in part, by partnering with “an independent group of physicians and genetic counselors.” And 23andMe says it clearly explains test limitations to users, noting prominently on its website that its BRCA1 and BRCA2 results, for example, test for only three variants of those genes.

CLAIM

4

THESE TESTS CAN TELL WHAT DIET IS BEST FOR YOU

HOW IT WORKS A number of DTC testing companies currently offer personalized diet recommendations based on your genetics. Some draw their advice from existing studies, such as those on how,

for instance, a specific genetic variation may be linked to a better response to a certain diet or exercise program.

THE DRAWBACKS The prospect of incorporating genetic information into dietary advice is exciting because it could allow dietitians to better tailor advice to individuals’ needs, says Amy Ellis, PhD, an associate professor in the department of human nutrition at the University of Alabama in Tuscaloosa.

But far more and better evidence is needed. Right now, the dietary advice from DTC companies may be based on incomplete evidence or could yield misleading conclusions, according to recent research. When a 2020 study in the journal *Nutrients* evaluated 45 DTC services offering personalized nutritional advice, it found a number of worrisome practices. For example, the authors say that more than 900 genetic variants may contribute to a person’s risk of obesity. Yet companies often provide weight-loss advice based on just a handful of variants.

What’s more, much of the research about gene variants and diet is observational and hasn’t been adequately replicated in other studies, says George Hindy, PhD, assistant professor of population medicine at Qatar University in Doha. Such research can suggest an association only between a variant and an outcome such as faster weight loss, not a cause-and-effect relationship. When it comes to a good diet, “genes ... are not a definitive answer,” says Yi Sherry Zhang, PhD, the founder of GenoPalate, which provides dietary advice based on genetic testing. “We need a holistic approach.”

SO, WHAT SHOULD YOU DO? In the end, DTC genetic tests can provide certain insights, but it’s important to be aware of their limitations. And keep in mind that your genetics are only one element of your overall well-being: Your environment, healthcare access, and behavior are also important, says James E. Rogers, PhD, CR’s director of food safety research and testing.