

Up to 40% of Consumer DNA Tests Are Inaccurate

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✓ Fact Checked

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STORY AT-A-GLANCE

- › Sales of direct-to-consumer DNA testing kits rose sharply in 2017, driven in part by large advertising budgets and a growing interest in discovering ancestral lineage
- › Although only one company is approved to share information about medical risk for some health conditions, nearly all companies share the raw data; research finds nearly 40% of medical data from these tests may be false positives
- › Clinical confirmation is vital before undergoing further tests or treatments based on DNA testing from at-home test kits as these tests were never intended for medical use
- › Your DNA contains profoundly personal information about your health, personality and family history, which may be used to discriminate against you in the workplace, affect your ability to acquire health insurance and is often sold to research companies

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The number of people requesting a direct-to-consumer DNA test during 2017 more than doubled. According to Ancestry.com, they claim the largest DNA database with more than 7 million people's DNA stored.¹ In combination with other companies collecting DNA data, the industry estimates over 12 million DNA profiles are on file.²

The popularity of these at-home DNA tests has only risen each year. According to David Mittelman, consumer genetics entrepreneur and cofounder of DNAGEeks,³ "The inflection point started in the summer of 2016 and from there it's gone into the stratosphere." However, while highly advertised and an incredibly popular way of

determining your ancestry, these at-home tests may provide false health information⁴ and place your privacy at risk.⁵

What's Being Tested?

DNA identification is a relatively new science. Before testing was available, other biological tools were used to help identify people and determine relationships. One of the first of those tools was blood typing.⁶

In the early 1920s, scientists were able to identify four different blood types. In the 1930s, researchers found other proteins on the surface of blood cells used to identify individuals. It wasn't until the mid-1970s when scientists discovered ways of tissue typing using the human leukocyte antigen (HLA) protein present on all tissue except red blood cells.

Then, in the 1980s the Restriction Fragment Length Polymorphism (RFLP) analysis technique was developed, becoming the first genetic test using DNA. Fast forward to 2010 and researchers are now using next-generation sequencing or massively parallel sequencing as the newest technique for genetic analysis.⁷ When a direct-to-consumer DNA test is requested, often what is tested is autosomal DNA,⁸ or one of 22 pairs of autosomal chromosomes inherited from your parents.

Also called genetic genealogy, DNA testing is used together with documentary evidence to define relationships between individuals. This genealogical DNA testing first became available on a commercial basis in 2000.⁹ Since then a number of companies have established private testing labs and databases, promising to help identify individuals in your family tree up to five generations back using data points compared against others in their database.¹⁰

DNA may potentially be used to map out your family tree, determine the ancestry of your dog, solve crimes or help your physician identify any genetic component to a health condition,¹¹ such as whether or not a woman carries the HER2 gene, knowledge of which will help focus the correct treatment.¹²

However, while solving crimes and identification of specific proteins to drive treatment protocols are completed in highly regulated labs set up to protect your privacy, direct-to-consumer DNA tests are not.

High Percentage of At-Home DNA Tests Provide False Results

In a limited study completed by Ambry Genetics,¹³ a small medical lab in California, researchers discovered using at-home DNA tests to assess for risk of certain diseases or other nonphenotypic traits, such as eye color, resulted in a 40% false positive reading.¹⁴ The information evaluated was about genetic makeup; looking for ancestral linkage was not the issue.

Not long ago, using a DNA sequencing test was an expensive undertaking, costing in the hundreds of thousands of dollars.¹⁵ However, today many of these home tests cost between \$100 and \$200, after which you reportedly have information about your ancestry and your risk of developing certain diseases.

Some companies even claim they can predict the type of wine you like to drink.¹⁶ During the study, Ambry evaluated the variants consumer test results identified associated with an increased risk of disease.

Ambry processes DNA for doctors and research institutions and found nearly 40% of test results from at-home DNA tests contained false positives. In other words, the direct-to-consumer test indicated there was a genetic variant increasing the individual's risk for disease, but according to Ambry's genetic sequencing, there wasn't.¹⁷ The study does not discount DNA testing, but rather points out raw data from consumer testing companies may not be as accurate as consumers hope.

What Ambry found was consumer labs test genotype DNA rather than sequencing it, and use just one method. Although this method is cheaper and quicker than clinical sequencing, it also is less effective and accurate than the clinical laboratory method of sequencing and using another test to confirm a positive variant.

A spokesperson from 23andMe, an at-home DNA test company, discussed the study with a reporter from Gizmodo,¹⁸ telling them 23andMe customers receive a warning the raw data is not necessarily accurate or appropriate for medical use. Contained in their terms of service is a warning that may not be entirely clear to consumers: “This data has undergone a general quality review; however, only a subset of markers have been individually validated for accuracy.”

Clinical Confirmation Needed Before Making Medical Decisions

Although these at-home genetic tests are popular as a relatively inexpensive way to track your ethnicity and genetic history, they are not intended for medical use. This means the data you are given is not a replacement for a real medical diagnosis, nor should the information be used to guide medical treatment.¹⁹

This warning refers to raw DNA data many companies send, containing a complicated list of genetic abnormalities implying you may have a greater likelihood to develop a disorder.

As 23andMe is the only company with U.S. Food and Drug Administration (FDA) approval to market genetic health risk tests for certain conditions, many companies simply offer raw data without an explanation and post a disclaimer that the results are not verified for accuracy. In other words, individuals are receiving information indicating they may be at risk for a significant health concern without any idea of what this actually means.

The authors of the featured study noted²⁰ false results about genetic disorders might lead people to better preventive care, but may also create needless anxiety over a nonexistent issue. The authors believe it is vital further testing is used to confirm a condition prior to designing a treatment protocol.

For instance, while you may have a genetic marker for a health condition, you may never develop it and vice versa. Genetics plays a role in disease development, but in many cases your lifestyle choices and environment play a larger role.

How Home DNA Testing Works

It appears the increase in interest for direct-to-consumer DNA testing could be a result of how much these companies are spending on advertising. In 2016, Ancestry.com spent \$109 million on TV and other advertising and was on track to spend even more in 2017.²¹ Most of the at-home kits work in a similar fashion. Since the company uses both documented evidence and DNA evidence to find your ancestry links, you'll first answer a few questions about yourself.

Once the kit has arrived you collect the sample as directed. Some DNA test kits use a vial of saliva, while others use scrapings from the inside of your cheek.²² Before sending the test kit back, you must register it online with the company. This is an important step as the kit is sent to the company without identifying information.

Registering the kit allows you to see your results online. Since some states have laws governing DNA testing by private companies when the tests are related to medical conditions, it is important to check the site's terms of service to see if there are any restrictions in your locale before ordering a kit.

It's important to note results will be different for men and women, as women, who have the XX chromosome, can only trace their ancestral history through the maternal line of the family. Since men have an XY chromosome, the test can trace both maternal and paternal lines, painting a more complete picture of ancestral lineage.²³

Companies keep their own database of DNA so software can search for DNA matches with as many people as share their results. If you're trying to build a family tree or look for relatives, this could be a useful feature.

Companies Finding Ways to Monetize Your Information

The sheer numbers of individuals who have data in DNA databases may catalyze the growth of other companies who reanalyze the data and monetize your information. For instance, Vinome²⁴ uses your genetic material to predict the type of wine you may find

most tasteful. Other companies use the information to provide a breakdown of nutrition or health risks.²⁵ Unfortunately, this occurs with little oversight from regulators.

In past years, direct-to-consumer DNA companies have worked hard to convince customers testing has value. The rising enthusiasm in genealogy has captured a growing interest in monetizing the platform. Harvard geneticist George Church, Ph.D., founded Nebula Genomics,²⁶ planning to sequence your DNA for under \$1,000, giving you insights about your health and at the same time secure the data.

In other words, when you take a DNA test through a direct-to-consumer company, the company has the right to sell your genetic data to third parties without passing you profit. On the other hand, when you pay Nebula to sequence your entire DNA, you own the data and may sell it to earn digital money.²⁷ Pharmaceutical and biotech companies use large DNA data sets in the development of new pharmaceutical drugs. This data is typically purchased for millions of dollars.

Nebula's goal is to eliminate the middleman so you have the opportunity to sell your own DNA data directly to drug companies and other data buyers. More startup companies are building platforms allowing you to sell your genetic information online, but they are not offering genome sequencing in the package. Although you may be intrigued with the idea of making money from a simple DNA test, it is critical you consider these future repercussions to your privacy.

Know This Before You Take a Direct-to-Consumer DNA Test

Your DNA is your most personal data. In an era where companies are unable to keep passwords and credit card numbers safe, is it realistic to believe they will be able to keep your DNA data safe?

In a paper published in 2013, Yaniv Erlich, Ph.D., computational biologist then working for Whitehead Institute for Biomedical Research, Massachusetts, demonstrated it was possible to discover identities of individuals who participated in genetic research

studies by cross-referencing their data with information freely available on the internet.²⁸

Past studies had demonstrated anonymous genetic databases could be exposed by matching data to a sample of DNA, but Erlich went one step further and found only publicly available information was required. Today, Erlich is on leave from Columbia and working as MyHeritage chief scientific officer, developing testing allowing DNA information to be overlaid on family trees in order to study the genetic implication of DNA and disease.²⁹

Although scientists are excited about the potential information revealed in a deep dive into DNA data, in an era of genetic research, this poses significant problems for your privacy. Hank Greely, director of the Center for Law and the Biosciences at Stanford School of Medicine, comments:³⁰

“There is no legal limit on what they could do other than the agreement that you enter into with them which they may or may not choose to follow. If they don’t follow it, the chance you would ever find out is very, very low. You cannot promise people absolute confidentiality.

The other side of it is that it’s possible that somebody will hack into a company database that does contain your information. My financial information has been hacked three times in two years. All that stuff is out there.”

Your DNA contain sensitive information about your health, personality and family history. In the fine print of nearly every testing company, the company claims ownership of your DNA, allowing third parties to access it and making it vulnerable to hackers.³¹

The direct-to-consumer DNA testing companies make it clear in their terms and conditions how they use your DNA, but these firms are not bound by HIPAA regulations, which means your personal information is unregulated. And, as with all data, the more places it can be found, the more chances are it can be leaked.

Even without an accidental leak, your genetic information may be used for workplace discrimination or in the acquisition of life insurance, long-term care or disability insurance. And, if you share your genetic information with your personal physician and the results are medically relevant, it may also negatively impact your health care coverage.³²

In fact, one company admitted in 2013 the real goal of the company was not to make money selling DNA testing, but instead to collect massive amounts of data they can use without any further consent.³³ So, while it may be tempting to forgo hours of research into developing your family tree by taking a simple DNA test, this test may turn out to be far from simple and cost far more than the original price in the long run.

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