

## Consumer Genetic Testing

June 14, 2019 – Volume 29, Issue 22

***Do the popular DNA tests offer useful information about health risks and heritage?***

By [Barbara Mantel](#)



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### Introduction

*Direct-to-consumer genetic testing, introduced in 2000, has seen explosive growth in recent years. In 2018, as many people purchased the testing kits as in all previous years combined. Companies such as Ancestry, FamilyTreeDNA and 23andMe provide genealogy information to consumers, and 23andMe also analyzes users' genetic risk for 12 diseases and health conditions. But critics say reports produced by the testing companies can be inaccurate, misleading and vulnerable to hacking. Others complain that government oversight is too weak to prevent genetic information from being used to discriminate against consumers or violate the privacy of relatives of people who submit their DNA for testing. Privacy concerns have grown especially acute after law enforcement officials began using some testing companies, such as GEDmatch and FamilyTreeDNA, to try to solve crimes. Testing companies defend the accuracy of their work and their privacy and security policies. They and their supporters, including some geneticists, say consumers have a right to their genetic information and that such data, stripped of identifying information, can help researchers find treatments for diseases.*



Randall Lorenz, of Reno, Nev., and Jerica Starkweather, of Emmett, Idaho, realized they may be close relatives after Lorenz, who learned late in life that he had been adopted as a baby, took a direct-to-consumer genetic test. He holds up a photo of Starkweather's mother, who could be his biological sister or cousin. By the end of 2018, more than 26 million people had used consumer genetic tests to learn more about their ancestry and health risks. (AP Photo/Idaho Statesman/Darin Oswald)

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### Overview

Three years ago, sisters Julie Lawson, 65, and Fredda Hurwitz, 52, took direct-to-consumer genetic ancestry tests and met at Hurwitz' Falls Church, Va., home to share results. Hurwitz' report included the name of an

unfamiliar man as a close genetic match. On Facebook, they found a picture and deduced from his age and resemblance to their father that he was a half-brother, a secret their father had never revealed.

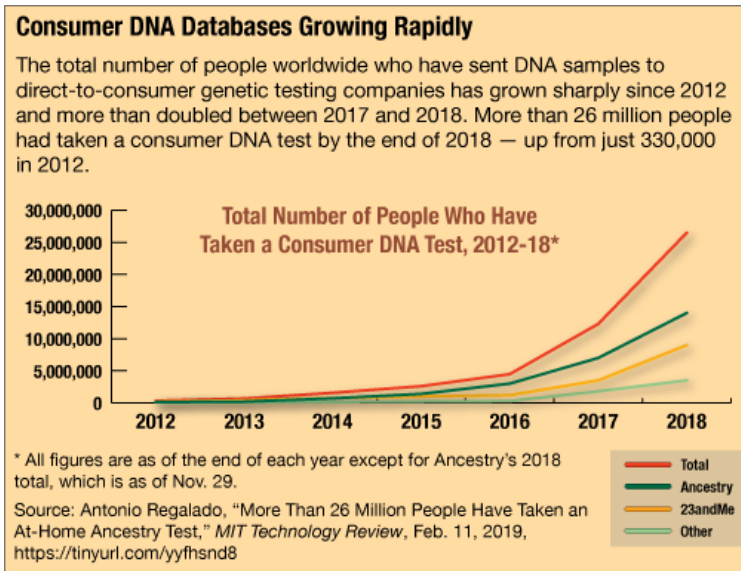
Lawson's report also contained a surprise. Her DNA showed no overlap with the man. It turned out Lawson was the product of a brief extramarital affair of her mother's. Julie and Fredda were half-sisters.



A reporter uses a swab to collect a saliva sample for genetic testing. While the tests can tell consumers about fun facts such as their sensitivity to sweet tastes, they can also reveal personal information such as the identities of birth parents or long-lost relatives. (Getty Images/Bloomberg/Cayce Clifford)

“We held each other,” Lawson said, “and we sobbed.” Because both parents are deceased, many questions remain unanswered. Yet the news allowed the women to forge relationships with newfound relatives.<sup>1</sup>

Since 2000, when FamilyTreeDNA of Houston first introduced direct-to-consumer genetic ancestry testing, the use of such services has exploded. “As many people purchased consumer DNA tests in 2018 as in all previous years combined,” according to the *MIT Technology Review*. That brought the total number of people worldwide whose DNA is in the industry's databases to more than 26 million, the publication said. At that pace, the total could reach 100 million people by the end of 2020, said biomedicine editor Antonio Regalado.<sup>2</sup>



### Long Description

The tests, which require consumers to spit or swab their cheeks and mail in the samples, are used for tracing ancestry, discovering relatives, finding out about amusing, genetically linked traits — such as earwax type or the likelihood of hating cilantro — and learning about one's genetic risk for certain diseases and health conditions, ranging from breast cancer to Parkinson's disease. Test kits range in price from \$59 for an ancestry test to more than \$1,000 for the most sophisticated health risk tests conducted by companies that market directly to consumers but require a doctor's order.

The industry's rapid growth has raised alarms among some legal experts, geneticists and bioethicists. They are concerned about false positives in the health data, the potential for confusion as consumers interpret health risk reports, insufficient government oversight of possible privacy violations and possible discrimination based on test results. Supporters, on the other hand, say consumers have a right to their genetic information and that the resulting data, stripped of personal identification, can help researchers find treatments and even cures for diseases.

"The main driver [of the rapid growth] is the popularity of genetic testing for ancestry discovery purposes," says Bruce Carlson, publisher of Kalorama Information, an Arlington, Va., health care market research company.

But people routinely misinterpret information about their genetic code, said Steven J. Heine, a psychology professor at the University of British Columbia and author of *DNA Is Not Destiny: The Remarkable, Completely Misunderstood Relationship Between You and Your Genes*. "We ... ascribe almost mystical powers to our genes," leading to a kind of genetic determinism, said Heine. <sup>3</sup>

For example, ancestry testing can foster a false sense of identity by "suggesting that key aspects of ... who we are, where we come from and where we belong in the world are rooted in our DNA," says Deborah Bolnick, an anthropological geneticist at the University of Connecticut. People's identities "really emerge from our lived experiences, the ways we're interacting with other people, the stories that we're being told from our families," says Bolnick, who adds she is particularly concerned about people who use genetic ancestry tests to claim Native American identity.

Geneticists say humans' DNA is mostly identical; only about 0.5 percent of a person's DNA is unique. Testing companies look for those differences. Some variants determine characteristics such as hair color or height. Others, often referred to as mutations, are strongly associated with diseases or health conditions.

The private genetic testing company Ancestry, located in Lehi, Utah, dominates the market, with the world's largest DNA database of any direct-to-consumer enterprise. It also has a trove of 20 billion digitized records used for traditional genealogy research, ranging from U.S. Census reports to ships' passenger lists. The company's DNA test surveys a person's genome at more than 700,000 locations on the 22 pairs of so-called autosomal chromosomes. It then delivers a report about a customer's ancestry, relatives and, for fun, traits such as sensitivity to sweet, savory and bitter tastes.<sup>4</sup>



Ancestry's website promotes the company's DNA tests, which cost as little as \$59 for a basic genealogy test. The direct-to-consumer DNA testing industry is growing rapidly, but critics are raising concerns that once the data is uploaded to a company's DNA database, consumers may not be able to control their private information. (AFP/Getty Images/Eric Baradat)

Besides Ancestry and FamilyTreeDNA, other large direct-to-consumer genetic testing companies include 23andMe in Mountain View, Calif., and MyHeritage in Israel. (CQ Researcher contacted the two largest companies for interviews: 23andMe did not respond and Ancestry declined to be interviewed.)

Tests can reveal information that treads on the privacy of others, such as the identities of birth parents or sperm donors who wish to remain anonymous. As Ancestry says in its privacy statement: "You may discover unexpected facts about yourself or your family when using our services. Once discoveries are made, we can't undo them."<sup>5</sup>

Sperm banks, in particular, face a quandary. For decades, donors have relied on sperm banks to protect their anonymity. But with direct-to-consumer genetic testing, sperm donors' anonymity will suffer "the same fate as the cassette tape," becoming a thing of the past, said Andrew Vorzimer, a reproductive law specialist in Woodland Hills, Calif.<sup>6</sup>

23andMe goes beyond its industry colleagues in its offerings. In April 2017, the company became the first, and so far only, direct-to-consumer genetic testing company authorized by the U.S. Food and Drug Administration (FDA) to market tests that, without a doctor's order, will provide genetic risk information for diseases or health conditions.<sup>7</sup> The FDA and the company stress that the tests are screening tools and should not be used for diagnosis. Positive results, they say, should be confirmed by a more comprehensive physician-ordered genetic test and medical evaluation.

23andMe's testing initially covered 10 conditions, including late-onset Alzheimer's, Parkinson's and celiac disease, which makes a person unable to digest gluten. Last year, the FDA approved a 23andMe test for

three genetic mutations, located on the BRCA1 and BRCA2 genes, known to significantly increase the risk of inherited breast and ovarian cancer. In January, the company gained approval to test for an inherited form of colorectal cancer.

But many geneticists question the health benefits of 23andMe's tests, especially for diseases for which there is no treatment. And because no doctor's order is required and no genetic counseling is offered with the results, consumers could be confused about how to decipher them, critics say.



Anne Wojcicki, CEO of the DNA testing company 23andMe, speaks during the TechCrunch Disrupt conference in September 2018 in San Francisco. Her company tests consumers' DNA for ancestry and for potential risks of developing about a dozen different diseases and health conditions. Because the tests can be ordered without a doctor's prescription, some geneticists question whether consumers can correctly understand the health data. (Getty Images/TechCrunch/Steve Jennings)

23andMe defends its tests. "We spent years proving to the FDA, through detailed analytical testing, that our Genetic Health Risk reports meet accuracy thresholds of 99 percent or higher," the company's CEO and co-founder Anne Wojcicki wrote to *The New York Times* in response to a critical editorial. "More equitable health care will come only if we can provide direct access — access without a medical professional barrier — in affordable ways."<sup>8</sup>

Direct-to-consumer genetic testing also raises privacy concerns, in part because DNA databases could be hacked. And a user could easily submit someone else's saliva sample and receive a full report of that person's ancestry and, if the testing company is 23andMe, health risks.

Moreover, consumer DNA testing companies are not bound by the federal Health Insurance Portability and Accountability Act (HIPAA), which requires clinical laboratories and health care providers to protect the privacy of health-related information, noted Thomas May, a bioethicist at the HudsonAlpha Institute for Biotechnology in Huntsville, Ala., which conducts genomics-based research. "Requiring HIPAA-style verification and privacy protection would cripple the [direct-to-consumer] testing industry," he said, "but some level of protection should be possible."<sup>9</sup>

Federal and state law is also silent on law enforcement's use of consumers' genetic information to help solve crimes, a practice that is spreading rapidly since California prosecutors used a DNA database last year to help catch a suspected serial killer who had eluded them for decades.

Privacy experts also raise the specter of some insurance companies using individuals' DNA information to discriminate when setting rates or determining coverage. The federal Genetic Information Nondiscrimination Act (GINA) of 2008 prohibits health insurers and employers from discriminating based on genetic information, but the act does not cover life, disability or long-term care insurers.<sup>10</sup>

At least 17 states have passed laws to fill that gap. California also extends protections “to prohibit genetic discrimination in emergency medical services, housing, mortgage lending, education and other state-funded programs,” according to the national law firm Carlton Fields.<sup>11</sup>

But in most states, insurers not covered by GINA or state laws “are going to increasingly, I think, ask you, ‘Have you taken any genetic tests?’ And if you say, ‘No,’ and you’ve taken one, now you have committed fraud,” says Mark Rothstein, director of the Institute for Bioethics, Health Policy and Law at the University of Louisville School of Medicine. “And if they could prove that, then your policy would be invalid.”

Non-health insurers have approached 23andMe for consumers' genetic information, Adriana Beach, corporate counsel for privacy, told a recent panel discussion on privacy. “We're not going to provide that information,” she said, adding that this policy is clearly stated on the company's website.

As the popularity of direct-to-consumer genetic testing continues to grow, here are some questions that bioethicists, legal experts, health care practitioners, geneticists and consumers are asking:

## Are direct-to-consumer genetic tests useful for assessing health risks?

It could not be easier to order a genetic health risk report from 23andMe. Click on the company website, pay \$149 — which also includes ancestry testing — and wait for the kit to arrive. Mail back your saliva sample, and three to five weeks later your results are ready to view online.

But many geneticists and other specialists say the health risk tests have no real value and can even threaten the health of consumers who do not understand their limitations. Allowing a direct-to-consumer company to assess health risks is “an unfortunate development that will likely cause considerable mischief,” said Dr. James Evans, a professor of genetics and medicine at the University of North Carolina, Chapel Hill.<sup>12</sup>

Other experts say 23andMe does an excellent job publicizing the uses and limitations of its tests, as the FDA requires, and it is up to consumers to educate themselves. “People have a right to their own genetic information, but with that right comes a responsibility,” said Dr. Robert Green, a medical geneticist at Harvard Medical School.<sup>13</sup>

One criticism involves the usefulness of some tests. For example, 23andMe tests for a variant in the APOE gene associated with late-onset Alzheimer's disease. But “many people with it will never get Alzheimer's,” says Michael Watson, executive director of the American College of Medical Genetics and Genomics, a professional membership organization in Bethesda, Md. And because no effective treatment exists, “I'm not sure how much utility [the test] may have,” he says.

### Questions to Ask Before Providing DNA

Before submitting DNA samples to a direct-to-consumer genetic testing company, consumers should study the company's privacy and research policies, usually available on its website, privacy experts say. Consumers should consider how comfortable they are with how such companies handle genetic information, which, unlike an email address or some other personal information, lasts for life. These are some of the questions experts recommend asking about DNA-testing companies:

- Will the company retain or destroy your DNA sample after analyzing it?
- Does the company anonymize and aggregate your genetic information before sharing it with third parties for research?
- Is your consent required before your genetic information, whether anonymized or identifiable, is used in research, and can you opt out of research at any time?
- Do you receive any compensation from allowing the company to use your data for research?
- Will the company inform you of changes in its privacy policy and allow you to remove your genetic information from its databases if you disagree with those changes?
- If the company is sold, can you remove all your data or any identifying information beforehand, in case the new owners have a different privacy policy?
- How does the company safeguard its genetic databases from security breaches?
- Would the company allow law enforcement agencies or life, disability or long-term care insurers to have access to its data?
- Does the company clearly explain the methodology used to determine ancestry and health risks?
- Does the company explain the limitations of its ancestry and health risk results?

— Barbara Mantel

### Long Description

Others warn that a negative test result may give users a false sense of assurance.

23andMe tests for three BRCA mutations, which occur mostly in the Ashkenazi Jewish population, but the company does not report on more than 1,000 other BRCA mutations associated with cancer risk. One study found that testing for only the three mutations would miss nearly 90 percent of people with BRCA mutations.

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“I worry that women who undertake testing from 23andMe could believe that they do not carry a mutation when in fact they do, and as a consequence could die of breast or ovarian cancers,” said Mary-Claire King, a University of Washington geneticist who helped discover the BRCA genes. 15

But Dr. Jeffrey Pollard, 23andMe's director of medical affairs, said the company focuses on the three mutations because they are among “the most well-studied [BRCA variants] and carry clear, documented risk for breast and ovarian cancer.” In addition, the company explains the test's limitations on its website and in results reports, he said. 16

Moreover, CEO Wojcicki said in her response to *The Times*, 23andMe's BRCA test flags people whose cancer risk might otherwise be missed. That's because some people with the three BRCA mutations either do not know their family medical history or have no family history of cancer, meaning a physician would have no reason to order genetic testing. 17

Critics also worry that, without the guidance of a genetic counselor or medical provider, customers may not understand that many factors besides heritable gene mutations help determine a person's overall likelihood of developing a disease or health condition.

Genetics is not always destiny, says Dr. Peter Hulick, medical director of the Neaman Center for Personalized Medicine at the NorthShore University HealthSystem in the Chicago suburbs. "It can be a strong component, but there are other factors that go into someone's overall risk."

Those include personal and family medical history, lifestyle, age, ethnicity and more, experts say. And many diseases, including 90 to 95 percent of cancers, are not the direct result of inherited genetic defects.<sup>18</sup>

Some experts also worry about false positive results based on raw DNA data. Customers seeking health information beyond 23andMe's authorized reports, for example, can download their raw DNA data and send it to a third-party website or app such as Promethease, Genetic Genie or LiveWello. These services also accept raw data from genetic ancestry sites such as Ancestry and FamilyTreeDNA. Their software combs through the scientific literature for information about health risks linked to the gene variants found in the raw data and issues a report. The services are not FDA regulated.

One study found that 40 percent of the genetic variants these services flagged were false positives that could not be confirmed by follow-up laboratory testing.<sup>19</sup>

"That's an exceedingly high number of individuals to stress out with a false positive result," said Catharine Wang, an associate professor of community health sciences at Boston University. The direct-to-consumer testing companies include disclaimers that their raw data have not been validated for accuracy, and they defend the practice of releasing the information to customers.<sup>20</sup>

Some testing companies that market and sell directly to consumers — such as Color Genomics in Burlingame, Calif., Genos in San Francisco and Veritas Genetics in Danvers, Mass. — analyze more of an individual's DNA than 23andMe, but their tests are more expensive and require a doctor's order. Because they are ordered by physicians, their tests do not need FDA approval. Such companies sell five times as many tests as 23andMe, according to Kalorama Information.<sup>21</sup>

Genos requires permission from a customer's doctor, while Color and Veritas allow customers to use the companies' independent network of physicians. That is a red flag for some observers. "There's a conflict," says Watson, who wonders whether physicians being paid by a laboratory, even if they are independent and not on staff, are incentivized to approve testing.

Color, which offers tests for 30 gene variants associated with breast, ovarian, uterine, colon, melanoma, pancreatic, stomach and prostate cancers, as well as 30 gene variants associated with heart disease, pays physicians a flat fee "that's not dependent on whether or not they approve testing or how many tests they approve," says Alicia Zhou, Color's vice president of research and scientific affairs.<sup>22</sup> "We do not want there to be a conflict of interest." Color also offers genetic counseling to customers.

Color, Genos, Veritas and 23andMe strongly recommend that customers discuss test results with their physicians. But Hulick, of the NorthShore University HealthSystem, says many customers never do, and "that's the biggest challenge for me." And even when customers share their results, he says, many primary care doctors are not prepared to interpret them. "They have limited training in genetics," he says.

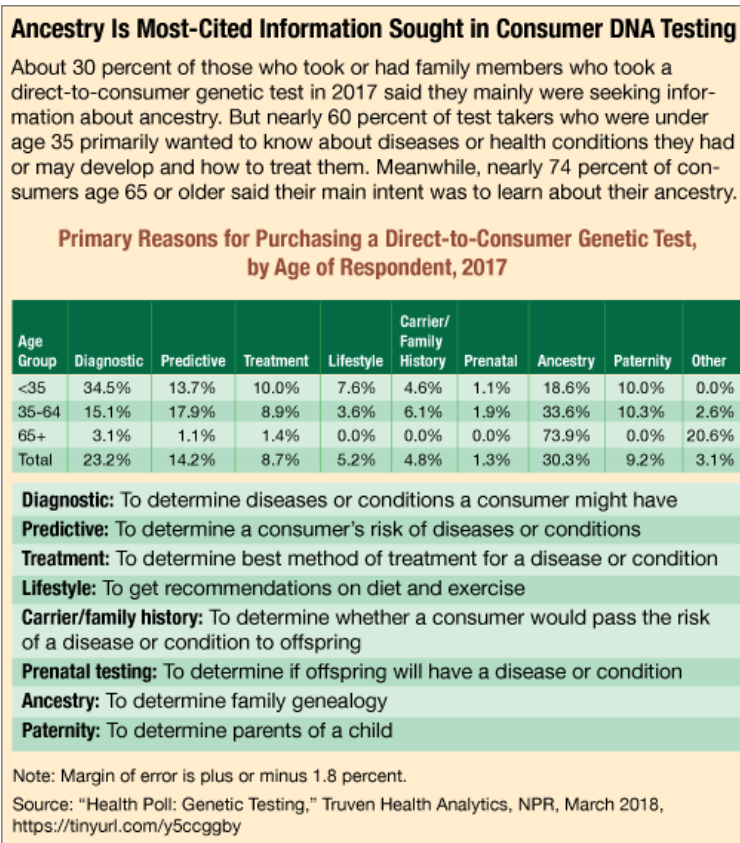
To address that lack, Hulick's department offers webinars on NorthShore's internal website, and he conducts frequent lectures for NorthShore specialists and primary care physicians.

Watson says his professional organization has begun to develop one-page documents for physicians that explain direct-to-consumer health risk tests.



## Do direct-to-consumer genetic tests provide accurate information about ancestry?

Millions of people have flocked to DNA testing websites that promise to “uncover your origins” (Ancestry), reveal “your unique heritage” (MyHeritage) or provide a breakdown of “your global ancestry by percentages” (23andMe). They may have shrugged if the results on the pie chart or map confirmed their own family tree research, or widened their eyes in surprise if the tests revealed something unknown.



### Long Description

But many geneticists and genealogists say consumers often do not understand the limits of what these tests can reveal.

“Most of the genealogists I know don't really take the results too seriously,” said Debbie Kennett, a British genealogist at the University College London, a public research institution.<sup>23</sup>

However, the companies do take their results seriously. “We're confident in the science and the results that we give to customers,” Ancestry said in a statement last year.<sup>24</sup>

Customers may believe their DNA is being compared to historical DNA, but that is not the case, said Adam Rutherford, a British geneticist and author of *A Brief History of Everyone Who Ever Lived: The Human Story Retold Through Our Genes*.

DNA ancestry tests are “not telling you where your DNA comes from in the past,” said Rutherford. “They're telling you where on Earth your DNA is from today.”<sup>25</sup> The testing companies compile reference databases

of DNA from customers the companies know live in and have roots in particular countries and regions. They then compare variants in a customer's DNA to variants in those databases. A customer is assigned some percentage of ancestry to a region or country depending on how much DNA he or she shares with current residents.

This may provide meaningful information going back four, five or even six generations, says Mark Thomas, a professor of evolutionary genetics at University College London, but not much beyond. That is because human history is full of migration and most people's ancestors did not stay put, he says.

Explanations of such limitations can be difficult to find on many company websites, although 23andMe displays a sample report with a chart mapping a customer's different geographic heritage to historical periods. In a statement last year, the company defended its reference databases, saying that it has “quality control mechanisms in place to ensure these data sets are sound.”<sup>26</sup>

Customers also may not realize their DNA contains genetic material from only a limited number of ancestors on their family tree. “An individual's DNA is inherited from recent ancestors in large random chunks, so the contribution of DNA from any particular ancestor can be nil after just a handful of generations,” British and Australian researchers wrote in a 2018 article in the scholarly journal *Genealogy*. “Therefore, each of us has inherited no DNA from the majority of our ancestors who lived just a few hundred years ago.”<sup>27</sup>

Nevertheless, Bolnick of the University of Connecticut says genetic ancestry testing can be valuable for people who lack much of a paper trail when trying to divine their family history. That includes members of African diaspora populations whose ancestors were brought to the Western Hemisphere in the trans-Atlantic slave trade. Often, “there were very deliberate efforts to erase their family histories and identities.” In such instances, genetic ancestry testing can provide some valuable insight into the past, she says.

The limitations of genetic ancestry testing are obvious to people who have sent DNA samples to several different companies and received conflicting results. That happens because each company looks at different genetic markers and uses its own reference databases and algorithms for analysis.

The algorithms are especially important, says Thomas. “Some [algorithms] are just nonsense, just scientifically unsound, some are quite explicit that they are telling you about more recent ancestry, some use the genetic information in really quite sophisticated ways, and others use it in very crude ways,” he says, without offering specifics.

In addition, customers can experience information whiplash as genetic testing companies continually expand their databases and work to improve their algorithms.

“I did a big, huge deep dive as soon as I saw the word ‘European Jewish,’” said Michelle St. George of Yakima, Wash., who got her DNA results back from Ancestry last August. She began to explore Jewish customs and culture, and the results “validated ... my love for people that are different from me, my need to be around different people. It validated who I was,” St. George told the *Detroit Free Press*.

But in September, Ancestry updated its methodology, and according to the revised results, St. George did not have Jewish ancestors.<sup>28</sup>

In a statement to the news outlet, Ancestry acknowledged that “the consumer genomics industry is in its early stages but is growing fast and we tell customers throughout the experience that their results are as accurate as possible for where the science is today, and that it may evolve over time as the resolution of DNA estimates improve.”

“For two months, I have done nothing but embracing what they're telling me my DNA was,” said St. George. But once she saw the new results, she stopped.<sup>29</sup>

## Is it ethical for law enforcement to use genetic genealogy to solve crimes?

Solving cold cases using genetic genealogy captured the public imagination in April 2018, after California prosecutors arrested Joseph DeAngelo, a suspected serial murderer known as the Golden State Killer who had escaped detection for more than 30 years. A retired police officer, DeAngelo is accused of scores of murders and rapes committed during the 1970s and '80s.

Sacramento County investigators linked the crimes to DeAngelo, age 72 when he was arrested, after they opened an account on GEDmatch, an open source genetic genealogy website that allows anyone to upload their genetic information from any of the private genetic testing companies, and uploaded crime scene DNA. GEDmatch's algorithm compared it to the DNA in its database and found a distant relative of DeAngelo. Law enforcement experts then painstakingly built a family tree and conducted more traditional detective work, allowing them to zero in on DeAngelo.<sup>30</sup>



Joseph James DeAngelo, allegedly the notorious Golden State Killer, appears in a Sacramento, Calif., courtroom in April 2018 during his arraignment. Investigators arrested DeAngelo after creating an account on GEDmatch and uploading crime scene DNA. The high-profile case has raised ethical questions about the use of genetic genealogy by law enforcement officials to help solve crimes. (Getty Images/The Sacramento Bee/Randy Pench)

Since then, law enforcement agencies around the country have resurrected long-dormant criminal investigations using the technique. Most are hiring the forensics consulting firm Parabon NanoLabs, based in Reston, Va., to do the genetic genealogy work and develop a suspect list. As of early May, Parabon said it had used the technology to help law enforcement agencies solve 55 cases in the past year.<sup>31</sup>

“The truth is, these cases wouldn't be solved if it weren't for this new, revolutionary [approach]. I mean, it's incredible,” says Sacramento County District Attorney Anne Marie Schubert.

A public opinion poll commissioned by the National Institutes of Health (NIH), a government health research agency in Bethesda, Md., found that the vast majority of respondents support solving violent crime using genetic genealogy databases containing everyday Americans' DNA information.<sup>32</sup> But some legal experts and genealogists are alarmed by the growing practice, based on concerns about privacy and potential misuse.

Natalie Ram, an assistant professor at the University of Baltimore School of Law, thinks the practice should be banned. “The real question is whether there is a public will to say privacy matters more than solving every possible crime,” says Ram.

In order to protect customers' privacy, Ancestry, 23andMe and most other large ancestry sites prohibit law enforcement agencies from opening accounts. (The exception is FamilyTreeDNA, which has allowed the FBI access to its database, causing a customer backlash.) (See [Current Situation](#).) But more than a million people have downloaded their raw DNA information from these and other ancestry sites and uploaded it to the 9-year-old GEDmatch website, which offers a broader search for ancestors and kin. Since the suspected Golden State Killer's arrest, GEDmatch has let users know that law enforcement uses its website.

Law enforcement has long compared DNA found at crime scenes with samples held in the FBI's Combined DNA Index System (CODIS), a collection of local, state and federal databases of DNA from convicted offenders and arrestees.<sup>33</sup> But GEDmatch includes the DNA of nonoffenders, and it contains more detailed genetic information for each individual: hundreds of thousands of genetic markers compared to 20 in CODIS.

“That gives us much more power to predict more distant relationships based on how much DNA any two people share,” says CeCe Moore, Parabon's chief genetic genealogist. Once relatives are identified, Moore combs through census, marriage, birth and death reports as well as newspaper archives and social media to create family trees and develop a list of suspects.

Law enforcement detectives then must build a case, including extracting a suspect's DNA from perhaps a discarded cigarette or coffee cup that can be compared to genetic material, such as blood or semen, left at a crime scene.

But critics say law enforcement's use of genetic genealogy may violate people's rights. They are particularly concerned about the privacy rights of relatives of people who upload genetic information to GEDmatch, saying those relatives had no say in the decision to share family DNA with the world and have no way to shield their own privacy.

“If I decide I don't want to be friends with you on Facebook anymore, I can unfriend you,” Ram says. “But I can't sever my genetic ties to my genetic relatives.”

Schubert says she “respectfully disagrees” with Ram. Forensic genetic genealogy narrows down suspects, allowing fewer innocent people to be investigated, says Schubert. “Isn't that good police work, as opposed to going down these rabbit holes that never produce anything?” she asks.

But Helen Wallace, director of GeneWatch UK, an online group in the United Kingdom that monitors developments in genetic technologies, is not persuaded. “What if a surveillance state misused such databases, not to track down criminals, but to identify political dissidents,” for example, by tracking them down through DNA left on coffee cups at a political meeting, asked Wallace.<sup>34</sup>

In recognition of the ethical and legal concerns, GEDmatch has said it allows law enforcement to use the website to investigate only homicides and sexual assaults. But New York University School of Law professor Erin Murphy said that is a slippery slope. “There's no [state or federal] rule saying police can only do this sort of genetic sleuthing if it's a homicide or rape,” said Murphy, who worries that police will begin using it for an array of lesser crimes.<sup>35</sup>

Last December, GEDmatch made an exception to its policy and allowed Utah detectives and Parabon to use the site to help find an assailant who had broken into a Mormon church and choked a 71-year-old woman playing the organ. She passed out but survived. In April, police arrested a 17-year-old suspect.

“This case was as close to a homicide as you can get,” said Curtis Rogers, the Florida retiree who runs GEDmatch, after a Utah newspaper revealed the company's role in the investigation in May. “The victim was reportedly in great fear that [the suspect] would return to end her life.”<sup>36</sup>

Almost immediately, there was a fierce backlash, and GEDmatch significantly curtailed the use of its website by law enforcement. From now on, the genetic information of a GEDmatch user will automatically be unsearchable by a law enforcement account unless a user elects to share it for that purpose.<sup>37</sup> At this point, it is unknown how many users will give their permission.

Meanwhile, Schubert's prosecutors have drafted a best-practices model that they share with law enforcement agencies nationwide. These practices include never making an arrest on a genealogy match alone, always obtaining a suspect's DNA sample and not using genetic genealogy data to investigate nonviolent crimes, says Schubert.

But Debbie Kennett of University College London said voluntary policies are not enough. She wants independent ethical committees to review law enforcement requests to upload crime scene DNA to genealogy websites.<sup>38</sup>

Ram prefers that states ban law enforcement's use of genetic genealogy websites for familial searches. Maryland and Washington, D.C., for instance, do not allow local law enforcement officials to search CODIS for familial matches over privacy concerns for family members, and Ram testified this year in favor of a Maryland bill that would have extended that prohibition to websites such as GEDmatch. The bill died in committee.



A Father Christmas doll holds a saliva collection kit for a 23andMe direct-to-consumer genetic test in a Washington, D.C., store in December 2018. Sales of the kits, which were a popular holiday item last year, have been booming. An analysis in the *MIT Technology Review* estimated that if the current sales growth rate continues, more than 100 million people will have their DNA data added to company databases by the end of 2020. (AFP/Getty Images/Eric Baradat)

In a few years, 90 percent of Americans of European descent, a demographic that dominates direct-to-consumer genetic testing databases, will be identifiable by name, based on the DNA data submitted to GEDmatch and further genealogy research, according to a study published in the journal *Science*. This includes people who never submitted saliva or cheek swabs for testing. A database such as GEDmatch needs to contain only 2 percent of a demographic for almost all related members of that demographic to be identifiable, the researchers said.<sup>39</sup>

## Background

### Early Advances in Genetics

On April 25, 1953, the journal *Nature* published a two-page letter from Cambridge University scientists James Watson and Francis Crick. “We wish to suggest a structure for the salt of deoxyribose nucleic acid (D.N.A.). This structure has novel features which are of considerable biologic interest,” they wrote. <sup>40</sup>

That brief and understated opening paragraph introduced “the most celebrated scientific discovery of the twentieth century,” said geneticist Kevin Davies. <sup>41</sup>

The *Nature* letter contained a simple diagram for the structure of DNA: the soon-to-be famous double helix, which looked like a twisting rope ladder. “The two ribbons symbolize the two phosphate-sugar chains, and the horizontal rods the pairs of bases holding the chains together,” the caption said. <sup>42</sup>

Swiss biochemist Johann Friedrich Miescher had discovered the DNA molecule in 1869. By the mid-20th century, scientists knew that the nucleus in cells contained chromosomes that contained genes, which determine heredity. Scientists also knew that chromosomes were composed of protein and DNA, which is passed from parent to offspring. They had a general idea of DNA's composition: four bases — adenine, cytosine, guanine and thymine — arranged around a phosphate-sugar chain. But they did not know DNA's exact structure. <sup>43</sup>

Watson's and Crick's discovery of that structure “immediately suggested that DNA — not a protein, as was widely imagined — was the master molecule that contains the genes,” according to the *Genome News Network*, an online magazine that covers developments in genomics research. In other words, genes are segments of DNA. <sup>44</sup>

At the end of their *Nature* letter, Watson and Crick briefly acknowledged the influence of the unpublished work of physicist Maurice Wilkins and his laboratory assistant, chemist Rosalind Franklin, at King's College London. Wilkins, without Franklin's permission, had shown Watson a photograph of DNA that Franklin had produced using a painstaking technique called x-ray crystallography. The photograph confirmed Watson's hunch that DNA was a helix — a smooth spiral — and provided some of the molecule's key dimensions. <sup>45</sup>



Cambridge University geneticist James Watson holds a model of a DNA molecule in Cambridge, Mass., in 1957. Four years earlier, he and colleague Francis Crick published a groundbreaking letter in *Nature* describing their pioneering work establishing the double-helix nature of DNA, which carries all genetic information. (Getty Images/The LIFE Picture Collection/Andreas Feininger)

In 1957, Crick gave a landmark address to the British Society of Experimental Biology, building on his work with Watson and the work of George Beadle and Edward Tatum, two Stanford University scientists. In 1941, Beadle and Tatum had discovered that the principal function of genes is to control the assembly of hundreds or thousands of amino acids into proteins, the complex molecules that are essential to the structure, function and regulation of the body's tissues and organs. But they could not explain the mechanism. In the speech, Crick hypothesized that a sequence of three DNA bases stands for one of 20 different amino acids. Each sequence within a gene instructs the cell to add another amino acid to a growing chain until that protein is complete.

By 1961, it was clear that Crick was correct. The next year, Watson, Crick and Wilkins shared the Nobel Prize in Physiology or Medicine. Franklin had died in 1958 from cancer, and the prize rules precluded a deceased individual from receiving the award. Over the years, Watson, who is now age 91, and Crick, who died in 2004, have been criticized for not giving more credit to Franklin's groundbreaking work.<sup>46</sup>

By 1966, scientists had determined the amino acid specified by each of the 64 possible three-base combinations, called codons. Most amino acids have more than one codon.<sup>47</sup>

Alterations in any of a gene's codons can have serious consequences. The first discovered disease-causing genetic mutation was for cystic fibrosis — a disease that primarily affects the lungs and digestive system — in 1989. In 1990, King, of the University of Washington, showed that breast cancer is inherited in some families, as the result of mutations in the gene she named BRCA1. In 1993 scientists in the United States and Venezuela discovered the gene mutation that causes Huntington's disease, which results in the death of brain cells.<sup>48</sup>

## Mapping the Human Genome

Finding one gene at a time worked well for diseases caused by a mutation, often inherited, of a single gene. But most common human diseases, including most cancers, arise from mutations of “multiple genes, spread diffusely throughout the human genome,” said Dr. Siddhartha Mukherjee, a cancer researcher and Pulitzer Prize-winning author.

“Gene by gene, a cell slouches toward cancer — acquiring one, two, four and then dozens of mutations that tip its physiology from controlled growth to dysregulated growth,” said Mukherjee. These mutations could be caused by environmental factors such as tobacco smoke, spontaneous errors during cell division or viruses. Some are inherited.<sup>49</sup>

This polygenetic nature of common diseases provided a major impetus for the quest to sequence all of human DNA, a task that took more than a decade to accomplish.

In 1990, the Human Genome Project formally began. It was coordinated by the NIH and U.S. Department of Energy, and conducted with research partners at universities in the United States, the United Kingdom, France, Germany, Japan and China. Its goal was monumental: to provide a complete and accurate sequence of the 3 billion base pairs that make up all human DNA and to identify the segments of DNA that comprise the estimated 20,000 to 25,000 human genes.<sup>50</sup>

“In April 2003, researchers announced that the Human Genome Project had completed a high-quality sequence of essentially the entire human genome.... It also identified the locations of many human genes and provided information about their structure and organization,” according to the NIH. The project then made the human genome sequence, along with tools to analyze the data, freely available on the internet.<sup>51</sup> Scientists hoped the results would allow them to more quickly identify gene variants linked to disease and develop genetic therapies.

The 2003 announcement came two years ahead of schedule, in part because of competition from the private sector. In 1998, a former NIH geneticist, Craig Venter, announced that he and a colleague were forming a company, later named Celera Genomics, to complete the human genome sequence by 2001. The company said it would use new sequencing machines and a faster sequencing technique than the one used by the Human Genome Project — although some scientists believed it was less accurate.

Faced with competition, the research centers associated with the government-led project also ordered the new machines. Sequencing the human genome became a race, and both public and private groups ultimately shared the credit when the final goal was reached.<sup>52</sup>

## Consumer Ancestry Testing

Meanwhile, laboratories had developed commercial genetic tests that physicians could order for patients whose family or personal medical histories indicated heightened risk for an inherited disease such as Huntington's.

Some entrepreneurs saw a business opportunity in allowing consumers to purchase genetic tests directly, without a doctor's order. By 2003, a few for-profit companies were advertising these direct-to-consumer genetic tests for health risks.<sup>53</sup>

In 2000, FamilyTreeDNA became the first company to market direct-to-consumer DNA testing for people seeking information about their family's genealogy. Ancestry had been founded in 1996 to provide historical documents, such as census reports and marriage documents, online. In 2007 it began offering DNA tests for ancestry.<sup>54</sup> Also that year, the new company 23andMe began offering consumers tests for both ancestry



and health risks, although the health tests were not as sophisticated as the clinical laboratory tests ordered by physicians.

About the same time, studies revealed public fears that insurers and employers might use genetic information to discriminate by, for example, denying insurance to individuals with a disease-related genetic mutation. As a result, consumers and physicians were reluctant to seek genetic tests.

In 2008, Congress passed GINA, prohibiting genetic discrimination in health insurance and employment. The 1996 HIPAA law barred group health insurance plans from charging discriminatory rates or denying coverage based on genetic information. GINA extended the prohibitions against genetic discrimination to individual health insurance plans and to employers. It also prohibited health insurers and employers from requiring or requesting genetic information or testing. But GINA does not cover life, disability or long-term care insurers, and several states have passed laws adding those protections.

By 2010, 30 companies around the world were marketing direct-to-consumer genetic tests for health risks, according to the Genetics and Public Policy Center at Johns Hopkins University. Besides 23andMe, those included deCODE in Reykjavik, Iceland; Navigenics in Foster City, Calif.; and Knome in Cambridge, Mass. <sup>55</sup> The types of tests varied widely and included testing for diseases linked to single gene mutations, such as Huntington's, as well as for susceptibility to disorders such as breast cancer, cardiovascular disease, depression, osteoporosis and Type 2 diabetes. <sup>56</sup>

However, only a small percentage of the U.S. population purchased the testing kits. In addition, scientists and physicians began questioning the accuracy and usefulness of direct-to-consumer genetic testing. <sup>57</sup> In 2010, the Government Accountability Office (GAO), which provides auditing, evaluation and investigative services for Congress, published a report called, "Direct-to-Consumer Genetic Tests: Misleading Test Results Are Further Complicated by Deceptive Marketing and Other Questionable Practices." <sup>58</sup>

GAO investigators created fictitious accounts and mailed DNA samples to four unnamed testing companies. The fictitious consumers received test results that were "misleading and of little or no practical use" and "DNA-based disease predictions that conflicted with their actual medical conditions," the agency said. In addition, the GAO found "10 egregious examples of deceptive marketing," including claims that the companies could use the test results "to create personalized supplements to cure diseases." <sup>59</sup>

Buffeted by criticism, low sales and falling prices, most direct-to-consumer genetic testing companies that offered medical information in the United States went out of business by 2012, and only 23andMe remained. However, DNA-based testing for ancestry was still commercially available. <sup>60</sup>

Until then, the FDA had not regulated direct-to-consumer genetic testing by companies offering health risk reports. But in November 2013 the agency changed course and sent a warning letter to 23andMe, instructing it to discontinue marketing of its personal genome service test until it received FDA clearance. 23andMe could continue to offer direct-to-consumer genetic testing for ancestry, the agency said, but not for health. <sup>61</sup>

Meanwhile, companies such as Color Genomics, founded in 2013, and Veritas Genetics, founded in 2014, entered the marketplace, selling genetic testing for medical conditions to consumers online but only with a physician's approval. Because of the physician oversight, their tests, which use different technology and examine much more of the human genome than 23andMe's tests, are not subject to FDA approval.

By 2017, 23andMe had submitted enough documentation to convince the FDA to allow it to offer direct-to-consumer test results for 10 diseases and health conditions. (Before the 2013 crackdown, the company had provided reports on hundreds of health conditions. <sup>62</sup>) They include late-onset Alzheimer's disease, Parkinson's, celiac disease and several rare conditions. In the past two years, the FDA has allowed the company to add reports for genes that indicate a risk for breast cancer and a form of colon cancer.

Yet, the controversy over such testing continues. Some health care practitioners and geneticists say the information is of little health benefit and could give consumers with a negative test result a false sense of security because the tests do not examine all possible genetic mutations associated with certain diseases. They also worry that consumers may not consult with their physicians and thus misunderstand their test results.

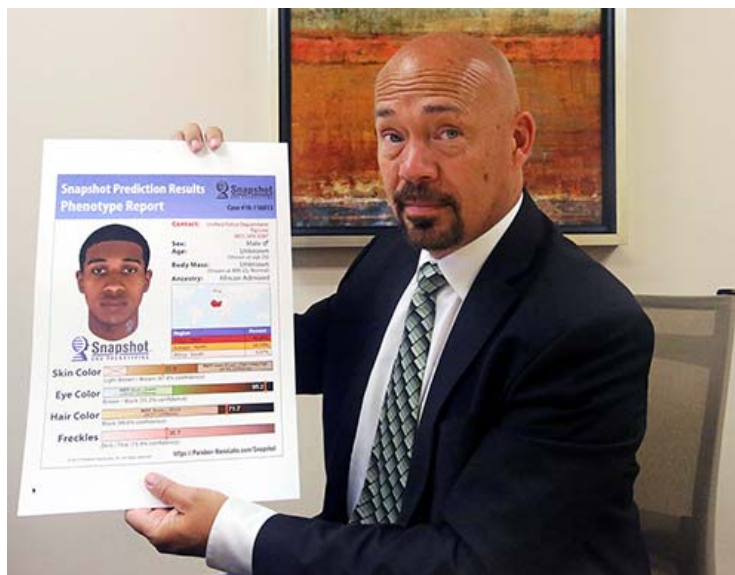
In addition, legal experts and bioethicists worry about privacy, as a growing number of consumers upload their genetic information to GEDmatch, the open access database increasingly used by law enforcement to help solve cold cases.

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## Current Situation

### FamilyTreeDNA and the FBI

This year has been one of controversy for FamilyTreeDNA. In late January, BuzzFeed News revealed that since December 2018, the company has allowed the FBI to create accounts and upload crime scene DNA in hopes of finding matches leading to suspects in unsolved rapes and murders. MyHeritage, Ancestry and 23andMe do not allow such accounts.



Private investigator Jason Jensen holds a phenotype report at his Salt Lake City office. Such reports use DNA to make predictions about hair and skin color, among other traits of criminal suspects. Some direct-to-consumer DNA companies have allowed law enforcement investigators to access their DNA databases to help solve criminal cases. (AP Photo/Rick Bowmer)

Many of FamilyTreeDNA's users were outraged because the company had never told them their data would be available for matching by law enforcement. "I feel they have violated my trust as a customer," said Leah Larkin, a genetic genealogist in Livermore, Calif. <sup>63</sup>

FamilyTreeDNA President Bennet Greenspan told BuzzFeed News, "We came to the conclusion that if law enforcement created accounts, with the same level of access to the database as the standard FamilyTreeDNA user, they would not be violating user privacy and confidentiality." <sup>64</sup>

But a few days later, Greenspan apologized to customers for not having revealed the agreement with the FBI. “I am genuinely sorry for not having handled our communications with you as we should have,” Greenspan told customers in an email. However, he defended the policy, saying, “We’ve received an incredible amount of support from those of you who believe this is an opportunity for honest, law-abiding citizens to help catch bad guys and bring closure to devastated families.”<sup>65</sup>

The FamilyTreeDNA episode highlights the lack of legal privacy protections in the United States for customers of genetic testing companies. “The main legal protections are [the companies’] terms of service and their privacy policies,” Robert I. Field, a professor of law and health management at Drexel University, said during a recent panel discussion on genetic testing and privacy.

In March, bowing to public criticism, FamilyTreeDNA allowed users to opt out of matching initiated by DNA accounts set up by law enforcement agencies.<sup>66</sup>

## State Legislation

As direct-to-consumer genetic testing grows in popularity, lawmakers in at least three states worry that federal and state laws do not adequately protect citizens’ privacy. For instance, life, disability or long-term care insurers could use genetic information to discriminate against applicants, and genetic testing companies are not properly notifying customers about sharing data with medical researchers, some legislators say.

In the past decade, at least 17 states have passed laws outlawing genetic discrimination by companies offering life, disability or long-term care insurance. And lawmakers in Connecticut, Florida and Illinois introduced measures this year to strengthen genetic privacy and protect against discrimination on the basis of genetic information, according to the National Conference of State Legislatures, which supports state legislatures with research and information.

“This is a huge step in the fight for Florida consumers and your right to genetic privacy,” Florida Chief Financial Officer Jimmy Patronis said in April, defending a Florida bill that would have required genetic testing companies to obtain written authorization from consumers before sharing or selling their results. “It is vital that DNA-testing companies allow ample opportunity for customers to protect their data.”<sup>67</sup>

The bill, which died in committee in May, also would have prevented life, disability and long-term care insurers from requiring genetic tests or considering genetic information when setting rates or denying coverage.<sup>68</sup>

The Connecticut and Illinois legislative sessions ended without a vote on those bills. The Connecticut measure would have prohibited consumer genetic testing companies from sharing “any personally identifiable genetic data or other personally identifiable information” about a person with any health carrier or life insurance company. The Illinois bill would have amended the state’s Genetic Information Privacy Act to include direct-to-consumer genetic testing in the definition of genetic testing.<sup>69</sup>

## New Diabetes Risk Score

23andMe is hoping to reach millions more customers with a new genetic health report it introduced in March, designed to assess an individual’s likelihood of developing Type 2 diabetes. But some experts are questioning its utility.

Type 2 diabetes is one of the most common chronic health conditions in the United States and the No. 1 cause of kidney failure, lower-limb amputations and adult blindness. More than 30 million U.S. adults have diabetes, almost all of them Type 2, according to the Centers for Disease Control and Prevention. An additional 84 million adults have prediabetes — blood sugar levels higher than normal — but 90 percent are

unaware of their condition. Left unaddressed by diet and lifestyle changes, prediabetes can become full blown diabetes.<sup>70</sup>

“Diabetes is a significant health issue in the United States that is expected to impact nearly half of the population,” said Wojcicki of 23andMe. “When customers learn about their genetic likelihood of developing Type 2 diabetes, we believe there is an opportunity to motivate them to change their lifestyle and ultimately to help them prevent the disease.”<sup>71</sup>

But Peter Kraft, a professor of epidemiology at Harvard University’s T.H. Chan School of Public Health, said, “It’s not clear that learning [about one’s] genetic risk helps people ‘get active,’ ‘eat healthy’ or stop smoking — the steps 23andMe recommends those at elevated risk take.”<sup>72</sup>

Others point out that traditional risk factors for diabetes are well known, including family history, lifestyle, age, weight and ethnicity. “I wonder if this test will be better for doctors than just seeing a patient’s family history or family tree,” said Dr. Aaron Neinstein, an endocrinologist at the University of California, San Francisco. “You can often tell a person’s risk just by seeing that.”<sup>73</sup> 23andMe encourages customers to share the results with their doctors, but the test is too new to know whether they do.

As with its 11 other “health predisposition reports,” 23andMe’s diabetes report notes its limitations. It “does not diagnose Type 2 diabetes or prediabetes and should not be used to make medical decisions,” according to the company website.

But the diabetes report differs from the company’s other health reports, which test for a few variants in just one or two genes. Type 2 diabetes and other common diseases, such as cardiovascular disease, are caused by a much larger number of gene variants. So 23andMe scans for more than 1,000 variants to produce an individual’s “polygenic risk score” for Type 2 diabetes and then adjusts it based on the person’s ethnicity and age.<sup>74</sup>

In addition, unlike its other genetic health tests, the diabetes test is not approved by the FDA. 23andMe said it does not need FDA approval because the diabetes test is a “wellness” product.<sup>75</sup>

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## Outlook

### Future Growth

The global market for direct-to-consumer genetic testing for disease risk is expected to grow by 25 percent or more a year over the next four years, according to Kalorama Information. The firm predicts that revenue from the sale of consumer test kits from 23andMe, which does not require a doctor’s order, and from the companies that require a physician’s permission, will rise from \$144 million this year to \$387 million in 2023.

“New technologies in testing will likely continue to fuel growth in combination with an aging population, increasing disease incidence and prevalence, a focus on prevention and early detection and new trends in personalized medicine,” the firm said in an April report.<sup>76</sup>

Hulick of the NorthShore University HealthSystem agrees that direct-to-consumer genetic testing for disease risk will continue to grow in popularity. “Many patients want to better understand their health,” he says.

But he would like to ensure that physicians are involved in “guiding patients through this journey,” so NorthShore has created a pilot program in partnership with the health technology company Color. Patients interested in their risk for certain diseases — regardless of their personal or family history — can get Color’s hereditary cancer and cardiovascular testing done with orders from NorthShore’s primary care physicians.

NorthShore geneticists and genetic counselors provide oversight and support, and specialists are available for follow-up if disease-associated mutations are detected.

Kalorama predicts that Ancestry, FamilyTreeDNA and My Heritage will also enter the burgeoning health market. [77](#)

Meanwhile, Thomas of University College London expects companies to improve how they market their ancestry testing, “including being more specific about when shared ancestors lived at particular locations,” he says. “But the cynic in me also anticipates more ‘genetic astrology,’ where claims are made about belonging to specific groups,” such as Native American communities.

Moore of Parabon NanoLabs says it is difficult to predict the future of forensic genetic genealogy because the technique has yet to be tested in the courts. “We’re still waiting on the precedent-setting decision from a judge that says, ‘Yes, this was appropriate. This was legal,’” she says. That could change in June, when the trial of William Earl Talbott II, accused of the 1987 murder of a young Canadian couple visiting Washington state, is scheduled to begin. Parabon did the genetic and genealogy analysis that led to Talbott’s arrest, and Moore is scheduled to testify at the trial.

If the Snohomish County Superior Court allows the technique, and states do not ban it, “the floodgates are going to open,” says Moore. “There are a lot of [law enforcement] agencies across the country that are waiting for that decision before they jump on board.”

Unlike Moore, Ram of the University of Baltimore School of Law would like states to ban the technique. But she is not hopeful.

Within five to 10 years, she predicts, “all or virtually all Americans will be identifiable, [and] the police will make increasing use of this technology,” using it to solve a wide variety of crimes. The allure of catching criminals and the influence of law enforcement agencies in state legislatures will outweigh privacy concerns, she predicts.

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## Pro/Con

Should women purchase direct-to-consumer genetic tests for breast cancer risk?

### Pro



#### Joel Eissenberg

Professor of Biochemistry and Molecular Biology, Saint Louis University. Written for *CQ Researcher*, June 2019

To answer this question, we need to unpack it a bit.

First, direct-to-consumer genetic tests assess breast cancer risk only in a limited sense. Currently, direct-to-consumer genomics companies offer tests for a few alleles (mutations) of the BRCA1 and BRCA2 genes known to elevate risk of breast and ovarian cancer. However, alleles of many other genes that are associated with increased cancer risk are not approved for direct-to-consumer testing. So women purchasing direct-to-consumer genetic tests for breast cancer risk may erroneously conclude they are free of elevated genetic risk because they receive advice based on a small subset of genetic

### Con



#### Fuki Hisama

Professor of Medical Genetics and Program Director of the Medical Genetics Residency Program, University of Washington. Written for *CQ Researcher*, June 2019

Millions of people have paid for direct-to-consumer (DTC) ancestry DNA testing or testing for ancestry, traits and the risk of developing specific health conditions. Unfortunately, I have seen many patients and physicians misunderstand the results of DTC testing. Last year, my colleagues and I published the story of a woman who had a strong family history of early onset breast cancer, yet her 23andMe DTC testing showed she had a “low risk” of breast cancer. Medical-grade genetic testing through our clinic showed the opposite: The woman has a mutation in the BRCA2 gene and is at significantly higher risk for breast and ovarian cancer.

risk factors. In addition, the cancer risk associated with most variants is currently unknown.

Another question is whether women should receive genetic test results for breast cancer risk absent any input from physicians and/or genetic counselors. At best, test results by themselves provide only a guide to action. Other than maintaining a healthy lifestyle — avoiding smoking and alcohol and maintaining a healthy body weight and diet — there is nothing a woman can do without consulting a physician to reduce her cancer risk if she learns she carries an increased genetic risk. To obtain a prophylactic mastectomy or oophorectomy (surgery to remove one or both ovaries) requires a physician.

So how is genomics testing information without input from a doctor or genetic counselor different from the other direct-to-consumer health tests that we take for granted? It isn't, really. Nobody asks whether women should be purchasing direct-to-consumer tests for fever (thermometers), high blood pressure (sphygmomanometers), obesity (bathroom scales) or pregnancy. Is a genetic test for breast cancer risk really different from these widely accepted tests that uncover health risks? To use knowledge of body temperature, blood pressure, weight or pregnancy status as a guide to action, women need to be informed about what this information means to them and seek physician care when appropriate.

Published research suggests that no lasting harm comes from giving people access to their genomics data. Accordingly, there is no justification for medical paternalism on the question of direct-to-consumer genomics testing. Knowledge is power, and in an open society, maximizing autonomy is a virtue. Of course, with freedom comes responsibility. In the example of direct-to-consumer testing for breast cancer risk, the responsibility properly falls on the consumer to be informed about what the test can and cannot say about risk, and to seek current and authoritative information.

The reason for the difference: Our testing detects thousands of variants in the BRCA1 and BRCA2 genes. The DTC test detects just three known mutations prevalent in the Ashkenazi Jewish population but rare to nonexistent in women from other ethnic backgrounds.

In April, *The New York Times* reported that 23andMe DTC testing misses nearly 90 percent of mutations for breast cancer. By contrast, genetic professionals do not merely test people. We also understand the limitations of such testing, and we explain to women with a genetic predisposition for cancer what they can do to lower their risks.

We referred the woman with the BRCA2 gene mutation to local, knowledgeable specialists. We also told her that even if her test results had been negative, she is at greater-than-average risk for breast cancer because of her family history, and we recommended high-risk breast cancer screening. DTC testing does not do any of that.

Some would argue that there are too few genetic physicians (about 1,000) and counselors (about 4,000) for the U.S. population, and that DTC testing makes genetics available to more people. However, genetic testing is not like a home pregnancy test, with high clinical validity and reliability for positive and negative results. It is nuanced and complex. A positive genetic test does not mean a woman has or will ever develop a disease, and a negative result does not mean she will not.

Genetic testing is best interpreted in the context of the patient's medical and family history. There are too few genetic professionals, but the solution is to provide more information about careers in genetics to students contemplating a health professions career — and to provide options for loan repayment and other financial support for medical students and genetic counseling students to increase the number of qualified genetic professionals.

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## Chronology

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**1940s–1970s** Scientists unravel the mysteries of DNA.

**1941**

Stanford University scientists George Beadle and Edward Tatum discover that each gene, which is a segment of the DNA found in most cells of all living things, tells cells how to manufacture a protein; proteins are required for the structure, function and regulation of the body's tissues and organs.

<b>1953</b>	Cambridge University scientists James Watson and Francis Crick publish their proposed double-helix structure for DNA in the journal <i>Nature</i> ; DNA looks like a twisting ladder, with each side made up of sugars and phosphate groups and the rungs formed by pairs of nitrogenous bases.
<b>1957</b>	Crick suggests that genes instruct cells to build proteins: An arrangement of three nitrogenous bases codes for each of 20 amino acids, the organic compounds that form proteins. His theory becomes the basis for DNA sequencing and modern biomedical research.
<b>1962</b>	Watson, Crick and physicist Maurice Wilkins share the Nobel Prize in Physiology or Medicine for their discovery of DNA's structure.
<b>1977</b>	American scientist Walter Gilbert and British scientist Frederick Sanger develop techniques for sequencing the order of the bases in DNA; DNA sequencing allows researchers to identify the location of genes, their variants and their associations with diseases.
<b>1980s–1990s</b>	The Human Genome Project to sequence the human genome gets underway.
<b>1986</b>	Discussions begin on how to sequence the human genome — the entire complement of human DNA.
<b>1989</b>	Scientists discover the genetic mutation that causes cystic fibrosis.
<b>1990</b>	The National Institutes of Health (NIH) and the Department of Energy begin coordinating researchers around the globe who help to decode the human genome.
<b>1993</b>	Geneticists discover the genetic mutation that causes Huntington's disease.
<b>1994</b>	Researchers identify BRCA1, the most common gene associated with breast cancer.
<b>1996</b>	Ancestry Publishing establishes <a href="http://Ancestry.com">Ancestry.com</a> , offering customers genealogy research using digitized historical documents.... The Health Insurance Portability and Accountability Act prohibits group — but not individual — health insurance plans from setting rates or denying coverage based on genetic information.
<b>1998</b>	Craig Venter, a former NIH geneticist, forms a company to compete with the Human Genome Project.
<b>2000 –Present</b>	Scientists map the entire human genome; private companies market direct-to-consumer genetic tests for ancestry and health risks.
<b>2000</b>	FamilyTreeDNA markets direct-to-consumer genetic testing for ancestry.
<b>2003</b>	The Human Genome Project completes a high-quality sequence of the human genome.
<b>2007</b>	Ancestry begins offering direct-to-consumer genetic testing for ancestry.... 23andMe offers consumers genetic testing for ancestry and health risks, without a doctor's order.
<b>2008</b>	The Genetic Information Nondiscrimination Act prohibits genetic discrimination by health insurers and employers.
<b>2010</b>	Thirty companies are marketing direct-to-consumer genetic tests for health risks, including 23andMe, deCODE in Reykjavík, Iceland, Navigenics in Foster City, Calif., and Knome in Cambridge, Mass.... A Government Accountability Office investigation of the industry finds deceptive marketing and misleading test results.
<b>2012</b>	Most direct-to-consumer genetic testing companies offering medical information in the United States, except for 23andMe, have gone out of business as a result of poor sales and public criticism.

2013

The Food and Drug Administration (FDA) instructs 23andMe to stop marketing its direct-to-consumer genetic tests for health risks.... Color Genomics is founded to market genetic testing for health risks directly to consumers but requires a doctor's order for such tests.

2017

After 23andMe submitted accuracy studies, the FDA approves the company's direct-to-consumer genetic tests for 10 health conditions, including Parkinson's disease.

2018

California prosecutors arrest Joseph DeAngelo, a retired police officer accused of rapes and murders in the 1970s and '80s; prosecutors matched crime scene DNA to that of a relative who had submitted their DNA to an open access genetic database.

2019

BuzzFeed News reveals that FamilyTreeDNA has been allowing the FBI to use its database to solve crimes without notifying customers (January).... Lawmakers introduce a bill to strengthen genetic privacy in Connecticut (January) and in Illinois (February).... 23andMe offers a direct-to-consumer test for Type 2 diabetes risk (March).

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## Short Features

### Ancestry Testing Raises Native American Identity Issues

*"Not a single tribe uses commercial ancestry testing to determine membership."*

For years, President Trump has taunted Sen. Elizabeth Warren, D-Mass., a former Harvard Law School professor, for listing herself in a law schools directory as a minority, based on family lore that she and her relatives in Norman, Okla., were of Native American ancestry.



A 2018 DNA test suggested that Sen. Elizabeth Warren, D-Mass., has a distant indigenous ancestor, although she has recently backed away from her earlier claims of Native American heritage. The use of such tests by people claiming to be descendants of Native Americans raises concerns of cultural appropriation. None of the 573 federally recognized tribes use commercial ancestry tests to establish tribal membership. (Getty Images/Bloomberg/Stefani Reynolds)



Last fall, Warren accepted Trump's challenge to take a genetic ancestry test. The results suggested she had an indigenous ancestor six to 10 generations ago.

Such testing cannot link ancestry to a particular tribe, says Deborah Bolnick, an anthropological geneticist at the University of Connecticut. "DNA markers just aren't localized to specific tribes," she says. "They're shared across many different communities because people don't just live and mate and stay within a single community over time."

Although Warren did not claim to belong to a particular tribe and did not identify herself as Native American when she released the findings in a five-minute video last October, the announcement stirred up an ongoing discussion of what it means to be Native American and the role of DNA in determining identity.<sup>1</sup>

The Cherokee Nation, based in Oklahoma, felt compelled to speak out.

"Using a DNA test to lay claim to any connection to the Cherokee Nation or any tribal nation, even vaguely, is inappropriate and wrong," Cherokee Nation Secretary of State Chuck Hoskin Jr. said in a statement. "Senator Warren is undermining tribal interests with her continued claims of tribal heritage."<sup>2</sup>

Showing Native American ancestry through a genetic test is "not the same as showing definitely that someone has the right to claim to be Native American as an identity," said Kim TallBear, a member of the Sisseton-Wahpeton Oyate tribe of South Dakota and author of *Native American DNA: Tribal Belonging and the False Promise of Genetic Science*. "There are very specific tribal enrollment rules from tribe to tribe. It's pretty complicated."<sup>3</sup>

Each of the 573 federally recognized tribes in the United States establishes what determines identity as a member of their community.<sup>4</sup>

"In our conversations with tribal communities in various parts of the United States, they're very clear that identity has many components," says Sara Chandros Hull, a bioethicist at the National Genome Research Institute at the National Institutes of Health (NIH), a government research organization in Bethesda, Md. "It has to do with a shared historical experience; it has to do with one's family and how one was raised; it has to do with one's values and religious beliefs. To my knowledge, there is not a single tribe that currently uses genetic testing of the sort we are talking about — commercial ancestry testing — in that determination."

Hull says she would encourage people who believe they may have Native American ancestry "to immerse themselves more deeply in the stories of Native Americans and the experiences that they lived, and live, in this country today." She would not recommend taking a direct-to-consumer genetic ancestry test.

Hull and a group of NIH colleagues examined the marketing of 25 genetic testing companies that offer customers a chance to see if they have Native American ancestry. The scientists, who published their findings in the journal *Genetics in Medicine* in December, found that many marketing campaigns conflate ancestry and identity.

For example, they wrote, an Ancestry advertisement on YouTube "depicts a customer describing her surprise at discovering a quarter percentage of Native American ancestry, talking among seemingly Native artifacts intended to reflect the customer's newfound heritage. Depictions and messages such as these oversimplify both the richer concept of what it means to be Native American and a tribe's sovereign power to establish what comprises such an identity."<sup>5</sup> Ancestry declined to be interviewed for this story.

In fact, genetic ancestry testing may threaten tribes' power to determine Native American identity, says Hull. With test results in hand, "people are claiming that they are Native American for purposes of gaining access to small-business grants for minorities or individual scholarships," she says. "It may reduce legitimate access to programs that are intended to help improve the socioeconomic status of tribal communities."

Robert Taylor, owner of an insurance company in Washington state, tried to apply for a state Disadvantaged Business Enterprise program based on genetic ancestry testing that said he was 6 percent Native American and 4 percent sub-Saharan African. After he was rejected, he sued the state. In December, the U.S. District Court for the Western District of Washington rejected Taylor's assertion that state officials had acted in "an arbitrary and capricious manner" in questioning his minority status.<sup>6</sup>

— **Barbara Mantel**

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## Consumer Genetic Testing Companies Use DNA Data for Research

*They make more money mining genetic databases than selling DNA kits.*

Scientists and outside partners of direct-to-consumer genetic testing companies are using the companies' ever-expanding DNA databases to study connections between genetic mutations and disease.

Consumers must provide their consent before their DNA can be used in such research, and their information is aggregated and anonymized before scientists comb through it. "More than 80 percent of our customers choose to participate in research at 23andMe," Adriana Beach, the company's corporate counsel for privacy, told an April journalism conference.

Lila D. Lecy gave her consent. The former flight attendant from South Carolina has Parkinson's disease, and 23andMe has used her genetic test results in at least 33 Parkinson's research studies. "I thought if I could help anybody avoid what I had to go through, I would be more than happy to help," said Lecy.<sup>7</sup>

Since late 2017, the Michael J. Fox Foundation for Parkinson's Research, an organization in New York City that seeks to find a cure, has provided an undisclosed amount of funding for 23andMe researchers to gain new genetic insights into the disease, using data provided since 2009 by 23andMe customers with Parkinson's.<sup>8</sup>

Groups teaming up with direct-to-consumer genetic testing companies on such research include nonprofits, university research departments, government scientists and pharmaceutical and biotechnology companies. Although the largest testing firms are privately held and do not disclose their financial information, analysts say the companies can make more money from mining their genetic databases for research than from selling kits to consumers.<sup>9</sup>

A 23andMe official suggested in 2013 that mining the databases had been the company's primary business model all along. "The long game here is not to make money selling kits, although the kits are essential to get the base level data," Patrick Chung, a 23andMe board member, said in 2013. "Once you have the data, [the company] does actually become the Google of personalized health care."<sup>10</sup>

23andMe collaborates with, among other for-profit firms, Pfizer, a pharmaceutical conglomerate in New York City; Biogen, a biotechnology company based in Cambridge, Mass.; and biotechnology company Genentech, in San Francisco.<sup>11</sup>

Last year, GlaxoSmithKline, a London-based pharmaceutical company, announced a four-year collaboration with 23andMe to use the company's "rich database and proprietary statistical analytics." The two companies will "share in the proceeds from new treatments and medicines arising from the collaboration." According to the research consent form 23andMe customers sign, none of those proceeds will go to those who contribute their DNA to the databases.<sup>12</sup> The pharmaceutical giant also invested \$300 million for an equity stake in 23andMe.<sup>13</sup>

After the GlaxoSmithKline announcement, some media commentators expressed concern that the company would have direct access to and control over 23andMe's genetic databases.<sup>14</sup> This is a "big misconception" said Beach at the journalism conference. Scientists who collaborate with 23andMe see only the results of analyses conducted by company scientists, she said.

According to its website, Ancestry has fewer collaborations with outside groups. One was a three-year contract for an undisclosed amount with the Google-funded biotechnology company Calico Life Sciences, which is researching the biology of the human lifespan. Its researchers were interested in the anonymized DNA of long-lived Ancestry customers. The collaboration ended last year, and so far, no research results have been made public.<sup>15</sup>

Some legal experts and bioethicists are concerned about these research collaborations. They note that consumers who agree to share anonymized DNA with researchers have no say over the kind of studies being conducted. "The researchers may engage in research that you really don't approve of, and they're not asking for your further permission," says Mark Rothstein, director of the Institute for Bioethics, Health Policy and Law at the University of Louisville School of Medicine.

Or researchers may engage in research that consumers do not understand. "We learn more about genetics every day," said George Annas, a legal scholar and bioethicist at Boston University who studies informed consent. "And so your consent is going to be more complicated tomorrow than it is today, just because there's more known about genetics."<sup>16</sup>

The direct-to-consumer genetic testing companies allow customers to opt out of research participation at any time, but the customers' DNA would continue to be used in ongoing studies.

Arthur Caplan, professor of bioethics at New York University's Langone Medical Center, points out another problem with using the databases for research: Data collected by the companies are not representative of the U.S. population. "Right now, the direct-to-consumer databases are heavily skewed toward people who can pay, people who have TVs or the internet" or healthy people who have the leisure to worry about their genetic health risks, says Caplan. "So far, 23andMe, for example, is mainly white upper- and middle-class people."

As a result, any disease-related genetic variants found through the databases will not necessarily be applicable to other populations, such as Korean Americans, Native Americans or African Americans, he says.

A better option for anyone interested in participating in genetic research, Caplan says, is the All of Us Research Program, initiated by the National Institutes of Health, a government research organization in

Bethesda, Md., in 2016. It aims to build an anonymized database of genetic information from at least 1 million participants, reflecting “the rich diversity of the U.S.” The data will be available for research under strict privacy protocols.

Partners include universities, health care systems, medical schools, research organizations and pharmaceutical companies. Individuals can contribute their DNA through clinical laboratories and drugstores.

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“Diversity is a hallmark of this effort,” said Eric Dishman, director of the All of Us Research Program. “We strive for diversity of people and also diversity of data types, so researchers can understand the many factors that influence health and health outcomes for each of us.” 18

— **Barbara Mantel**

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## The Next Step

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## Contacts

### 23andMe

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A direct-to-consumer genetic testing company that provides information about genetically linked traits, health risks and ancestry and conducts medical research using its DNA database in partnership with nonprofits and pharmaceutical companies.

### American College of Medical Genetics and Genomics

7101 Wisconsin Ave., Suite 1101, Bethesda, MD 20814

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Membership organization representing clinical geneticists, clinical laboratory geneticists and genetic counselors.

### American Society of Human Genetics

6120 Executive Blvd., Suite 500, Rockville, MD 20852

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[www.ashg.org](http://www.ashg.org)

Membership organization for human genetic specialists.

### **Ancestry**

1300 W. Traverse Parkway, Lehi, UT 84043

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A genealogy research and genetic testing company that maintains the world's largest genetic database of any direct-to-consumer genetic testing enterprise.

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Federal department that examines ethical questions related to biomedical research, including the ethics of genetics research.

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Publisher of market research on health care, medical devices and biotechnology, including genetic testing.

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Federal agency that conducts and funds genomic research and studies the impact of genomics on society.

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Company that provides genetic genealogy services to law enforcement officials to solve criminal cases.

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## Document APA Citation

Mantel, B. (2019, June 14). Consumer genetic testing. *CQ researcher*, 29, 1-54. Retrieved from <http://library.cqpress.com/>

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