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How Much Can You Learn From a Home DNA Test?

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One reporter has her DNA analyzed and finds that genetic testing isn't an exact science.

by Boonsri Dickinson; photography by Rudy Archuleta



This story is as intimate as I will get with you. In it, I unzip my genes and give you a sneak peek at my biological destiny. I took three DNA tests from three different DNA testing companies—one from a firm called [Navigenics](#), one from [23andMe](#), and one from [deCODE](#) genetics.

I was so hyped up about getting my DNA analyzed, I didn't really think about the consequences of having all that information at my fingertips. Perhaps that's typical of a 25-year-old. I'm working at my first job out of college and living in New York City. There's plenty in the here and now to worry about: The homeless man deliberately running into me on my way to work. Living on a shoestring budget. Finding Mr. Right.

Some of the test results from the three companies matched up with one another.

Some didn't.

But I'm getting ahead of myself. Let me tell you what these tests actually test. Our DNA is made up of 3 billion base pairs, with myriad possible sequences of the four chemicals (represented as A, T, G, and C) that form the "instruction book" for a human being, geneticist Francis Collins explains. Sometimes a mistake gets introduced into the DNA sequence. Collins calls these mutations "misspellings"; when a large number of people share the same mutation, they're known as [single nucleotide polymorphisms](#) (SNPs, pronounced "snips"). DNA testing companies use the SNPs to calculate people's genetic risk of developing complex diseases. Broadly speaking, Navigenics, 23andMe, and deCODE genetics each say that they interpret genetic research gathered from scientific papers, apply it to a customer's DNA sequence, and deliver the results online. The companies expect that most people will be interested in buying the test, everyone from the average joe to a college student to a politician to me—a half-Asian, half-European girl from Florida.

I'm at risk of developing: sausage-like swelling, ?morning stiffness, inability to identify people's faces, fat rolls on my stomach, and bloody stools.

Navigenics

I initially heard about Navigenics at a charity event when I spoke to [William A. Haseltine](#), founder and former chairman and CEO of Human Genome Sciences, about genetic testing. The next day, I called Navigenics to ask if I could get tested for free. Normally the cost is \$2,500. Product manager Christine Lin said that I could, but only if I could turn around my spit sample quickly. The company was beta testing, and I could be part of the first group.

When the DNA kit arrived in a FedEx package November 11, 2007, I felt a rush. The kit included instructions for how to collect my spit. I harvested the necessary amount of saliva in just five minutes.

But before mailing the spit, I filled out a questionnaire. I penned in basic information like my birthday, address, and details about my ancestry. Interestingly, age and race do not get factored into the calculations—the formula assumes I'm an American of European descent (well, at least half of me is). The fine print that came with the kit stated that I wasn't going to get medical advice from them and sought permission to use my sample for (gasp) scientific research. Samples are sent directly back to Navigenics' outsourced

lab, [Affymetrix](#) in West Sacramento, California. Normally it would take three to four weeks to get your DNA results online. I had to wait two months to get mine because Navigenics wanted to give me the results in person when I visited their headquarters in Redwood Shores, California.

Last January I escaped the bitter bite of the New York winter and flew to California to visit Navigenics and 23andMe. When I arrived at Navigenics' waterfront office, I was jet-lagged and anxious. Regular customers have the option of calling a toll-free number to talk to a genetic counselor, but I got to speak to one in person (the benefits of being a reporter). I expected the counseling session to be therapeutic—like a scene out of [The Sopranos](#), when Tony goes to his shrink to talk about his anxieties. My genetic counseling session wasn't like that at all.

Instead, I sat in an office chair with one of Navigenics' in-house genetic counselors, Elissa Levin, ready to look at my results for the first time. I didn't want this test to change my life. I just wanted some insight. My eyes glazed over while examining the results on a computer screen: I might go blind (macular degeneration). My joints might ache (osteoarthritis). I might put on weight (obesity). I might have dry skin (psoriasis). I might have diarrhea and constipation issues ([Crohn's disease](#)). I took a deep breath.

Oh, no, I thought.

I have Crohn's disease.

I HAVE Crohn's disease?

I have Crohn's disease....

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Out of the list of potential problems, I focused on the one illness I had a symptom of. I used to have a sensitive stomach, but I started avoiding foods with wheat and dairy two years ago. Since then, the recurring episodes of bloating and irritable bowel problems have virtually disappeared.

"There's such a spectrum of gastrointestinal disease," Levin said. "A lot of people go a long time being batted around from specialist to specialist and get a whole battery of tests." The DNA test might at least point me in the right direction, she added.

We discussed what little I knew about my family history, addressed concerns I had, and talked about the possibility of following up with my doctor. I knew I wouldn't go home and ask my doctor for a colonoscopy, especially since my intestinal problem subsided years ago.

Next I spoke to the Navigenics CEO, Mari Baker. She, of course, had taken the test and was lucky, in the sense that hers had led to an actual diagnosis. "There might be a low lifetime risk of developing a disease, but if I'm the one genetically loaded for it, maybe I'm the person who is going to get it," Baker said. Her risk for [celiac disease](#) was higher than in the average population, so she asked her doctor for follow-up testing, which showed she did have the illness. Now she cuts gluten out of her diet to prevent uncomfortable symptoms.

Baker's diagnosis was just one example of how Navigenics can empower people. The company is the brainchild of a doctor and a geneticist. [David Agus](#), the research director of the Louis Warschaw Prostate Cancer Center and director of the Spielberg Family Center for Applied Proteomics, both at Cedars-Sinai Medical Center in Los Angeles, spent most of his time with end-of-life cancer patients. He told a friend how he wished there were a better system in place to screen people for genetic risks, so they could prevent cancer. That friend introduced him to Dietrich Stephan, now the chief science officer of Navigenics, who had been studying the genetics of disorders such as autism and exercise-induced heart attacks at the [Translational Genomics Research Institute](#) in Arizona. After a few phone conversations, Stephan and Agus raised enough capital to create Navigenics.

Now I had some information about my genetic profile, but there were still two other tests to consider. I wanted to see if everything matched up.

In the meantime, I got a little depressed when I printed out a 236-page document—a book of Boonsri—and read about some of the symptoms of the diseases I'm at risk of developing: sausalike swelling, morning stiffness, inability to identify people's faces, fat rolls on my stomach, and bloody stools.

I felt helpless. I didn't know what action to take or if I should be taking any at all. I followed up with a second Navigenics genetic counselor, Shannon Keiran. We focused on the diseases I'm at risk for and talked about what I should do to prevent them. Watch out for obesity, she said. That's easy, I thought. I run every day. While my risk for Crohn's was higher than most, my chance of getting it was still less than 1 percent (I'd say those are pretty good odds). When it comes to [macular degeneration](#), genes play a big part. Wearing sunglasses may help, Keiran said. For osteoarthritis, make sure you don't wear high heel shoes because it places strain on your knees, she said. I asked about alcohol consumption. Psoriasis may be affected by alcohol. And of course, she added, "moderation is key."



Would the next two tests assuage my fears—and end my occasional bouts of hypochondria?

23andMe

DISCOVER paid 23andMe \$999 for a DNA test (no reporter's comp this time). I rushed through the fine print: "If you are asked by an insurance company whether you have learned genetic information about health conditions and you do not disclose this to them, this may be considered to be fraud.... You may learn information about yourself that you do not anticipate. This information may evoke strong emotions and has the potential to alter your life and worldview. You may discover things...e.g., your father is not genetically your father."

No problem.

The next day, on December 12, 2007, a second FedEx box arrived at my office. I opened the bright green package at my desk and prepared for spitting episode number two. The [DNA test kit](#) included a vial and instructions. It was easier the second time. I mailed the sample to 23andMe's contracted lab, Illumina in San Diego. (They use a different lab now.) Four weeks later, in January, I signed on to my 23andMe online account to "unlock the secrets of [my] own DNA."

23andMe, like Navigenics, took my markers and calculated my genetic risks for certain diseases. I began surfing my genome, all 580,442 SNPs they had sequenced. In addition to telling me my risk for certain diseases, 23andMe provided ancestry information, social networking functions, and more (details below). Of the 70 different conditions to choose from, let me start with the silly ones. I have dry earwax (true). Apparently my sense of smell stinks (true) thanks to a genotype of TT on my 14th chromosome. One of the most fascinating things I learned is that my blood type is made up of more than 25 blood groups. There's even information available regarding histocompatibility. (I can see if a potential date is compatible with me immunologically.)

I checked my risk for incurable diseases like [lupus](#). I had access to my relative risks of psychiatric diseases such as bipolar disorder and schizophrenia. I clicked on lactose intolerance. 23andMe said I should have the enzyme to digest milk. But I know I can't digest it. Believe me, I've tried.

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23andMe's feature called 23andWe allowed me to share this milk indigestion problem with them. On the lactose intolerance page, I took a survey about drinking milk and checked the option: "Yes, I'm pretty sure I am [lactose intolerant]. Milk shakes are my enemy." The point? This information "could lead to major insights when combined with genetic information from the 23andMe database," the Web site read. I wished for a moment that all this educational material had been available when I was taking genetics in high school. I clicked on the section "Genetics 101" and watched one of the animated videos: [A banana has 11 pairs of chromosomes](#), one video said. The side dish of ancestry information enabled me to trace my maternal line back 35,000 years.

When it comes to pinning down your risk of more complex diseases, it's a bit challenging for someone of mixed ethnicity. You have the option to pick only one ethnicity. So when I clicked on European, the results didn't quite match up to what I saw when I clicked on Asian. My risk for type 2 diabetes and rheumatoid arthritis was both low and high, depending on which ethnicity I chose. So what happens when you're both, like me? I printed out the discrepancies to ask the 23andMe founders in person.

After going to Navigenics in January, I went to 23andMe. I took a 10-minute cab ride to 23andMe's office, enjoying [the Silicon Valley scene](#): the smell of fresh air, suburban tree-lined streets, bikers in spandex on their way to work, and men in suits standing on street corners with laptop bags in hand. I entered 23andMe's nondescript building and waited in the lobby, playing with the toys there: fingers flashing red lights, pins that say "I spat," and stress balls on the coffee table. Sweaty employees jogged down the stairs in their gym clothes, returning from personal training sessions and making their way to a kitchen stocked full of snacks.

One of the cofounders of 23andMe, Linda Avey, told me the company wants to create the world's largest database of DNA information. Glad I helped contribute to their plans for world (DNA) domination, I found myself thinking. What is halting progress is the lack of people involved in genetic studies, not the technology, Avey said. By translating thousands of scientific discoveries and interpreting what the discoveries mean to an individual, they hope to "break down the barriers between the general public and what is going on in academic science," she explained.

The idea of starting 23andMe evolved out of cubicle chatter. Avey had thought about collecting people's DNA when she was working at her former job. She'd sit around with coworkers and talk about how great it would be if "we had more genetic information on more people so that we could learn more about why people get a disease." At this point [Anne Wojcicki](#), cofounder of 23andMe, entered our conversation full of energy. Wojcicki's interest began when she was working in the biotech investment field. "All this money goes into research, and very little comes out of it," she said. "One of the biggest challenges is getting enough data." Getting more data means more people will have to spit. Google has bought into this notion: So far, the company has invested \$3.9 million in 23andMe and an undisclosed amount in Navigenics. (Google's cofounder, [Sergey Brin, is married to Wojcicki.](#))

Frustration over mounds of genetic information never making it out of genetic research cliques is understandable, but I had my own frustration to address. You get the best results if you are of European descent. I told the founders I was an Asian-European mix, and when I clicked on the different options on the Web site, I got conflicting results. I wanted to know why. "Research needs to go a significant step further," Wojcicki admitted. "23andMe's goal is to advance genetics research in this area. So 12 months from now, when you're logging on to the site every day, we hope the data is more reflective of your [ethnic] background."

Later that night in my hotel room, I logged on to my 23andMe account. I noticed a new addition to the FAQs; it was about mixed ethnic backgrounds. So I clicked on it: "Because genetic association studies are generally performed in populations of a particular ethnic background [European, Asian, African], we cannot know whether the associations will also apply to those of a mixed background.... Feel free to look at the different incidence estimates the 23andMe Odds Calculator provides for different ethnicities, but take the estimates with even more grains of salt than usual."

DeCODEme

Like any good medical consumer, I got a third opinion. DeCODE genetics is known for [discovering genetic risk factors](#) for type 2 diabetes, early-onset heart attack, and breast cancer, among others. For this unscientific reason alone, I decided to use the company's test—named deCODEme—to validate the other two.

I bought the test for a dollar—the best dollar I've ever spent. Normally it costs \$985. (I gave in to temptation and accepted the reporter's discount.) A few days later, I received my "buccal DNA collector" in the mail. By popping this stick in and out of my mouth for two minutes, I collected cheek cells—certainly a nice change from spitting. I mailed my sample off to deCODE genetics' in-house testing lab in Reykjavik, Iceland.

There's something to be said for taking a test three times. Seeing my genetic risks displayed online seemed perfectly normal now—just as normal as checking my e-mail. When I received a message in my in-box telling me that my analysis was ready, I signed on to my deCODEme account to view my results. First I adjusted the settings to my sex and age and even checked an option to say please contact me if there's a chance to participate in genetic research. I had to pick Asian or European—again, no dual ethnic function existed. Finally I explored the 29 diseases and traits available at my fingertips.

First I pretended to be fully Asian and saw that I was at risk of developing type 2 diabetes, [colorectal cancer](#), and rheumatoid arthritis. Next I was a European female. This time I was at risk for age-related macular degeneration, atrial fibrillation, rheumatoid arthritis, and Crohn's disease. Yikes.

The CEO of deCODE genetics, [Kári Stefánsson](#), interpreted my results for me. It is not the norm to have the CEO look at your results. Usually you'd receive your results online, and it would largely be up to you to interpret them for yourself. Stefánsson is the famous gene hunter who spearheaded the effort that mapped 65 percent of Iceland's genome. I printed out my two summary reports—the Asian and the European—for him to look at.

I walked from my office to meet Stefánsson in the lobby of his hotel in Manhattan. Dressed all in black, he intimidated me. It wasn't because at 6 foot 5 he towered over me or that he made fun of my wimpy handshake. What intimidated me was his confidence. If there were such a thing as a preacher for genetic testing, Stefánsson would be converting a lot of people. But after talking to him for hours, I realized Stefánsson was a lighthearted, matter-of-fact, all-around amazing guy—someone you'd invite to a dinner party to guarantee interesting conversation.

"There's a paradigm shift from intervention to preventive medicine as we speak," he said. "It happened when people started to download information about diseases. This didn't happen before. Doctors used to be omnipotent; no one ever questioned them." Knowledge is power.

I asked him to read my genetic horoscope. He looked at the European results first.

"You're at high risk for developing [atrial fibrillation](#), so cut back on how much alcohol and coffee you drink," he said, ever so calmly. A person with atrial fibrillation has irregular heartbeats that can cause blood clots and lead to strokes. "The biggest alarm here is the possibility of developing macular degeneration." I know I've mentioned this before, but seriously, I'm at risk of going blind in, like, 35 years. And that worries me, since I can't see crap without my glasses now.

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Then I asked him to look at my Asian readout. Stefánsson didn't know why the Asian readout was so sparse, so he called a colleague in Iceland. Now that's customer service at its best. Very little is known about the risks in Asians, the colleague said over the phone. This, however, might change soon. Stefánsson is establishing research collaboration in Hong Kong, with the hope of expanding deCODEme to the Far East.

Can I trust what is there now? "There's always a risk that information is wrong," Stefánsson said. "It is at least as accurate as any medical tests are. We are the guys who discovered most of the genes. We are like fish in water. This is what we've been doing for more than a decade."

Why find out about the risks of diseases when there is no cure? "If you don't want to [look at Alzheimer's](#) because you're afraid that you would be unable to face the fact that you're likely to lose the little God gave you, then you just don't click on it," Stefánsson said. Why should someone in her early twenties worry about old-people diseases? "Do you want to live a life of a youngster, with the illusion of immortality?" he asked. He answered my questions with questions. Stefánsson told me to think of the tests as an instrument to learn something about myself.

Stefánsson shared his own results with me. He's at higher-than-average risk of [developing prostate cancer](#), so he should probably check with a urologist on a regular basis. "I was a medical doctor for 25 years, and I don't like doctors," he told me. "I rarely go to see them. Most people I know who have died, have died fairly soon after seeing a doctor or have died in a hospital. I tell the people I care about to stay away from hospitals," Stefánsson joked.

I left the hotel feeling pretty good about my genes for the most part. But my eyes throbbed the rest of the day. Perhaps I just needed to change my contacts. Where do you draw the line between knowing risks and believing you'll get sick? The baggage of knowing my genetic outlook was taking its toll, momentarily tricking me into thinking my risk of macular degeneration was an actual medical diagnosis.

In addition to giving me varying reports about my potential risk of illness, I found that each company had a totally different personality. Like overprotective parents, Navigenics told me only the risks they thought I should know. With enthusiasm their genetic counselors took care of me. Once the results are available, you have the option to call. Like a night out on the town with a popular clubbing buddy, 23andMe knew how to entertain me. Playing on its Web site was almost as much fun as surfing Facebook. I even made some friends along the way (including 23andMe director and investor Esther Dyson). By the time I used deCODEme, the tests' novelty had worn off. Still, deCODE genetics was authoritative. My first impression came entirely from its CEO—idealistic, likable, and thoughtful.

The Critics

So what do the critics of these tests say? "If [the companies] say you have a certain gene variant, can you believe the results?" asks [Muin J. Khoury](#), director of the National Office of Public Health Genomics at the Centers for Disease Control and Prevention. Our understanding of how each gene, each cigarette, and the environment interact and how they contribute to disease is comparable to the state of computing in the 1980s. "Your family history reflects the shared environment and the sharing of many genes," Khoury says. Family history is your best predictor, he argues.

Seeing my genetic risks displayed online seemed perfectly normal now—just as normal as checking my e-mail.

Alan E. Guttmacher, now acting director of the [National Human Genome Research Institute](#) at the National Institutes of Health, agrees with Khoury. "If you read the fine print carefully, these Web sites don't usually make false claims, but I think a lot of people

will have false hopes. The information is incomplete at best and, in some cases, very likely wrong," Guttmacher says. "The results look scientific," he adds, but don't be fooled. "I would have been more surprised if the results were consistent."

Both Khoury and Guttmacher have aired their concerns. Khoury, coauthor of a recent paper on the subject published in *The New England Journal of Medicine*, clearly warns that this technology is not ready for prime time.

"The FDA isn't currently regulating the tests," says [Gail Javitt](#), the law and policy director at the Genetics & Public Policy Center at Johns Hopkins University. "Even though they have the authority to, they're not enforcing it." Because the tests were made in-house by the labs, it's up to the labs to maintain standards. "There's a lack of oversight, but what's more troubling is there's no health-care intermediary," Javitt says. "It might misinform customers about their risk, and they will go on to take actions that aren't warranted."

This story is a kind of consumer report on DNA testing. In 2006 the General Accounting Office (GAO) [did its own report](#), focusing on the related field of nutrigenetic testing, in which companies test your DNA and then use that information to give you diet and lifestyle recommendations. But the GAO did not cover the broader range of DNA tests available directly to consumers. The GAO sent two people's spit to different companies but made up 14 different profiles to pretend the samples represented fictitious people of various age groups, weights, and lifestyles. "The 14 results we received do contain predictions that a consumer may interpret as diagnoses," the report read. "Even though all of the genetic information contained in the test results based on a single source should be identical, we received disparate results for a sample from the same source."

Me

While the results were sometimes conflicting, the advice was basically the same: Stop smoking, lose weight, exercise more, and control blood pressure. Something tells me I should be doing all these things anyway.

I'm glad I took the tests. I went on a trip into my past, present, and future. It's an experience that gave me a new perspective on life. For six months I visited uncharted genetic territory in a whole new world (unexplored by most people). To some, my DNA diary may seem like the confessions of a journalist discovering in the end that her genetic information didn't lead to a diagnosis of any kind. But maybe it will help me someday, when I'm trying to piece together my medical mysteries.

For now, as I sit in my office with an iced coffee in hand and log on to Navigenics, I check out what I can do to prevent multiple conditions (as if it's a luxury): Exercise. I run every day. Don't smoke. Never have. Healthy diet. I think so. Healthy weight. I hope so. Vitamins. Um, I should start. Lower blood pressure. No clue what mine is. Lower cholesterol. Oh, man, I don't know that either. Time to see my doctor. Avoid hormones. Can't. Reduce stress. OK, after this story. Get enough sleep. Ditto. Avoid alcohol. I have, thanks to the DNA tests. [Don't wear high heels](#). What, really?

I click "sign out." I try to forget about the risks and live life the best I can. Whatever my biological destiny is, at least I think I can change it a little.

For more information see the [interactive feature](#).