

Technology

For \$4,000, This Genetic Counselor Will Screen Your DNA for Thousands of Diseases

At Boston's Brigham and Women's Hospital, healthy patients can get tested to gauge their risk for developing a myriad of conditions.

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The Genetic Counselor With a \$4,000 DNA Test

In late 2015, Kristine Trudeau was waiting inside Brigham and Women's Hospital in Boston to get some blood drawn when a research assistant approached her. Would she be interested in donating her blood for genetics research?

"I thought, that's a good thing, right?" recalled Trudeau, who was 51 at the time. "Anything I can do to help." She signed some papers and forgot about it.

A few years later, Trudeau received a letter from the researchers. They had sequenced her DNA and found an anomaly. She called the number listed and was connected to Carrie Blout, a genetic counselor. Blout told Trudeau that she had an elevated risk of developing breast and ovarian cancers—but that they should run another test to be sure.

Trudeau opted to take Blout's recommendation to see a specialist, and soon she learned the breast cancer wasn't just a hypothetical risk: She already had it. The doctors found the tumor so early—the malignancy had yet to spread to her lymph nodes—that she was able to avoid

debilitating treatments like chemotherapy. Research suggested she was more likely than not to develop breast cancer again by age 70, so she opted to have her breasts removed. And to reduce her risk of developing ovarian cancer, she got her ovaries and fallopian tubes removed as well.

It's cases like these that prompted Brigham and Women's Hospital to quietly open its Preventive Genomics Clinic last November. The goal is to catch unsuspected conditions early while they're still treatable—or to prevent conditions from ever developing in the first place.

While several commercial labs now offer to sequence customers' genomes for similarly preventative purposes, Brigham's clinic, led by medical geneticist Robert Green, is one of the world's first to do so for patients at a hospital. Blout helped Green set up the clinic, and has counseled its first few patients—including me.

Genetic counselors have traditionally seen sick patients who are awaiting a diagnosis, but Blout is a new type of counselor. Her role is the focus of the latest episode of *Next Jobs*, a mini-documentary series about professions of the future. With this new clinic, Blout sees seemingly healthy patients who are looking for everything. The difference may seem small, but the switch offers nuanced challenges. How do you disclose an alarming finding to someone who wasn't already expecting it? With medicine's still-evolving understanding of the genetic underpinnings of disease, how do you explain to the patient that she may develop a certain condition in the future, but she could also never develop it at all? Blout has counseled more than a hundred patients on unexpected genetic findings through various research studies, and has offered recommendations and feedback to medical providers who have delivered results to many more. She may know more than anyone else in the world when it comes to this emerging niche of her profession.

She said that many genetic counselors worry about the prospect of delivering test results to otherwise healthy patients. "Sometimes I'm telling them really scary stuff they weren't anticipating, like you're at risk to develop cancer, or you're at risk to develop a pretty serious cardiac condition," Blout said. "But what I've learned is that people generally do really well with it."

The clinic's prices range from \$250 to \$2,950 just for the lab work; the more money you pay, the more thorough your results. I opted for the priciest one, which would sequence 97.9% of my genome, focusing on about 3,700 genes that scientists have linked to more than 2,500 conditions. Compare that with the popular at-home DNA testing kit from 23andMe Inc. For \$199, 23andme examines about 0.01% of the genome to assess one's risk of developing 13 conditions.





Genetic counselor Carrie Blout, right, explains the Preventive Genomic Clinic's various testing options. *Bloomberg*

Blout then made sure I understood the limitations of the test: Virtually no finding could tell me that I would develop any condition with 100% certainty. She asked me if I was prepared for the possibility I was at risk of developing something I couldn't treat or prevent. Then, I signed a lot of papers.

In June, I returned to Boston for my follow-up. As soon as we sat down, Blout told me they found nothing serious. What they did find was that I was a carrier for three rare and recessive conditions I could pass on to my kids—including a sensitivity to light so severe that even mild sun exposure could make the skin blister. But it was very unlikely that I would.

It was a tremendous relief. However, the total bill for the clinic visit and the lab came out to \$4,045, and only a portion of that was covered by insurance. For me, was it worth the cost? If my situation were closer to Kristine Trudeau's, in which the information helped me get help early for a treatable illness, I would certainly feel differently. But there's another possibility—that the test gives you knowledge of a potentially impending disease you can't fight.

A woman who joined a genome sequencing study in 2014 found herself in that scenario. Her test revealed she had a genetic variant linked to Alzheimer's, for which there's currently neither a cure nor a treatment to stop or slow the disease's progression. The woman, who is 28 today and asked to remain anonymous to protect her medical privacy, is not exhibiting any of Alzheimer's symptoms. She's thriving in her job as a landscape designer. She's in a relationship with a boyfriend she loves. She may never develop Alzheimer's—but her odds are far worse than that of the general population's. Other patients with her variant seem to be developing the disease as early as their mid-60s—and she's seen its debilitating effects, in her own grandfather before he died, and now, she suspects in her father, too, even though he hasn't been officially diagnosed.

And still, she's glad she knows.

“Having that information now, there are things that I can do to start looking for symptoms or be on top of early symptoms,” she said. “I imagine that I'll start seeing a neurologist probably in my fifties. I've been thinking a lot about getting involved in long-term studies on late-onset Alzheimer's. There are small things I can do, and certainly there isn't a cure or anything, but it does give me something to hold on to.”

Not everyone will take such devastating news with grace. And that's one of the reasons why genome sequencing for disease prevention is controversial; when she presents her research at genetics conferences, Blout fields all kinds of questions from skeptical peers. Will worrying test results make patients anxious or depressed? Will they prompt patients to take unnecessary medication, or opt for surgeries that end up harming them? And would the cost savings that come from early diagnosis and prevention be worth the money this testing currently costs? There are many drawbacks that the research is still parsing out.

“So far it seems like it's not increasing anxiety. It seems like we're finding important things. It seems like we're saving lives with a lot of our research. But for me to say, yes everyone should get screened for sure, is a little premature,” Blout said. “Not everybody wants this type of information.”

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