https://www.usatoday.com/story/news/2016/06/05/uvm-medical-center-forefront-genome-sequencing/84781390/

USA TODAY **Greg Merhar diagnosis himself with FMF after genome sequencing**

UVM Medical Center blazes genetic mapping trail

Dan D'Ambrosio, Free Press Staff Writer Published June 5, 2016 | Updated June 7, 2016

Michael Towle made the long walk down Colchester Avenue to get the results of his genetic testing on a dreary, drizzly day in April.

"I was more anxious than I thought I would be," Towle said. "On the way there I was thinking, 'Do I really want to go to this appointment?'"

The cause of Towle's angst was simple: The results of sequencing his genome could reveal he's likely destined to get Alzheimer's disease or some other life-altering malady.

Towle, network business director at the University of Vermont Medical Center, is one of 73 volunteers from the hospital staff to take part in a pilot project intended to launch UVM Medical Center's new initiative to embrace genomic screening as a path to better medical care.

Towle had the option to only learn about conditions he could do something about, or to learn it all. He chose the latter.

"I came back very clean, which is great," Towle said. "I left there very relieved."

Six of the volunteers were less fortunate. They received "difficult" news, according to hospital spokesman Michael Carrese. None of the six were willing to talk about what they had learned, because they were still absorbing the news, and in some cases, still needed to talk to family members, Carrese said.

The weight of genetic knowledge includes not only diseases and conditions you are likely to get, but also which mutated genes you could pass on to your children, resulting in diseases such as cystic fibrosis. Towle prefers to see this knowledge as power rather than tragedy, allowing him to make more informed decisions.

The results of genomic screening are not all heavy either. Towle, for example, learned he is unable to taste bitterness, which explained his predilection for IPA beers.

"They don't taste bitter to me," he said.

Welcome to the future of medicine.

'I almost fell out of my chair'

Dr. Debra Leonard is the director of the Genomic Medicine Program at the University of Vermont College of Medicine. She came to Burlington three years ago from Weill Cornell Medical College in New York, after, she said, she realized that hospital didn't share her vision for comprehensive genomic testing. Leonard received a very different reception from Dr. John Brumsted, president and CEO of The UVM Medical Center and The UVM Health Network.

Leonard told Brumsted she wanted to develop a program she called "Genomes for All." To do that, she would need a big lab.

"I said, 'Every lab I've worked in so far, as soon as it's built it's too small already,'" Leonard remembered. "I said, 'What I am imagining is we are preparing for the time in 10 years when we will be sequencing the genome of every patient who comes to the hospital.'"

Leonard prefaced her remarks by warning, "Don't fall out of your chair." Brumsted remained in his chair, and told Leonard he agreed with her.

"At which point I almost fell out of my chair," Leonard said. "Because I'd never had someone who understands how fundamental the genetic information is."

Taking a patient's genetic information is no different than taking her blood pressure, temperature, heart rate, height and weight, Leonard said.

"It provides basic information that for some people is useful now," she said. "For other people we don't know enough yet to understand what it might mean, especially for healthy people, but for those with disease risks driven by genetic differences in their genomes, it can be helpful."

A shift in foundation

Brumsted sees Leonard's vision as nothing less than a shift in the foundation on which the hospital practices medicine and delivers care.

"Take a step back and look at where health care is going," Brumsted said. "We've long been set up to take care of individuals with a problem, a chronic illness, cancer, a heart ailment. We need to keep doing that really well. But we also need to move much more into keeping people healthy, preventing them from ending up with a malady."

Brumsted believes genomics will play a big role in that shift.

"We're already starting to see a move toward more personalized care of individuals, first in cancer," he said. "We can take bits of a tumor or cancer and based on the genetic makeup of the individual and that cancer you can tell which chemotherapy might be advantageous, which could be detrimental, and how much chemotherapy to use."

The scope of disorders for which the hospital will be able to tailor its therapy to individuals will increase rapidly, in Brumsted's opinion. One example: Rather than recommending colonoscopies for everyone over the age of 50 — now the standard of care — genetic screening could lead to perhaps only one-third or one-half of the population requiring colonoscopies, based on an analysis of their genomes.

"Think of the savings in health care dollars," Brumsted said.

Genomes for All could also help doctors identify the health risks their patients face in advance, allowing patients to change their lifestyles in hopes of preventing disease.

"That's keeping the population healthy," Brumsted said. "All of that has to be done on a backdrop of privacy of information, just like we have now, with a lot of rules and regulations around how we handle personal health information."

Construction on the $2.5 million lab Leonard wanted begins in two weeks within existing space at the hospital, Brumsted said. The lab should be up and running by the end of the year.

Sequenced at birth

Leonard enlisted the help of Illumina, based in San Diego, for the pilot genome project at UVM Medical Center. A publicly traded company with some 5,000 employees and offices from Singapore to Sao Paolo, Illumina generated revenues of $2.2 billion in 2015.

Dawn Barry, Illumina's vice president of applied genomics, said no other hospital she knows of is planning quite the push into genomics as UVM Medical Center is, including the use of genetic counselors to make sure patients understand the context of their results.

Barry envisions a future when people will have their genomes sequenced at birth, and then use that information throughout their lives to fend off illness and better treat disease.

Illumina subsidized the cost of genomic screening for UVM Medical Center's pilot project at $2,900 each, compared to the $7,000 to $10,000 price tag common in the marketplace, Barry said.

"We think it's very important to allow professionals the opportunity to put themselves in patients' shoes," she said.

Currently, insurance doesn't cover the cost of sequencing an entire genome, according to Towle, the UVM Medical Network business director. Insurance often will cover certain sequencing if there's a clinical reason for it, such as determining whether someone carries the genes for breast cancer.

Towle believes that with more sophisticated data, and more proof that genomic screening can lower the cost of health care, insurance companies could come around to covering complete genome sequencing.

A new normal

Dr. Leonard and her husband, photographer Greg Merhar, were among Illumina's first clients; they gave each other genomic screenings as Christmas gifts in 2014 at a cost of $5,000 each. The investment paid off handsomely, when Merhar, a serious runner, learned that a lifetime of struggles with stomach problems and swollen Achilles tendons and ankles, among other ailments, was because of a condition known as Familial Mediterranean fever.

Merhar's condition had gone undiagnosed, or misdiagnosed, for decades. One orthopedist wanted to cut off his heel bone, move it over and screw it back on to fix his chronically swollen ankle. Merhar opted for a brace on his ankle for a year to take down the swelling.

Once his genome revealed the true cause of his trouble, Merhar began taking small doses of a prescription drug, Colchicine, which has been used for centuries to treat gout.

"I took a dose on a Friday night," Merhar said. "Honestly I was feeling way better the next morning. After two days, I was pretty much pain-free."

After several weeks, Merhar's Achilles tendons looked normal. He hasn't had an abdominal attack — which he described as like swallowing a basketball — since last August, when he started taking Colchicine. He no longer fidgets from constant pain in his back and hips.

"Now I feel what normal is like," Merhar said. "Going from where I was to normal is a miracle to me."

This story has been updated to correct Illumina's annual revenue in 2015 as $2.2 billion.

This story was first published on June 5, 2016. Contact Dan D'Ambrosio at 660-1841 or ddambrosio@freepressmedia.com. Follow him on Twitter at www.twitter.com/DanDambrosioVT.