THE $1,000 GENOME

< Scientists See Upside And Downside Of Sequencing Their Own Genes

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RENEE MONTAGNE, HOST:

To begin our series The $1,000 Genome, we heard yesterday how decoding the entire human genetic blueprint could revolutionize how doctors treat sick people. Today, we're going to hear what it may tell us about healthy people. NPR's Rob Stein explores the experience of two prominent geneticists who had their DNA deciphered to find out for themselves.

ROB STEIN, BYLINE: The quest to harness the power of our DNA began with a few pioneers. Listen to this '70s documentary.

(SOUNDBITE OF MOVIE)

STEIN: That's Watson, as in Watson and Crick. They won a Nobel Prize for discovering the double helix structure of DNA. That's that thing that looks like a spiral staircase, built with what scientists call base pairs. Here's Watson recalling a crucial moment in that discovery.

(SOUNDBITE OF MOVIE)

STEIN: These days...

: I'm 84 years old.

STEIN: Watson's still play a pioneering role of sorts in the world of genetics. When scientists were looking for the first person to test the new super-fast way of deciphering someone's entire genetic blueprint, who did they ask to volunteer?

: They had to sequence someone, so they got me.

STEIN: Watson gave them a blood sample and the scientists went at it. When they showed him the results, Watson discovered they had solved a mystery for him. He never understood why he was having so much trouble controlling his blood pressure with drugs called beta blockers.

: I had tried to take beta blockers but they put me to sleep.

STEIN: After being sequenced, his doctors figured out why. He had genes that made him more sensitive than most people to the drug he was taking.

: Now I can take them once a week, so my blood pressure is better controlled now because of my genome.

STEIN: This is one of the big things that doctors think they'll get from sequencing: a whole new way to figure out which drugs work for their patients, which don't, which are safe, and which are dangerous. Robert Green is a Harvard geneticist.

ROBERT GREEN: The doctors of the future, when you start to prescribe a drug for which you have a genomic variation that would give you a side effect, a flag will pop up and say maybe you ought to consider another drug.

STEIN: Overall, though, Watson didn't end up learning all that much from his DNA.

: Really, nothing came up.

STEIN: Skeptics say that's probably the case for most people at this point. Scientists still know so little about how to interpret the data that most people will get little, if any, useful information out of it. That's one reason some scientists are sequencing themselves - to learn how to use this flood of information. We'll get to more about Watson's genome later. But first we turn to another scientist.

MICHAEL SNYDER: Michael Snyder. I'm chair of the genetics department at Stanford University.

STEIN: Snyder decided to become his own sequencing guinea pig. But this time the results were dramatic. One of the first things that popped out was a shocker. Snyder was at risk for Type 2 diabetes. But here's the thing. It just didn't seem to make any sense. Snyder had no family history of diabetes. He wasn't overweight. There was no reason to think he should get the disease. But just to be on the safe side, Snyder asked a colleague who specializes in diabetes to start watching his blood sugar levels. At first she was skeptical.

SNYDER: The person doing the test said there's no way you're at risk for Type 2 diabetes. And I said, well, I don't think so either. But my genome says that there's something interesting about my glucose metabolism, so I think we should do this test.

STEIN: So everyone was stunned when his blood sugar started rising - and then kept rising. Within months, it spiked. They had literally watched him become a diabetic in real time.

SNYDER: And sure enough, she classified me as diabetic. So in fact my genome then did predict I was at risk for a disease, which by following the various markers for that

disease, I did discover I did get.

STEIN: Snyder jumped on it. He totally transformed his diet and kicked up his exercise. After about six months, his blood sugar gradually fell back to normal.

SNYDER: That's the power of genomics, is to help you catch things as early as possible. So some people might say that actually my genome saved my life.

STEIN: Snyder's story is the kind of thing that's feeding the buzz about sequencing and raising the prospect of sequencing becoming routine - as routine as, say, testing your cholesterol. Here's Robert Green again. He's the Harvard geneticist.

GREEN: I think it's going to be part of everyday medicine sooner than most of us can actually imagine.

STEIN: Genome sequencing could spot who's going to get breast cancer, escape prostate cancer, need to watch their heart.

GREEN: So I think there's lots of ways in which genomics will be integrated into medicine in the coming years. I think it's one of the most exciting frontiers in science and society.

STEIN: Now, let's get back to one of the early guinea pigs in this frontier: James Watson. He agrees sequencing people's genomes is a good idea.

: I'm all for, you know, moving through life with knowledge.

STEIN: But when scientists asked him to volunteer, he had one condition.

: I didn't want know its prediction for Alzheimer's.

STEIN: Watson had watched his grandmother die from Alzheimer's.

: You can't do anything preventive, so why should you want to know?

STEIN: So he told the geneticists they could tell him everything else that showed up in his genes, but not that. This is actually one of the big debates about sequencing. For all that we could potentially learn, how much do we really want to know?

JAMES EVANS: Your genome is a complex and not necessarily a real warm and fuzzy place.

STEIN: James Evans studies genetics at the University of North Carolina. He says some people may want to know everything, but a lot of people may not. And what do doctors do if they stumble across something they weren't even looking for? Plus, Evans says, there's plenty of chances of getting results that could end up doing more harm than good.

EVANS: You're dealing with a medical test. And medical tests have the power to help. They have the power to hurt. They have the power to confuse.

STEIN: In Watson's case, the first interpretation of his genome indicated he should already be dead, killed by a terminal illness.

: It was a nasty condition. I purposefully didn't think about it, because I figured I would wonder why I wasn't sick. Well, it turns out I shouldn't have been.

STEIN: Doctors quickly realized they had misread it. And luckily Watson hadn't gotten too worked up about what they had gotten wrong. But there are also potential drawbacks to getting it right. Michael Snyder, the scientist from Stanford, got a glimpse of that himself. After he was sequenced and found out he was at high risk for diabetes, his wife tried to increase his life insurance. But because of his diabetes risk, the price shot through the roof.

SNYDER: So the bottom line is that an increase in life insurance essentially became

prohibitively expensive.

STEIN: Federal law bans health insurance companies and employers from penalizing people based on genetic information, but it doesn't cover life insurance or long-term care insurance, leaving people like Snyder vulnerable to discrimination. Despite that, Snyder is convinced sequencing has more upsides than downsides. And despite his false alarm, DNA pioneer James Watson agrees.

: I think many people would benefit from having their DNA told, and that would more than compensate for the occasional mistakes.

STEIN: Or the unexplored pitfalls. Others aren't so sure. They wonder - will sequencing finally realize the promise of genetics? Or is it far too soon for most people to venture into the dark corners of their DNA? Rob Stein, NPR News.

(SOUNDBITE OF MUSIC)

MONTAGNE: Next week, how sequencing is already helping patients. This is NPR News.