Medical Xpress

Research shows hope for personalized genome sequencing

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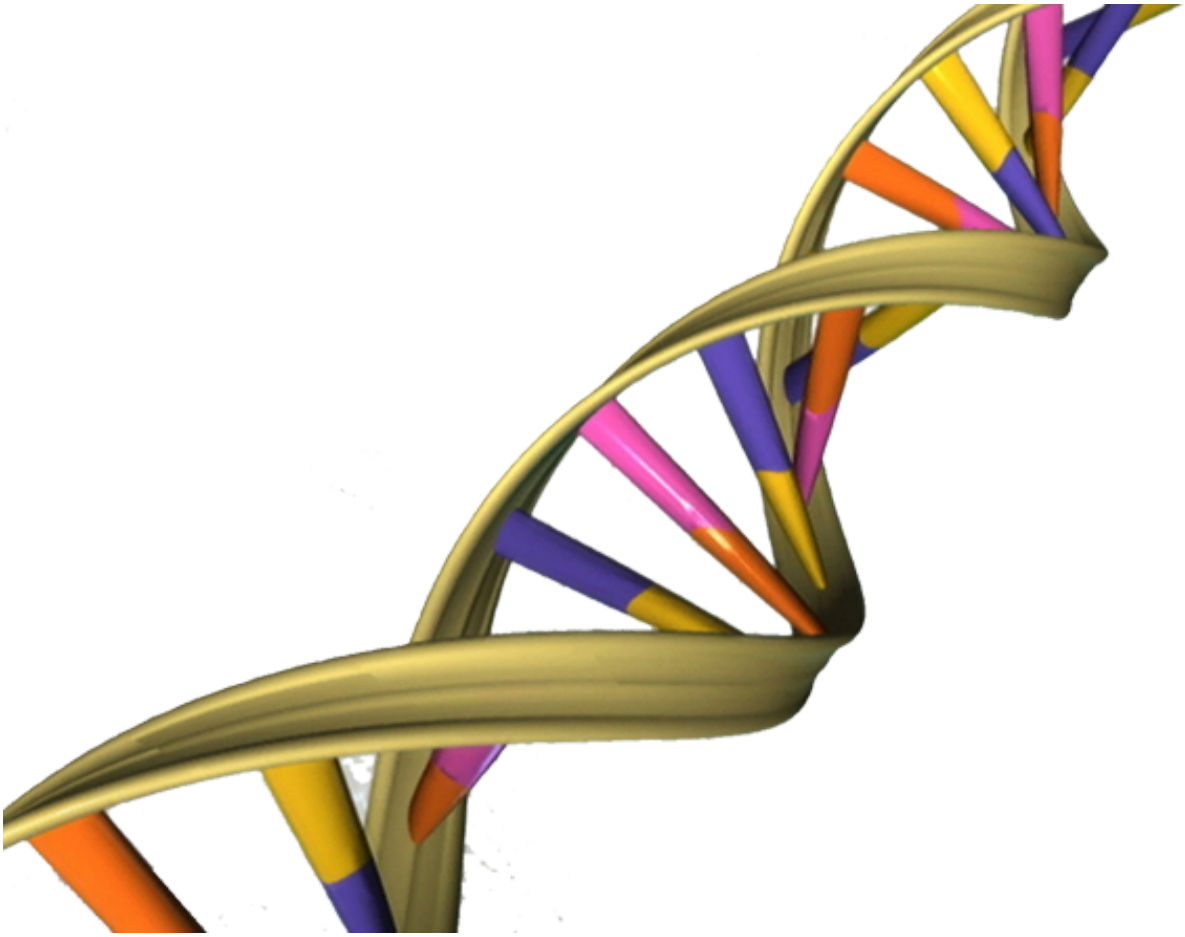

A depiction of the double helical structure of DNA. Its four coding units (A, T, C, G) are color-coded in pink, orange, purple and yellow. Credit: NHGRI

Imagine if you could carry a credit card-size record of all the 3 billion A's, T's C's, and G's that make up the alphabet soup of your genome. A simple swipe of the card could inform your physician right away if a drug being considered will help you - or even hurt you.

This is the kind of promise behind President Barack Obama's $215 million initiative to develop personalized medicine.

"We've arrived at the point where this could happen, and is going to happen," Francis Collins, director of the National Institutes of Health, said at a recent biotechnology conference in Philadelphia.

A newly published study by researchers at the University of Pennsylvania is another step on the path.

The study, which appears this month in the journal *Cell*, came from a curious case regarding a class of anti-diabetes drugs known as thiazolidinediones, or TZDs. TZDs are highly effective - in some people. But for 20 percent to 30 percent of patients, they are useless and can even cause serious side effects.

The Penn team had a hunch that the variation could have something to do with small differences in the regions of the genome that control whether a gene will be switched off or on, much like a light switch. These areas, called regulatory regions, work by lighting up genes when a molecule known as a nuclear receptor attaches to DNA.

Many drugs on the market, such as TZDs, work by binding to nuclear receptors, which regulate whether genes are turned on. The Penn researchers found that one change in the sequence of base pairs - adenine, thymine, cytosine and guanine, those A's, T's, C's and G's - in the light switch regions of the genome may ultimately predict how those drugs will affect a particular patient.

"Every drug has a risk of some sort," said Mitchell Lazar, professor of medicine and genetics at Penn's Perelman School of Medicine and senior author on the study. Figuring out how those risks are related to a person's genetic code is "one of the principles of personalized medicine."

"The cost of determining a person's genome is coming down to the point where it's widely predicted that in five to 10 years, every person will be able to have their genome sequenced," Lazar said. "Epidemiologists and statisticians will be able to correlate individual difference(s) in the genome and ask, 'Was the drug effective?' "

Finding the mutations that matter is the hardest part, said Tim Reddy, assistant professor of biostatistics and bioinformatics at Duke University, who was not involved in the Penn study. However, he said, projects like this, which identify the genetic predictors of people who respond to medications vs. people who don't, are the first steps toward making personalized medicine a reality.

And, Reddy said, like any good study, "this opens up a lot more questions and a lot more opportunities. Nuclear receptors are the targets of countless drugs for other diseases. I'm really excited to see how the principles that were revealed here can be generalized for other drugs that target" these molecules.

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