What Is the Future of Medical Genetics?

Researchers Gather to Learn Newest Developments

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More genetic knowledge has not always been a good thing.

When the first disease gene for Huntington's was discovered, it didn't lead to better treatment right away. But several patients who found out about their risk committed suicide.

"How human beings will take a piece of information may be different before they know the test and after they know the test," said Aravinda Chakravarti, a Johns Hopkins researcher.

He explained that many people want to know more about their genome, or hereditary information encoded in their DNA, only to find that once they have that information they can do little to prevent the disease it predicts.

As doctors and researchers gather at The Jackson Laboratory in Bar Harbor, Maine, for the 49th annual Short Course on Medical and Experimental Mammalian Genetics, they will discuss the latest advances in genetics, as well as an overview of changes showing how patients are treated in clinics by doctors. As the doses of the drugs we take are determined increasingly through a test of our genes, having someone who can explain the results of these genetic tests -- many of which are now available by mail order -- is more important than ever.

That may sound like a simple-sounding proposition, perhaps. But the number of tests for genetic diseases outnumbers biochemists trained in genetics in the United States by more than 3 to 1, and entire states are without a single physician who can explain the results of a genetic test.

In recent years, many sources have offered testing directly to consumers, without the promise of genetic counseling to accompany it.

"It's the Wild West right now, really, in this area," said Dr. Robert Nussbaum, the chief of medical genetics at the University of California, San Francisco.

Ada Hamosh, clinical director of the Institute for Genetic Medicine at Johns Hopkins, noted that the abundance of information can be a concern for physicians trained in genetics -- even more so for someone who has his or her test results but no way to interpret them.

"It's an enormous amount of partial knowledge. It's very overwhelming, from a clinical perspective," she said. "We're going to spend the next 50 years, at least, trying to figure all this stuff out."
The Doctor Will See Your Genes Now

Hamosh noted that it is important that patients bring their family history to their doctor so that their physician will know if there is a need for genetic testing. She said that even patients with a genetic diagnosis with no cure should still see a geneticist to help manage their condition, because of the speed of progress.

The thinking is, Hamosh says, "If I don't know something today, I will know more tomorrow."

Hamosh added that she never dismisses a patient who comes to see her, no matter how little is known about the condition. The only time she will stop seeing patients is if they see her about a condition their parents had that they clearly have not inherited.

In addition to finding new ways to treat genetic disorders, physicians also expressed hope for their use in determining drug dosing and delivery.

One example is Warfarin, the generic name of the anticoagulant known as Coumadin. Two to three million new prescriptions are written for it each year, according to UCSF's Nussbaum.

He explains that the proper dosing can range from 1 milligram to 15 milligrams, and too much of the drug can lead to serious bleeding -- bleeding that sends 30,000 people to the emergency room each year.

As for how the dose is determined, "it's all chasing your tail now," Nussbaum said. Dosing begins at 5 milligrams and is then adjusted to the patient's needs. If a problem arises, doctors back off. It's classic trial and error.

But that may soon change. In the first major trial for dosing a drug based on a gene profile, physicians hope to find if they can predict the proper dosage based on a person's genes. Run by the University of Pennsylvania, pharmacists involved in the trial will determine the dosage based on the genetic profile, and neither doctors nor patients will know exactly how much is being administered.

Nussbaum says his group at UCSF plans to be among those medical groups whose patients are a part of the trial.

Can Laws Keep Up?

But as genetics develops new ways of diagnosing and treating patients, many are worried about whether laws and policies will keep pace.

Hamosh notes that she and genetic counselors in her clinic frequently have to fight to get insurance to pay for gene testing and some of the treatments that follow, which many insurance companies refuse to cover.

On the flip side, however, many are worrying about what will happen once all their genetic information is available, whether it will mean insurance companies denying medical coverage or the government using it to track everywhere they go.

Johns Hopkins' Chakravarti notes that genetic testing has many applications for preventive medicine that could ultimately save in health-care costs, but the only way to ensure that they are not used against a patient is through the courts.
"The only way in which we can prevent wide-scale discrimination base on someone's blueprint will be by legislation," he said.

But as Chakravarti points out, not all genetic information is harmful if released.

For example, he said, the first widespread genetic testing people underwent was for blood type in order to avoid problems when they underwent transfusions.

People are willing to tell other people their blood type without concern, he said, because it can be beneficial in many instances.

This, said Chakravarti, is the direction genetics research should head in future years -- tests with a clear benefit to the patient and with no negative uses.

"Ultimately, if there are genetic tests that are accurate and can be used for a well-defined medical need, we won't be afraid to share our medical information," he said.

But he notes that patients who worry how their DNA may be used have valid concerns, especially with the speed of technologies that are making sequencing easier and easier.

Governments may insist on collecting more information on citizens, he notes, and people will have to decide whether their motives are malicious or benign.

"You can't fake your DNA profile," he said. "Maybe that's your future identity."

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