Off-the-Shelf Genetic Testing On Display

The emerging market of direct-to-consumer genetic testing gets down to business.

By Emily Singer

Want to share your genome online with friends and family? Find out how well you metabolize B vitamins? Determine if you're genetically susceptible to forming blood clots on long flights? All of this is possible with a credit card and an Internet connection, thanks to the growing field of direct-to-consumer genetic testing, which aims to move genetic tests out of the doctor's office and into the hands of individuals.

The first annual Consumer Genetics Show, which started today in Boston, highlights some of what's available to today's consumer. A number of genomics startups have booths lined up along the conference hall, offering everything from genetic tests that give individuals insight into their heart health, nutritional requirements, and optimal weight-loss strategies to full genome sequences. Inside the hall, meanwhile, scientists, physicians, and entrepreneurs are grappling with some of the controversies that have grown along with the field. Foremost among these concerns: How good is the information delivered by these tests? How well can people understand the results? And how effectively can they help people manage their health?

To date, medical genetics has been largely limited to tests ordered by physicians for rare disorders triggered by defects in single genes, such as cystic fibrosis. But as the price of genetic technologies has plummeted--the cost of sequencing a genome has dropped 10,000-fold in the past four years--the possibilities for genetic testing have grown. Scientists have identified hundreds of genetic variations that can raise the risk of common diseases, such as diabetes and heart disease; point to particular ancestries; and even influence traits, such as height.

A number of companies offering direct-to-consumer genetic testing have cropped up in the past two years to capitalize on these advances, from 23andMe and Navigenics, which offer genome-wide scans to identify specific disease-linked genetic variations, to Knome, which offers whole-genome sequencing to the wealthy. Any doubts that personal genomics is making its way into the mainstream can be assuaged with a look
at Interleukin genetics, a startup that sells genetic tests for heart-disease risk, B vitamin metabolism, and other factors through Amway, the direct-sales company. "With minimal advertising, these companies have captured the imagination of the public," says Robert Green, a neurologist at Boston University and one of the conference organizers. "The public is eager to know more about genetics."

Along with this burst in genetic knowledge, however, comes a number of concerns. Scientists question how useful the currently available information is in managing one's health. "The most important issues in consumer genomics are around consumer utility and whether the kind of information available right now is useful to consumers medically and personally, and whether it has any potential for harm, either through misleading people, misunderstanding, or false reassurance," says Green. At this point, nongenetic factors, such as family history, body mass index, and history of smoking, often provide a better predictor of disease risk than does genetics, he says.

A second worry is how genetic information is delivered to consumers. "The effort is to shift the focus from the physician, who usually focuses on specific problems, to the consumer, who is looking for a more broad-based view," says Jorge Conde, president of Knome, which is based in Cambridge, MA. "There are a lot of questions around how to do that in a way that is digestible and relevant and responsible for someone not trained in genetics."

Different companies handle this different ways, some providing information online, others offering genetic counseling. "I think genetic counseling is very important because lots of people make no distinction between single-gene disorders, like cystic fibrosis, and genetic factors that may elevate their risk for, say, heart disease in a modest way," says Michael Christman, president of the Coriell Institute for Medical Research, a nonprofit research center in Camden, NJ. Common complex diseases, such as heart disease, Alzheimer's, and type 2 diabetes, are caused by a combination of genetic and environmental factors, making it difficult to predict the impact of a single linked genetic variation in an individual patient. "In the absence of someone very knowledgeable to explain this, there is the potential for gross interpretation of what it really means," says Christman.

Most direct-to-consumer genetic-testing companies have declined to release sales figures, so despite the public attention, it's unclear whether their business models are succeeding. But most people seemed unconcerned. "If you look at the first commercial transactions on the Internet, few of the early companies necessarily survived intact, but the ideas they invented became the industry," says James Heywood, cofounder of
PatientsLikeMe, a company based in Cambridge, MA, that collects, shares, and analyzes data on patients with different diseases. "It's like going back to the invention of the computer: was it the computer that was important, or was it the applications that were invented to use on it? Who knows what application will effectively build this new market?"

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