Direct-to-consumer genomics: Harmful or empowering?

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Thanks to recent scientific advances and plunging costs in genetic sequencing, consumers now can order simple, inexpensive, mail-in

genetic tests to learn more about health risks, inherited traits and ancestry. But, is it a good idea to bypass your doctor's office when it comes to interpreting health risks?

New technology means grappling with new questions of ethics, best practices and access to information. In an article published in *Missouri Medicine*, Joel Eissenberg, Ph.D., professor of biochemistry and molecular biology at Saint Louis University, explores questions that stem from these advances, which have the effect of separating the physician-patient relationship from access to new personal health data.

Some companies offer DNA tests via a mail-in saliva sample. Once the sample is tested, the company provides consumers with a report showing whether they are a carrier for any of over 35 diseases, as well as information about traits and ancestry. Participants might learn, for example, that they carry a genetic variation for cystic fibrosis or hereditary hearing loss. Carriers may pass along these genetic variations to a child, who could develop the condition if both parents carry and pass on the variation.

Other tests might find that a consumer carries a genetic variation that increases the risk of breast cancer or Alzheimer's disease. In the case of breast cancer, preventive measures, like regular screening via mammography or preventive surgery, may offer a chance to limit the risk of developing cancer. On the other hand, there is no proven medical treatment for Alzheimer's disease.

In the article, Eissenberg notes several areas of concern for consumers as they attempt to interpret their personal information without the help of a physician or genetic counselor, including a limited understanding of genetics, confusion about disease risk and anxiety caused by new information in the absence of guidance from a medical professional.

Eissenberg concludes, however, that individuals should be able to access their own personal genome data.

"In an open society, maximizing autonomy is a virtue," Eissenberg said. "Knowledge is power."

He notes that, in many ways, we've already moved along the path of patient-initiated testing, with digital thermometers, blood pressure cuffs, blood sugar monitors and pregnancy testing kits that give the public direct access to health information.

In theory, understanding genetic risk could help guide life decisions, such as encouraging regular cancer screenings, a healthy diet and exercise to offset some genetic risks for some illnesses. (Though, some research has found that communicating DNA-based disease risk has little or no effect on smoking and physical activity.)

"At its best, direct-to-consumer genomics testing could eventually become like other forms of home medical testing - another way for people to take personal control of their health and wellness," Eissenberg said.

At the same time, he urges caution in how these tests are marketed, noting that currently they are not able to offer useful predictions or recommendations for many diseases.

"Today, however, genomics testing to assess risk for complex diseases rests on a weak foundation of clinical validation. Thus, in most cases, genomics data cannot serve as a guide to action."

Finally, while Eissenberg does not believe medical paternalism is warranted in restricting access to direct-to-consumer tests, he shares several instances in which consumers may benefit from consulting with

their physician or a genetic counselor: to prepare for adverse news, to help interpret risk, to prescribe a risk response and to counsel caution in over interpretation.

But the bottom line?

"It is important to stress that genetic risk is not the same as genetic destiny," Eissenberg said.

Provided by Saint Louis University