Online tool helps patients demystify the 'Pandora's box' of genomic sequencing

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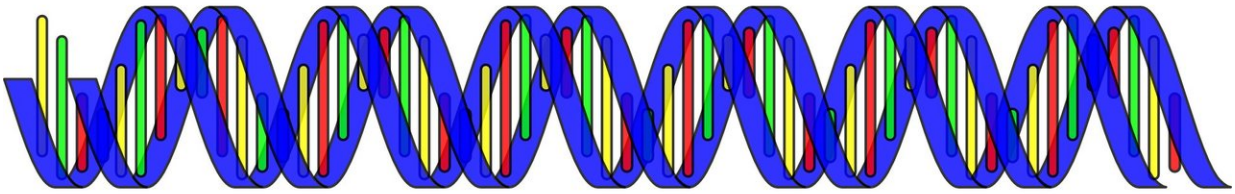

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A decision aid developed to support patients undergoing genomic sequencing can reduce the amount of time patients spend speaking with overburdened genetic counselors while helping them were more knowledgeable about the benefits of sequencing, suggests a study from St. Michael's Hospital.

Published in *Genetics in Medicine*, the official journal of the American

College of Medical Genetics and Genomics (ACMG), the study evaluated the effectiveness of the Genomics ADvISER, an interactive online decision aid designed to guide patients who have had their genome sequenced and are being offered additional medical information revealed by the sequencing.

Generally, patients undergo genomic sequencing to learn about a particular condition they may have—but the test also reveals secondary or incidental findings, a slew of other data that may or may not present troubling news about a person's risk to develop additional health issues.

For example, a person could have their genome sequenced to evaluate heart disease and uncover that they have a neuromuscular disease as well as a high risk for heart issues. Another might have their genome sequenced as part of a study on Cystic Fibrosis—a genetic lung disease—and find out they also have alterations in a gene that influences cancer.

"When you start to open the Pandora's box to find one thing you did want to know about your genome, you might find more than you bargained for," said Dr. Yvonne Bombard, a scientist at the Li Ka Shing Knowledge Institute of St. Michael's Hospital, who has led the creation of the Genomics ADvISER.

"Clinical guidelines recommend that clinicians engage in shared decision-making with patients about receiving secondary findings before sequencing. With existing constraints on genetic counselors' time, there is a need for less resource-intense ways to provide education and decision-making support."

Dr. Bombard's study found that patients who used the Genomics ADvISER needed to spend 24 minutes less speaking with a genetic counselor than those who did not access the tool. Those who used the

Genomics ADvISER also said they were more knowledgeable about the benefits of sequencing.

This is the first known randomized controlled trial to evaluate a decision aid that guides adult patients' selection of secondary findings from genomic sequencing. One hundred and thirty three patients were randomized. The intervention arm used the Genomics ADvISER to select the categories of secondary findings they wished to receive, whereas control participants spoke to a genetic counselor to make their selection.

While the decision aid did not decrease decisional conflict compared with genetic counseling alone, it did result in shorter conversations with counselors and a higher knowledge of sequencing benefits.

"With this study, we have evidence that our decision aid has the potential to address a critical gap that exists in genomic sequencing," Dr. Bombard said.

"As genomic sequencing becomes more popular in a variety of specialties, achieving improved genomics knowledge with fewer resources and significantly less time spent with genetic counselors is a crucial benefit the Genomics ADvISER provides."

Dr. Bombard and her team have started to look at ways to develop tailored methods to support patient needs during and after genomic sequencing. They are working on the 2.0 version of the Genomics ADvISER, which will transform aid into an adaptable, interactive, patient-centered digital decision support tool using artificial intelligence to customize it based on patient preferences, values, health literacy and experiences.

Provided by St. Michael's Hospital