Direct-to-consumer genetic testing kits vary in predictions of disease risk

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(Medical Xpress)—An in-depth analysis and comparison study conducted by investigators at Emory's Rollins School of Public Health demonstrated variations in predicted disease risks by companies that offer direct-to-consumer personal genome testing.

Led by Cecile Janssens, PhD, research professor of Epidemiology, the

team investigated DNA test results from three "direct- to-consumer" genome testing companies (23andMe, deCODEme, and Navigenics) to assess and compare their predictive abilities. Results show that predicted risks differed among the companies and were contradictory for certain traits in certain individuals.

The complete findings are available in the online edition of Genetics in Medicine.

"Although two of the companies that we studied are no longer operating, genotyping and sequencing is becoming less expensive and testing such as this is increasingly popular," explains Janssens. "The methods used for predicting these types of results are of important concern."

Test results provided by the three companies indicated an individual's risks for a large number of diseases. The study was conducted by creating DNA data for a hypothetical population of 100,000 individuals which is a less expensive and equally valid method for demonstrating the variations in predicted risk among the companies.

Predicted risks were calculated using the prediction methods of the three companies, obtained from their websites. Predicted risks were assessed and compared for six diseases: type 2 diabetes, prostate cancer, celiac disease, Crohn disease, age-related vision loss, and abnormal heart rhythm.

The variations in predicted risks were explained by three factors:

- The companies considered a different number of genetic variants in the risk calculations. Generally a larger difference in the number of variants implied more variation in predicted risks.
- All three companies used an estimate for the average population

disease risk as a starting point for their predictions. Differences in average risks affect predicted risks of all consumers to the same extent. For example, when the average odds is two times higher, all predicted odds are also two times higher.

- The companies applied different mathematical formulas. The formulas of two companies led to an overestimation of risks when predicted risks were higher and even predicted risks that were higher than 100 percent.

"Our study provides insight into the methodology and performance of risk estimation for personal genome tests," Janssens explains. "Future efforts to design predictive models will benefit from understanding the strengths and limitations of these current models and formulas."

More information: www.nature.com/gim/journal/vao … full/gim201380a.html

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