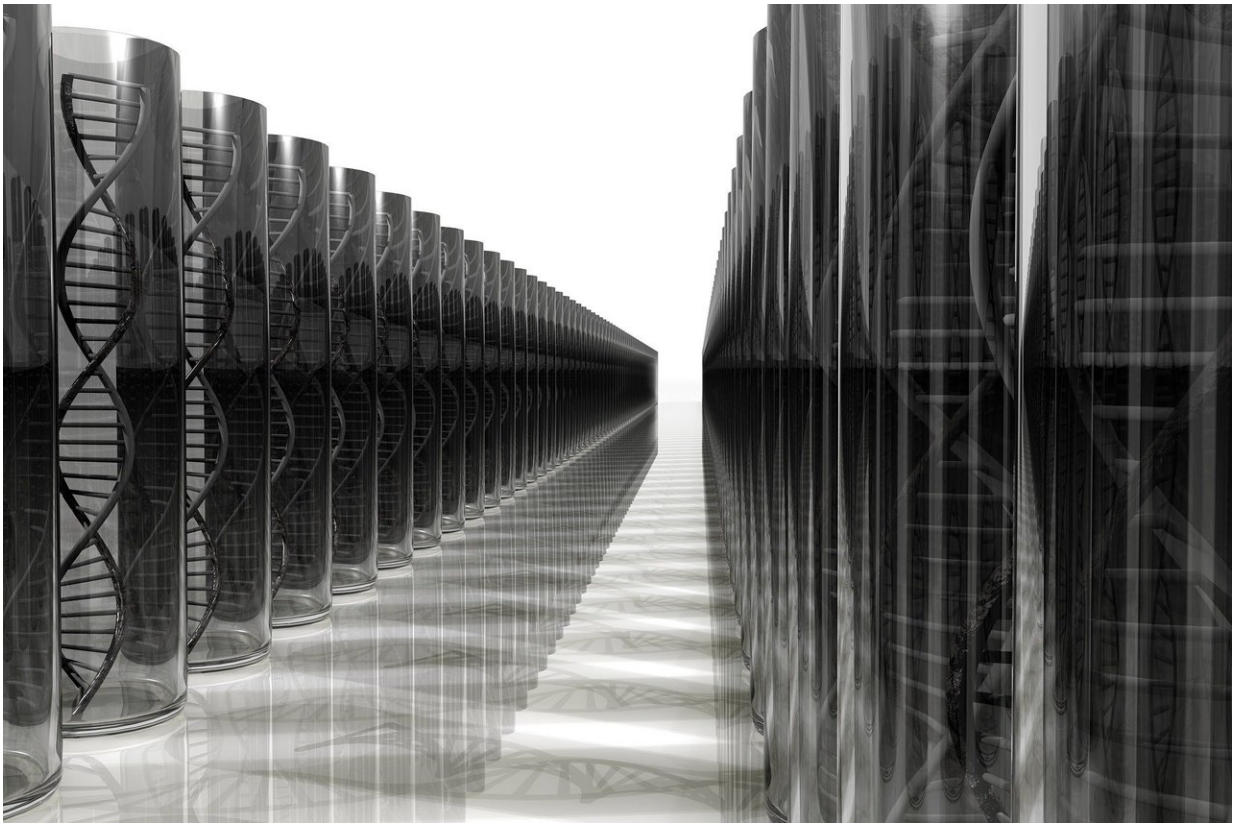


'Consider a number of factors' before buying an at-home genetic testing kit

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Direct-to-consumer laboratories now offer at-home testing kits that allow individuals to mail in a saliva sample and receive a variety of genetic test results. These at-home kits had an unprecedented year in

2017. According to MIT Technology Review, more people took genetic tests last year than in all previous years combined. Research has estimated that the market for genetic health testing could nearly triple—from \$99 million in 2017 to \$310 million in 2022.

Even though direct-to-consumer testing kits are topping many holiday gift guides for 2018, the kits come with caution from the clinical genetics community. Many of the concerns are related to interpretation, privacy and accuracy of results. VCU News spoke with Tahnee Causey, assistant director of the Virginia Commonwealth University School of Medicine's genetic counseling program, about [genetic counseling](#) and direct-to-consumer testing kits.

What is a genetic counselor?

A genetic counselor is a medical professional trained to help individuals who have a genetic condition or may be at risk for one. By testing for and interpreting complex genetic information, genetic counselors help families adapt to a diagnosis or an inherited trait in themselves or their relatives.

A medical geneticist is a doctor who performs physical exams for genetic conditions and works with other [medical professionals](#) to make recommendations about management and treatment.

While physicians, nurses and other health care partners receive education and exposure to genetics, genetic counselors and medical geneticists are the most qualified to provide testing due to the complexity and rapid evolution of testing and our understanding of genetics.

For example, there are more than 50,000 genetic tests that can be ordered worldwide, compared to just a little more than 1,000 tests just

six years ago. Genetic counselors and medical geneticists can help patients decide which [test](#) to order and interpret results.

How do labs determine familial relation?

Each individual has a unique genetic "blueprint." The closer the degree of relationship between two individuals, the more common genetic markers these two will share. Therefore, a parent and child will share more genetic markers than do first cousins.

Labs store the "genetic fingerprints" obtained from people tested in large computer databases. By determining the percentage of shared genetic information, the lab can show a patient other family in the database and determine the level of relationship.

Some laboratories allow patients to opt in or opt out of relationship testing. We encourage consumers to think carefully before they choose to receive relationship information. It is important to understand that if someone opts in, they may learn new information about themselves and their biological relationships. For example, siblings they may not be aware of due to alternative parental relationships (different fathers or mothers).

What about tests that can determine ancestry?

One of the most popular products in direct-to-consumer testing is ancestry testing. By using a variety of genetic markers, a lab can determine an individual's ancestral roots. For example, the lab can establish that an individual is Northern European, African, Jewish or Asian. In some cases, more specific markers can provide even more specific countries of origin. Some labs will even determine how much Neanderthal DNA an individual has!

Is there anything a buyer should be aware of when purchasing a direct-to-consumer genetic test?

We encourage our patients to consider a number of factors. First, many of these labs do not have any genetic counselors on staff, which means that an individual may have to decipher complex genetic results on their own.

Second, some labs encourage consumers to upload their genetic information into publicly accessible databases. This upload again forces an individual to interpret these results independently, and often incorrectly.

Third, many labs perform genotyping. This testing looks only for specific changes in a gene and does not sequence or read the gene in its entirety. These results may not provide a comprehensive risk analysis. Without a genetic counselor or clinical geneticist helping with the interpretation, most consumers would be unaware of a possible risk.

Fourth, many direct-to-consumer laboratories collect a great deal of medical information in the forms completed at the time of sample submission. These labs ask for permission to save an individual's DNA when testing is complete. Many labs continue to do research on the saved DNA samples, and occasionally, they may learn valuable information about a genetic trait or condition that could be medically important. In these circumstances, labs are not obligated to re-contact the patient or share profits made from a genetic discovery.

Finally, there is emerging medical literature that suggests the results obtained from direct-to-consumer labs may not be accurate and are not validated in a regular clinical genetics lab.

Is there anything VCU Health can offer that an at-home genetic testing kit cannot?

Yes, genetic counselors. VCU Health has 15 genetic counselors and two clinical geneticists on staff who provide services in all areas of genetic medicine, including prenatal/preconceptional, cancer, pediatrics, adults, neurology, cardiology, ophthalmology, hematology and metabolics.

Provided by Virginia Commonwealth University

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