

What your patients must know about direct-to-consumer lab tests

NOV 20, 2018

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More than 12 million people have had their DNA analyzed with direct-to-consumer (DTC) genetic genealogy tests, according to estimates from the industry, with use of tests from companies such as AncestryDNA doubling in 2017. These and other DTC laboratory tests—often conducted without the involvement of a physician, with results reported directly to the patient—may lead patients to potentially harmful misunderstandings.

The AMA offers a helpful explainer on direct-to-consumer genetic testing. Results of these kinds of tests can be challenging to interpret, the AMA says.

“A positive result does not always indicate a clinical diagnosis. Instead, it may indicate an increased risk for developing a disease or condition. Similarly, a negative result is not indicative of the absence of disease risk. These concepts can be difficult for consumers to understand without a physician or genetic counselor to fully explain them,” the AMA website says.

The concern is not just about DTC genetic testing, as explained in a *JAMA* Viewpoint article by physician-attorney Kimberly Lovett Rockwell, MD, JD. She cited industry estimates that the value of the DTC testing market will reach \$350 million by 2020.

Most DTC testing companies “offer these tests widely to the public without any reference to evidence-based guidelines or the appropriateness of testing in their advertisements,” the *JAMA* Viewpoint says. “Moreover, advertisements tend to entice consumers by appealing to fears of contracting common disorders such as cardiac disease, stroke and various cancers. Most of these medical tests, however, are of low or negative value for a large segment of the consuming public.”

The AMA has communicated with the Food and Drug Administration about the need for vigilant oversight of DTC laboratory testing, and continues to work closely with the FDA and Congress to ensure appropriate oversight of clinical testing services.

The AMA advocates modernization and reform of the oversight of clinical laboratory testing, including DTC testing. The Association has provided technical support concerning various legislative reform models that address the need for appropriate oversight of DTC testing.

If your patient asks about DTC testing options, you should talk with them about the risks and benefits of these tests, along with the risks of interpreting such test results without input from you or another qualified health professional.

Among the risks are that DTC lab tests may:

- | Overstate the impact of particular findings, for which little or no evidence may be available, to support questionable recommendations such as those regarding nutritional choices.
- | Not be set up with the appropriate sensitivity and specificity needed for population screening, which raises the potential for a high rate of false-positive results, and may not cover all risk factors for the condition.
- | Avoid regulatory oversight, particularly when lacking health or medical claims.

Because of these and other limitations, DTC results should not be used for diagnosis or treatment decisions, and should not be used as a substitute for a physician's clinical experience and guidance.

Experts at the Centers for Disease Control and Prevention's Office of Public Health Genomics have laid out other potential harms of consumer genetic testing and noted the critical questions that should be asked. The CDC's message to consumers? "Think before you spit."

DTC genetic testing should be distinguished from the techniques and innovations in genetics and personalized medicine that can dramatically improve patient care. Explore how genetics and personalized medicine lets physicians deliver treatment and prevention options based on a patient's genome sequence, health history and lifestyle.