# How physicians should follow up on direct-to-consumer genetic tests

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Primary care physicians are increasingly asked by patients to interpret and follow up on the results of direct-to-consumer (DTC) genetic tests, placing significant new demands on their time and their skills. Two physician experts provide insights into these challenges and describe how physicians may look to traditional practice guidelines to help patients understand and respond to DTC genetic testing results.

Following are highlights from an article published in the *AMA Journal of Ethics*® (@JournalofEthics) by Kyle B. Brothers, MD, PhD, a pediatrician and bioethicist, and Esther E. Knapp, MD, MBE, a pediatric hematologist-oncologist, both at the University of Louisville School of Medicine.

Using a composite case of a 34-year-old woman who received test results indicating increased risk of disease and carrier status of a pathogenic gene variant, the authors outline the top-level concerns with DTC genetic testing and provide a plan for responding to requests for counseling and follow-up diagnostics.

## Potential problems

One significant implication of the growing popularity of DTC genetic testing is the demand it puts on physicians to simply understand the results.

“While primary care clinicians typically possess the necessary history and physical exam skills, physicians typically do not have sufficient expertise to interpret and assess risk conferred by individual genetic variants and to develop either a diagnostic or a surveillance program tailored to a patient’s particular needs,” the authors wrote. “Even subspecialty-trained physicians can feel reluctant to interpret such test results.”

Moreover, counseling on DTC genetic testing results typically requires time-intensive visits, which most primary care practices are not set up to provide.

In addition, “DTC genetic testing companies vary widely in their laboratory practices, including which genotyping technologies they use and the techniques used to validate results,” the authors noted, citing a study that showed 40% of genetic variants identified in DTC laboratories were not confirmed using a more rigorous testing method.

## How to respond

“If there is a first rule of medicine, it is that physicians should never order a test unless there is a foreseeable benefit from ordering that test. No test is completely risk free," the authors wrote, noting that even noninvasive tests carry risks of false positives and false negatives. “Because so many physicians strive to prevent harms to their patients by following this rule, DTC genetic testing results can seem out-of-place in clinical contexts.”

But while physicians may be tempted to either disavow an obligation to discuss DTC genetic test results or try to convince patients to ignore the results, this can put patients at risk of harm by discouraging them from revealing DTC testing or treatments they are receiving.

“For the present, then, primary care clinicians will need to be aware of what they do not (indeed, cannot) know about genetic testing,” the authors wrote. “They can initially respond to patients’ requests for counseling by explaining possible quality problems with DTC genetic testing and welcoming their questions. For now, most primary care clinicians should refer their patients to appropriate experts to interpret and further evaluate DTC test results to ensure their patients receive the best care possible.”

However, the current workforce of geneticists and genetic counselors is insufficient to meet the projected demand for counseling, the authors added.

“The day will soon come, then, when practical constraints will force many primary care clinicians to learn more and begin counseling patients about DTC genetic results without involving genetics specialists,” they wrote.

The?September 2018 issue?of *AMA Journal of Ethics* further explores, in print and podcast, ethics in precision health.

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