# At-Home Genetic Testing: The Good, The Bad, and The Ugly

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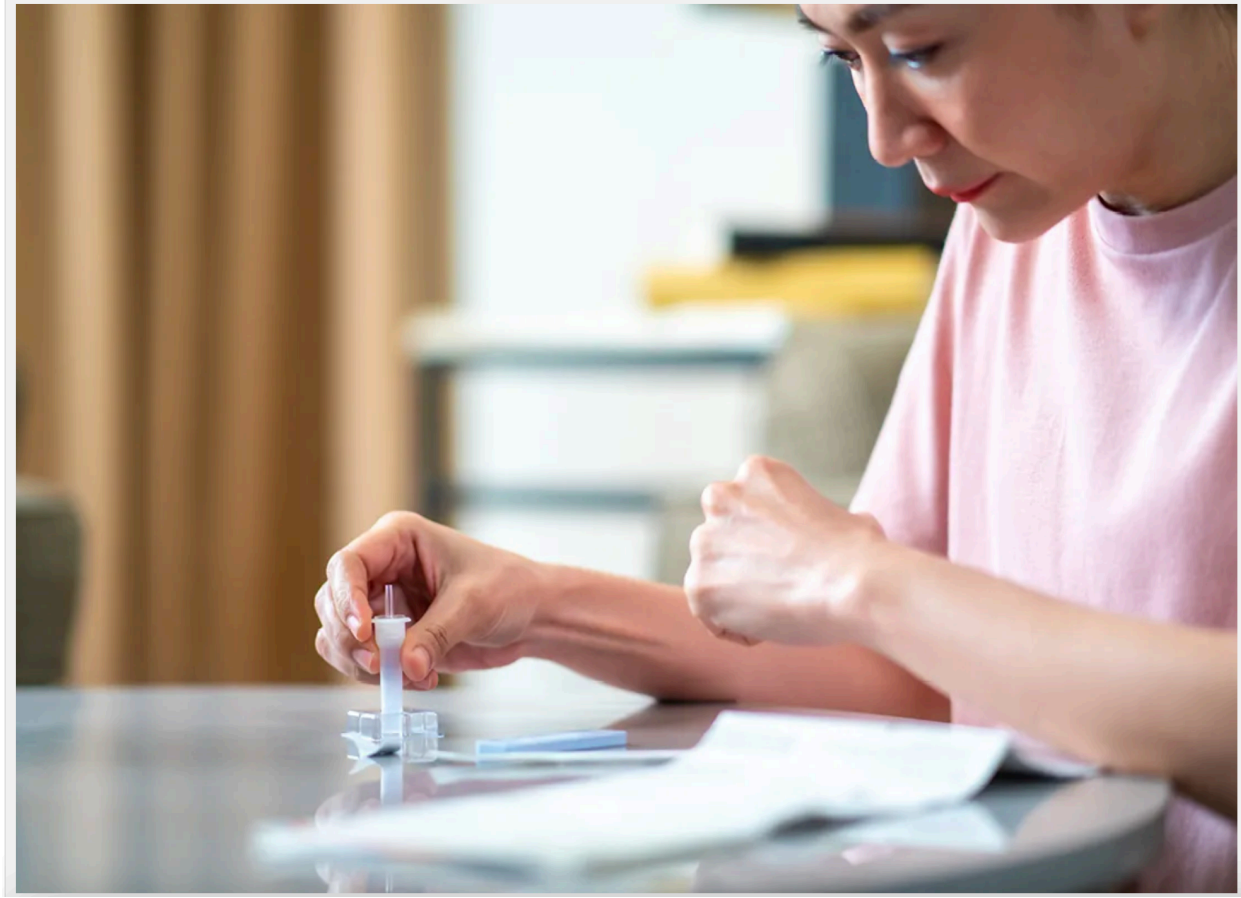
Direct-to-consumer (DTC) genetic testing has risen in popularity in recent years—but is it all it's cracked up to be?



Genetic testing is a powerful tool in the arsenal of modern medicine, offering valuable insights into our genetic makeup, predispositions to certain diseases, and potential future health risks. These tests have traditionally taken place in clinical settings with professional interpretation and proper patient support.

However, with the emergence of direct-to-consumer (DTC) genetic testing companies in the early 2000s—and an estimated market size of $1.98 billion as of 2023[1]—genetic testing for disease risk is now accessible from the comfort of one's home.

For genetic testing labs, DTC will either come as a challenge or a business opportunity depending on local market conditions and the awareness of laboratory leaders. Observant lab professionals will note the good, bad, and ugly about DTC testing as they determine what those factors mean for their laboratories.

## The good: Greater access to genetic testing options

DTC offers a huge advantage for consumers: People can order tests online, collect the samples at home, and receive their results within a few weeks.

"This is the first time in human history that we have access to our genetic data essentially at the push of a button," says Bethany Zettler, a genetic counselor at Brigham and Women's Hospital.

This convenience can give people more autonomy over their health information and decisions—from proactively changing their lifestyle habits to accessing further testing or care.[2] Furthermore, DTC testing is often more affordable than tests ordered through a clinic and results are returned faster.

For those who don't want to undergo invasive testing to learn more about their genetics, DTC testing can be an inviting route because samples are usually collected through non-invasive methods—such as a saliva sample that the consumer then mails back to the lab—rather than a blood test at the hospital.

## The bad: Potential false hope from DTC results

This democratized access to genetic information can be "empowering," says Zettler, but DTC genetic testing results can also impact individuals in other ways. "Some people learn about serious health risks outside of the medical mainstream without built-in support or follow-up, while others might dive deeper into their raw data and explore findings that are not necessarily valid but can be confusing and alarming."

While the presence of a single genetic marker does not guarantee a patient will develop a disease in their lifetime, the absence of one does not guarantee they won't develop it, either. DTC results are not conclusive and do not consider lifestyle and environmental factors, leaving individuals to interpret results themselves or seek out a genetic counselor.[3]

Conversely, patients who undergo clinical testing at a preventive genomics clinic receive support from genetic counselors who guide them through the process, interpret the results within the context of the patient's medical and family history, and provide support in light of the results.

"We have already met with them to discuss risks, benefits, limitations, and potential results of testing, and once the results are available, we know they are medically valid and can be used to inform care," Zettler says.

## The ugly: Inconsistencies across the board

DTC companies may test for different variants or use various techniques to analyze samples, which could lead to discrepancies in the results reported by different companies for the same individual. Furthermore, unlike clinical genetic tests, which are subject to rigorous validation and quality control processes, the certifications for quality and accuracy can vary across DTC labs (23andMe remains the only company with FDA authorization for certain genetic tests[4]).

According to Zettler, DNA microarray testing is the most commonly used test for genotyping in DTC labs, but she says it is "one of the cheapest and least comprehensive options."

"Microarray testing does not screen for any new or unique variants and is not clinically interpreted by a human—it's a computer output," she continues. "Some DTC labs are validated to report certain variants—meaning they have shown they can reliably detect these findings—and some of those variants can have important implications for health. However, these results are not intended for medical management, and both the FDA and DTC labs recommend that results be clinically confirmed."

A study from 2018 showed that 40 percent of genetic testing results from DTC companies were false positives, and some genetic variants that were labeled as "increased risk" in the DTC raw data were actually considered benign by several clinical laboratories.[5] The study's sample size was modest—which 23andMe employees highlighted when they criticized the paper[6]—and genetic testing technology is rapidly evolving and improving as time goes on, but it demonstrates the importance of confirming DTC raw data variants in clinical laboratories.

Until 2023, 23andMe was only authorized by the FDA to test for three mutations in *BRCA1/2*.[7] These mutations are most common in the Ashkenazi Jewish (AJ) population but are rare in the general population, and researchers have shown that DTC testing of only three *BRCA1/2* founder mutations misses over 90 percent of *BRCA1/2* pathogenic or likely pathogenic variants in people of non-AJ ancestry and around 10 percent of variants in people of AJ ancestry.[8] In 2023, the FDA authorized 23andMe to test for 41 more *BRCA1/2* variants, but the agency stated that these variants "do not represent the majority of the *BRCA1/BRCA2* variants in people of most ethnicities."[9]

According to Zettler, clinical laboratories have more safeguards against these types of issues. "Clinical labs use a wide range of appropriate tests to analyze the DNA in various ways, depending on what the clinician recommends and what the patient is motivated to learn," she says. "DTC testing is like skimming the SparkNotes of your DNA, while clinical testing is like reading the whole book."

## What's the solution for labs?

Accuracy, expert support, and accessibility are the key foundations to genetic testing—but is there a way to achieve all three? Zettler says that telemedicine—which has increased significantly since COVID-19—could increase accessibility for patients unable to travel to clinics.

"Additionally, some labs offer consumer-initiated, physician-mediated testing, which is similar to DTC testing whereby the individual can order genetic tests online; however, a key difference is that they can access clinically valid testing with a medical professional reviewing the order for appropriateness," she says. "This approach helps expand accessibility, but the ordering provider isn't directly involved in the patient's ongoing medical care, so there is no built-in support or follow-up."

Additionally, policymakers are stepping up, such as with the proposed Access to Genetic Counselor Services Act in the US Senate.[10]

"This bill would drastically expand access to genetic testing and support for the public by formally recognizing genetic counselors as healthcare providers and allowing for direct reimbursement by Medicare," says Zettler. "This recognition would help reduce healthcare disparities by ensuring that more individuals, especially those with limited resources, can benefit from genetic counseling and testing."

Based on these developments, genetic testing labs face a series of deliberations when it comes to DTC:

- If laboratories see DTC testing as a challenge to their market position, they must figure out a way to combat consumer fascination with at-home testing. This aspect is tricky as more and more healthcare services migrate from the waiting room to the living room.
- If labs view DTC testing as a business opportunity, they must determine how to best market genetic testing services to consumers who have questions about the at-home results. In this way, DTC might be considered a way to drum up new business.
- If genetic testing labs support laws that allow greater reimbursement for these tests, they must get in front of their representatives and senators to voice that opinion.

There's no doubt that at-home genetic testing has increased accessibility to genetic information, but this should not come at the cost of accurate results and proper follow-up support. The public's interest in DTC genetic testing will only continue to grow—and companies will need to be more transparent about the purpose and clinical significance of their tests while providing the information that will help patients respond appropriately to their results.

## References:


1. Precedence Research. Direct-To-Consumer Genetic Testing Market Size, Share, and Trends 2024 to 2034. April 2024. https://www.precedenceresearch.com/direct-to-consumer-genetic-testing-market.

2. Oh B. Direct-to-consumer genetic testing: advantages and pitfalls. *Genomics Inform*. 2019;17(3):e33. doi:10.5808/GI.2019.17.3.e33.

3. Marzulla T et al. Genetic counseling following direct-to consumer genetic testing: Consumer perspectives. *J Genet Couns*. 2021;30(1):329–334. doi:10.1002/jgc4.1309.

4. U.S. Food & Drug Administration. Direct-to-Consumer Tests. December 20, 2019. https://www.fda.gov/medical-devices/in-vitro-diagnostics/direct-consumer-tests#list.

5. Tandy-Connor S et al. False-positive results released by direct-to-consumer genetic tests highlight the importance of clinical confirmation testing for appropriate patient care. *Genet Med*. 2018;20(12):1515–1521. doi:10.1038/gim.2018.38.

6. Wu S et al. Addressing the accuracy of direct-to-consumer genetic testing. *Genet Med*. 2019;21(3):758–759. doi: 10.1038/s41436-018-0094-5.

7. U.S. Food & Drug Administration. FDA authorizes, with special controls, direct-to-consumer test that reports three mutations in the BRCA breast cancer genes. March 6, 2018. https://www.fda.gov/news-events/press-announcements/fda-authorizes-special-controls-direct-consumer-test-reports-three-mutations-brca-breast-cancer.

8. Desai NV et al. Retrospective Cohort Study on the Limitations of Direct-to-Consumer Genetic Screening in Hereditary Breast and Ovarian Cancer. *JCO Precis Oncol*. 2023;e2200695. doi:10.1200/PO.22.00695.

9. U.S. Food & Drug Administration. FDA Roundup: September 1, 2023. September 1, 2023. https://www.fda.gov/news-events/press-announcements/fda-roundup-september-1-2023.

10. National Society of Genetic Counselors. Access to Genetic Counselor Services Act Organization Support Letter. June 2024. https://www.nsgc.org/POLICY/Federal-Advocacy/Bill-Letter-Signature.