



Article: *In-Depth Confirmatory Testing, Insurance Coverage Growing Alongside DTC Genetic Testing* has been updated.

Confirmatory Testing, Insurance Coverage Growing Alongside DTC Genetic Testing

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NEW YORK (GenomeWeb) – Recognizing the growing popularity of direct-to-consumer (DTC) genetic testing services like 23andMe, clinical labs are being asked to verify results that indicate individuals have an increased risk for diseases like cancer, and some commercial insurers are even paying for this confirmatory testing.

A company called My Gene Counsel this week launched a pilot program that will provide education, facilitate genetic counseling, and if needed, offer confirmatory testing to individuals with a personal or family history of cancer who have taken a DTC cancer risk test but still have unanswered questions. My Gene Counsel is partnering with telegenetics firm GeneMatters to provide counseling and clinical testing lab Ambry Genetics for confirmatory testing.

The launch of the multi-pronged program also coincides with a growing willingness among insurers to pay for confirmatory testing for those who have gotten DTC testing in certain instances. So far, at least three insurers, Blue Shield of California, Anthem, and Aetna, have updated their BRCA testing policies to cover this repeat testing.

These are the latest examples of how the healthcare system is adapting to consumers' growing appetite for genetic health risk information. "We know that these tests yield meaningful information and that healthcare providers can no longer afford to brush these aside and say, 'Throw them in the trash,'" said Ellen Matloff, an experienced genetic counselor and founder of My Gene Counsel. "We're trying to build that bridge back."

'This must change'

Genetics experts began recognizing the need for confirmatory testing services a year ago, soon after [23andMe garnered](#) US Food and Drug Administration permission to sell a test directly to consumers that gauges three mutations in BRCA1 and BRCA2 genes associated with high risk of breast, ovarian, and prostate cancers and that tend to show up in individuals of Ashkenazi Jewish descent. Since then, the company has also gotten [FDA clearance](#) for its DTC test for two MUTYH variants associated with high risk of a colorectal cancer syndrome, but hasn't launched this test yet. However, the agency's authorization for these tests comes with the caveat that the results shouldn't be used to make medical decisions without confirmation by a clinical grade test.

Although 23andMe doesn't provide genetic counseling as part of its service, the online health risk reports contain language highlighting that these tests gauge a limited number of variants associated with cancer

risk, that individuals with positive or negative reports should get confirmatory testing before taking medical action, and that individuals with a family history should consider other kinds of tests. Despite this language, genetic counselors and patient advocates have continued to be concerned that customers of DTC testing companies are not receiving the support they need to fully understand what they're getting and not getting from these services.

They are particularly worried about individuals with a strong personal or family history of cancer receiving false-negative results from DTC testing and not realizing that they are still at risk. "That subsection of the population is not receiving the focus," said Matloff.



Screenshot of My Gene Counsel video explaining why after DTC genetic testing some people may need confirmatory testing.

DTC testing can also yield false positives especially when customers of companies like 23andMe or Ancestry.com upload their raw data files to data-interpretation services, such as Promethease, which analyze those files and reports disease-linked SNPs from the literature.

Researchers at Ambry [published a study](#) last year in *Genetics in Medicine* involving 49 customers of a DTC testing firm or a data interpretation service, and reported a 40 percent false-positive rate. With respect to BRCA1/2, specifically, Ambry confirmed all nine cases where DTC testing identified one of the three Ashkenazi Jewish founder mutations, and also confirmed four other BRCA1/2 variants, but could not confirm eight variants in these genes.

[Another study](#), involving around 100 patients who got confirmatory testing after DTC screening from molecular testing lab Invitae, reported a false-positive rate of around 50 percent. In this study, around two-thirds had learned their risk through a DTC testing company and one-third had submitted their data to a data interpretation service.

In response to these studies, 23andMe has defended that its tests are well validated and FDA authorized. The company has also been highlighting the stories of its customers who have learned genetic health risk

information from DTC testing and have been able to act on it. 23andMe has also pointed out that these studies include customers of DTC companies and third-party data interpretation services, and has distanced its services from the latter.

However, many customers of 23andMe and other DTC testing companies turn to data interpretation services thinking they can learn even more about their health risks, but open themselves up for more confusion and false findings. The studies conducted by Invitae and Ambry point to the limitations of both types of services and need for confirmatory testing.

"We know it's not a black and white situation. You can get really good information [from DTC tests] that could save your life," Matloff acknowledged. "But there are also a fair amount of these tests that prove to be false positive. And so, we just need to find a quality, reliable way to separate the two."

Matloff recently recounted in *Forbes* her experience uploading her raw data file from a genetic genealogy testing company to a third-party data analysis service and receiving a false-positive result indicating she had a pathogenic mutation associated with Lynch syndrome. As an experienced genetic counselor who had counseled many people with hereditary cancer syndromes, she knew her family history didn't precisely align with Lynch syndrome, which increases the risk of colorectal, uterine, ovarian and other cancers. She suspected this may likely be a false positive, and yet, upon seeing the result "calm, cold panic set in," she wrote.

With the help of a genetic counseling colleague she navigated the process to verify her result, which indeed turned out to be a false positive. "I learned a lot from having Lynch syndrome for 30 hours. I learned, firsthand, how terrifying genetic testing can be, even when you're doing it just for kicks," she wrote. "I learned how challenging it can be for even a certified genetic counselor to navigate the pathway to verify a consumer test result in a medical-grade laboratory. This must change."

Carving a path

Matloff is hoping to bring about this change through the confirmatory testing program she has launched with Ambry. There is a need for this type of service, since the Konica Minolta subsidiary has noted an uptick in samples coming in for confirmatory testing following DTC testing.

Color is another lab that [launched a program](#) last year for individuals who have a positive BRCA test results from a DTC company. Qualifying customers can get tested on Color's \$250 panel test that gauges 30 cancer-linked genes, including BRCA1/2, for \$50.

Although there may be more opportunities for confirmatory evaluations after DTC testing, there are gaps in the system that make it hard to measure how prevalent this is and how many people should be tested but are not. DTC testing services are outside the healthcare system, and there isn't an established way of tracking when DTC testing customers are coming back into the healthcare system to verify their results after learning their risks for cancer and other diseases online. And there is no tracking of DTC testing customers who should come in for verification testing and further counseling but are not, which means that a subset of people may be missing opportunities for early diagnosis and disease prevention.

Following the FDA's authorization of 23andMe's BRCA test a year ago, several groups began thinking about how to advance mechanisms and resources to ensure that those who need follow up medical services receive it. The National Society of Genetic Counselors and another group called the Center for Genomic Interpretation [began urging insurers](#) to update coverage policies to include confirmatory testing for those who have gotten a positive result from 23andMe's FDA authorized BRCA test.

The concerns of stakeholders struck a chord with Blue Shield of California, according to Stephanie Gandomi, who is a genetic counselor and manages the insurer's clinical genetics programs. In its [most](#)

[recent BRCA testing coverage policy](#), updated in January, Blue Shield of California states that confirmatory testing is "medically necessary" for individuals who have a pathogenic BRCA1/2 mutation from an FDA approved, over-the-counter genetic test (i.e. 23andMe).

"We felt we needed to be in alignment with the NSGC's position and the FDA," Gandomi said. "We felt this was the most ethical and member-centered approach we could take as a health plan. It was an easy decision for us." Insurers typically prefer providers use in-network labs when testing patients, and Blue Shield of California's in-network labs that could provide confirmatory BRCA testing include Ambry, Invitae, Myriad Genetics, GeneDx, Color, Progenity, Laboratory Corporation of America, and Quest Diagnostics. Gandomi noted out-of-network labs that are CLIA certified could also do the confirmatory testing.

Aetna, meanwhile, [now covers](#) confirmatory testing for people who have tested positive for the three BRCA mutations by 23andMe's test. Anthem's [test request form indicates](#) it will cover confirmatory testing for individuals with a family history of cancer who have had a positive result from an FDA-authorized BRCA DTC test.

Matloff recognized insurers' updated policies supporting confirmatory testing as a step in the right direction. "We want to carve a path so that direct-to-consumer [testing] customers who need repeat testing can get the testing they need and their insurance companies will pay for programs like ours in the future," she said.

My Gene Counsel's offering is currently aimed at individuals who have positive or negative results following a DTC cancer risk assessment. However, only those who also have a strong personal or family history of cancer will get a free report from My Gene Counsel that explains their DTC test results in greater detail, the difference between a DTC test and a clinical grade genetic test, and why verification of a DTC test result is necessary. These individuals will also receive a free 30-minute pre-test genetic counseling session from GeneMatters.

Following an evaluation of an individual's personal and family history of cancer, a genetic counselor will recommend confirmatory testing if needed. "We're not going to recommend testing for everybody," Matloff said, adding that two customers have already gone through the evaluation process and been told they don't need confirmatory testing.

But if a counselor does recommend testing, then the patient can take Ambry's test requisition form and ask her own doctor to order the verification analysis or pay \$50 to a telemedicine company, called InteleLabs, to order the testing. Based on the confirmatory test results, My Gene Counsel will issue a report to the patient to explain the findings.

Currently, commercial insurers pay for testing for cancer risk genes, such as BRCA1/2, when individuals have a personal or family history of cancer, while Medicare covers testing only for people who currently have or had cancer in the past. For this reason, My Gene Counsel's pilot program is starting by focusing on people who meet these criteria, so they are more likely to qualify for covered testing, even if the insurer doesn't have a specific policy around confirmatory analysis.

Cody Buhler, Ambry's director of business development, noted that the company's tests are broadly covered by insurers and four out five patients tested by the lab don't pay anything for testing. Still, in cases where individuals still don't meet coverage criteria, Ambry's cash price is \$399 and the company also has a patient assistance program based on financial need.

For this initial pilot, Ambry is covering the cost of My Gene Counsel's reports to patients and one genetic counseling session. Based on how this effort goes, Matloff hopes that other genetic testing companies, insurers, and wellness groups will similarly partner with My Gene Counsel to launch other confirmatory testing programs.

There seems to be a demand for this. Although the pilot has only been live for only a week, My Gene Counsel has already heard from individuals who have gotten DTC testing for non-cancer indications and want confirmatory testing. As more FDA-approved DTC tests become available to consumers and require confirmation, Gandomi said that Blue Shield of California would certainly consider expanding coverage policies to include verification of results based on a review of the evidence.

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