

# Ambry Genetics and Lake Medical Imaging Increase Access to Cancer Risk Assessments and Genetic Testing

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## Summary

Genetic testing can offer valuable information to help health care providers better tailor treatment plans to individual patients, but some physicians may not have knowledge of the resources available to help them offer hereditary cancer risk assessment and genetic testing. Ambry Genetics' CARE (Comprehensive Assessment Risk & Education) Program™, which uses a comprehensive digital platform, was created to identify more high-risk patients, educate them on their risks for potential inherited cancer, and provide them with the option to undergo genetic testing.

With multiple locations in Central Florida, Lake Medical Imaging has been a leader in mammography for more than 50 years. Lake Medical Imaging's incorporation of Ambry Genetics' CARE Program showed how cancer risk assessment and genetic testing can be efficiently implemented or expanded within a local community to provide comprehensive health information at scale. In the first 12 months of implementing the CARE Program, Lake Medical Imaging successfully screened more than 23,000 patients and 25% met National Comprehensive Cancer Network® (NCCN®) criteria for genetic testing for potential hereditary cancer risk based on NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®).

After implementing the CARE Program, Lake Medical Imaging saw a five-fold increase in the identification of high-risk patients and was recognized with the 2022 Radiology Business Management Association RAD Honors Innovation Award for broadening access to genetic cancer risk assessments and testing for their patients.

This white paper highlights how the CARE Program can make it easier for medical practices to provide their patients proactive cancer screenings, genetic risk assessments, genetic testing and access to genetic counseling.



2022 RAD Honors Innovation Award from the Radiology Business Management Association.

## Introduction: More Access to Genetic Testing is Needed

Early detection of cancer leads to better treatment outcomes and lower costs according to the World Health Organization (WHO).<sup>1</sup> Identifying patients with a predisposition to cancer leads to tailored care plans, including options to manage cancer risk and detect signs of cancer earlier. One way to do this is using genetic testing to screen for hereditary cancer risk. However, genetic testing is often hard to scale. A medical group needs to identify genetic testing candidates, offer testing access, provide patient education, and interpret and apply the test results.

Unfortunately, many patients who could benefit from genetic testing are not offered the chance. It is estimated that 80 percent of women with a history of breast cancer who meet criteria for hereditary cancer testing do not receive genetic testing.<sup>2</sup> Women who harbor a pathogenic variant may have a higher for breast and/or other cancer types. Going untested means they do not have the opportunity to learn of and consider interventions to mitigate these risks.

When genetic testing is not included in a health assessment there can be a detrimental impact on the patient. For example, pathogenic variants in dozens of genes are associated with cancer predisposition: BRCA-related breast and/or ovarian cancer is one of the best-known examples. The National Cancer Institute estimates that 1 in 400 people in the U.S. have a pathogenic variant in the *BRCA1* or *BRCA2* genes, which increases to 1 in 40 for those with Ashkenazi Jewish heritage.<sup>4</sup> Women with a pathogenic variant, or mutation, in the *BRCA1* or *BRCA2* genes have greater than 60% lifetime risk of breast cancer.<sup>5</sup> Men with a variation in the *BRCA1* or *BRCA2* genes are also at risk. Prostate cancer occurs in about 1 in 8 men in their lifetime<sup>6</sup> with hereditary prostate cancer making up about 5 percent of all cases.<sup>7</sup> However, men with a variant in the *BRCA1* or *BRCA2* genes are 7 to 8 times more likely to develop prostate cancer than those without.<sup>8</sup>

## Traditional Approaches to Family History Assessment Challenging

Many health care professionals use personal and family history to screen patients for breast cancer risk. Family histories as provided by patients can be incomplete depending on the circumstances and access to the data, and providers need both resources and time to be able to collect the information and determine NCCN eligibility for genetic testing. These challenges can be compounded when patients are asked to recall and report family histories during appointments. Because CARE assessments are sent to patients ahead of their appointments, they have time to discuss their family histories with relatives, potentially making reported family histories more accurate and complete.

Many of these health care providers want to add genetic information to their current screening process to help them create cancer risk management plans for their patients, but lack the tools and process to accomplish it. Access to genetic testing is a challenge even in insured, higher-income communities, but the challenge is amplified in underserved patient populations. Many would benefit from genetic testing, but are not given the opportunity for a variety of reasons. Ambry Genetics wants to change that.

## Reasons People Don't Pursue Genetic Testing



## Solution: Increasing Access to Genetic Testing Through Ambry's CARE Program

The Ambry CARE Program™ proactively helps identifies patients at increased risk for cancer. It works on a digital platform that allows health care providers to easily provide cancer risk assessment and education on genetic testing, order genetic testing, give the results and provide access to genetic counselors. Genetic testing may include the analysis of genes linked with hereditary predisposition to a variety of cancers, including breast, ovarian, colorectal, uterine, pancreatic, prostate, and stomach cancers, as well as melanoma and others.

Applying a standardized risk assessment platform like the CARE Program enables health partners to equitably offer risk assessment to patients across their system's patient population. Based on internal Ambry data, applying at a rate standardized risk assessment like that offered through the CARE Program enables health partners to consistently offer risk assessment to patients, which is nine to ten times greater than the national average.

## Lake Medical Imaging Integrated the CARE Program

Lake Medical Imaging, a full-service, fully accredited radiology practice with half a dozen busy locations in Central Florida, offers digital mammography with tomosynthesis, automated breast ultrasound, breast MRI, contrast enhanced mammography and breast biopsy. By combining these technologies, Lake Medical Imaging had the highest breast cancer detection rate per 1,000 patients in the Southeastern United States, in recent years.<sup>11</sup> Lake Medical Imaging's goal was to identify more women at increased risk for breast cancer, before they were diagnosed, in order to best tailor breast cancer screening for the individual patient and thereby diagnose more cases of breast cancer at early stages.

Lake Medical Imaging – which produces approximately 200 mammograms each day—implemented the CARE Program in their practice in 2020. To start the implementation, Ambry Genetics and Lake Medical Imaging first met on June 6, 2020. All contracting, workflow assignments, and Information Technology discussions were completed in less than three months and the program launched on September 9, 2020. Adopting the program in the Lake Medical Imaging offices was simplified with materials for provider and patient education. A programmatic approach allowed for streamlined introduction, contracting, implementation and expansion. It also allowed for continued refinement and customization of the workflow to meet practice needs.

When the program launched, Lake Medical Imaging offered a genetic testing panel through the CARE Program that analyzed genes linked to an inherited risk of eight different cancers, including breast cancer. The patient questionnaire also applied the Tyrer-Cuzick (TC) risk model to estimate lifetime breast cancer risk, to provide additional information affecting breast imaging recommendations.

Incorporating a new process into a high-volume workflow required collaboration and communication. The patient process was as follows:

- Patient schedules a mammogram
- A week before her appointment, personal/family history data is collected digitally by CARE through text or email-initiated risk assessment
- An automated risk assessment is performed based on this history. The patient's TC score and NCCN eligibility for genetic testing are determined and provided to the patient.
- The patient receives education through the CARE platform.
- If the patient meets NCCN criteria for genetic testing, the process and cost are explained.
- If the patient elects to proceed, her blood sample is drawn at the time of her mammogram.
- Genetic test results are returned by a provider or the CARE platform.
- Patients are also connected with post-test education, including access to telehealth genetic counseling at no additional cost.
- The practice is provided documentation of each step of the patient journey (clinical summary, virtual assistant transcript, genetic test results, genetic counseling note).

## Results and Clinical Impact

Prior to the implementation of the CARE Program, Lake Medical Imaging did not offer comprehensive breast cancer risk assessment or genetic testing, instead, they relied solely on traditional methods of risk assessment such as reviewing family history. Traditional methods resulted in **fewer than 5%** of patients being counseled about their options for changes in breast cancer screening management due to a potential increased breast cancer risk. They considered other options before choosing to work with Ambry Genetics and the CARE Program. Following implementation of the CARE Program, however, the combined Lake Medical Imaging sites saw nearly a **five-fold** increase in patients who were determined to be at increased risk. This proactive process earned high marks for patient satisfaction and engagement. See Appendix A for the breakdown of statistics.

## Best Practices for Replication

Lake Medical Imaging's risk assessment and genetic testing program provides a pathway for other practices that want to implement a similar program. Their experience showed the speed at which such a program can be installed and the difference it can make within a year. Lake Medical Imaging found success through the following aspects of the CARE program:

- *Use of digital educational materials on the CARE Program for health partners.* The CARE Program provides access to a library of infographics available for website use and practice implementation. (See Figure 1.)
- *Training for clinicians.* The CARE Program provides education to clinicians and practices on the benefits of cancer risk assessment and genetic testing, and how implementing these practices enables individualized cancer risk management for their patients.
- *Affordable testing without significant increases in clinic resources.* In this initiative, genetic testing was shown to be affordable for Lake Medical Imaging patients and did not drain the clinic's resources.
- *Use of technology, including text or email-initiated risk assessment, digital patient education, and telegenetic counseling—especially in rural areas.* Access to the CARE Program via smartphone allowed the patients ease of use. Following genetic testing, patients could access genetic counseling by phone if needed.

Ambry is encouraged by Lake Medical Imaging's results, as they reflect similar trends from other CARE Program partners on a national scale.



Figure 1. An example of supplemental material available for marketing and web use.

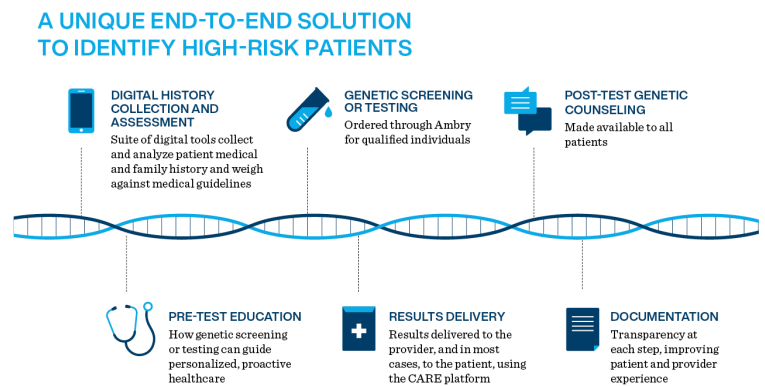


Figure 2. The CARE Program Overview

## Conclusion

The partnership between Lake Medical Imaging and Ambry Genetics shows that offering cancer risk assessment, education, and genetic testing benefits patients and health care partners and is a feasible option for a practice to adopt. This is a health care success story that was recognized and awarded one of the 2022 Radiology Business Management Association's (RBMA) PaRADigm awards. The RBMA RAD Honors Innovation Award was developed "to recognize radiology practices and vendors who have demonstrated the highest degree of creativity, innovation, and exemplary leadership."<sup>12</sup>

Lake Medical Imaging's success is an inspiration: it shows how cancer risk assessment and genetic testing can be efficiently implemented or expanded within a local community to provide comprehensive health information at scale. By partnering with Ambry Genetics and using the CARE Program, more patients can be connected to care that is standardized, proactive, and equitable.

## Appendix A: Comprehensive Statistics of Patient Participation

In the first 12 months of implementing the CARE Program, Lake Medical Imaging successfully screened more than 23,000 patients and more than 25% met NCCN criteria for genetic testing for potential hereditary cancer risk. More than 2,000 patients proceeded with genetic testing, and over 90 were identified as having a pathogenic genetic variant linked with increased cancer risk.

Total invitation to the program was over 30,000 individuals, with 73% completing the digital assessment (23,402). Twenty-eight percent of those who filled out the assessment met NCCN Guidelines® (6,624 patients) criteria for genetic testing, and 14% (949) had a Tyrer-Cuzick score greater than 20. A total of 1,753 moved on to genetic testing, and 6% (90 patients) tested positive for a pathogenic genetic variant. These results were similar to national percentages (see Table 1).

### Lake Medical Imaging CARE Analytics: September 2020 – August 2021

Assessments Sent	Assessments Completed	Met NCCN Criteria	TC Score > 20	Tests Ordered
31,849	23,402 (73%)	6,624 (28%)	949 (14%)	1,907 (29%)

### National CARE Analytics: September 2020 – August 2021

Assessments Sent	Assessments Completed	Met NCCN Criteria	TC Score > 20	Tests Ordered
244,297	174,572 (71%)	53,049 (30%)	17,765 (10%)	15,694 (30%)

Table 1. Lake Medical Imaging Analytics vs. National September 2020 – August 2021

Across the six testing sites, results were also similar, with approximately three-quarters of patients responding to the assessment, approximately a third of those meeting NCCN criteria for genetic testing, and a third of those moving forward with genetic testing. Table 2 shows the breakdown.

### Site by Site Comparison

Site	Assessments Sent	Assessments Completed	Met Criteria for Testing	Genetic Tests Ordered
Brownwood	5,133	4,086 (80%)	1,179 (29%)	454 (39%)
Lake Sumter Landing	5,455	4,063 (74%)	1,088 (27%)	289 (27%)
Mulberry Grove	3,677	2,379 (65%)	729 (31%)	263 (36%)
Colony Plaza	5,950	4,113 (69%)	1,138 (28%)	289 (25%)
Sharon Morse	5,779	4,638 (80%)	1,259 (27%)	322 (26%)
Leesburg	5,855	4,123 (70%)	1,231 (30%)	290 (24%)
<b>TOTALS</b>	<b>31,849</b>	<b>23,402 (73%)</b>	<b>6,624 (28%)</b>	<b>1,907 (29%)</b>

Table 2. Site-by-Site Comparison

Genetic counseling was accessed by over 80% of patients found to have a pathogenic genetic variant. The identification of pathogenic genetic variants led to the following changes in recommended management. (See Table 3.)

**Imaging Management Impact for Positive Patients**

Recommendations	Frequency	# Patients
Clinical breast exam	Q6 months	49
Breast MRI/Contrast enhancing mammography	Annually	49
Risk-reducing mastectomy	Once	19
Risk-reducing Salpingo-oophorectomy	Once	21
Colonoscopy	Q1-5 years	48
Other high-risk screening/surgery	Variable	26

Table 3. Imaging Management Impact for Positive Patients

Ambry is encouraged by Lake Medical Imaging’s results, as they reflect similar trends from other CARE Program partners on a national scale. Since the CARE Program began in 2020, over a half a million patients have been enrolled, with approximately 75% completing the CARE risk assessment, 28% meeting criteria for genetic testing and an additional 10% having Tyrer-Cuzick scores greater than 20, with recommendations for medical management changes. Program benefits are clear when considering the patients who have discovered they have a pathogenic variant linked with hereditary cancer predisposition and those who have been identified as high-risk and referred for advanced screening.

## Appendix B

Visit Lake Medical Imaging’s webpage for the program at

<https://lakemedicalimaging.com/services/womens-imaging/genetic-testing-for-breast-cancer/>.

## Acknowledgments

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## Endnotes

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