Poster #13

Integrating Genetic Testing, Combined Polygenic Risk, and Family History Assessment to Improve High Risk Breast Cancer Identification in a Community Breast Imaging Center

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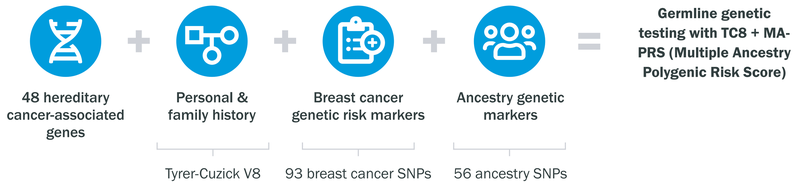
Background

- This initiative evaluated whether the Tyrer–Cuzick v8 (TC) model alone is sufficient for breast cancer (BC) risk assessment compared to a comprehensive strategy incorporating detailed family history intake, germline multigene panel testing, and a combined risk score (CRS) that integrates TC with a polygenic score validated for all ancestries.
- The aim was to determine whether this integrated approach more accurately identifies individuals at increased risk for breast and other cancers in a community breast imaging center and better supports personalized screening and management decisions.

Methods

- Beginning in November 2020, Singing River Health System implemented a comprehensive breast risk assessment program that combined TC modeling, multigene germline testing, and CRS into its breast imaging program.
- During routine mammography, patients completed TC and were screened for genetic testing eligibility based on NCCN guidelines.
- Eligible patients received a telephone-based family history review and genetic education, followed by same-day sample collection after consenting to testing (**Figure 2**).
- Testing included a germline multigene panel and CRS to determine individualized BC risk (**Figure 1**).
- After results were available, a nurse navigator reviewed results and guideline-based recommendations with patients.
- Program outcomes were analyzed to determine clinical management implications and to identify individuals who would not have met high-risk criteria using TC alone.

Figure 1. Multigene Panel and CRS

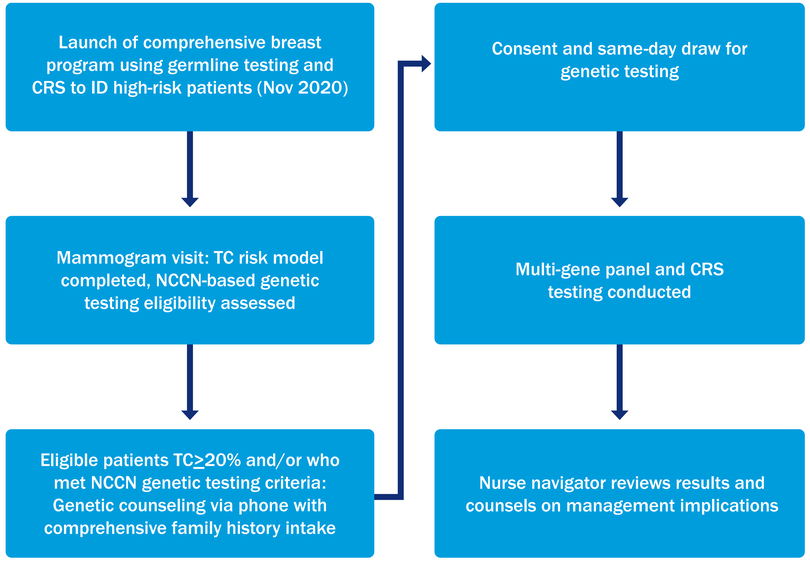


Results

From November 2020 through December 2025, TC results were determined for 3,017 patients; eligible patients underwent germline genetic testing and CRS. Among these, 895 (30%) required changes in medical management despite having a TC-estimated lifetime BC risk <20% as follows:

- 75 (8.4%) carried a pathogenic variant (**Figure 3**).
- 186 (20.8%) had lifetime BC risk ≥20% based on CRS (**Figure 3**).
- 634 (70.8%) required modified management after expanded family history was obtained during counseling (**Figure 3**).

Figure 2. Risk Assessment Clinic Flow Chart



Downstream clinical actions that have the potential for meaningful impact on breast cancer outcomes included:

- 175 (19.6%) risk-reduction counseling visits (**Figure 4**).
- 103 (11.5%) breast MRIs for patients whose CRS revealed a lifetime BC risk ≥20% (**Figure 4**).
- 63 (7.0%) gastroenterology consults resulting in 26 colonoscopies (**Figure 4**).

Figure 3. Patients with TC<20% with Management Changes

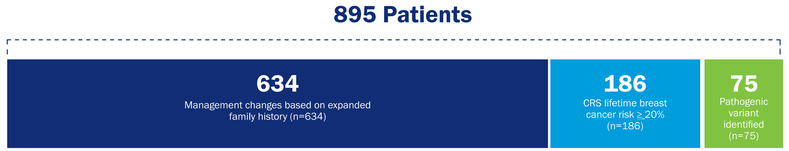
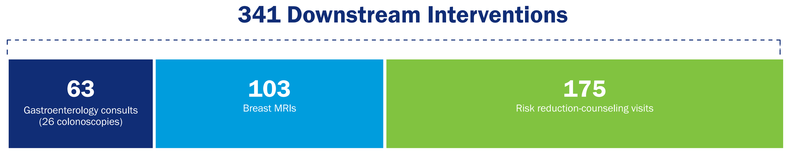


Figure 4. Integrated risk assessment led to meaningful downstream clinical actions.



Conclusions

- A multifaceted risk assessment program integrating TC with CRS and germline genetic testing identified substantially more individuals requiring high-risk management than TC alone.
- This approach revealed patients who benefited from guideline-recommended surveillance, preventive interventions, and specialty referrals, many of whom would have been missed using TC as a standalone assessment.
- These findings support the value of integrated genetic, polygenic, and family history-based evaluation within community imaging centers.

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