Implementation of Universal Germline Hereditary Cancer Genetic Testing for Targeted Therapy and High-Risk Screening

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Introduction

- Patients referred for germline genetic testing (GT) have traditionally been selected using guidelines based on personal and family history of cancer, age, and ancestry.
- Current guidelines may miss many people with pathogenic variants (PV) that can benefit from associated management and treatment recommendations.
- Lack of awareness of GT guidelines by referring healthcare providers also contributes to missed opportunities for patients.
- Miami Cancer Institute launched a pilot to evaluate the implementation and outcome of a universal germline hereditary cancer GT model integrated into a new patient intake at a hybrid community-academic cancer center.
- Effectiveness in identifying individuals with PV who may not meet GT criteria was assessed.

Methods

- Data was collected from Dec 1, 2024, to August 31, 2025.
- All new patients at the hybrid cancer center received a pre-visit link to complete a genetics questionnaire, review education, and consent to testing.
- Eligibility included adults ≥18 years old with no bone marrow transplant, no active hematologic malignancy, or prior GT after 2013.
- A genetics software platform automated the workflow, including cancer risk assessment (CRA) generation, test ordering, and result delivery.
- Patients meeting traditional criteria were informed of likely insurance coverage; others were advised of potential self-pay.
- Blood samples were collected post-visit, and testing was performed using a 76- or 90-gene panel.
- Results were integrated into the electronic medical record and disclosed via phone or platform, depending on result type.

Results

Universal GT Workflow: December 2024 – August 2025

7740 Patients Contacted

6084 (79%) Activated Link

4859 (80%) Completed Cancer Risk Assessment

250 (5%) Ineligible for GT

4609 (95%) Eligible for GT

1657 (36%) Consented

1188 (72%) GT Ordered

1130 (95%) Results Returned

80 (7%) P/LP

45 (4%) Carrier

346 (31%) VUS

659 (58%) Negative

Figure 1. Pre-Visit Questionnaire and Education

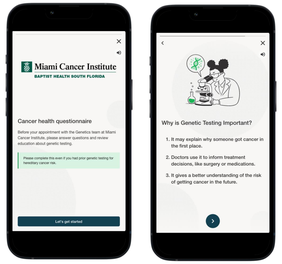


Figure 2. Return of Results

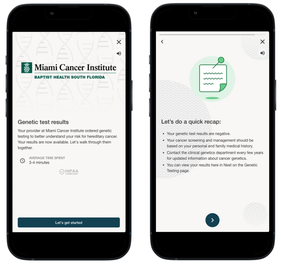


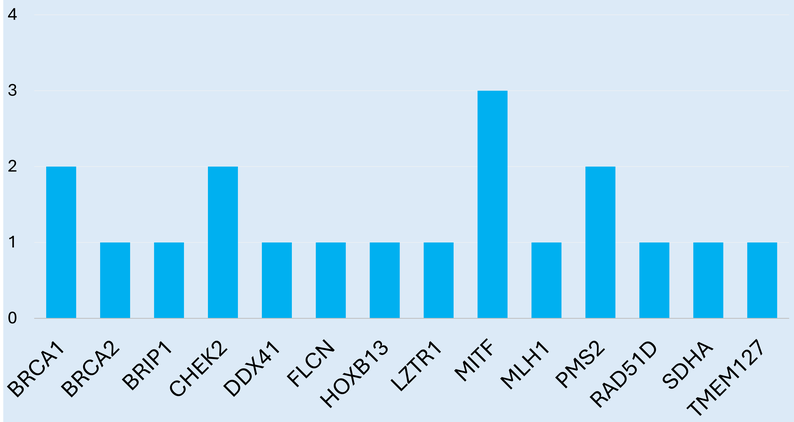
Table 1: Demographics of Patients Completing Pre-Visit Questionnaire

Sex	Female	3209 (66%)
	Male	1649 (34%)
	Other	1 (<1%)
Race	Non-Hispanic White	1116 (23%)
	Non-Hispanic Black	247 (5%)
	Hispanic/ Latino	2841 (58%)
	Other	655 (13%)
Age	18-39	696 (14%)
	40-59	1724 (35%)
	60+	2439 (51%)
Language	English	3729 (77%)
	Spanish	1130 (23%)

Table 2: P/LP Results by Affected Status and GT Criteria Met / Not Met

P/LP	Affected, met criteria	41 (51%)
	Unaffected, met criteria	21 (26%)
	Affected, criteria not met	10 (13%)
	Unaffected, criteria not met	8 (10%)

Figure 3. Frequency of Gene Identification in Positive Patients Not Meeting GT Criteria



Conclusion

- These findings highlight a significant gap in screening practices and support the need for more inclusive testing strategies to improve access to precision oncology and preventative care.
- With the right workflow, universal germline genetic testing is both feasible and effective in identifying at-risk individuals missed by guideline-based approaches.
- Purpose-built software can provide the automation and coordination necessary to implement this type of workflow at scale.
- Future study directions include addressing the gap between patients who consented and those for whom genetic testing was ultimately ordered by adding reminders for sample collection and improving data collection to minimize excluded patients.
- Further data review will help identify opportunities to optimize follow-through and strengthen the service delivery model.

References

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