

ABSTRACTS

Patterns and Predictors of Germline Testing Completion in Early-Onset Colorectal Cancer: Underuse, Delayed Testing, and Racial Differences in a Multihospital Cohort

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Objectives

Germline testing is recommended for patients with early-onset colorectal cancer (EOCRC), but real-world completion and pathway gaps are incompletely characterized. We evaluated germline testing completion, timing, hereditary evaluation milestones, and predictors of testing completion in a multihospital cohort.

Methods

We conducted a retrospective cohort study of patients younger than 50 years who were diagnosed with colorectal adenocarcinoma and seen within a multihospital health system in Washington, DC, from 2020 to 2025. Patients were identified through the electronic medical record and were required to have at least 2 visits within the selected clinics. The primary outcome was completion of germline genetic testing. Secondary outcomes included documented genetic referral, completion of genetic counseling, timing of testing, recorded germline results, and demographic and clinical factors associated with testing completion. Categorical variables were compared using Fisher exact tests, and continuous variables using Wilcoxon rank-sum tests. Multivariable logistic regression was used to identify factors independently associated with germline testing completion. Covariates included age, sex, race, diagnosis year range (2020-2022 or 2023-2025), family history of colorectal cancer, and stage at diagnosis. Diagnosis during 2020-2022 was the reference category for diagnosis year. Adjusted odds ratios (aORs) with 95% confidence intervals (CIs) were reported.

Results

Among 403 patients with EOCRC, 164 (40.7%) completed germline testing, and 239 (59.3%) did not. Only 3 patients (1.8% of tested patients) underwent testing before diagnosis. Of 164 patients who completed testing, 113 (68.9%) had an accessible germline result report; 51 lacked an available report. Missing reports reflected incomplete documentation, testing performed outside the health system without an uploaded report, or documentation of testing completion without the final result. Documented referral for genetic evaluation

was identified in 79 patients (19.6%), and genetic counseling completion was documented in 89 patients (22.1%). Among 113 patients with results, a negative test result was the most common outcome (60 [53.1%]). Testing completion differed significantly across race categories ($P=.003$), with completion highest among Asian patients (14 [82.4%] of 17), followed by White patients (71 [44.9%] of 158), Black patients (59 [38.8%] of 152), and patients classified in the other race category (16 [28.6%] of 56). The other category represented a heterogeneous residual race classification in the electronic medical record. In multivariable analysis, male sex was associated with lower odds of testing completion (aOR, 0.59; 95% CI, 0.36-0.96; $P=.04$) as was classification in the other race category (aOR, 0.37; 95% CI, 0.17-0.77; $P=.008$). Diagnosis during 2023-2025 was associated with higher odds of testing completion compared with 2020-2022 (aOR, 4.24; 95% CI, 1.11-16.24; $P=.04$).

Conclusions

Germline testing remained underused in EOCRC, with rare prediagnosis testing and gaps across referral, counseling, and testing milestones. Testing completion improved in more recent years, but racial differences persisted. Standardizing the timing of germline testing relative to diagnosis, along with consistent tracking and documentation of results, may improve timely hereditary evaluation in patients with EOCRC.

Methods

- General: Methods do not state which tests were used. Recommend adding a brief statement about what statistical methods were used (eg, chi-square, multivariable logistic regression).
- General: Were cancer stage, insurance status, or site of care included as covariates for this health equity analysis?
- Line 14-15: Where is this multihospital health system generally located?
- Line 22-23: “Among tested patients, 113 had recorded germline results.” The abstract states, “164 (40.7%) completed germline testing” but “113 had recorded germline results” is presented ambiguously. Reviewers suggest modifying the Methods to state: “Of 164 tested patients, results were recorded for 113,” making the denominator explicit.
- Line 22-23: Reviewers also ask the author to consider adding one sentence explaining why 51 tested patients (31%) lack recorded results (164->113).

- Line 26-28: The multivariable finding states, “male sex and other race were independently associated with lower odds of testing completion” but does not include numbers. The word limit for abstracts submitted to *GMR* is 500 words, so reviewers encourage the author to add adjusted odds ratios with 95% CIs (and ideally *P* values) for male sex and other race.

Results

- Line 26: Reviewers request clarification of the term “other race.” Is this a defined census-style category or a residual/heterogeneous group in the study? An independent association with a potentially poorly defined “other” category is difficult to interpret.

Conclusion

- Line 30-31: Recommend modifying “Standardized hereditary evaluation workflows may improve testing completion and equity” to “Standardized timing of testing relative to diagnosis” for clarity.

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