

Disparities in hypermethylation testing for MLH1-deficient endometrial cancer



Introduction

- MLH1-deficient endometrial cancer may result from sporadic mutations or heritable Lynch syndrome.
- Hypermethylation testing can help distinguish between these causes.
- Inconsistent reporting of hypermethylation results can affect treatment decisions and delay Lynch syndrome identification.
- Lynch syndrome increases the risk of multiple malignancies, and because it is heritable, early detection is critical for both patients and families.

Methods

- We conducted a retrospective cohort study on patients diagnosed with endometrial cancer.
- Patient demographics, cancer information, and outcomes were collected from a gynecologic cancer database.
- Hypermethylation testing reported within 40 days of the surgery/biopsy for patients with an MLH1 deficiency was considered adequate.
- Race, age and cancer stage were compared between patients to determine if there were any risk factors for delays in receiving test results.
- The groups were analyzed using Chi-square tests.

Results

Turnaround Time For Test Results

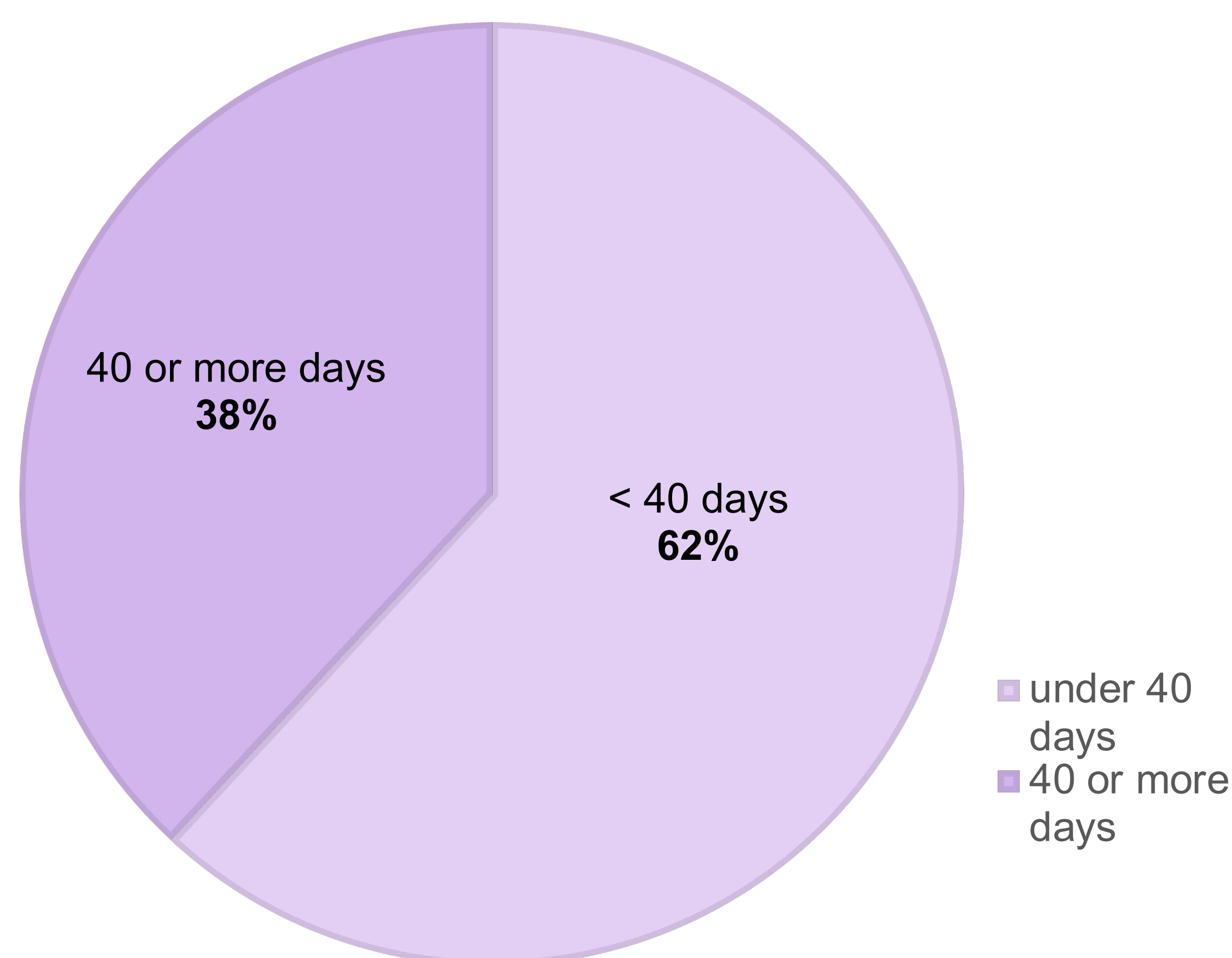


Figure 1

Delay in Hypermethylation Testing for White and Non-White Women

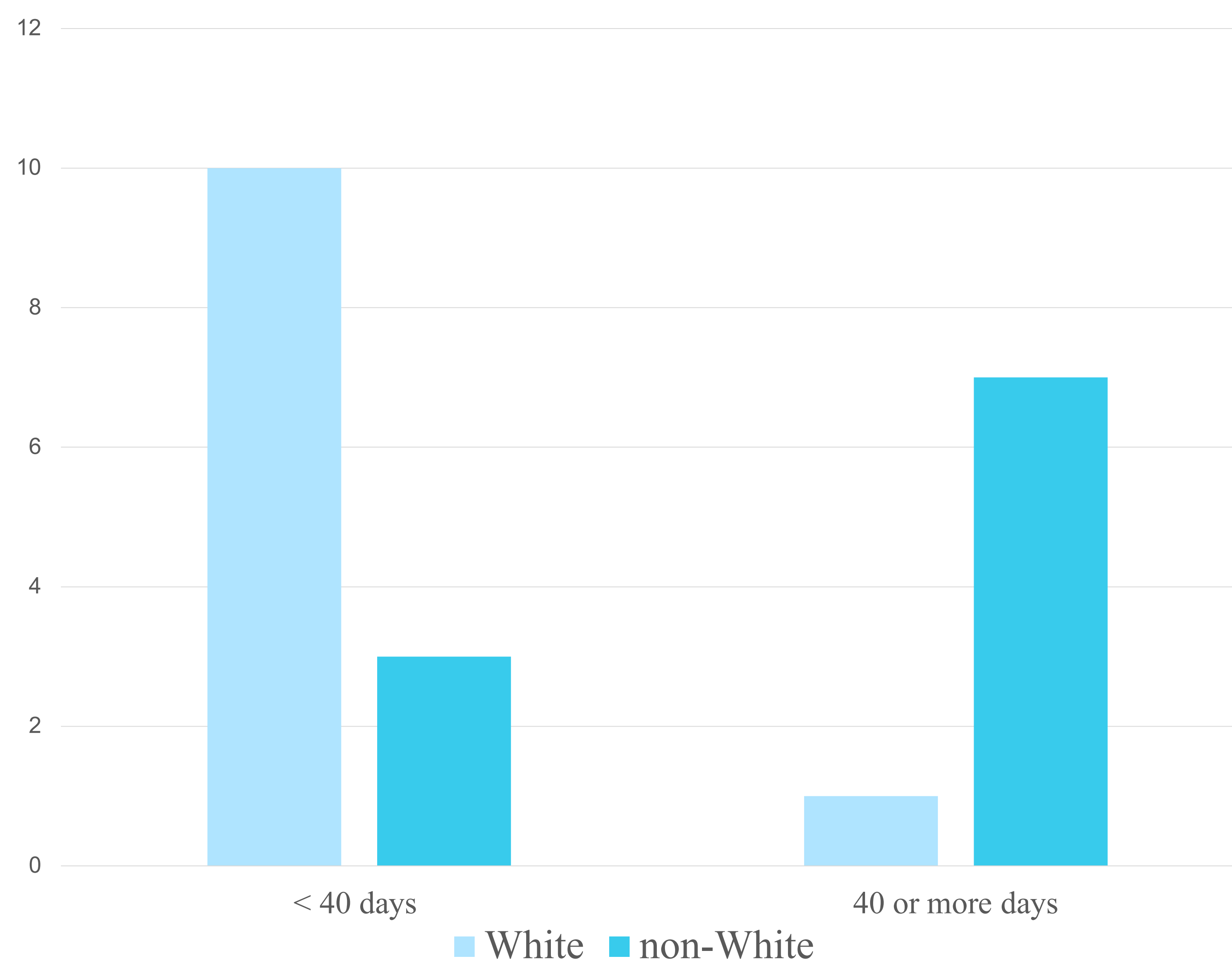


Figure 2

Figure 1. Adequate (n=13) and delayed (n=8) test result distribution for women diagnosed with MLH1 deficient endometrial cancer (n=21).

Figure 2. Comparison of timely vs. delayed testing for White and non-White women. A significant difference was found between the groups (p = 0.02).

Results

- Overall, 25 endometrial cancer patients were included in the study with 21 of the patients having completed hypermethylation testing.
- The four other patients received genetic testing and were reported to be negative for Lynch syndrome.
- 62% of the 21 patients received their tests results back within 40 days while 38% received the results after 40 days.
- White patients were more likely to receive the report back in an adequate amount of time then non-White patients (p = 0.02).
- A comparison of patients younger than 65 to those old than 65 yielded no significant result.
- Comparison between early (I, II) and late (III, IV) stage cancer did not reveal any significant findings.

Conclusion

- The purpose of the study was to identify disparities in timely hypermethylation testing among subgroups.
- A disparity was identified in adequate testing for non-White women diagnosed with MLH1-deficient endometrial cancer.
- Faster turnaround for test results could improve the plan of care and affect the physician's decisions.
- The study was limited in its sample size; however, these findings highlight the need for efficient and equitable hypermethylation testing.