Late-stage breast cancer in Young Appalachian women: missed hereditary risk and persistent disparities in a community health setting.

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Introduction

- The incidence of breast cancer among young women is on the rise.
- In rural regions, a combination of socioeconomic disadvantages, poor health literacy, and limited access to specialized care exacerbates diagnostic delays, even more so in patients with hereditary risk.
- This study investigates the stage of presentation, family history, genetic testing uptake, and social determinants of health among young women diagnosed with breast cancer in a community health setting in Appalachia.

Methods

- This retrospective cohort study included 1,359 patients diagnosed with breast cancer at Tennova North Knoxville Medical Center from 2020 to 2024.
- Patients were stratified by age, i.e. young patients (<45 years) versus (vs) average-onset patients (age ≥45), stage at diagnosis, insurance status, urban vs rural residence, educational attainment, reported family history of cancer, and whether they underwent genetic testing and/or counseling (if positive family history of any cancer).
- Descriptive statistics were calculated, and bivariate comparisons were performed using Chi-square & t-tests. Logistic regression models were constructed to identify predictors of advanced-stage disease (stages III and IV) at diagnosis and the likelihood of undergoing genetic testing.

Results

- Of the 1,359 patients included, 16.3% (n=222) patients were < 45 years old at diagnosis. Among these young patients, 80.2% (n=178) presented with stage III or IV disease, while 6.3% (n=14) and 13.5% (n=30) were diagnosed at stages I and II, respectively.
- 81.1% (n=180) reported a first-degree family history of cancer, yet only 16.2% (n=36) received genetic testing and/or counseling.
- In bivariate analysis, patients with Medicaid or no insurance were significantly more likely to present with advanced stage breast cancer compared to those with private insurance (OR 4.5; 95% CI: 2.6-7.8, $p < 0.001$).
- Rural residence was also significantly associated with late-stage diagnosis (OR 3.7; 95% CI: 2.0-6.9, $p = 0.002$).
- Lower educational attainment (less than high school diploma) was associated with decreased likelihood of receiving genetic testing (OR 0.34; 95% CI: 0.15-0.78, $p = 0.01$).
- In multivariate logistic regression, both uninsured status ($p < 0.001$) and absence of genetic counseling among patients with positive family history of cancer ($p = 0.002$) independently predicted advanced-stage presentation at diagnosis.

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Conclusion

- This study highlights a troubling pattern of late-stage breast cancer diagnosis in young Appalachian women, despite the presence of family history of cancer in > 80% of cases.
- Profound disparities in genetic testing utilization, driven by socioeconomic status, rurality, and health literacy, likely contribute to missed opportunities for early detection.
- These findings underscore the urgent need for community-based hereditary risk screening programs, access to genetic counseling, and culturally competent education strategies within rural healthcare systems.

