Germline and somatic genetic testing trends among women with epithelial ovarian cancer

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Topic: Financial Toxicity and Disparities

Objectives
Identifying homologous recombination deficiency (HRD) mutations has significant treatment implications for women with epithelial ovarian cancer (EOC). While germline testing strategies have been well catalogued, best-practice strategies for obtaining somatic, germline, or combination testing remain unclear. We aimed to trend rates and temporal relationships of germline and somatic genetic testing at an academic institution to identify gaps in equitable access to precision testing.

Methods
All patients with EOC were identified from the electronic medical record (EMR) using diagnosis codes at a single urban academic gynecologic oncology group from 1/2015 to 12/2020. IRB approval was obtained. EMR review was performed to analyze trends in germline and somatic genetic testing. Descriptive statistics and chi-square tests were performed.

Results
431 women with EOC were identified. Patients were predominantly white (88%), older women (mean age 68), who had insurance (97%), advanced stage disease (80%), and high-grade serous histology (77%). 80% of patients underwent germline testing, and 15% had BRCA1/2 mutations identified. 50% underwent somatic testing with a detection rate of 29.5% for HRD mutations. Somatic and combination testing rates increased each contact year (Figure 1A/1B). Patients with negative germline testing were more likely to undergo somatic testing (60%) than those with germline BRCA1 (29%) or BRCA2 (37%) mutations (p = 0.0012). 12.5% of patients with negative germline testing had a somatic mutation in either BRCA1/2 or loss of heterozygosity. Of patients without germline genetic testing, only 27% had somatic testing performed (22/81). 30% had somatic testing performed after recurrence; 36% of patients with a recurrence never underwent somatic testing.

Conclusions
Somatic testing resulted in actionable HRD mutations in 1 in 3 patients in this cohort, with 1 in 8 patients having somatic HRD mutations despite negative germline testing. This illustrates the therapeutic potential of comprehensive genetic profiling and represents how variations in practice may result in missed therapeutic interventions. As somatic testing rates continue to increase, efforts to define barriers and create streamlined, guideline-based strategies for universal testing are warranted to better align with current guidelines.

Abstract Table or Graph
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