Poster 14: Influence of genetic risk awareness on high grade serous fallopian tube carcinoma diagnoses

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Topic: Genetics

Objectives

High-grade serous carcinoma originates in the fallopian tubes of patients with hereditary gynecologic cancer risk. We sought to evaluate genetic testing patterns and the contribution of germline pathogenic variants (PV) in patients diagnosed with high-grade serous fallopian tube cancer (HGSFTCA).

Methods

This IRB-approved retrospective cohort study included patients with HGSFTCA diagnosed between March 2008 and March 2023 in a large health care system within a single state. Demographic, clinical, pathologic, and germline genetic testing data were abstracted from the electronic medical record.

Results

Among 279 HGSFTCA cases, 7.5% (n=21) were stage I, 9.7% (n=27) were stage II, 55.6% (n=155) were stage III, and 27.2% (n=76) were stage IV. Germline genetic testing was completed in 86.0% (n=240). Genetic testing was performed prior to diagnosis in 7.2% (n=20), after diagnosis in 69.9% (n=195), timing was unknown in 9.0% (n=25), testing was recommended but not completed in 7.5% (n=21), and no testing was discussed or completed in 6.5% (n=18). Germline PVs were present in 21.9% (n=61) of the overall cohort, including in BRCA1 in 10.4% (n=27 BRCA1 alone, n=2 BRCA1 with a second PV), BRCA2 in 6.5% (n=18), BRIP1 in 0.4% (n=1), RAD51C in 0.7% (n=2), and other cancer susceptibility genes in 3.9% (n=11). PVs were found at higher rates in patients with early stage compared to advanced stage disease (38.1% in stage I; 37.0% in stage II; 18.1% in stage III; 19.7% in stage IV). Among patients with a germline PV, the timing of genetic testing prior to HGSFTCA diagnosis occurred in 100% with stage I, 30% with stage II, 7.1% with stage III, and 0% with stage IV.

Conclusions

In this cohort of HGSFTCA with high rates of genetic testing, germline PVs are more frequently found in early-stage compared to advanced-stage disease. In early-stage patients, finding a PV prior to a HGSFTCA diagnosis may have contributed to earlier detection of these cancers. This underscores the importance of genetic awareness in providing patients with the opportunity to undergo proactive cancer risk reducing strategies.