Poster 13: Genetic testing patterns and pathogenic variant rates in patients with epithelial ovarian cancer

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Topic
Genetics

Objectives
While it is recommended that all patients with epithelial ovarian cancer (EOC) undergo genetic testing, the national testing rate is reported to be less than 40%. We aim to describe germline pathogenic variant (PV) prevalence in a cohort of EOC patients with an above average testing rate, focusing on demographic and clinical characteristics, including ethnicity, disease stage, and histology.

Methods
Genetic testing data from EOC patients diagnosed at a network of hospitals in a single state between 2021-2024 were analyzed. Statistical analyses were conducted using GraphPad Prism, with chi-square tests for categorical variables and t-tests for continuous variables.

Results
Among 1,308 patients with EOC, 1,007 (77.0%) underwent genetic testing and 236 (18.0%) had a PV. PV carriers were younger (median age 62.0 vs. 68.0 years, p< 0.0001). Groups with higher PV rates included those identifying as Hispanic (30.0%), Ashkenazi Jewish (25.7%), and Jewish not otherwise specified (NOS) (35.71%). PV were more common in Stage III (19.5%) and IV (20.2%) disease and in high-grade serous (HGS) (21.8%) and adenocarcinoma NOS (28.6%) histologies. The 236 PV were found most frequently in BRCA1 (n=110, 46.6%%), BRCA2 (n= 62, 26.3%), BRIP1 (n=8,3.4%), RAD51C (n=5,2.1%), MUTYH (n=9,3.8%), ATM (n=8,3.4%), and CHEK2 (n=8,3.4%). The BRCA1 185delAG Ashkenazi Jewish founder PV was found in 6 patients that identified as Hispanic. There were no differentiating characteristics among the 110 (8.4%) that were offered but declined testing or the 191 (14.6%) for whom testing was never discussed.

Conclusions
In this dataset of EOC patients with high rate of genetic testing, we found higher rates of PV prevalence in Hispanic and Jewish patients. Mutations were also associated with younger age, advanced disease stage, and HGS histology. This highlights the importance of equitable access to genetic testing, particularly for diverse populations disproportionately affected by hereditary EOC.

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