Poster 15: A Comparative Study of Ovarian Cancer Family History by Germline Genetic Testing Results

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Topic: Genetics

Objectives

Our team set out to study differences in the frequency of ovarian cancer in the family history of unaffected women who underwent germline genetic testing, with the goal of exploring the relationship of family history to results of genetic testing.

Methods

We performed a retrospective cohort study looking at all unaffected women in our hospital system who underwent germline genetic testing for cancer risk in 2022 and 2023. Reviewing results for genes that impact ovarian cancer risk, namely BRCA1, BRCA2, BRIP1, MSH6, PMS2, ATM, and CHEK2, we divided the cohort into those with a pathogenic variant, those with a variant of uncertain significance, and those who tested negative. Analysis of variance was used to see if there were any significant differences in their family histories across testing results.

Results

We found that patients with pathogenic variants reported ovarian cancer in their family history more frequently than patients with variants of unknown significance and patients with negative testing ($F=4.11$, $p< 0.05$), and no difference in family history between patients with variants of unknown significance and patients with negative testing. This effect was especially strong when we limit the family history to first degree relatives. 14% of pathogenic patients had at least one first degree family member with ovarian cancer, compared to just 6.7% for patients with variants of unknown significance and 6.3% for patients with negative testing ($F=9.79$, $p< 0.0001$).

Conclusions

Analysis of unaffected women presenting for cancer genetic testing in our patient population over the last two years indicate that those with a family history of ovarian cancer, and especially those with first-degree relatives with ovarian cancer, are over twice as likely to have a pathogenic variant in a gene associated with ovarian cancer. There was no difference in family history between patients with variants of unknown significance and those who tested negative. This supports existing evidence for the need to regularly assess family history, and proactively screen and test patients for cancer genetics.

Abstract Table or Graph

	Results of Genetic Testing			
	Pathogenic Variant	Variant of Unknown Significance	Negative	Total
Patients Tested	231	333	4138	4702
Patients Tested with Any Family History of Ovarian Cancer	55	57	590	702
Patients Tested with First-Degree Relative(s) with Ovarian Cancer	32	22	226	280
Patients Tested with Second-Degree Relative(s) with Ovarian Cancer	26	33	361	420