Research uncovers more inherited genetic mutations linked to ovarian cancer

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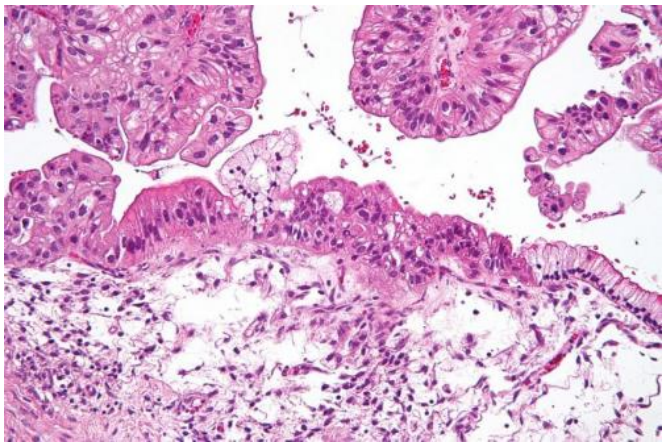

Intermediate magnification micrograph of a low malignant potential (LMP) mucinous ovarian tumour. H&E stain. The micrograph shows: Simple mucinous epithelium (right) and mucinous epithelium that pseudo-stratifies (left - diagnostic of a LMP tumour). Epithelium in a frond-like architecture is seen at the top of image. Credit: Nephron /Wikipedia. CC BY-SA 3.0

Previous research has established a link between genetic mutations in the BRCA1 and BRCA2 genes to an increased risk of developing ovarian, fallopian tube or peritoneal cancer in women. A recent publication documents the efforts of a team of researchers affiliated with the Gynecologic Oncology Group (GOG) to determine if inherited genetic mutations other than BRCA1 and BRCA2 can also put a woman at risk of developing these diseases.

The team - which includes Paul DiSilvestro, MD, head of research with the Program in Women's Oncology at Women & Infants Hospital and professor of obstetrics and gynecology at The Warren Alpert Medical School of Brown University - published their findings in the article "Inherited Mutations in Women with Ovarian Carcinoma" in the recent issue of the *Journal of the American Medical Society*.

"Descriptions of the identity of these genes and their frequency was lacking in the medical literature," Dr. DiSilvestro explains. "The goal of this research was to better define these issues."

More than 1,900 women with ovarian cancer who were identified through the University of Washington gynecologic tissue bank and from various GOG clinical trials made up the study population. Information about mutation frequencies were compared with the National Heart, Lung and Blood Institute GO Exome Sequencing Project and the Exome Aggregation Consortium. Clinical characteristics and survival rates were assessed by mutation status.

What the evaluations revealed was that 18 percent of the women with ovarian cancer carried mutations in genes associated with ovarian cancer risk beyond the BRCA1 and BRCA2 genes.

"The results of this trial expanded our knowledge of the genes that we suspect cause hereditary ovarian cancer, bringing the total to 11," Dr. DiSilvestro says, adding that, "Genetic testing should now begin screening for these nine additional genetic mutations so women carrying the genes can make educated decisions about their health care future."

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