

Early diagnosis of high-grade serous ovarian cancer feasible

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Early diagnosis of high-grade serous ovarian cancer (HGSOC) seems feasible through analysis of genomic alterations in DNA from Papanicolaou (Pap) test smears, according to a study published online Dec. 6 in *Science Translational Medicine*.

Lara Paracchini, Ph.D., from Humanitas University in Milan, and colleagues examined a retrospective and multicentric cohort of 250 archival Pap [test](#) smears collected during routine gynecological screening. Samples were taken from 113 presymptomatic women who were subsequently diagnosed with HGSOC (pre-HGSOC) and from 77

healthy women at different time points (from one month to 13.5 years before diagnosis). Low-pass whole-genome sequencing of DNA derived from Pap test samples in terms of copy number profile abnormality (CPA) was used to detect genome instability.

The researchers found that the CPA values of DNA extracted from Pap samples were significantly higher from pre-HGSOC women versus healthy women. This assay could detect HGSOC presence up to nine years before diagnosis, consistent with the longitudinal analysis of clonal pathogenic *TP53* mutations. Integration of the CPA score into the EVA (early [ovarian cancer](#)) test resulted in sensitivity, specificity, and accuracy of 75, 96, and 81 percent, respectively.

"We consider the [data](#) sufficiently convincing to warrant prospective clinical investigations aimed at verifying whether the longitudinal analysis of CPA in Pap test smears renders the prediction of HGSOC possible," the authors write.

More information: Lara Paracchini et al, Genomic instability analysis in DNA from Papanicolaou test provides proof-of-principle early diagnosis of high-grade serous ovarian cancer, *Science Translational Medicine* (2023). [DOI: 10.1126/scitranslmed.adi2556](https://doi.org/10.1126/scitranslmed.adi2556)

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