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Title: Multigene panel testing in gynaecological malignancies: A study in a cohort of over 1000 patients
Topic : 02. Diagnostics

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Introduction/Background
Gynaecological malignancies, specifically ovarian and endometrial cancer, are

often attributed to an inherited predisposition. A variety of genes has been associated with oncogenesis; thus, multigene panel testing is recommended to patients. Detecting pathogenic alterations can lead to proactive measures such as improved screening and preventive actions, or redefinition of their clinical management through personalized health care.

Methodology

Between 2020 and 2025, over 1000 women were referred for genetic testing at Genekor Medical S.A., with a diagnosis of ovarian and/or endometrial cancer. Next Generation Sequencing was performed in DNA extracted from all patients' peripheral blood, targeting 52 genes. Analysis was performed for both single nucleotide variations (SNVs) and copy number variations (CNVs), to detect pathogenic/likely pathogenic variants in cancer susceptibility genes.

Results

Among patients tested, 166 were diagnosed with endometrial cancer, while 958 had malignancy of the ovaries. In 27% and 28% of cases, respectively, a pathogenic/likely pathogenic variant was identified. In endometrial cancer cases, SNVs were identified in clinically important genes like MSH6 (16.2%), MLH1 (10.8%), MSH2 (10.8%), BRCA2 (5.4%), PMS2 (5.4%), EPCAM (2.7%). CNVs were mainly found in MSH2, followed by MLH1 and PMS2. In ovarian malignancies, the most common altered genes were BRCA1 (39.9%), BRCA2 (14.0%), RAD51C (5.6%), PALB2 (2.4%), MLH1 (2.4%), RAD51D (1.7%), BRIP1 (1.4%) for SNVs and BRCA1 for CNVs. Alterations were also identified in ATM, CHEK2, MUTYH, FANCA, FANCM and in some clinically important genes that have not yet been associated with the development of gynaecological tumors (e.g. TP53, CDH1, CDKN2A). Variants of Uncertain Significance were identified in 37% and 36% of ovarian and endometrial cancer cases, respectivelly.

Conclusion

Comprehensive multigene genetic testing is necessary for appropriate clinical management of pathogenic variants' carriers. Alterations detected in gynaecological tumors may be associated with other malignancies as well, thus giving the opportunity to physicians to manage patients holistically.