#383 BIOENDOCAR: IDENTIFYING CANDIDATE BIOMARKERS FOR DIAGNOSIS AND PROGNOSIS OF ENDOMETRIAL CARCINOMA USING MACHINE LEARNING AND ARTIFICIAL INTELLIGENCE

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Introduction/Background Endometrial carcinoma (EC) is the most common gynaecological malignancy in the developed world. Currently, no valid non-invasive diagnostic or prognostic methods exist, making diagnosis and treatment rely on histopathological and surgical findings. The clinical study 'Biomarkers for Diagnosis and Prognosis of Endometrial Carcinoma' (BioEndoCar; NCT03553589) addresses this issue.

Methodology A prospective observational case-control study was conducted at six medical centres across Europe. Plasma samples from women with diagnosed EC and controls were examined using non-targeted/targeted metabolicomic and semi-quantitative immune-based proteomic approaches. The blood metabolomics (>850 metabolites) and proteomics (>900 proteins) data together with clinical and epidemiological data, were analysed using advanced artificial intelligence (AI) and machine learning (ML) methods to develop new diagnostic/prognostic models for early EC diagnosis and identifying patients with low/high risk for cancer progression and recurrence.

Results BioEndoCar has recruited over 440 patients, with strict standard operating procedures for sample collection, processing, and storage. The diagnostic/prognostic models based on all data developed using AI/ML methods showed promising characteristics with a repeated k-fold cross-validation AUC > 0.8. The developed models will undergo further validation using both statistical (AI/ML) approaches to confirm which subset of proteomic and metabolomic data could serve as diagnostic and prognostic biomarkers in endometrial cancer.

Conclusion The BioEndoCar study has completed the initial phase of identifying and validating diagnostic/prognostic models for early EC diagnosis and identifying patients with low/high risk for cancer progression and recurrence using artificial intelligence and machine learning methods. If validated, the models including a subset of proteomic and metabolomic data could serve as a foundation for developing valuable non-invasive tools for the diagnosis and prognosis of EC.

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#432 SPECTRUM OF BRCA1, BRCA2, PALB2, ATM AND TP53 PATHOGENIC VARIANTS IN BREAST AND OVARIAN CANCER PATIENTS OF TATAR AND BASHKIR ETHNIC ORIGIN

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Introduction/Background Tatarstan and Bashkortostan are large republics located nearby Volga river. Although they joined Russia about five hundred years ago, they managed to preserve national authenticity over centuries.

Methodology The study included 132 ovarian cancer (OC) patients and 277 women with breast cancer (BC), who reside in Tatarstan or Bashkortostan and identify themselves as ethnic Tatars or Bashkirs. In order to enrich the analyzed groups by carriers of cancer-predisposing pathogenic variants (PVs), OC were selected on the basis of high-grade serous histology, and BC were represented mainly by early-onset and/or family-history positive and/or bilateral and/or receptor triple-negative cases. Coding sequences and 5’- and 3’-UTRs of BRCA1, BRCA2, PALB2, ATM and TP53 genes were analyzed by next generation sequencing.

Results The frequency of BRCA1/2 mutations was 25/132 (19%) in OC and 43/277 (16%) in BC. BRCA1 PVs accounted for 47/68 (69%) cases with mutation. A significant share of BRCA1 PVs detected in ethnic Tatars and Bashkirs was represented by Slavic founder alleles (c.5266dupC (5382insC): n = 14; C61G: n = 4; c.3700_3704delGTGAAA: n = 3; c.4034delA: n = 2; c.3756_3759del: n = 1). Seven patients carried BRCA1 c.5161C>T [Q1721X] allele, which is an ethnicity-specific mutation characteristic for this region. There were two recurrent BRCA2 PVs, c.39–1_-39delGA (n = 6) and c.468dupT (n = 4). One patient carried PV in PALB2 (c.221delA). No instances of ATM or TP53 heterozygosity was observed.

Conclusion Despite well-preserved national identity of Tatars and Bashkirs, Slavic BRCA1 founder PVs are common among patients from these ethnic groups. In addition, one BRCA1 and two BRCA2 ethnicity-specific PVs were identified in this study. Recurrent BRCA2 c.39–1_-39delGA allele deserves particular attention, because it is located not in coding but in regulatory region of the gene.

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#502 ADDITIONAL INHIBITION OF PHOSPHOR-S6 KINASE IMPROVES THE THERAPEUTIC EFFECT OF CARBOPLATIN IN OVARIAN CANCER

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Introduction/Background S6 kinase is a protein kinase that is involved in signal transduction. S6 kinase is thought to play an important role in the compensatory adaptive response of various cancers against anti-cancer drug. We examined whether varied patients with low/high risk for cancer progression and recurrence using artificial intelligence and machine learning methods.