

Results Among 150 HGSC samples, we identified 44 samples (29.3%) with reportable variants with variant allele frequencies from 10.3% - 99.4%. These included 35 point mutations/insertions/deletions, 7 exon/whole gene deletions, and 2 BRCA1 exon 13 duplications. A subset (26) of these variants were then confirmed by targeted assays using Sanger and MLPA.

Conclusions Utilizing NGS technology, we reliably identified BRCA mutations in FFPE tumor samples. A validated NGS pipeline provides a valuable clinical tool to conduct Traceback initiatives to the families of deceased ovarian cancer patients never tested for germline mutations.

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234 CLONALITY ANALYSIS OF SYNCHRONOUS ENDOMETRIAL AND OVARIAN CARCINOMAS IN PATIENTS WITH LYNCH SYNDROME

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Objectives Sporadic synchronous endometrial (ECs) and ovarian cancers (OCs) have been shown to be clonally-related and to likely constitute metastases from each other. We sought to define whether synchronous ECs/OCs in patients with Lynch syndrome would be clonally-related or independent tumors.

Methods We subjected synchronous ECs/OCs from four patients with clinically confirmed Lynch syndrome to massively parallel sequencing targeting 468 cancer-related genes. Somatic mutations, copy number alterations, clonal relatedness and clonal decomposition were performed using state-of-the-art bioinformatics methods.

Results All synchronous ECs/OCs were considered independent primaries based on clinico-pathologic criteria. Sequencing analysis revealed that the ECs and OCs of three cases harbored strikingly similar repertoires of somatic mutations and gene copy number alterations and were clonally related. Specifically, in one case (LS2), a subset of subclonal mutations in the EC became clonal in the OC, suggesting that the ovarian tumor originated from the endometrial tumor. In contrast, in another case (LS5), the EC and OC harbored distinct somatic mutation profiles with no shared mutations; consistent with them constituting two independent primary tumors. In this case (LS5), a *PTEN* mutation and loss of protein expression were found to be restricted to the EC.

Conclusions Akin to sporadic synchronous ECs/OCs, the majority of Lynch syndrome-related synchronous ECs/OCs originate from a single primary tumor at variance with their clinical-pathologic diagnosis. Given that in the context of Lynch syndrome, synchronous ECs/OCs may be independent primary tumors, Lynch syndrome testing should be considered when synchronous ECs/OCs present with distinct genetic or immunohistochemical profiles.

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235 ABO, RH, KELL AND MN SYSTEMS WITHIN UTERINE CANCER

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Objectives The blood group antigens are found in several human cell types. There is a variable expression of histo-blood groups antigens within normal endometrium, which are also involved in hormonal regulation of glycosyltransferase activity. We aimed to investigate ABO, RH-hR, Kel and MN systems in women with Uterine cancer (UCa) in Adjara (Georgia) population.

Methods Internationally recognized immunoserology methods were used to reveal the erythrocyte group antigens. The obtained results were statistically processed by using appropriate formulas.

Results From ABO system alleles, r and q alleles frequencies were higher within the patients with UCa compared with control group of patients. From Rh-Hr system, the distribution frequencies of D, c and e alleles, Cc, Ee and EE genotypes, cDe, cDE haplotypes are increased in UCa. From Kell q(k) and p (K) alleles, p(k) allele tends towards to UCa in the tumor. Notably, MN system p (M) and q(N) alleles, revealed q(N) allele increased frequency in UCa compared to control group.

Conclusions Based on our study, ABO, RH-hR, Kell and MN systems antigens may be useful for UCa predisposition and development.

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236 BEHAVIORAL AND PSYCHOLOGICAL IMPACT OF ONCOGENETIC COUNSELING FOR HEREDITARY BREAST CANCER

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Objectives Oncogenetic counseling for hereditary breast cancer is an important tool for populational cancer risk screening. Individuals submitted to oncogenetic counseling must be aware about the social and emotional impact of genetic test result in their lives.

Methods Ten years follow-up of a cohort submitted to oncogenetic counseling in Brazil was evaluated for the health perception, depressive or anxious symptoms and life habits/behaviour before and after the oncogenetic counseling. Survival analyzes were performed using the Kaplan-Meier method and the Log-Rank test was used to verify the existence of significant differences.

Results 139 patients have been submitted to oncogenetic counselling, 135 women and 4 men; 113 patients had early onset