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Anais: 3° Simpósio de Especialidades Oncológicas e Seminário de Iniciação Científica do Instituto Mário Penna

IDENTIFICATION OF GENETIC VARIANTS ASSOCIATED WITH TRIPLE-NEGATIVE BREAST CANCER AND HIGH-GRADE SEROUS OVARIAN CANCER

IDENTIFICAÇÃO DE VARIANTES GENÉTICAS ASSOCIADAS AO CÂNCER DE MAMA TRIPLO-NEGATIVO E CÂNCER DE OVÁRIO SEROSO DE ALTO GRAU

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ABSTRACT

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Breast and Ovarian Cancer represents challenging diseases for clinical practice nowadays. While the first one is the most common neoplasm affecting the female population, the second, even with a lower incidence, presents elevated mortality[2,3]. Both diseases have different subtypes. Triple Negative Breast Cancer (TNBC) is the most complex since it does not have any specific target for treatment[4]. In the same manner, High-Grade Serous Ovarian Cancer (HGSOC) is the more frequent and complex subtype due to its lack of adequate diagnosis[3]. Different subtypes also require different treatments. Thus, it becomes clear the need for more effective diagnostic methods and the search for new personalized treatments. Therefore,

the importance of searching for possible biomarkers for those diseases stands out[5]. The present study aims to identify potential predictive or prognostic tumor biomarkers actionable to clinical practice in patients with TNBC and HGSOC. For that, blood samples were collected from 10 patients from the Tumor Biobank Mário Penna Institute (CAAE - 82703418.8.0000.5121). Out of these, six were related to TNBC and four to HGSOC. The samples were processed, and the plasma was separated through the standard laboratory procedures. cell-free DNA (cfDNA) was extracted utilizing Quick-cfDNA Serum & Plasma (Zymo). cfDNA library was prepared utilizing xGen DNA Library Prep MC Kit (IDT) and the hybridization with xGen Hybridization capture of DNA Libraries Kit (IDT), conform manufacture instructions. The quantification and quality of the library were assessed utilizing a Qubit fluorometer and TapeStation System, respectively. The final library displayed the expected cfDNA profile. Libraries were sequenced using the 300-cycle high-output kit on the NextSeq 550 (Illumina). Generated more than 30 million reads for each sample. Mapping analyses to the human reference genome (GRCh38), variant calling, and correlation of findings with patient's clinical data are ongoing and will provide the genetic profile of our studied cohort. This study is in initial phase. However, it has a great potential to identify new prognostic-biomarkers candidates in TNBC and HGSOC[1]. Furthermore, this study may contribute to expanding knowledge about the factors that influence prognostic outcomes in these diseases, which may favor the development of more personalized therapies and diagnostics based on liquid biopsy[1], allowing treating these conditions more quickly and efficiently.

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NOTAS

CONFLITOS DE INTERESSE

Não há conflitos de interesse financeiros ou de outra natureza por parte dos autores.

CONTRIBUIÇÃO

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