genetic screening to search for pathogenic variants in these genes in search of strategies, mainly for risk management and individualized treatment. However, in Brazil, access to genetic tests is not accessible to everyone and many regions of the country still need to be explored. Therefore, the aim of the study was to investigate germline pathogenic variants in BRCA1 and BRCA2 in women with ovarian cancer in Brazilian Northeastern.

Methodology Molecular evaluation to search for germline pathogenic variants in the BRCA1 and BRCA2 genes through Next Generation Sequencing – NGS was performed in 40 women with high-grade serous epithelial ovarian cancer. All patients were registered in the Pernambuco Public Health System’s Hereditary Cancer Program

Results Thirteen germline pathogenic variants were identified, eleven in BRCA1 and two in BRCA2, and one variant of uncertain significance in BRCA2. The median age in this group was 48 years. The pathogenic variant in BRCA1 c.5266dupC, originally described as founder of Ashkenazi Jews, was identified in three patients and all were from the Northeast region of Brazil.

Conclusion The data are unprecedented for this region of Brazil in patients with ovarian cancer and show the great heterogeneity of ancestors in the formation of the Brazilian population. Germline pathogenic variants in BRCA1 and BRCA2 in women with ovarian cancer in Brazilian Northeastern is common and should be offer for every case. They also corroborate previous data on the founder effect of the variant described in the country and show the need to assess the molecular profile of patients with hereditary cancer syndromes.