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ANALYSIS OF THE PREVALENCE OF THE C.156_157INSALU PATHOGENIC VARIANT IN BRCA2 GENE IN CENTRAL BRAZIL PATIENTS WITH SUSPECTED HEREDITARY BREAST AND OVARY CANCER SYNDROME (HBOC)

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The pathogenic variant c.156_157insAlu has a founding effect in Portugal, accounting for half of the cases of hereditary breast cancer in the country, and has been appearing frequently in studies conducted in Brazil. Considering the absence of published data on the frequency of the *BRCA2* variant for the Midwest region of the country and the importance of Portuguese colonization for the genetic composition of the Brazilian population, the present study aimed to identify the prevalence of variant c.156_157insAlu in the *BRCA2* gene in patients with breast cancer in a population from Central Brazil. We evaluated 100 patients with breast cancer who met NCCN criteria for HBOC syndrome and who were treated at the Advanced Center for Breast Diagnosis at Clinical Hospital, UFG. 4 mL of blood was collected for DNA extraction, which was then submitted to the PCR technique for exon 3 of the *BRCA2* gene and then performed on agarose gel electrophoresis (2%). Then, the samples that showed amplification for the variant studied were sent for Sanger sequencing in ABI3730 Genetic Analyzer apparatus for confirmation of the presence of the Alu insert and the MLPA test using the SALSA MLPA P045 *BRCA2/CHEK2* kit from lot C1- 0416. Only one patient (1%) presented variant c.156_157insAlu in exon 3 of the *BRCA2* gene. The patient was female, had invasive ductal carcinoma of the luminal A subtype, diagnosed at age 41 with a family history of breast, lung, liver, throat, pancreas and intestine cancer, sarcoma, Hodgkin's lymphoma and melanoma. Our results suggest that, despite the low incidence observed, the c.156_157insAlu variant in exon 3 of the *BRCA2* gene should be screened for all patients in the Midwest region of Brazil with suspected HBOC syndrome, as well as the study of their relatives.