

Investigation of germline variants in patients with luminal A and triple-negative breast cancer



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Background: Pathogenic and likely pathogenic germline variants in genes associated with breast cancer (BC) play an important role in the development, progression, and response to therapy. Therefore, the identification of these variants can provide valuable insights into the mechanisms of BC pathogenesis and improve early disease detection, treatment strategies, and patient outcomes. **Material and Methods:** Whole-exome sequencing (WES) was performed on 52 women diagnosed with luminal A or triple-negative BC at the Hospital of Lithuanian University of Health Sciences Kaunas Clinics. The study used genomic DNA extracted from peripheral blood samples. WES data were analyzed using the Franklin platform by Genoox. Pathogenic and likely pathogenic variants were assessed in 81 genes, as defined by the ACMG Secondary Findings V3.2 (2023), and in 18 genes associated with BC. **Results:** A total of ten pathogenic and four likely pathogenic germline variants were identified in the study group. Of these, five variants were detected in the *BRCA1* gene, three in the *BRCA2* and *CHECK2* genes, and one each in the *NBN*, *ATM*, and *MUTYH* genes. Overall, all variants were found in 19 patients, each of whom had only one clinically significant variant. Nine different variants were identified in patients with luminal A breast cancer, and four - in triple-negative breast cancer. Only one pathogenetic variant was identified in both groups. **Conclusion:** In conclusion, 14 clinically significant germline variants were identified in patients with luminal A or triple-negative breast cancer that may potentially affect tumor pathogenesis.